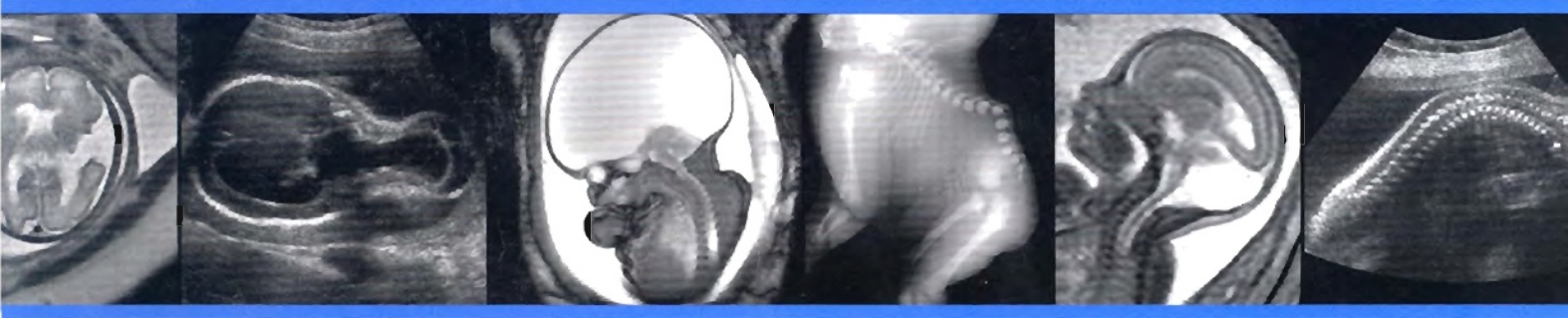
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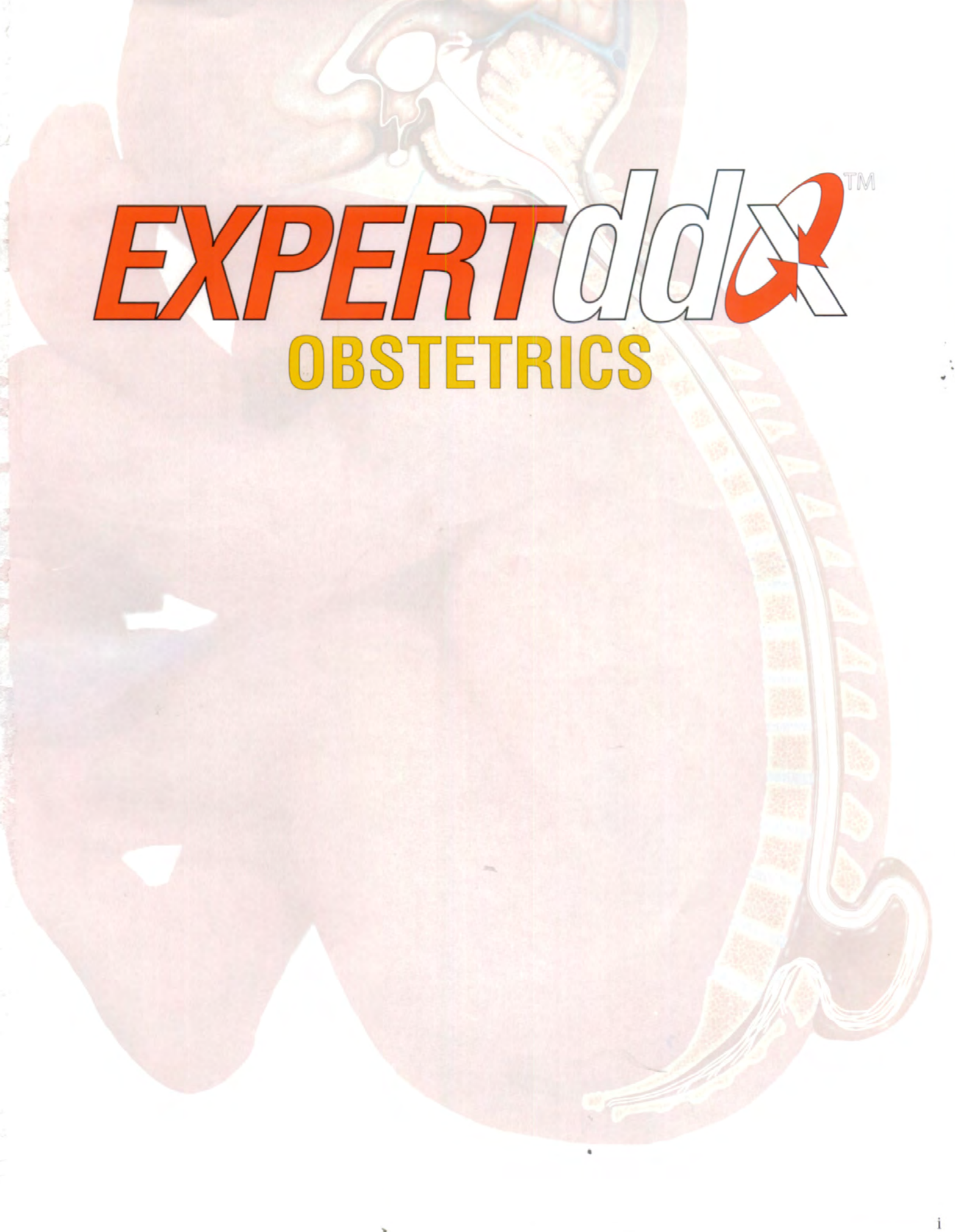




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OBSTETRICS



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OBSTETRICS

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We dedicate this book to our patients, the strong and brave pregnant women who trust us during difficult times. We owe them gratitude, compassion, and expertise.

And to Robert
And the world's loves lost and refound – it is better the second time around!

PJW



EXPERTddx™
OBSTETRICS

Once the appropriate technical protocols have been delineated, the best quality images obtained, and the cases queued up on PACS, the diagnostic responsibility reaches the radiology reading room. The radiologist must do more than simply "lay words on" but reach a real conclusion. If we cannot reach a definitive diagnosis, we must offer a reasonable differential diagnosis. A list that's too long is useless; a list that's too short may be misleading. To be useful, a differential must be more than a rote recitation from some dusty book or a mnemonic from a lecture way back when. Instead, we must take into account key imaging findings and relevant clinical information.

With these considerations in mind, we at Amirsys designed our Expert Differential Diagnoses series—EXPERTddx for short. Leading experts in every subspecialty of radiology identified the top differential diagnoses in their respective fields, encompassing specific anatomic locations, generic imaging findings, modality-specific findings, and clinically based indications. Our experts gathered multiple images, both typical and variant, for each EXPERTddx. Each features at least eight beautiful images that illustrate the possible diagnoses, accompanied by captions that highlight the pertinent imaging findings. Hundreds more are available in the eBook feature that accompanies every book. In classic Amirsys fashion, each EXPERTddx includes bulleted text that distills the available information to the essentials. You'll find helpful clues for diagnoses, ranked by prevalence as Common, Less Common, and Rare but Important.

Our EXPERTddx series is designed to help radiologists reach reliable—indeed, expert—conclusions. Whether you are a practicing radiologist or a resident/fellow in training, we think the EXPERTddx series will quickly become your practical "go-to" reference.

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PREFACE

When we wrote our first book, *Diagnostic Imaging: Obstetrics*, we set out to create a comprehensive resource for fetal imaging. "Our baby" was very well received (thank you!) so why do we need another book? Although a comprehensive text can give a wealth of in depth information on a given entity, seldom does the patient present saying "I think my baby has XYZ syndrome". The reality is you see an abnormality and now must formulate an appropriate differential diagnosis. *EXPERTddx: Obstetrics* was designed with that very thought in mind and approaches fetal imaging from a working, clinical perspective.

There is an old saying in radiology that "the best way to miss a finding is to make a finding". One becomes so fixated on the obvious finding that the subtle one goes unnoticed, and it is this second finding that may be the key to the specific diagnosis. We wrote *EXPERTddx: Obstetrics* to serve as a practical guide to fetal diagnosis, pointing out which ancillary features are critical to evaluate. It includes many of the most commonly encountered and frustrating findings with which we are confronted. A fetus has a mildly short femur, now what? A patient is referred for an elevated MSAFP; after ruling out a neural tube defect, now what? The face looks funny; if it isn't a proboscis what could it be? Our author team has endeavored to answer these and over a hundred additional important imaging and clinical presentations encountered by the interpreting physician.

For each differential there are imaging and clinical pearls helping to distinguish the various diagnoses; but most importantly, each differential is heavily illustrated, including not only ultrasound, but also fetal MR and clinical/pathologic correlation. As a group, we pored over these lists and literally thousands of cases, endeavoring to make them as practical and inclusive as possible, painstakingly choosing illustrations to best demonstrate the finding. The book is supplemented by an eBook companion that has hundreds more images, creating one of most comprehensive resources for differential diagnoses.

We are excited about the addition of our "new baby" to the family. As was true with its older sibling, it was a labor of love.

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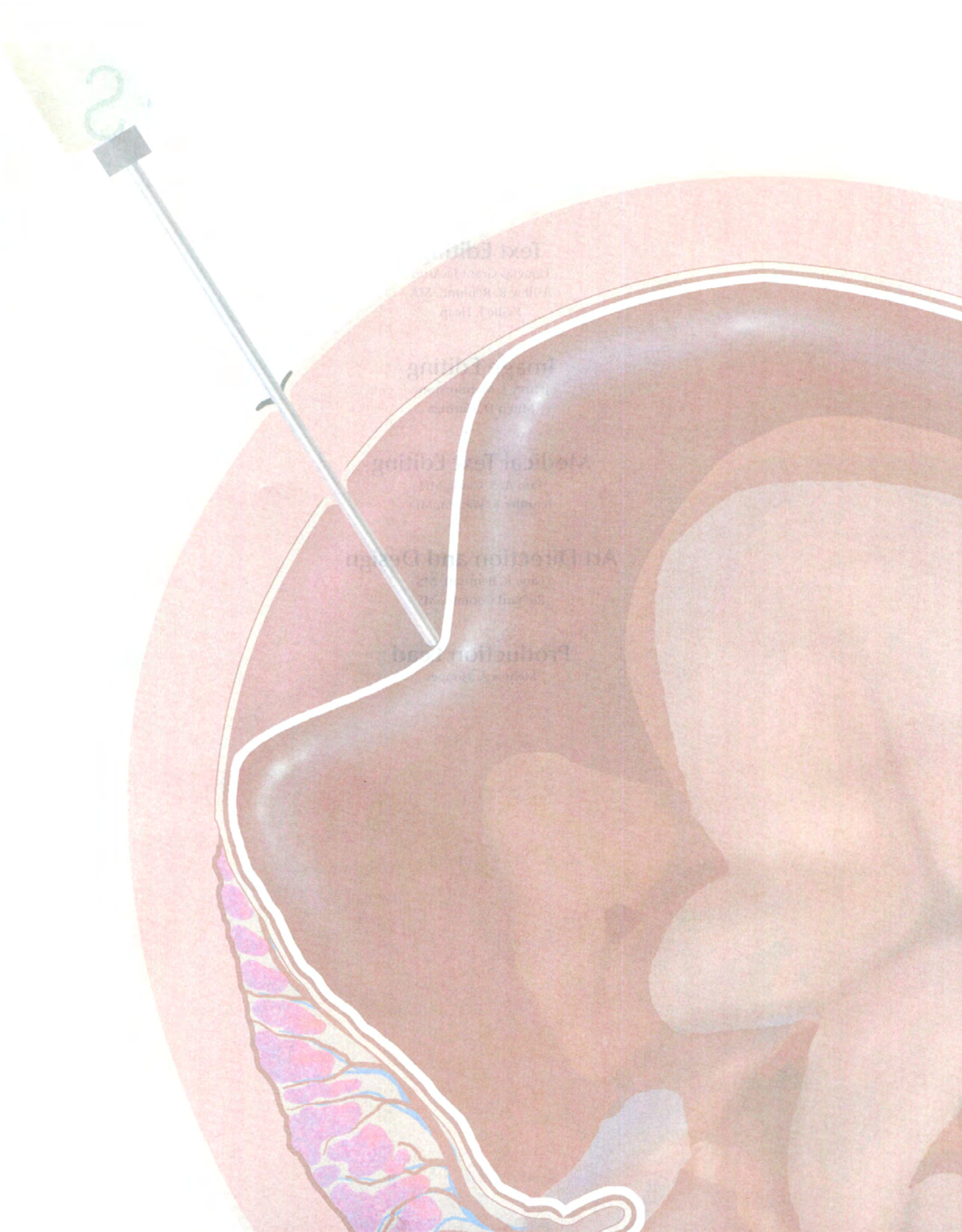
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Central Nervous System

Face/Neck

Chest

Cardiac

Abdomen

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Multiple Gestations

Placenta & Umbilical Cord

Fluid

Growth and Well Being

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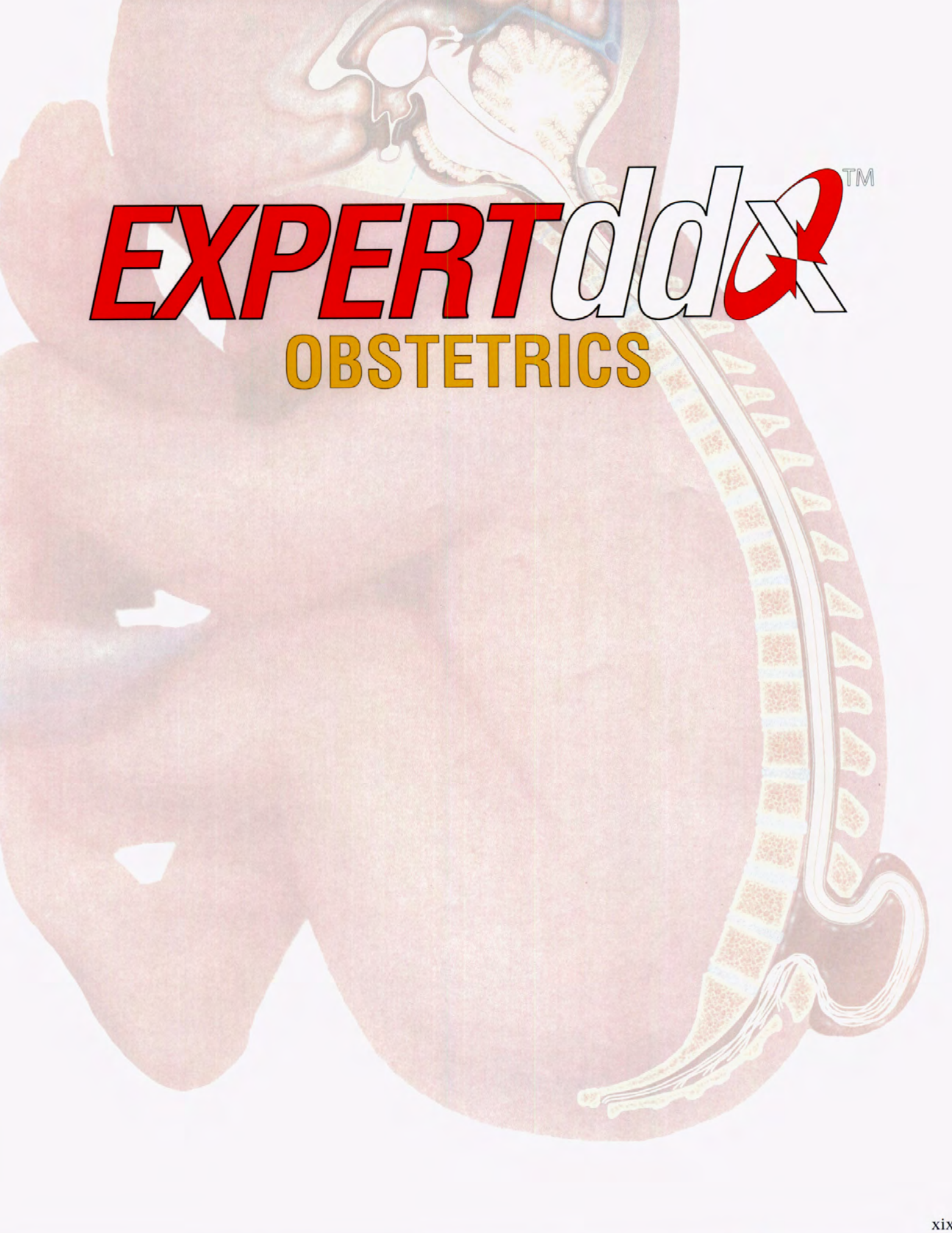
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BLEEDING WITH IUP

DIFFERENTIAL DIAGNOSIS

Common
- Failed Pregnancy
- Perigestational Hemorrhage
- Early Normal Pregnancy
- Anembryonic Pregnancy

Less Common
- Partial Mole
- Twin Demise

Rare but Important
- Interstitial Ectopic
- Cervical Ectopic
- C-section Scar Ectopic
- Heterotopic Pregnancy

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is there a gestational sac?
  - Imperative to differentiate a normal early gestational sac from a pseudosac seen in ectopic pregnancy
  - If no intrauterine pregnancy (IUP) look for adnexal mass, echogenic fluid in cul-de-sac
- Where is the sac located?
  - Compare to prior studies if available
- Has there been appropriate interval development?
  - Must know normal developmental milestones
- Normal sac development
  - Intradecidual sac sign (IDSS) earliest sign of IUP
  - IDSS seen by 4-4.5 weeks after last menstrual period (LMP)
  - Gestational sac "burrows" into endometrium
  - Echogenic ring is eccentric to linear interface of endometrial surfaces
    - Must follow to confirm appropriate growth/milestones
  - Double decidual sac sign (DDSS) seen by 5-5.5 weeks post LMP
    - Initially described as first reliable transabdominal sign of IUP
    - Decidua parietalis (endometrium) surrounds decidua capsularis (gestational sac) = two echogenic rings
- Pseudosac associated with ectopic pregnancy
  - No DDSS
  - Flatter shape than normal gestational sac
  - Central in cavity rather than eccentric
- Even if normal IUP seen beware heterotopic pregnancy if patient symptomatic/has risk factors
- Placenta previa/abruption are NOT first trimester diagnoses
  - Placenta often covers internal os in first trimester
  - Placenta large relative to uterine size
  - Lower uterine segment (LUS) elongates after 28 weeks
  - Placental trophotropism results in migration of placenta away from cervix/LUS as pregnancy progresses

Helpful Clues for Common Diagnoses
- **Failed Pregnancy**
  - Cardiac activity will be absent
  - Sac being expelled from uterus, may contain yolk sac or embryo
  - Sac often flattened/irregular shape
  - Sac in endometrial/cervical canal not implanted into uterus
  - Color Doppler shows lack of normal trophoblastic flow
- **Perigestational Hemorrhage**
  - May be asymptomatic or present with vaginal bleeding
  - Echogenic fluid deep to chorion
    - Becomes hypoechoic over time
  - Normal gestational sac contents
- **Early Normal Pregnancy**
  - Bleeding in pregnancy before visualization of gestational sac (presumed to be implantation bleeding)
    - Follow all apparent IDSS to ensure normal developmental milestones
    - Beware tiny cystic structures in endometrium, may be dilated endometrial glands
  - Idiopathic bleeding: Normal sac/embryo seen but no obvious collection of blood
- **Anembryonic Pregnancy**
  - No visible embryo in gestational sac with diameter ≥ 18 mm by transvaginal exam
  - Look for "empty amnion" sign

Helpful Clues for Less Common Diagnoses
- **Partial Mole**
  - Vaginal bleeding
  - Abnormal embryo/fetus
  - Placenta may look normal or appear cystic

- "Chorionic bump" correlates strongly with partial mole
- **Twin Demise**
  - One normal gestational sac
  - One sac with variety of appearances
    - Perigestational hemorrhage
    - Embryonic demise
    - Anembryonic gestation

Helpful Clues for Rare Diagnoses

- **Interstitial Ectopic**
  - Look for interstitial line sign
  - Eccentric placement of sac in relation to uterine cavity
  - < 5 mm of surrounding myometrium very suggestive
- **Cervical Ectopic**
  - Sac implanted in cervical stroma
  - Look for rim of tissue around sac rather than sac within endocervical canal
  - Sac positioned low in uterus but still perfused (compared to abortion in progress with flattened sac, lack of perfusion)
- **C-section Scar Ectopic**
  - Gestational sac implants into scar from prior C-section, extends to serosa
- **Heterotopic Pregnancy**
  - Intrauterine & ectopic pregnancy
  - Look for adnexal mass, echogenic fluid in addition to IUP
    - Differential: Normal IUP with hemorrhagic corpus luteum
  - Risk factors for heterotopic pregnancy
    - Assisted reproduction
    - Intrauterine contraceptive device
    - History of pelvic inflammatory disease
    - History of endometriosis

Other Essential Information

- Important to recognize unusual (other than tubal) ectopics
  - Pregnancy is "in uterus" but not in correct place
  - C-section scar ectopic treated with systemic methotrexate or percutaneous injection
  - Risk of torrential bleeding/emergent hysterectomy if curettage attempted in cervical ectopic
  - Interstitial ectopic also best treated conservatively if patient stable

Alternative Differential Approaches

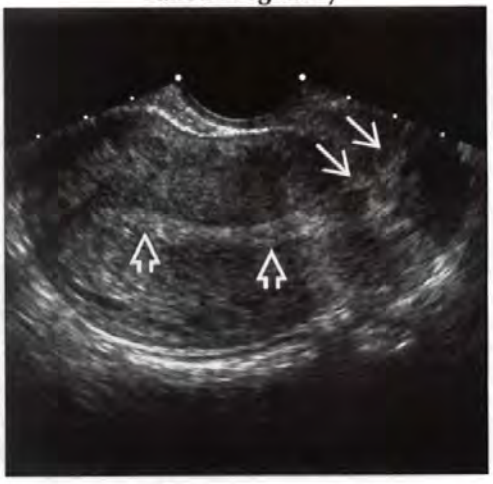
- Bleeding with intrauterine sac but no fetal pole
  - Anembryonic pregnancy
  - Pseudosac from ectopic pregnancy
  - Perigestational hemorrhage
- Bleeding with an embryo
  - Perigestational hemorrhage
  - Idiopathic (no obvious collection of blood)
- Bleeding with no visible IUP
  - Very early normal pregnancy
  - Complete abortion
  - Tubal ectopic pregnancy

Failed Pregnancy



Sagittal transabdominal ultrasound shows an apparently empty uterus with a small amount of fluid ➡ in the region of the cervix. The patient had an earlier scan documenting a live IUP.

Failed Pregnancy



Sagittal transvaginal ultrasound in the same case as previous image shows an empty endometrial cavity ➡ but some mixed echogenicity material ➡ in the cervix.

BLEEDING WITH IUP

(Left) *Sagittal transvaginal ultrasound in the same case "zoomed" on the cervical area shows the collapsed gestational sac ➡ containing a 6 mm dead embryo (calipers) and a yolk sac ➡. This spontaneously passed shortly after the exam.*
(Right) *Sagittal ultrasound shows an IUP with a yolk sac ➡ and embryo ➡. There is an adjacent perigestational hemorrhage ➡. This pregnancy failed.*

Failed Pregnancy



Perigestational Hemorrhage



(Left) *Sagittal ultrasound shows a large perigestational hemorrhage ➡ at the end of the first trimester.* ***(Right)*** *Axial ultrasound shows that the perigestational hemorrhage ➡ covers at least 50% of the gestational sac circumference. Despite the large size and repeated episodes of spotting, this patient delivered a healthy term infant.*

Perigestational Hemorrhage



Perigestational Hemorrhage



(Left) *Sagittal transvaginal ultrasound shows a thick, decidualized endometrium, with a tiny fluid-filled structure ➡ low in the uterus.* ***(Right)*** *Color Doppler shows the same fluid-filled structure ➡. This proved not to be the gestational sac, as an IUP was later seen at the fundus. Presumed etiology for spotting was implantation bleeding. Follow-up is often required to differentiate an early IUP from cystic dilatation of endometrial glands.*

Early Normal Pregnancy



Early Normal Pregnancy



BLEEDING WITH IUP

Anembryonic Pregnancy

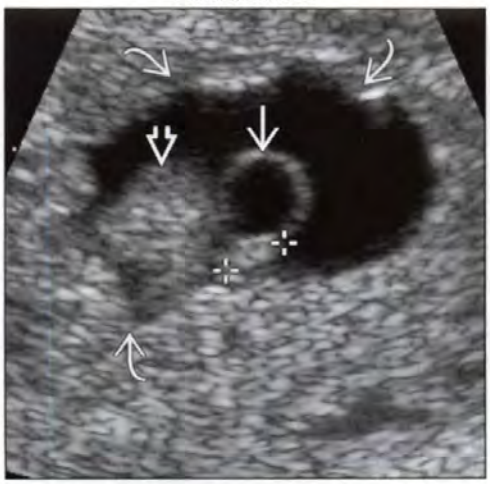


Anembryonic Pregnancy



(Left) *Transabdominal ultrasound shows the "empty amnion" sign. Note also the irregular shape of the gestational sac and small echogenic yolk sac.* ***(Right)*** *Sagittal oblique transabdominal ultrasound in an HIV-positive patient shows a flattened empty gestational sac (calipers). Mean sac diameter was 31 mm, and TV sonography confirmed the absence of an embryo and yolk sac.*

Partial Mole



Partial Mole



(Left) *Transvaginal ultrasound shows a dead embryo (calipers), irregular gestational sac, and large yolk sac. Note the echogenic material adjacent to the embryo. This is a chorionic bump.* ***(Right)*** *Transvaginal ultrasound in the same case as previous image shows multiple protrusions of the chorion (i.e., chorionic bumps). Partial mole was confirmed after D&C. There is a strong association of chorionic bumps with partial mole.*

Twin Demise



Twin Demise



(Left) *Transabdominal ultrasound shows a dichorionic twin gestation with failure of the right-sided gestation. The left-sided sac contains a normal embryo and yolk sac.* ***(Right)*** *Transabdominal ultrasound shows a normal second trimester fetus on the right and a small tissue mass on the left, representing a dead embryo. Typically the failed pregnancy resorbs over time and may disappear completely.*

BLEEDING WITH IUP

Interstitial Ectopic



Interstitial Ectopic

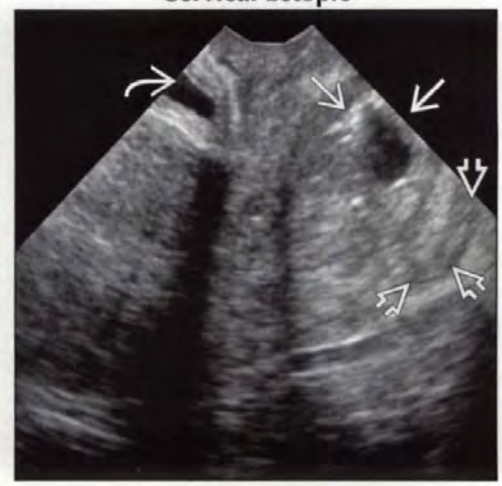


(Left) *Axial oblique transvaginal ultrasound shows the "interstitial line sign" ➡ leading from the uterine cavity to the gestational sac ➡, which is implanted far lateral in the right cornual region.* ***(Right)*** *Sagittal oblique transvaginal ultrasound in the same case as previous image shows a very thin rim of myometrium ➡ that surrounds the gestational sac (< 5 mm of surrounding myometrium is highly suggestive of an interstitial ectopic pregnancy).*

Cervical Ectopic



Cervical Ectopic



(Left) *Sagittal ultrasound shows an "empty" uterus with a thin endometrial echo complex ➡. Note the fluid containing structure in the region of the cervix ➡.* ***(Right)*** *Sagittal transvaginal ultrasound in the same case as previous image shows the gestational sac ➡ implanted in the anterior cervical stroma. The bladder ➡ and posterior cervical stroma ➡ are also seen.*

Cervical Ectopic



C-section Scar Ectopic



(Left) *Axial ultrasound through the cervix in the same case shows an embryo ➡ within the gestational sac ➡, which is implanted within the anterior cervical lip (➡ indicates posterior lip). Doppler evaluation showed cardiac activity.* ***(Right)*** *Sagittal transvaginal ultrasound shows blood products in the endometrial cavity ➡. The gestational sac is irregular in shape, and the chorionic tissue ➡ extends into the anterior myometrium at the site of the old hysterotomy.*

BLEEDING WITH IUP

C-section Scar Ectopic

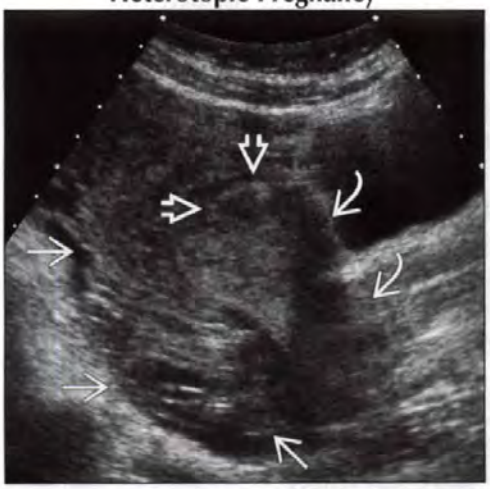


Heterotopic Pregnancy



(Left) *Color Doppler ultrasound from the prior case shows increased blood flow in the region of the scar ➡. Note that the sac is not implanted in the cervix nor is it within the cervical canal ➡, as would be seen with an abortion in progress.* ***(Right)*** *Axial oblique transvaginal ultrasound in a patient following assisted reproduction shows an IUP ➡ with another gestational sac ➡ seen in the left adnexa. Salpingostomy was successful, and the IUP went to term.*

Heterotopic Pregnancy

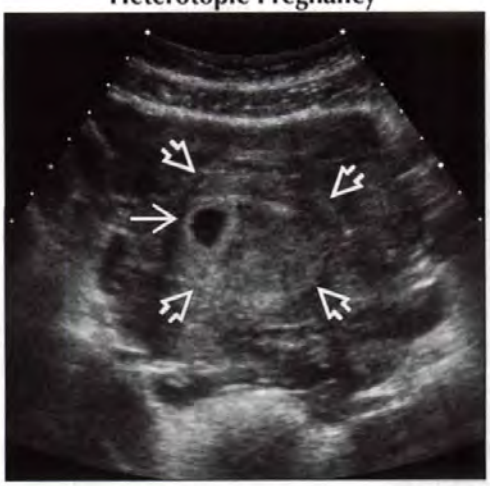


Heterotopic Pregnancy

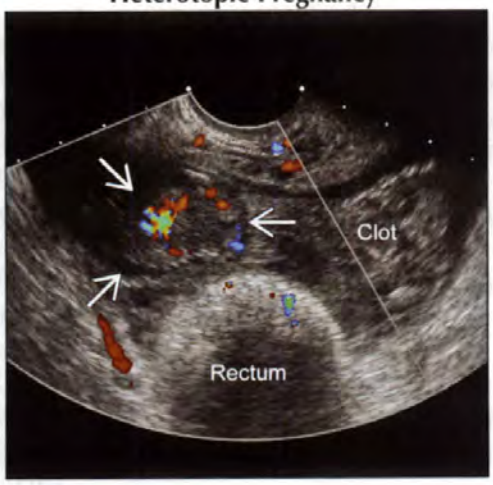


(Left) *Sagittal transabdominal ultrasound shows an anteposition uterus ➡ and a large volume of clot in the cul-de-sac ➡. An echogenic ring in the fundus ➡ was a possible intrauterine pregnancy.* ***(Right)*** *Coronal transabdominal ultrasound shows fluid in the hepatorenal fossa ➡, confirming a large volume intraperitoneal bleed.*

Heterotopic Pregnancy



Heterotopic Pregnancy



(Left) *Axial ultrasound confirms an IUP ➡. Pelvic thrombus obscures normal uterine serosal echoes ➡; therefore, careful imaging is required to separate the uterus from adnexal structures.* ***(Right)*** *Transvaginal ultrasound of the right adnexa shows a hemorrhagic mass ➡ with surrounding flow separate from the uterus and IUP. Ruptured right tubal ectopic was confirmed at surgery; thus this is a heterotopic pregnancy.*

BLEEDING WITHOUT VISIBLE IUP

DIFFERENTIAL DIAGNOSIS

Common
- Complete Abortion
- Very Early Normal Pregnancy
- Retained Products of Conception
- Tubal Ectopic

Less Common
- Complete Hydatidiform Mole

Rare but Important
- Abdominal Ectopic

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- It is essential to confirm patient is actually pregnant
  - Ruptured hemorrhagic cyst can present in similar manner to ectopic pregnancy
  - Most cyst ruptures are managed conservatively
  - Bleeding ectopic pregnancy requires surgery
  - Check urine pregnancy test ± quantitative beta hCG in all cases
- Tubal ectopic must be ruled out in every case
  - If patient clinically unstable laparoscopy may be required to establish diagnosis
    - Blood in pelvis obscures serosal margins
    - Patients are often in pain and cannot tolerate transducer pressure
    - Positive pregnancy test & blood in pelvis & unstable patient requires laparoscopy
  - If stable then "wait and see" approach is reasonable
    - Follow serial beta hCG at 48 hr intervals
    - Repeat ultrasound when beta hCG reaches threshold levels or if symptomatic change
  - Avoid presumptive diagnosis of ectopic
    - Must know normal early pregnancy development and milestones to avoid this mistake
    - Do not give methotrexate to a patient with a possible early intrauterine pregnancy (IUP)
    - Therapeutic abortion required if methotrexate spontaneous abortion does not occur

Helpful Clues for Common Diagnoses
- **Complete Abortion**
  - Uterus empty with os closed
  - Beta hCG will decrease on follow-up
- **Very Early Normal Pregnancy**
  - Bleeding in pregnancy before visualization of gestational sac (presumed to be implantation bleeding)
  - Commercial pregnancy tests now very sensitive: Positive on day 1 of missed menses
  - At 4-4.5 weeks menstrual age may not see any sonographic evidence of pregnancy
    - Transvaginal (TV) sonography mandatory for best resolution
    - Beware tiny cystic structures in endometrium, may be dilated endometrial glands
    - Follow all apparent IDSS to ensure normal developmental milestones
  - If quantitative beta hCG low and patient stable repeat at 48 hr intervals
    - If doubling normally likely early IUP
    - Repeat scan once beta hCG > 2,000 IU
- **Retained Products of Conception**
  - Material in endometrial cavity
    - Echogenic more suspicious than hypoechoic
  - Look for feeding vessels on color Doppler
- **Tubal Ectopic**
  - Beta hCG at level where IUP should be seen (> 2,000 IU)
  - No intrauterine gestation sac
  - Look for adnexal mass
    - Tubal ring moves separately from ovary with transducer pressure
    - "Ring of fire" on color Doppler interrogation due to trophoblastic flow
    - Remember corpus luteum also has "ring of fire" but will be in or move with ovary
  - Look for hematosalpinx
    - Tubular adnexal structure with echogenic, non-perfused contents
    - Due to hemorrhage into lumen of tube
    - Occurs before tubal rupture leads to intraperitoneal bleeding
  - Beware pseudosac in uterus
    - Fluid collection central in uterine cavity
    - Normal gestation "burrows" into endometrium, therefore eccentric or intradecidual

- No intradecidual sac sign (IDDS)
- No double decidual sac sign (DDSS)
- Flattened shape
- No recognizable internal structures: May contain low echogenicity amorphous
- Normal IUP has yolk sac ± embryo

◦ Echogenic fluid in cul-de-sac correlates strongly with bleeding
- Pus also creates echogenic fluid but pregnancy test negative, patient presents with fever/elevated white cell count

Helpful Clues for Less Common Diagnoses

• **Complete Hydatidiform Mole**
◦ Technically "product of conception" but abnormal so no embryo
◦ Uterus distended by mass with hydropic chorionic villi creating a "bunch of grapes" or snowstorm appearance
◦ Beta hCG may be very high
◦ Look for theca lutein cysts in ovaries
- Seen in 25% of cases

◦ Often large for dates with hyperemesis ± hypertension

Helpful Clues for Rare Diagnoses

• **Abdominal Ectopic**
◦ Gestation implanted outside uterus within peritoneal cavity
◦ Amniotic sac intact therefore embryo/fetus surrounded by fluid
◦ May see multiple placental implantation sites
◦ Uterus is empty and deep in maternal pelvis
◦ ALWAYS make sure that the fetus is IN the uterus
- Follow cervix to anterior and posterior myometrium

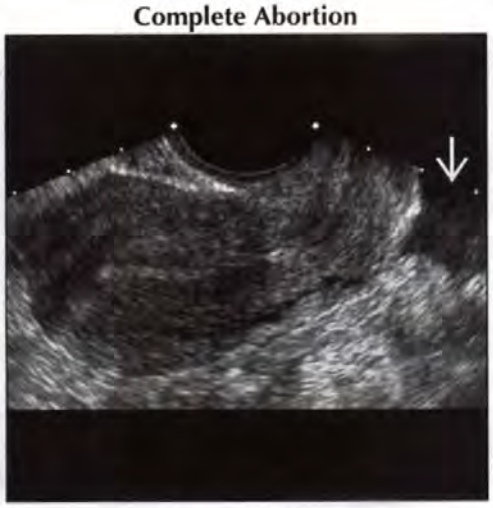
Other Essential Information

• Time line, clinical history and prior lab/imaging information are important
◦ When did urinary pregnancy test become positive?
◦ Cervix open or closed?
◦ Are there any prior scans?
- If confirmed prior IUP with empty uterus now complete abortion is most likely diagnosis

◦ Is there a quantitative beta hCG?
- Doubling normally → probable early IUP
- Increasing abnormally → abnormal IUP or ectopic
- Decreasing → failed IUP, complete abortion, resolving ectopic

• Discriminatory beta hCG
◦ Variable numbers in literature from 1,500 → 2,000
◦ Most institutions decide on a number; author's institution uses 2,000
◦ Once the threshold is reached an IUP should be demonstrated
- Regular increase and no IUP ⇒ ectopic most likely diagnosis
- If single data point and no IUP ⇒ ectopic or complete abortion are possible, management determined by clinical condition

Complete Abortion



Sagittal transvaginal ultrasound shows an empty uterus in a patient with a positive urine pregnancy test and pelvic pain. She had passed "tissue" and was clinically stable, though in pain. Note echogenic fluid ➡.

Complete Abortion



Ultrasound in the same case shows a hemorrhagic right adnexal mass ➡. Her pain settled; repeat scan showed resolution of clot. Presumed diagnosis was complete abortion and ruptured hemorrhagic corpus luteum.

BLEEDING WITHOUT VISIBLE IUP

Very Early Normal Pregnancy

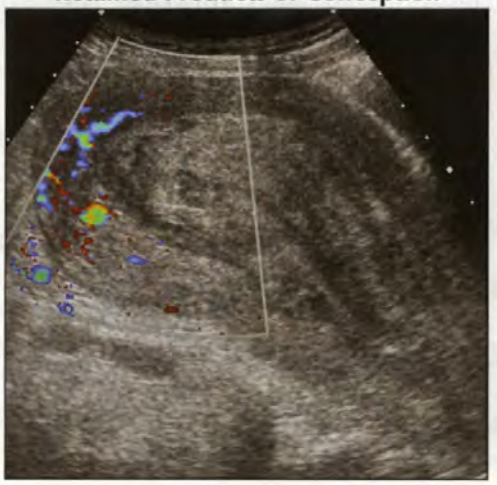


Very Early Normal Pregnancy

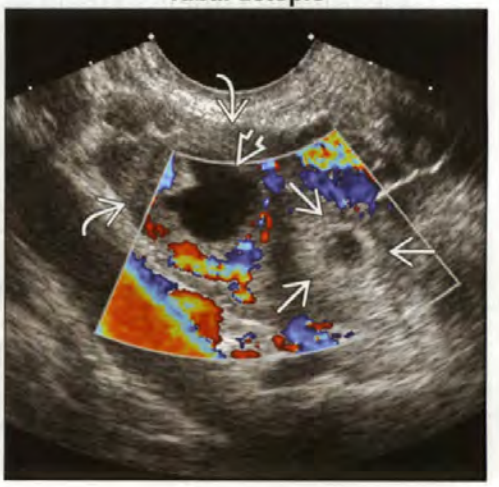


(Left) *Sagittal transvaginal ultrasound shows fluid in the cavity, decidualized endometrium, and a questionable echogenic ring, suggestive of the intradecidual sac sign. The patient was clinically stable.* ***(Right)*** *Axial transvaginal ultrasound in the same case 10 days later shows interval development of a normal IUP with an embryo. Short term follow-up scans are very useful in early pregnancy.*

Retained Products of Conception



Tubal Ectopic

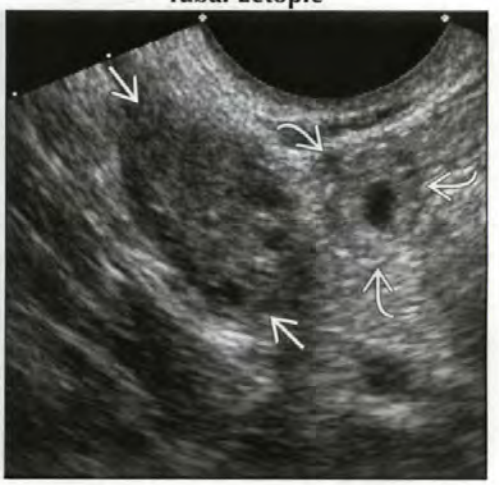


(Left) *Sagittal color Doppler ultrasound shows a large amount of echogenic material in the uterus in a patient with a prior ultrasound confirming a live IUP. This is the typical appearance of retained products of conception.* ***(Right)*** *Axial oblique color Doppler ultrasound shows an adnexal ring adjacent to the right ovary, which has a corpus luteum cyst within it. Other images showed no evidence of an IUP. Surgery confirmed right tubal ectopic.*

Tubal Ectopic



Tubal Ectopic



(Left) *Sagittal transvaginal ultrasound in a different case shows thick inhomogeneous endometrium but no evidence of a gestational sac.* ***(Right)*** *Axial oblique transvaginal ultrasound in the same patient shows a normal ovary and a ring-like adnexal mass. There was no evidence of intraperitoneal bleeding, and the patient was successfully treated with methotrexate.*

BLEEDING WITHOUT VISIBLE IUP

Tubal Ectopic

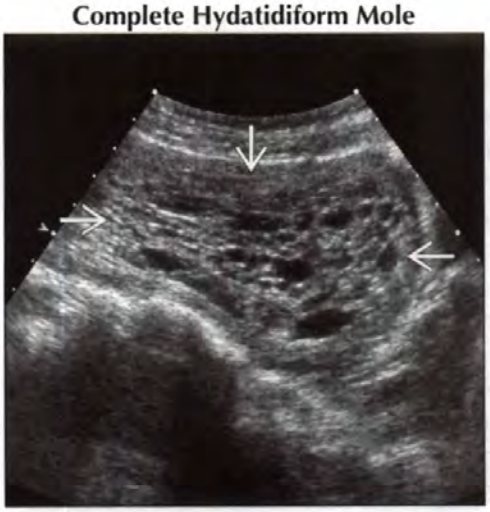


Tubal Ectopic

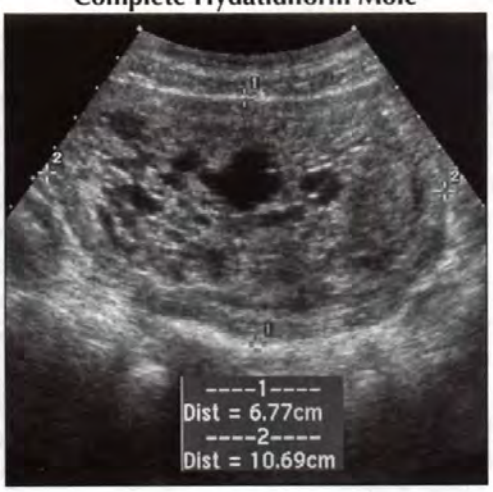


(Left) *Sagittal transvaginal ultrasound shows an irregularly shaped "sac" with a single echogenic ring and internal debris. Note echogenic fluid in the cul-de-sac. The patient refused intervention as this was a highly desired pregnancy.* ***(Right)*** *Power Doppler ultrasound 48 hours later shows increased intraperitoneal bleeding and a hemorrhagic left adnexal mass. Surgery confirmed ruptured tubal ectopic. Uterus negative for products of conception.*

Complete Hydatidiform Mole

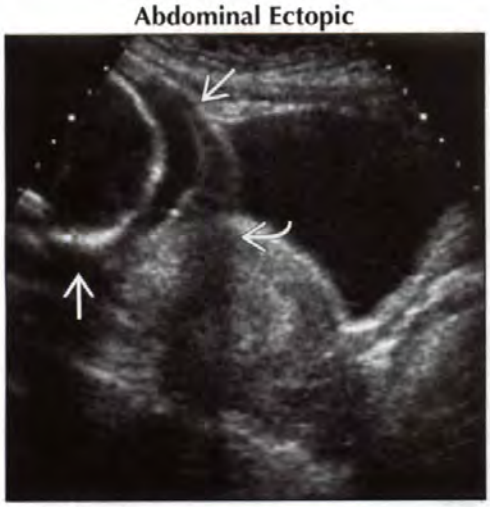


Complete Hydatidiform Mole

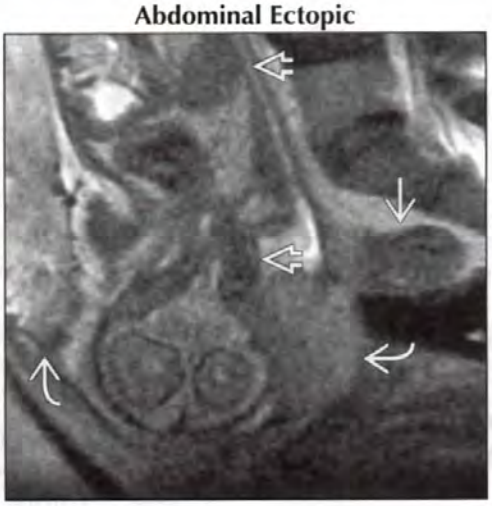


(Left) *Sagittal transabdominal ultrasound shows a multicystic mass in the uterus. There is no recognizable fetus.* ***(Right)*** *Axial transabdominal ultrasound in the same patient confirms a large uterus filled with a multicystic mass. The beta hCG was elevated. She was treated with suction curettage. Pathology confirmed complete hydatidiform mole.*

Abdominal Ectopic



Abdominal Ectopic



(Left) *Sagittal transabdominal ultrasound in a patient with abdominal pain at 24 weeks shows that the gestational sac is implanted on the fundus, not inside the uterus. She had three prior scans at which this finding was missed.* ***(Right)*** *Sagittal T2WI MR performed for surgical planning shows two placental masses and the fetus without surrounding myometrium. The cervix could be followed to an empty uterus in adjoining scan planes.*

ABNORMAL SAC CONTENTS

DIFFERENTIAL DIAGNOSIS

Common
- Anembryonic Pregnancy
- Abnormal Yolk Sac
- Embryonic Demise
- Pseudosac in Ectopic Pregnancy

Less Common
- Retained Products of Conception
- Gestational Trophoblastic Disease
  - Complete Hydatidiform Mole
  - Partial Mole
  - Invasive Mole
- Chorionic Bump
- Endometrial Polyp in Pregnancy

Rare but Important
- Abnormal Fetus

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Be familiar with normal appearances of early pregnancy (i.e., normal sac contents)
  - Intradecidual sac sign (IDSS)
    - Earliest described sign of intrauterine pregnancy (IUP)
    - Cystic structure within endometrium, eccentric in relation to cavity
  - Double decidual sac sign (DDSS)
    - Earliest transabdominal finding of IUP
  - Double bleb sign
    - Seen for short period in early pregnancy
    - Double bleb = yolk sac + amnion on either side of embryonic disc
- Extra-embryonic coelom (chorionic cavity) = space between amnion and chorion
  - Normally echogenic compared to anechoic fluid inside amnion
- Normal IUP growth/milestones
  - Normal gestational sac grows ~ 1 mm per day
  - When mean sac diameter (MSD) ≥ 10 mm yolk sac should be visible
  - When MSD ≥ 18 mm embryo should be visible
  - "Five alive" rule: Embryo with crown rump length ≥ 5 mm must have cardiac activity

Helpful Clues for Common Diagnoses
- **Anembryonic Pregnancy**
  - Empty amnion sign
  - Gestational sac with MSD > 18 mm, visible amnion ± yolk sac but no embryo
    - Failure of embryo to develop
    - Early embryonic demise and resorption
- **Abnormal Yolk Sac**
  - Flattened
  - Calcified
  - > 6mm diameter
- Yolk sac supports pregnancy prior to placental development
  - Normally ≤ 6 mm in diameter, spherical in shape
- **Embryonic Demise**
  - Absence of cardiac activity in an embryo with crown rump length > 5 mm
    - Use endovaginal sonography
    - Two independent observers
  - Beware of confusing maternal pulse with embryonic cardiac activity
    - Embryonic heart rate is usually faster than maternal (120-160 vs. 80 beats per minute)
    - If possible embryonic bradycardia feel maternal pulse while watching embryonic heart rate
- **Pseudosac in Ectopic Pregnancy**
  - Oval or flat shape
  - Central in cavity
  - Not surrounded by double, echogenic, chorionic ring
  - Possible pseudosac merits careful search for ectopic pregnancy
  - Tubal ectopic most common association with pseudosac
    - Cornual, cervical, C-section ectopics should be evident

Helpful Clues for Less Common Diagnoses
- **Retained Products of Conception**
  - RPOC: Irregular collapsed sac or echogenic chorionic remnants
  - Blood flow on color Doppler implies RPOC rather than blood clot
- **Complete Hydatidiform Mole**
  - Typical clinical presentation
    - Hyperemesis, hypertension, size > dates, vaginal bleeding
  - Cavity distended by mass with multiple "cysts" → "bunch of grapes" or "snowstorm" appearance
  - May be associated with ovarian theca lutein cysts

ABNORMAL SAC CONTENTS

- **Partial Mole**
  - First trimester: Unusual looking sac/chorionic bump
  - Second trimester presentation depends on source of third set of chromosomes
    - Large, cystic placenta if paternal
    - Small placenta if maternal
- **Invasive Mole**
  - No embryo
  - Complex intrauterine vascular mass
  - Loss of endometrial myometrial interface due to invasion into myometrium
- **Chorionic Bump**
  - Focal protuberance from chorionic surface
  - First described among in vitro fertilization (IVF) population but also seen with spontaneous conception
  - 50% loss rate documented in IVF patients
  - Etiology unknown; possible early bleed
  - Strong association with partial mole
- **Endometrial Polyp in Pregnancy**
  - < 2 cm unlikely to impact pregnancy rate/success in IVF population
  - Modern approach to IVF patients indicates polypectomy justifiable if polyp discovered in work-up

Helpful Clues for Rare Diagnoses

- **Abnormal Fetus**
  - First trimester detection of fetal anomaly is possible
    - Increased nuchal translucency
    - Cystic hygroma
    - Limb reduction defects
    - Central nervous system: Holoprosencephaly, hydranencephaly, exencephaly
    - Abdominal wall defects: Gastroschisis, omphalocele

Other Essential Information

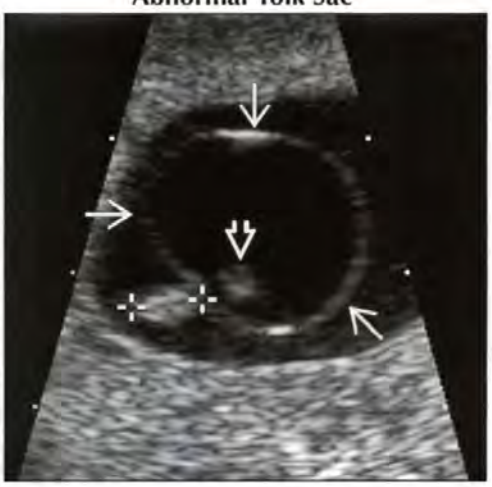
- Differentiate abnormal sac contents from abnormality adjacent to sac
  - Perigestational hemorrhage
    - Mixed echogenicity between chorion and myometrium
    - Crescentic shape
    - Often associated with vaginal bleeding
  - Failed twin
    - Smaller MSD than normal twin
    - Irregular shape
    - Diminished chorionic echogenicity
    - Decrease in size on follow-up
  - Uterine myomata
    - Hypoechoic mass adjacent to sac
    - Posterior submucosal myomata associated with recurrent abortion
    - Degenerated fibroid may appear similar to sac with echogenic fluid content
- Beware pseudosac of ectopic pregnancy
  - Ruptured ectopic still important cause of maternal morbidity and mortality
  - Know signs of intra-uterine pregnancy (IUP)
    - IDSS: Fertilized ovum burrows into decidualized endometrium
    - DDSS: Two concentric echogenic rings
    - Presence of a yolk sac confirms IUP

Anembryonic Pregnancy



Coronal transvaginal ultrasound shows an abnormal calcified yolk sac ➡ outside the amnion ➡. At this mean sac diameter a living embryo should have been present. This is the "empty amnion" sign.

Abnormal Yolk Sac



Transvaginal ultrasound shows a structure (calipers) mistaken for the embryo. It is a pyknotic yolk sac lying outside the amnion ➡. The embryo ➡ is inside the amnion. Follow-up confirmed embryonic demise.

ABNORMAL SAC CONTENTS

Abnormal Yolk Sac



Abnormal Yolk Sac



(Left) *Transvaginal ultrasound shows an enlarged yolk sac (calipers) adjacent to the misshapen amnion. The embryo had no cardiac activity.* ***(Right)*** *Transvaginal ultrasound shows an intrauterine pregnancy with a triangular yolk sac. The embryo had bradycardia with heart rate of 90 bpm. Spontaneous miscarriage occurred before the follow-up examination.*

Embryonic Demise

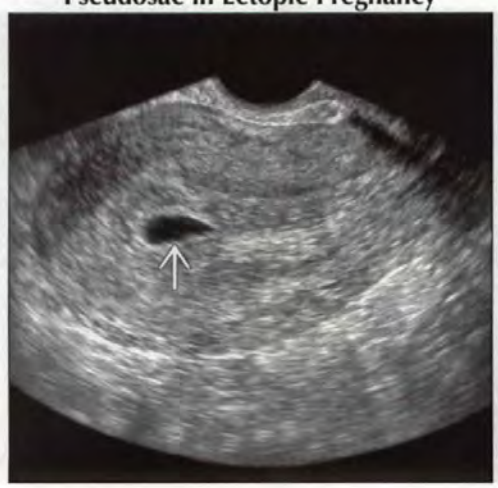


Embryonic Demise



(Left) *Color Doppler ultrasound shows a residual dead embryo in a sac full of echogenic debris. Initial images two weeks earlier had shown a live embryo, yolk sac and chorionic bump.* ***(Right)*** *M-mode ultrasound shows no detectable cardiac activity for the embryo, consistent with embryonic demise. A markedly enlarged yolk sac is seen next to the embryo.*

Pseudosac in Ectopic Pregnancy



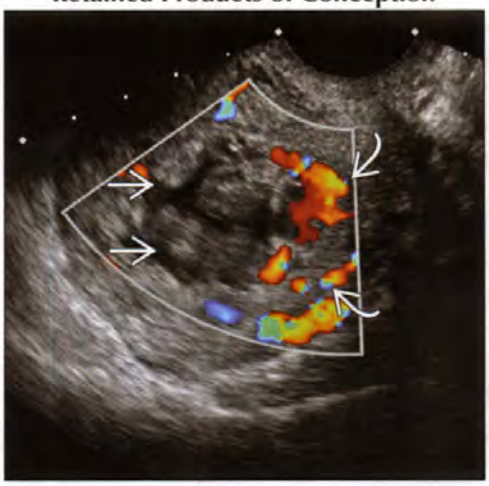
Pseudosac in Ectopic Pregnancy



(Left) *Sagittal ultrasound shows a small fluid collection in the middle of the endometrial cavity. This should not be confused with an IUP. Note the lack of a double decidual sac sign. An ectopic pregnancy was confirmed.* ***(Right)*** *Transvaginal ultrasound shows a "sac" with irregular echogenic debris. There is a large amount of blood within the cul-de-sac. Surgery confirmed a ruptured tubal ectopic pregnancy.*

ABNORMAL SAC CONTENTS

Retained Products of Conception

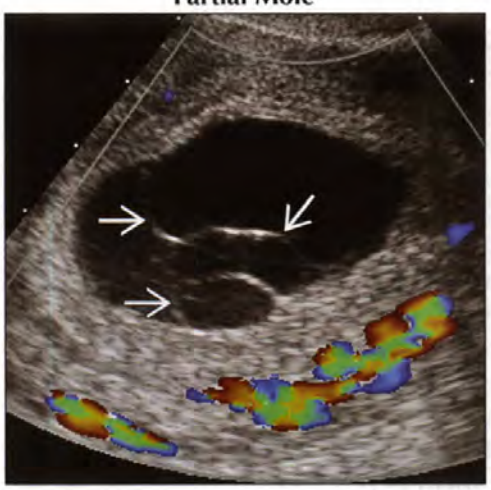


Complete Hydatidiform Mole

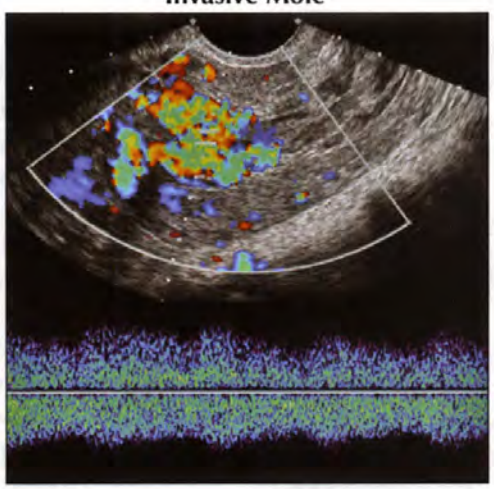


(Left) *Color Doppler ultrasound in a patient with pain and bleeding shows mixed echogenicity material ➡ with increased flow ➡. Earlier scans had shown a live embryo. D&C confirmed RPOC.* ***(Right)*** *Transabdominal ultrasound shows the uterus distended by a multicystic mass ➡. No normal fetal parts were seen. The "cysts", which are hydropic chorionic villi, create the characteristic "bunch of grapes" appearance.*

Partial Mole

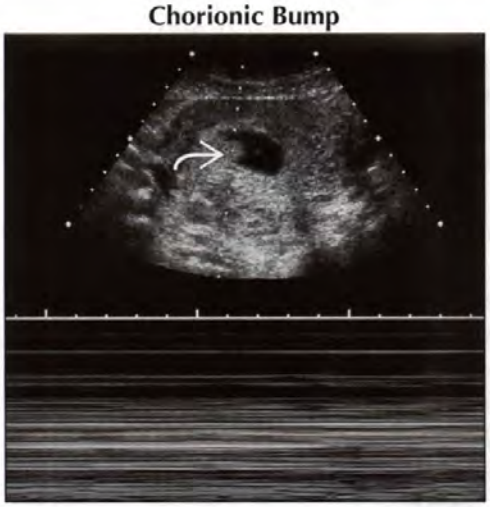


Invasive Mole

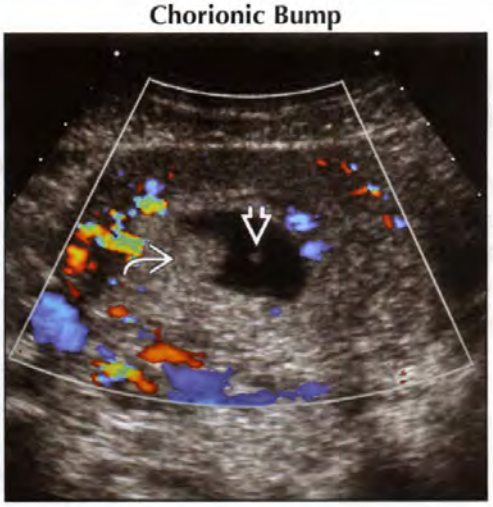


(Left) *Transvaginal ultrasound shows an intrauterine sac containing fine avascular membranes ➡ but neither embryo nor yolk sac. Tissue diagnosis after D&C was partial mole.* ***(Right)*** *Color Doppler ultrasound shows a hypervascular mass with no clear distinction with the myometrium, very concerning for invasion. Pulsed Doppler shows high velocity, low resistance flow. Medical treatment was successful.*

Chorionic Bump



Chorionic Bump



(Left) *M-mode ultrasound not surprisingly shows no cardiac activity in this chorionic bump ➡ mistaken for an embryo by an inexperienced sonographer. An anembryonic pregnancy was confirmed on follow-up.* ***(Right)*** *Color Doppler ultrasound in the same case shows no flow within the chorionic bump ➡. A small echogenic yolk sac ➡ is also seen. Chorionic bumps have been associated with both triploidy and early pregnancy loss.*

ABNORMAL SAC POSITION

DIFFERENTIAL DIAGNOSIS

Common
- Spontaneous Abortion
- Tubal Ectopic

Less Common
- Uterine Duplication
- Interstitial Ectopic
- Cervical Ectopic
- C-Section Scar Ectopic

Rare but Important
- Heterotopic Pregnancy
- Abdominal Ectopic

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Confirm intrauterine pregnancy (IUP)
  - Exclude ectopic pregnancy
    - Beware of pseudosac: Fluid centrally located in endometrial canal
  - Should have typical signs of early IUP depending on gestational age
    - Intradecidual sac sign: Echogenic ring around sac within endometrium
    - Double decidual sac sign: Paired echogenic rings around early sac
    - Double bleb sign: Yolk sac and amniotic sac
    - Diamond ring sign: Yolk sac with associated early embryo
- Clarify location of implantation site
  - Should be eccentric within endometrium
  - Sac should be within the body of the uterus above internal os
  - Myometrium should be completely surrounding sac

Helpful Clues for Common Diagnoses
- **Spontaneous Abortion**
  - Use color Doppler to differentiate ectopic implantation from abortion in progress
    - Early IUP has increased surrounding flow; "ring of fire" appearance due to trophoblastic tissue
    - Spontaneous abortion is much less vascular
  - If embryo visualized, assess for heart beat
    - If heart rate detected, usually indicates an implanted pregnancy
  - Evaluate internal os
    - Will be open for abortion in progress
    - Closed in cervical ectopic
  - Correlate with serial hCG
    - Should be decreasing with spontaneous abortion
- **Tubal Ectopic**
  - Most specific diagnostic clue: Adnexal gestational sac ± embryo
  - Most common presentation: No IUP, tubal/adnexal mass, echogenic free fluid in cul-de-sac
    - Decidual reaction in uterus
    - May have pseudosac
    - Heterogeneous tubal hematoma
    - Pulsed Doppler shows low resistance flow in tubal pregnancy
  - Ectopic often on same side as corpus luteum
  - Ultrasound negative in 5-10% of cases

Helpful Clues for Less Common Diagnoses
- **Uterine Duplication**
  - May give the appearance of ectopic implantation
  - Implantation actually within one horn of uterine anomaly
    - Didelphys: 2 separate endometrial cavities
    - Bicornuate: 2 separate uterine horns with concave outer uterine contour
    - Septate: Variable length of septum separating cavities, normal outer uterine contour
  - Myometrium completely surrounds sac as implantation is normal
- **Interstitial Ectopic**
  - Look for interstitial line sign
    - Echogenic line from endometrium to ectopic sac
  - Myometrium around sac should be at least 5 mm thick
  - Can grow to be larger than tubal ectopic as blood supply better
    - Within intramural portion of fallopian tube
  - May present as echogenic mass within cornua without sac
    - Mass is combination of trophoblastic tissue and hematoma
- **Cervical Ectopic**
  - Prior instrumentation of uterus considered key risk factor

- Assess for "hourglass" shape of uterus
  - Cervix distended but internal os is closed
  - Transabdominal ultrasound helpful to evaluate landmarks and shape
- Eccentric implantation into wall of cervix
  - Distinguishes from spontaneous abortion in progress which is central

- **C-Section Scar Ectopic**
  - Multiple prior C-sections may increase risk
    - Look for implantation near scar and thinned/absent anterior myometrium
  - Assess for other associated complications if presenting later in pregnancy
    - Placenta accreta, increta, percreta
    - Placenta previa
    - Placental abruption
  - Trophoblastic tissue in scar may invade into bladder

Helpful Clues for Rare Diagnoses

- **Heterotopic Pregnancy**
  - Correlate with clinical history
    - < 1:30,000 in spontaneous pregnancies
    - Much more common if history of assisted reproduction
    - Damage to endometrium or fallopian tubes predisposes to ectopic implantation
  - IUP identified but adnexal mass seen
  - Beware of misdiagnosis due to "ring of fire" around corpus luteum
    - Intraovarian ectopics exceedingly rare
    - Tissue around corpus luteum can normally be hypervascular

- **Abdominal Ectopic**
  - No IUP identified but sac or embryo/fetus seen outside the uterus
  - Lack of normal hypoechoic myometrial rim around pregnancy
  - Look for echogenic free fluid in abdomen and pelvis
  - If ruptured or early may only see hematoma

Other Essential Information

- Always correlate with serum human chorionic gonadotropin (hCG)
  - If hCG > 2000 IU should see IUP
    - No IUP → missed or spontaneous abortion vs. ectopic pregnancy
    - If no IUP and patient stable, can follow serial hCG ± ultrasound
  - If hCG < 2000 IU may be too early to see IUP
    - Differential includes early IUP, spontaneous abortion and ectopic pregnancy
    - Look for signs of ruptured ectopic: Echogenic fluid, adnexal "mass", pain, anemia
    - If patient stable, can follow with serial hCG and ultrasound
- Normal hCG should double every 2-3 days in the first trimester
  - Ectopic pregnancies usually rise more slowly than normal IUP
  - Abnormally developing early IUP can also have slower increase in hCG

Spontaneous Abortion



Sagittal transvaginal ultrasound shows an amorphous 8 week embryo and sac in the cervix. The internal os is open, indicating this is an abortion in progress rather than a cervical ectopic.

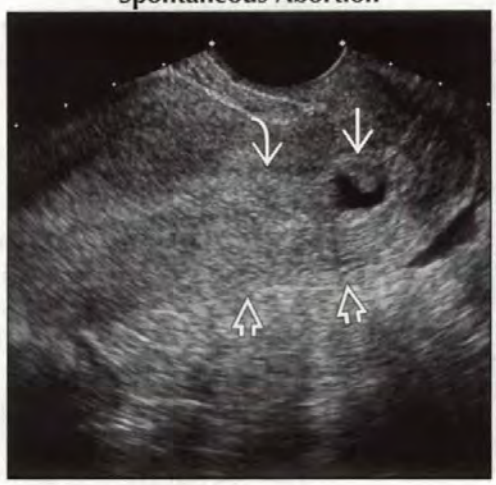
Spontaneous Abortion



Sagittal transvaginal ultrasound shows a flat gestational sac and decidual tissue in the cervix of a patient with an incomplete spontaneous abortion. D&C showed products of conception.

ABNORMAL SAC POSITION

Spontaneous Abortion

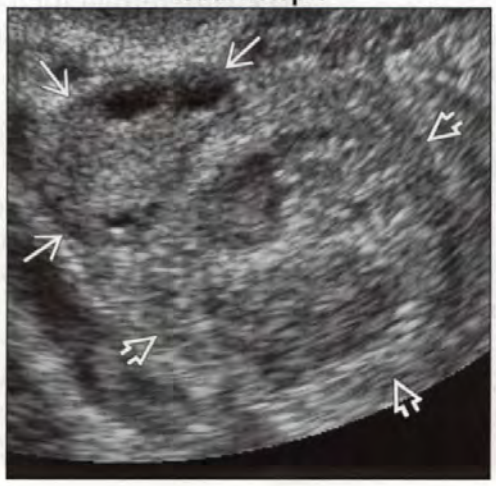


(Left) *Sagittal transvaginal ultrasound in the first trimester shows a passing embryo and sac in the cervix. The internal os is open causing the external contour to be relatively flat compared to a cervical ectopic which can create an "hourglass" contour.* ***(Right)*** *Axial transvaginal ultrasound shows an ectopic gestational sac within the ampullary portion of the tube. The tube is distended with blood and is well seen. The endometrial cavity is empty.*

Tubal Ectopic



Tubal Ectopic

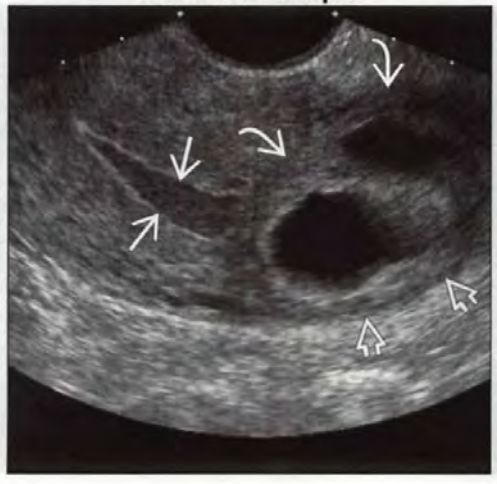


(Left) *Sagittal transvaginal ultrasound shows a normal right ovary and an adjacent heterogeneous ruptured tubal ectopic pregnancy.* ***(Right)*** *Transverse transabdominal ultrasound in a patient with known bicornuate uterus shows a first trimester pregnancy in the right horn, and decidual reaction in the left horn.*

Uterine Duplication



Interstitial Ectopic



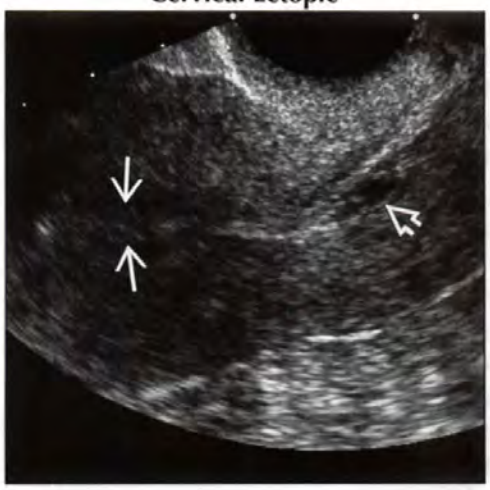
(Left) *Transvaginal ultrasound shows the endometrial cavity distended with blood products and an eccentric gestational sac with minimal myometrial coverage. At surgery, an unruptured interstitial ectopic was resected.* ***(Right)*** *Sagittal transabdominal ultrasound shows an empty uterus and a cervical gestational sac.*

Cervical Ectopic

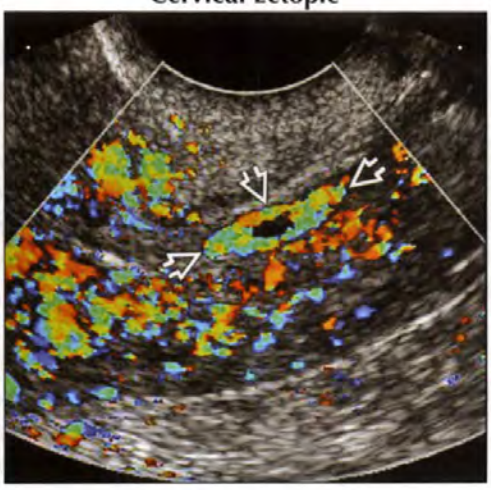


ABNORMAL SAC POSITION

Cervical Ectopic



Cervical Ectopic



(Left) *Sagittal transvaginal ultrasound shows an empty uterus with a thin endometrium and an eccentric sac in the cervix.* ***(Right)*** *Sagittal color Doppler ultrasound of the same patient shows that the ectopic sac is very vascular for its size. A sac in a spontaneous abortion would not exhibit this degree of vascularity.*

C-Section Scar Ectopic



Heterotopic Pregnancy



(Left) *Sagittal ultrasound shows an anechoic gestational sac at the site of a C-section scar. There is a surrounding decidual reaction within the myometrium. The endometrium is seen centrally within the uterus.* ***(Right)*** *Transverse transvaginal ultrasound of a heterotopic pregnancy shows an intrauterine pregnancy at 6 weeks 4 days. In addition, there is an associated left adnexal gestational sac and live embryo.*

Abdominal Ectopic



Abdominal Ectopic



(Left) *Sagittal T2WI MR shows an early second trimester fetus and placenta are located outside of the uterus. There is a decidual reaction within the uterus.* ***(Right)*** *Sagittal transvaginal ultrasound shows an empty uterus and a sac with fetal parts in the posterior cul-de-sac. This was confirmed with MR.*

INCREASED NUCHAL TRANSLUCENCY

DIFFERENTIAL DIAGNOSIS

Common

- Chromosome Abnormality
  - Trisomy 21
  - Trisomy 18
  - Trisomy 13
  - Turner Syndrome (XO)
- Congenital Heart Defects
  - Ventricular Septal Defect
  - Hypoplastic Left Heart
  - Atrioventricular Septal Defect
- Cystic Hygroma
  - Hydrops
- Normal Amnion (Mimic)

Less Common

- Nuchal Cord (Mimic)
- Fetal Demise

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Nuchal translucency (NT) measurement recommended in all 11-14 wk pregnancies
- Larger NT associated with worst prognosis
- Look for other first trimester aneuploidy markers at time of NT assessment
  - Absent nasal bone (NB)
  - Ductus venosus (DV) assessment
    - Normal DV with antegrade triphasic flow
    - Abnormal DV with retrograde "A" portion of wave
- Look for first trimester anomalies
  - Transvaginal ultrasound helpful
- Correlate NT with first trimester maternal serum results
  - Free β-human chorionic gonadotropin (β-hCG)
  - Pregnancy-associated plasma protein A (PAPP-A)
  - T21 pattern is ↑ β-hCG, ↓ PAPP-A
- Offer chorionic villus sampling (CVS)

Helpful Clues for Common Diagnoses

- **Trisomy 21**
  - Absent NB
    - Absent NB with a crown rump length (CRL) 45-64 mm = 17x ↑ T21 risk
    - Absent NB with a CRL 65-84 mm = 44-48x ↑ T21 risk
    - As NT increases, incidence of absent NB also ↑
  - Abnormal DV waveform
    - Retrograde "A" wave
  - Look for cardiac defect with transvaginal ultrasound
    - Atrioventricular septal defect
  - ↑ NT may evolve into nuchal skin fold thickening in 2nd trimester
- **Trisomy 18**
  - T18: Second most common chromosome abnormality
  - Some major anomalies are detectable in first trimester
    - Omphalocele
    - Cardiac defects
    - Musculoskeletal anomalies
- **Trisomy 13**
  - T13: Third most common chromosome abnormality
  - Some major anomalies are detectable in first trimester
    - Holoprosencephaly
    - Cardiac defects
    - Omphalocele (omphaloceles containing bowel have higher incidence of chromosomal abnormalities)
- **Turner Syndrome (XO)**
  - Largest NTs are seen with TS
  - Cystic hygroma
    - ↑ NT with septations
  - Hydrops
    - ↑ NT + fluid in any one other compartment
    - Pleural effusion
    - Body wall edema
    - Ascites
  - Cardiac defects
    - Hypoplastic left heart
- **Congenital Heart Defects**
  - ↑ NT seen with isolated congenital heart defects
    - Normal CVS
    - Normal maternal serum testing
  - Ductus venosus assessment helpful, however not always abnormal
  - Early echocardiography with transvaginal scanning
    - Color Doppler important for small septal defects
  - Second trimester echocardiography
    - If first trimester echo is inconclusive or normal

INCREASED NUCHAL TRANSLUCENCY

- **Cystic Hygroma**
  - Larger septated mass
  - Increased incidence in Turner syndrome but may occur in chromosomally normal fetuses
  - May develop hydrops
    - Incurs very poor prognosis
- **Normal Amnion (Mimic)**
  - Must see both amnion and skin to measure NT
    - Do not confuse amnion for nuchal skin
  - Fetus lies on amnion
    - Obscures the nuchal skin line
    - Wait for fetus to move away from amnion
  - Amnion and chorion normally fuse by 14 wks

Helpful Clues for Less Common Diagnoses

- **Nuchal Cord (Mimic)**
  - Umbilical cord behind neck in first trimester
    - Finding is not clinically significant
  - Measuring cord + neck will falsely increases NT
  - Cord can compress skin with focal increased NT
  - Consider bringing patient back on another day
- **Fetal Demise**
  - Obvious diagnosis during real time evaluation

Other Essential Information

- NT measurement technique
  - Timing
    - 11-14 wks menstrual age
    - CRL of 45-84 mm
  - Fetal position
    - Midsagittal plane
    - Head in neutral position
    - Beam perpendicular to skin
  - Image appropriately magnified
    - Head, neck, upper chest occupies ≥ 75% of image
  - Calipers must be placed correctly
    - Use "+" not "x" calipers
    - Crosshatch borders fluid
    - True measurement of anechoic fluid only
  - Sonographer accreditation recommended
    - Didactic course
    - Submit images for review
    - Audit every 6 months
- Maternal serum testing + NT
  - Increases sensitivity for detection of aneuploidy
    - > 90% detection rates reported
  - Sequential screening most commonly offered
    - Nuchal translucency
    - First trimester blood test
    - Second trimester blood test
    - 18-20 week anomaly screen

Trisomy 21



Sagittal ultrasound shows increased NT (calipers) and absent nasal bone ➡. The nuchal skin is seen separate from the amnion ➡. The calipers are placed to measure only fluid.

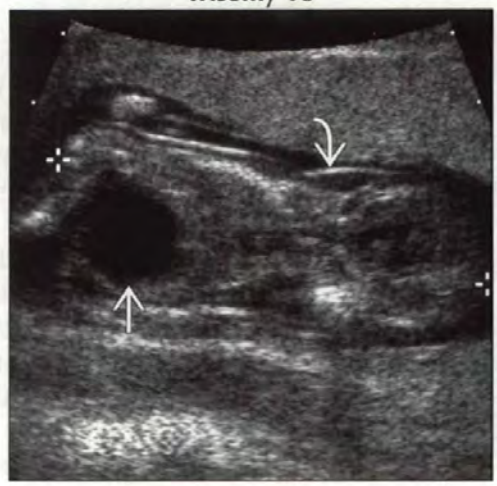
Trisomy 21



Axial ultrasound shows second trimester follow-up in the same fetus. The nuchal skin fold is now thick ➡. Increased NT often becomes increased nuchal fold thickening, both markers for T21.

INCREASED NUCHAL TRANSLUCENCY

Trisomy 18



Trisomy 18



(Left) *Sagittal ultrasound shows increased NT and a markedly enlarged bladder. **(Right)** Coronal transvaginal ultrasound shows a club foot. Many major anomalies are detectable in the first trimester, especially when transvaginal ultrasound is used to assess fetal anatomy.*

Trisomy 13



Trisomy 13

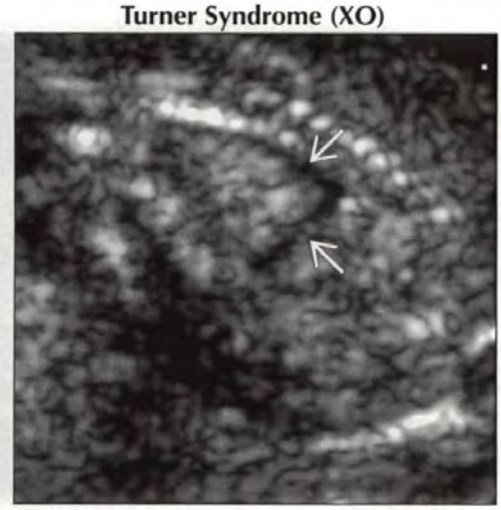


(Left) *Sagittal ultrasound shows a markedly increased nuchal translucency. Further evaluation of the fetal anatomy was performed by transvaginal ultrasound. **(Right)** Axial transvaginal ultrasound shows first trimester alobar holoprosencephaly. The thalami are fused and there is a single monoventricle. A mantle of brain crosses the midline and there is no falx.*

Turner Syndrome (XO)



Turner Syndrome (XO)



(Left) *Axial ultrasound shows a cystic hygroma with a single thin septation. **(Right)** Coronal ultrasound shows a pleural effusion. The largest NTs are associated with hydrops and Turner syndrome.*

INCREASED NUCHAL TRANSLUCENCY

Ventricular Septal Defect

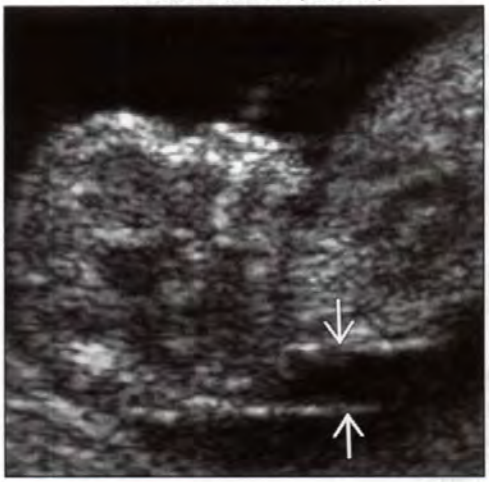


Ventricular Septal Defect

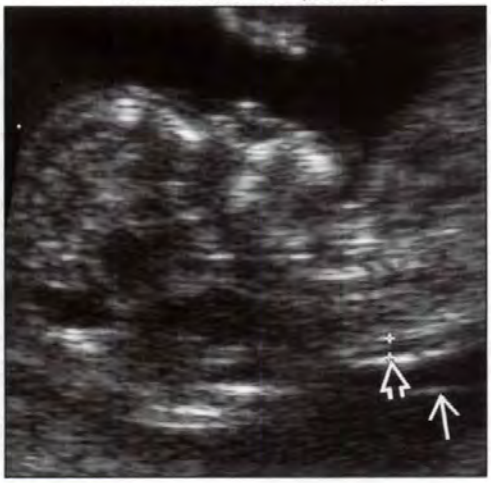


(Left) *Sagittal ultrasound shows increased nuchal translucency. The patient's chorionic villus sampling results were normal so she came back at 14 weeks for an early fetal cardiac study.* ***(Right)*** *Axial color Doppler ultrasound performed with transvaginal scanning shows a ventricular septal defect. The left ventricle and right ventricle are otherwise normal.*

Normal Amnion (Mimic)



Normal Amnion (Mimic)



(Left) *Sagittal ultrasound shows increased fluid behind the neck; however, a separate amnion and nuchal skin line are not seen on this image.* ***(Right)*** *Sagittal ultrasound shows better magnification in the same case. The NT is normal. The nuchal skin line is seen separate from the amnion and therefore the amnion no longer mimics the skin line.*

Nuchal Cord (Mimic)



Nuchal Cord (Mimic)



(Left) *Sagittal ultrasound shows increased NT (calipers) and a nuchal cord. The cord can cause spurious increased NT measurement because of mass effect on the skin and should not be included in the measurement.* ***(Right)*** *Sagittal pulsed Doppler ultrasound confirms the presence of the umbilical cord behind the neck. The maternal serum screen results and the baby were normal.*

ABNORMAL FIRST TRIMESTER FETUS

DIFFERENTIAL DIAGNOSIS

Common
- Increased Nuchal Translucency
- Cystic Hygroma
- Central Nervous System Anomalies, Severe
- Congenital Heart Defects

Less Common
- Absent Nasal Bone
- Gastroschisis
- Omphalocele
- Conjoined Twins
- Twin Reversed Arterial Perfusion

Rare but Important
- Autosomal Recessive Syndromes

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Must know normal developmental anatomy to avoid erroneous diagnosis of anomaly
  - Brain: Infratentorial
    - Rhombencephalon = precursor of cerebellum and brain stem
    - Appears hypoechoic
    - Not to be mistaken for posterior fossa cyst
  - Brain: Supratentorial
    - Presence of complete falx excludes alobar/semilobar holoprosencephaly
    - Look for "butterfly" sign of choroids to exclude alobar/semilobar holoprosencephaly
  - Skull vault
    - Ossification visible by ~ 12 weeks
  - Abdomen
    - Physiologic herniation of bowel is a normal embryological process
    - Bowel leaves peritoneum → base of umbilical cord → rotates 270° then re-enters abdominal cavity
    - Re-entry complete by 11.2 weeks gestational age
    - Never normal to see liver in base of cord
  - Limbs
    - Limb buds develop by 9 weeks
    - Femur can be measured by 13 weeks, visible earlier
    - Hands and feet fully formed by 13 weeks
  - Amniotic fluid
    - Produced by membranes in first trimester
    - Renal function does not account for majority of fluid volume until 16-17 weeks
    - Cannot exclude renal agenesis, autosomal recessive polycystic kidney disease on basis of normal fluid in first trimester

Helpful Clues for Common Diagnoses
- **Increased Nuchal Translucency**
  - Check ductus venosus (DV) waveform
  - Abnormal DV waveform = ↑ risk of adverse outcome even if chromosomes normal
  - Trisomy 21
    - Look for associated absent nasal bone, atrioventricular septal defect
    - Cystic hygroma/skin edema in Down syndrome is truncal
  - Trisomy 18
    - Look for associated omphalocele, complex congenital heart disease
  - Trisomy 13
    - Look for associated alobar holoprosencephaly, cyclopia, proboscis
  - Turner syndrome
    - Fetuses often hydropic
    - Look for "domed" extremity edema, Down syndrome edema more truncal
- **Cystic Hygroma**
  - Look for internal septations on axial images
  - Look for other stigmata of Down/Turner syndrome
- **Central Nervous System Anomalies, Severe**
  - Exencephaly
    - No skull vault echo
    - Brain seen "too well" initially with eventual destruction
    - Look for amniotic bands as etiology
  - Anencephaly
    - Look for "frog eye" appearance
    - No skull or brain above orbits
  - Occipital encephalocele
    - Confirm defect from different scan planes to avoid confusion with cystic hygroma
  - Alobar holoprosencephaly
    - Butterfly sign of choroid will be absent
    - Look for monoventricle/fused thalami
- **Congenital Heart Defects**
  - Use color Doppler as well as high resolution grayscale imaging

- All fetuses with increased NT should have formal 2nd trimester echocardiography

Helpful Clues for Less Common Diagnoses

- **Absent Nasal Bone**
  - Midsagittal plane
  - Normal appearance is 2 bright echoes, one from skin, shorter, brighter echo from bone
- **Gastroschisis**
  - Bowel loops free in amniotic fluid, no surrounding membrane
  - Cord inserted on abdominal wall, defect usually to the right
- **Omphalocele**
  - Membrane bound defect
  - Cord inserted at apex of defect
  - **Never** normal to see liver involved in physiologic bowel herniation into base of cord
- **Conjoined Twins**
  - Monochorionic monoamniotic gestation
  - Fixed relationship of embryos/fetuses with contiguous skin covering
- **Twin Reversed Arterial Perfusion**
  - TRAP: One normal "pump" twin
  - One anomalous twin
    - Diffuse truncal edema
    - Often subcutaneous cysts in edematous tissues
    - Absent or rudimentary cranial vault
  - Always check direction of flow in umbilical artery of an anomalous twin
    - Will be toward the anomalous fetus in TRAP

Helpful Clues for Rare Diagnoses

- **Autosomal Recessive Syndromes**
  - 25% recurrence risk, early diagnosis allows intervention for poor outcome conditions
  - Meckel Gruber Syndrome
    - Occipital encephalocele, renal cystic dysplasia, polydactyly
  - Achondrogenesis 1A,1B
    - Severe micromelia, poor spine ossification, hydrops

Other Essential Information

- NT measurement technique
  - Midsagittal scan plane
  - Neutral head position
  - Use (+) not (x) cursors
  - Show amnion separate from nuchal skin
- Transvaginal ultrasound mandatory for adequate resolution of anomalies
- Normal first trimester scan does not exclude all anomalies
  - Some entities change progressively over time
    - Coarctation/aortic stenosis may not have significant hemodynamic effects until third trimester
    - Aqueductal stenosis often presents as hydrocephalus in 3rd trimester

Increased Nuchal Translucency



Sagittal ultrasound shows typical increased nuchal translucency (calipers) of 3 mm in a fetus with trisomy 21. In the second trimester nuchal fold skin thickening and brachycephaly developed.

Increased Nuchal Translucency



Sagittal ultrasound at 13 weeks shows increased nuchal translucency ➡ and pleural effusions (not shown). Subsequently a cystic hygroma developed and genetic amniocentesis revealed Turner syndrome.

ABNORMAL FIRST TRIMESTER FETUS

Increased Nuchal Translucency



(Left) *Pulsed Doppler ultrasound at 9 weeks shows abnormal ductus venosus flow with reversal during the A wave. Follow-up at 13 weeks showed ↑ nuchal translucency. At birth infant had a non-lethal, short-limbed, skeletal dysplasia.* ***(Right)*** *Axial transvaginal ultrasound in a fetus with cystic hygroma shows only right globe & suggests hypoplastic left midface. The brain also looked abnormal. Pregnancy termination revealed both trisomy 21 & trisomy 9.*

Cystic Hygroma



Cystic Hygroma

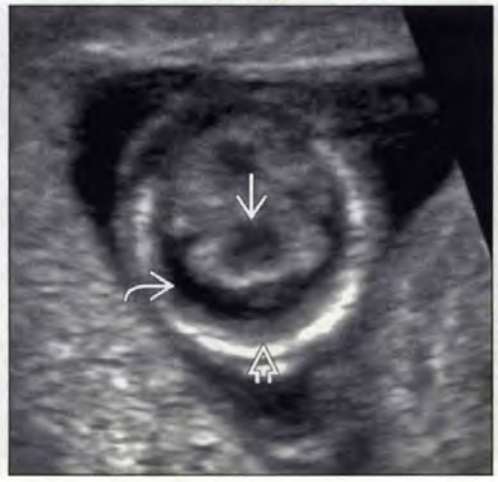


(Left) *Transvaginal ultrasound shows a large septated cystic hygroma.* ***(Right)*** *Transabdominal ultrasound two weeks later at 15 weeks shows skin edema and bilateral pleural effusions indicating hydrops. Fetal demise occurred but the couple declined autopsy. Development of hydrops in a fetus with cystic hygroma confers a dismal prognosis.*

Cystic Hygroma

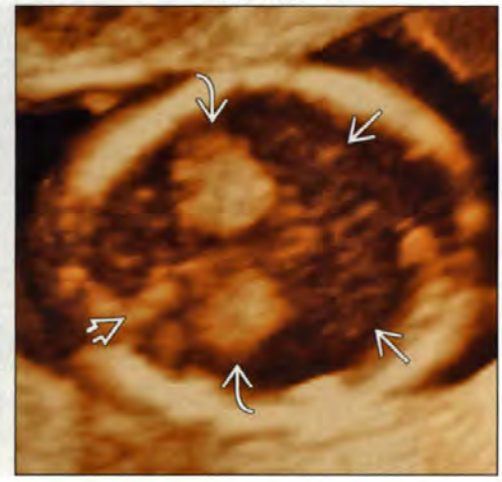


Central Nervous System Anomalies, Severe



(Left) *Axial ultrasound of the fetal head shows fused thalami, monoventricle, and thin brain mantle, consistent with alobar holoprosencephaly. Chorionic villus sampling revealed trisomy 13.* ***(Right)*** *Axial 3D ultrasound shows two hemispheres posteriorly with a falx and choroid echoes. Anteriorly the falx is absent, and there is a monoventricle indicating semilobar holoprosencephaly in a 12 week fetus.*

Central Nervous System Anomalies, Severe



ABNORMAL FIRST TRIMESTER FETUS

Central Nervous System Anomalies, Severe



Central Nervous System Anomalies, Severe



(Left) *Coronal transvaginal ultrasound shows progression of findings of exencephaly between 9 and 12.5 wks. At 9 wks there is a definite head but the shape is somewhat irregular.* ***(Right)*** *Sagittal transabdominal ultrasound in the same fetus as the prior image at 12.5 wks shows exencephaly with absent calvarium and exposed neural tissue.*

Central Nervous System Anomalies, Severe

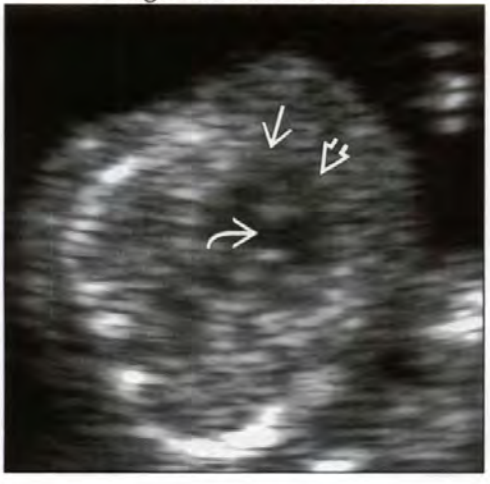


Central Nervous System Anomalies, Severe



(Left) *Coronal transvaginal ultrasound of another exencephaly shows amorphous tissue without a surrounding ossified skull vault. Bony orbits are visible in the face.* ***(Right)*** *Coronal ultrasound shows complete lack of cerebral tissue but an intact falx. Brainstem herniation into the supratentorial space can be confused with thalamic fusion in holoprosencephaly, however the presence of a falx verifies the diagnosis of hydranencephaly.*

Congenital Heart Defects



Congenital Heart Defects



(Left) *Axial transvaginal ultrasound in a fetus with increased nuchal translucency and normal chromosomes shows a ventricular septal defect between the left and the right ventricles.* ***(Right)*** *Axial color Doppler ultrasound in the same fetus as the previous image confirms the diagnosis of a ventricular septal defect in the late first trimester.*

ABNORMAL FIRST TRIMESTER FETUS

Congenital Heart Defects





(Left) *Axial ultrasound at 13 weeks shows a single ventricle ➡. The spine is indicated by the ➡. The fetus also had increased nuchal translucency.* ***(Right)*** *Axial transvaginal ultrasound in another fetus with increased NT but normal chromosomes shows a hypoplastic left heart. The left ventricle ➡ is much smaller than the right ➡. It does not extend to the apex of the heart and did not have blood flow on color Doppler evaluation.*

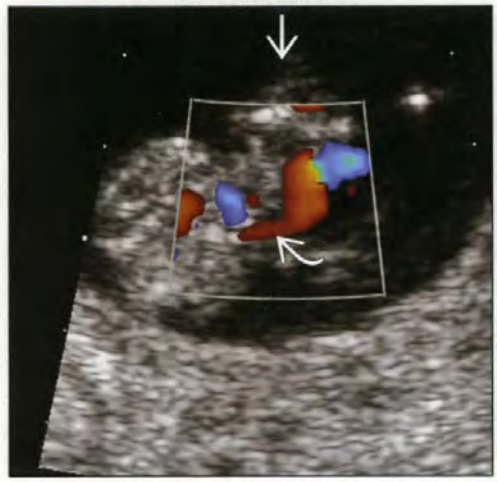
Absent Nasal Bone





(Left) *Sagittal ultrasound fails to show a nasal bone ➡. Nuchal translucency measured > 95th percentile and ductus venosus flow was abnormal. Chorionic villus sampling revealed trisomy 21.* ***(Right)*** *Sagittal ultrasound shows no identifiable nasal bone ➡. The nuchal translucency measurement was normal. The patient was 40 years old and opted for amniocentesis. The results were normal male, and the neonate was normal.*

Gastroschisis



Omphalocele



(Left) *Axial color Doppler ultrasound shows bowel loops ➡ exterior to the abdomen adjacent to a normal cord insertion site ➡, typical of gastroschisis.* ***(Right)*** *Sagittal transabdominal ultrasound in a fetus with a cystic hygroma ➡ shows solid tissue ➡ in an abdominal wall defect consistent with an omphalocele. The patient elected to terminate the pregnancy without chromosome analysis.*

Conjoined Twins



Conjoined Twins

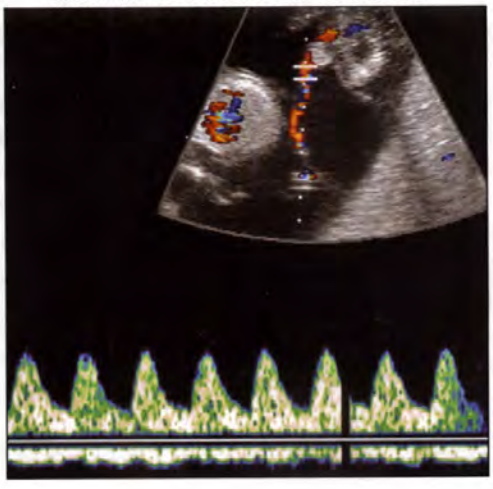


(Left) *Transvaginal ultrasound shows conjoined twins in the first trimester. There is a single chorionic sac ➡ with two embryos (calipers) seen in close apposition. There is only one yolk sac ➡ and no dividing membrane.* ***(Right)*** *Ultrasound shows cranial fusion in another case. The profile of one twin ➡ is seen fused to an axial plane ➡ of the other twin's head. Common vessels were seen between the brains as well as a contiguous bone and skin covering.*

Twin Reversed Arterial Perfusion

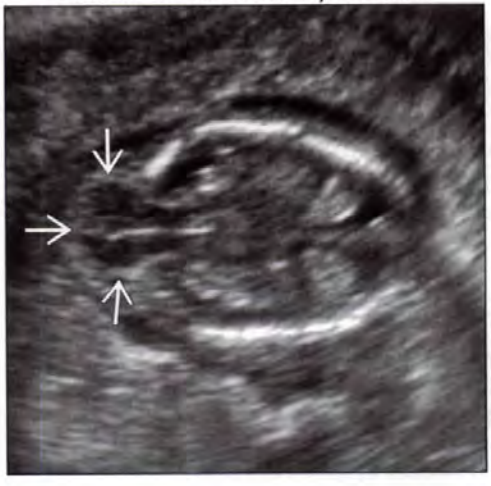


Twin Reversed Arterial Perfusion



(Left) *Sagittal transvaginal ultrasound shows an abnormal, grossly edematous twin fetus with absent cranial structures ➡. Umbilical artery flow toward the placenta from the normal twin ➡ was seen.* ***(Right)*** *Pulsed Doppler ultrasound in the same case as the previous image shows umbilical artery flow is towards the abnormal fetus proving the diagnosis of twin reversed arterial perfusion.*

Autosomal Recessive Syndromes



Autosomal Recessive Syndromes



(Left) *Axial transvaginal ultrasound showed a large occipital encephalocele with herniation of the cerebellum ➡.* ***(Right)*** *Coronal transvaginal ultrasound in the same case as previous image shows bilateral, enlarged, cystic kidneys (calipers and ➡). These are typical features of Meckel Gruber syndrome, an autosomal recessive disorder with a 25% recurrence risk.*

FIRST TRIMESTER MEMBRANES

DIFFERENTIAL DIAGNOSIS

Common
- Normal Chorioamniotic Development
- Dichorionic Diamniotic Twins
- Monochorionic Diamniotic Twins
- Monochorionic Monoamniotic Twins

Less Common
- Triplets and Beyond
- Septate Uterus (Mimic)
- Resolving Perigestational Hemorrhage
- Twin Demise
- Increased Nuchal Translucency (Mimic)

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Must know normal early pregnancy development
- Chorion
  - Thick, echogenic ring visible as double decidual sac sign (DDSS)
    - Inner sac is gestational sac (decidua basalis, decidua capsularis)
    - Outer sac is endometrium (decidual parietalis)
  - Chorionic frondosum
    - Normal focal thickening in chorion at 8-13 weeks; first trimester placenta
- Amnion
  - Thin delicate membrane inside echogenic chorionic ring
  - Yolk sac (YS) develops later but thick echogenic wall → visible earlier
  - In multiple gestations infer amnionicity from number of yolk sacs
  - "Double bleb" sign: Yolk sac + amnion on either side of embryonic disc
  - Amnion expands rapidly to surround embryo
  - Embryo inside amnion, YS outside
  - Amnion abuts chorion by 14-16 weeks, membranes no longer visible as separate layers
- Space between amnion and chorion = extra embryonic coelom space (chorionic cavity)
  - Fluid echogenic due to protein content
    - Do not confuse with perigestational hemorrhage (blood between chorion and myometrium)
  - Fluid inside amnion is normally anechoic

Helpful Clues for Common Diagnoses
- **Dichorionic Diamniotic Twins**
  - Two complete echogenic rings created by two chorions
    - Thick echogenic membrane between two sacs
    - Creates twin peak membrane in second trimester
  - Two amnions
  - Two YS
  - Two embryos
- **Monochorionic Diamniotic Twins**
  - One echogenic ring = one chorion
  - Two amnions
    - Thin membrane between sacs
    - No twin peak sign
  - Two YS
  - Two embryos
- **Monochorionic Monoamniotic Twins**
  - One echogenic ring = one chorion
  - One amnion
  - One YS
  - Two embryos
    - Ensure embryos are separate to avoid missing conjoined twins
- If apparent monochorionic twins on first trimester transabdominal scan always do transvaginal scan to look for membrane

Helpful Clues for Less Common Diagnoses
- **Triplets and Beyond**
  - Count echogenic chorionic rings
  - Count amnions or YS if very early
  - Count embryos once cardiac activity visible
  - Determine chorionicity/amnionicity
    - Same impact on prognosis in higher order multiples as in twins
- **Septate Uterus (Mimic)**
  - Patient may be aware or have suggestive history
    - Recurrent abortion, preterm labor
  - Gestational sac may appear eccentric in location
  - Apparent "membrane" will be thick
    - Echogenicity similar to myometrium
  - 3D scans useful
    - Evaluate fundal contour
    - Look for continuity of septum with fundal myometrium
    - Assess orientation/location of sac to exclude cornual ectopic

- **Resolving Perigestational Hemorrhage**
  - Often history of spotting/vaginal bleeding
  - Hypoechoic material deep to echogenic chorion
    - Shape, echogenicity and volume change over time
  - Crescentic shape
    - Membranes are spherical
- **Twin Demise**
  - One gestational sac stops growing
  - Cardiac activity ceases
  - Embryo may resorb completely ("disappearing twin")
- **Increased Nuchal Translucency (Mimic)**
  - Key element in measurement is identification of amnion distinct from nuchal translucency (NT)
    - Watch for fetus to move away from amnion
    - Midsagittal plane

Other Essential Information

- Best imaging tool
  - Use transvaginal ultrasound for maximum resolution
- Prognosis in multiple gestation depends on chorionicity
  - First trimester is best time to determine chorionicity
  - Monochorionic twin complications
    - Twin-twin transfusion syndrome
    - Twin reversed arterial perfusion
    - Unequal placental sharing
    - In case of twin demise significant morbidity to surviving twin
  - Monoamniotic twins at additional risk for cord accidents
  - Early identification allows appropriate monitoring with early intervention for complications

Alternative Differential Approaches

- Abnormal First Trimester Membranes
  - Empty amnion sign
    - Reliable indicator of failed first trimester pregnancy
    - Mean sac diameter > 18 mm without visible embryo inside amnion = "empty" amnion
    - Yolk sac may be seen between amnion and chorion
  - Chorionic bump
    - Abnormal focal protuberance from chorionic surface
    - Associated with 50% loss rate in assisted reproduction population
    - Do not mistake "chorionic bump" for embryo: Embryo should be inside amnion
  - Chorioamniotic separation
    - Persistent unfused amnion and chorion > 14-16 wks (not truly a first trimester diagnosis)
    - Associated with aneuploidy/fetal anomalies

Normal Chorioamniotic Development



Axial transvaginal ultrasound shows shows the double decidual sac sign. The inner ring ➡ is the decidua capsularis and the outer ring ➡ is the decidua parietalis. The yolk sac ➡ is a specific sign of IUP.

Normal Chorioamniotic Development



Transvaginal ultrasound shows the "double bleb" appearance created by adjacent amnion ➡ and yolk sac ➡ within the chorionic sac ➡.

FIRST TRIMESTER MEMBRANES

Normal Chorioamniotic Development



(Left) *Transvaginal ultrasound shows the embryo (calipers) within the enlarging amnion. The yolk sac is outside the amnion and will eventually be obliterated as the amnion abuts the chorion.* ***(Right)*** *Transvaginal ultrasound shows echogenic fluid in the chorionic cavity around the anechoic fluid within the amniotic sac (chorion indicated by).* *Compare to perigestational hemorrhage which occurs deep to the chorion.*

Normal Chorioamniotic Development



Dichorionic Diamniotic Twins



(Left) *Transvaginal ultrasound shows two chorionic sacs forming a thick inter-twin membrane . One yolk sac is seen; the second was present on other scan planes.* ***(Right)*** *Transvaginal ultrasound shows two amnions within a single chorion . The amnions will come together to form a thin inter-twin membrane with no intervening chorion. Note embryos are inside the amnions, while yolk sacs are outside (one yolk sac is partially visible).*

Monochorionic Diamniotic Twins



Monochorionic Monoamniotic Twins



(Left) *Transvaginal ultrasound show two fetuses surrounded by a single amnion . Only one yolk sac is seen. This indicates monoamniotic twinning.* ***(Right)*** *Sagittal oblique transvaginal ultrasound shows two chorionic rings with three yolk sacs . Follow-up images confirmed the presence of two amnions in the anterior sac. Therefore this is a dichorionic, triamniotic triplet pregnancy.*

Triplets and Beyond



FIRST TRIMESTER MEMBRANES

Septate Uterus (Mimic)



Septate Uterus (Mimic)



(Left) *Transvaginal ultrasound at 6 weeks post LMP shows a gestational sac ➡ to the left side of the uterus in a patient with a known septum ➡. **(Right)** Transvaginal ultrasound at 11 weeks in the same case shows the fetus ➡ to the left of the septum ➡ with decidualized endometrium ➡ to the right of the septum. Note straight fundal contour.*

Resolving Perigestational Hemorrhage



Resolving Perigestational Hemorrhage



(Left) *Axial transvaginal ultrasound shows two intrauterine fluid collections (➡, ➡) that may be confused for two gestational sacs. The true gestational sac ➡ is thick-walled and contained a yolk sac on other views. The apparent membrane ➡ is the wall of the gestational sac. **(Right)** Sagittal transvaginal ultrasound in the same patient as the previous image shows that the hemorrhage ➡ is flattened or oblong in shape and does not have a thick wall.*

Twin Demise



Increased Nuchal Translucency (Mimic)



(Left) *Transvaginal ultrasound shows two gestational sacs ➡ in a dichorionic twin pregnancy. The smaller sac failed to grow from earlier scans, and there was no embryo within it, consistent with twin demise. **(Right)** Sagittal ultrasound with the fetus away from the amnion ➡ clearly shows increased NT ➡. Chorionic villus sampling results were normal but at 14 weeks EV sonography showed a ventricular septal defect.*

FIRST TRIMESTER PELVIC PAIN

DIFFERENTIAL DIAGNOSIS

Common
- Hemorrhagic Cyst
- Corpus Luteum Cyst
- Ectopic Pregnancy

Less Common
- Adnexal Torsion
- Appendicitis

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- First rule out ectopic pregnancy (EP)
  - Is there an intrauterine pregnancy (IUP)?
    - Presence of IUP best evidence against EP
    - Heterotopic ectopics are rare
  - Is the human chorionic gonadotropin (hCG) level high enough to see an IUP?
    - Should see IUP if hCG levels are > 2000 mIU/mL IRP (international reference preparation)
  - Lack of IUP at low hCG levels does not rule out EP
    - EP are not normal pregnancies and may have lower hCG levels
  - Look for blood in cul-de-sac
  - Look for an adnexal mass
    - EP itself or a hematoma
    - Look for EP on same side as the corpus luteum
- Evaluate ovary on side of pain
  - Is the ovary enlarged?
    - Consider torsion
  - Is there a mass or mass-like lesion?
    - Characterize mass wall
    - Evaluate internal morphology
  - Doppler ultrasound
    - Variable findings with torsion
    - Compare with other ovary
    - Peripheral flow in EP ("ring of fire") and ovarian cysts
    - Internal flow in neoplasm
- Rule out other causes for pain
  - Gastrointestinal
    - Compression ultrasound for appendicitis
    - Inflamed bowel
  - Genitourinary
    - Hydronephrosis
    - Ureteral calculi
    - Pyelonephritis

Helpful Clues for Common Diagnoses
- **Hemorrhagic Cyst**
  - Usually from hemorrhage into a corpus luteum
  - Acute and subacute findings
    - Reticular lace-like pattern
    - Fibrin strands not true septations
    - Retracting blood clot
  - Findings on follow-up
    - Fluid-fluid level
    - Anechoic cyst
  - Mass should resolve completely with time
    - 6 week follow-up recommended
  - Role of Doppler
    - No internal flow
    - Rule out associated torsion
- **Corpus Luteum Cyst**
  - Abbreviated CL
  - Variable wall appearance
    - Thick, hyperechoic wall most common
    - Thin wall if large cyst
  - Variable internal echoes
    - Anechoic
    - Complex if hemorrhagic
    - Solid appearing if thick wall and no fluid
  - Doppler findings
    - Low resistive vascular flow in cyst wall
    - No internal flow
  - Rule out associated torsion
- **Ectopic Pregnancy**
  - Uterine findings
    - Thin or thick endometrial cavity
    - "Pseudogestational sac" from blood in endometrial cavity
    - Endometrial cysts can mimic early IUP
    - Rare heterotopic pregnancy (IUP + EP)
  - Variable adnexal findings with tubal EP
    - Tubal distention or hematoma
    - Tubal gestational sac separate from ovary
    - EP on same side as CL in 85%
  - Doppler findings
    - Low resistive, high velocity flow
    - "Ring of fire"
  - Peritoneal blood
    - Echogenic fluid in cul-de-sac
    - Look higher up for abdominal blood
  - Unusual EP locations
    - Interstitial/cornual
    - Cervical
    - Ovarian
    - Abdominal

Helpful Clues for Less Common Diagnoses

- **Adnexal Torsion**
  - Usually involves both ovary and tube
  - Ovary is almost always enlarged
    - Right > left (3:2)
    - Peripheral cysts from central edema
    - Fluid-debris levels in cysts seen with high resolution scanning
  - Doppler findings
    - Venous flow lost first
    - Variable arterial findings secondary to dual blood supply to ovaries
  - Associated adnexal mass common
    - Dermoid
    - Corpus luteum
    - Tubal cyst
    - Serous cystadenoma
  - Free fluid in cul-de-sac
    - Mostly anechoic
- **Appendicitis**
  - Appendix may be in an atypical position
    - Gravid uterus lifts appendix out of pelvis
    - Have patient point to focal area of tenderness
  - Graded compression ultrasound with high resolution linear probe
  - Blind-ending tubular structure
    - > 6 mm outer to outer wall diameter
    - Periappendiceal fluid
    - Echogenic fat surrounds inflamed appendix
    - Inflammatory mass if ruptured
    - Associated adjacent lymphadenopathy
  - Other imaging may be necessary
    - MR
    - CT

Other Essential Information

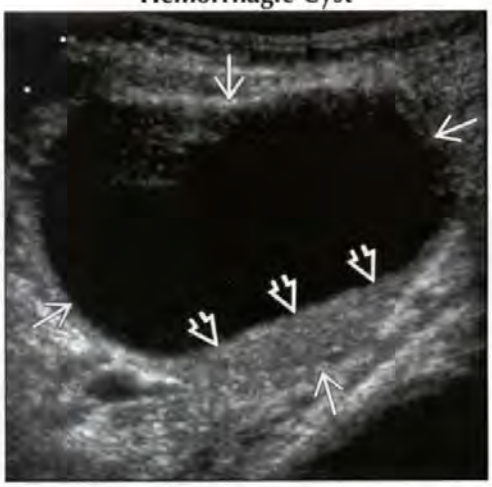
- Clinical differentiation between causes of acute pelvic pain is difficult and good imaging is essential
  - Appendicitis and torsion both have increased white count
- Rule out diagnoses sequentially
  - First rule out EP
  - Next rule out ovarian causes for pain
  - Finally rule out appendicitis
- Ovarian mass may be difficult to differentiate from adnexal mass near the ovary
  - Use pressure from transvaginal probe to separate ovary from mass
- Other imaging may be necessary
  - Try to avoid CT in first trimester
  - Must weigh maternal status with need for imaging
  - High morbidity to patient and fetus if missed or delayed diagnoses
  - Consider MR for appendicitis
- All complex ovarian masses must be followed to resolution
  - Ovarian neoplasm diagnosed during first trimester often removed surgically in second trimester

Hemorrhagic Cyst



Transvaginal ultrasound shows a large cyst with internal lace-like echoes from fibrin strands ➡, an appearance consistent with a retracting clot.

Hemorrhagic Cyst



Axial ultrasound of the same ovary two weeks later shows the cyst ➡ is almost completely anechoic, with the exception of a fluid-debris level ➡. Eventually, the cyst completely resolved.

FIRST TRIMESTER PELVIC PAIN

Corpus Luteum Cyst



(Left) Transverse ultrasound shows a large, thin-walled, anechoic cyst arising from the ovary. The 14 week gravid uterus is anterior to the cyst. (Right) Sagittal ultrasound shows a corpus luteum cyst within the right ovary. It has a more typical thick-walled appearance with minimal internal echoes.

Corpus Luteum Cyst



Ectopic Pregnancy



(Left) Sagittal transvaginal ultrasound shows an empty uterine cavity with a thick decidual reaction in a patient with an hCG level of 3000 mIU/mL. (Right) Coronal transvaginal ultrasound shows an ectopic gestational sac adjacent to an ovary, which contains a hypoechoic corpus luteum. This unruptured ectopic was successfully treated with methotrexate.

Ectopic Pregnancy

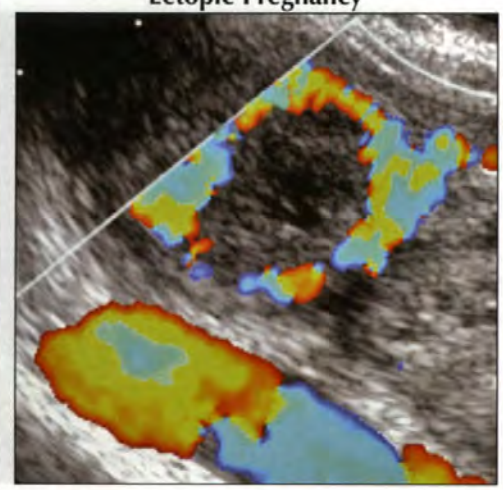


Ectopic Pregnancy



(Left) Coronal ultrasound shows a tubal gestational sac adjacent to the right ovary. (Right) Coronal color Doppler ultrasound in the same patient as previous image, shows the classic "ring of fire" appearance of ectopic pregnancy. The gestational sac wall contains vascular trophoblastic tissue and "lights up" with color Doppler.

Ectopic Pregnancy



Ectopic Pregnancy

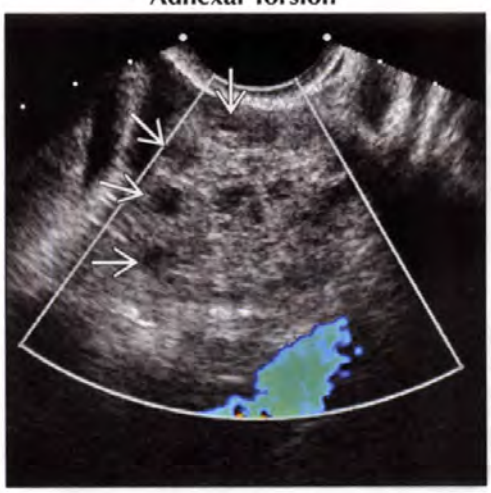


Ectopic Pregnancy



(Left) *Sagittal transvaginal ultrasound shows a "pseudo gestational sac"* ➡. *Endometrial fluid may mimic an intrauterine pregnancy.* ***(Right)*** *Coronal ultrasound shows a large complex adnexal mass* ➡ *and echogenic fluid* ➡ *in the same patient as the previous image. A ruptured ectopic pregnancy was found at surgery.*

Adnexal Torsion



Adnexal Torsion

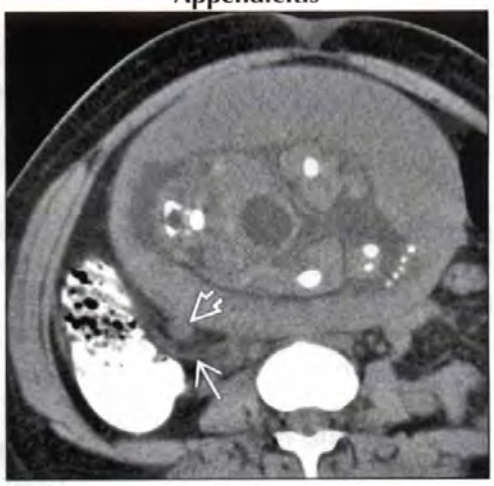


(Left) *Sagittal oblique color Doppler ultrasound shows an enlarged ovary with peripherally displaced follicles* ➡ *and no significant blood flow.* ***(Right)*** *Transverse ultrasound in the same case, shows a midline large unilocular cyst* ➡ *anterior to the cervix* ➡. *At surgery, adnexal torsion involving a paraovarian cyst was diagnosed.*

Appendicitis



Appendicitis



(Left) *Axial transabdominal ultrasound shows a non-compressible, distended appendix* ➡. *Note that the tubular structure is blind ending* ➡ *and surrounded by echogenic fat.* ***(Right)*** *Axial NECT in another patient shows a distended appendix* ➡ *and mesenteric inflammation* ➡. *The appendix is behind the gravid uterus and was not seen with ultrasound.*

ADNEXAL MASS IN FIRST TRIMESTER

DIFFERENTIAL DIAGNOSIS

Common
- Corpus Luteum Cyst
- Ovarian Teratoma
- Hyperstimulation Syndrome
- Theca Lutein Cysts
- Endometrioma
- Ovarian Neoplasm

Less Common
- Pedunculated Fibroid
- Paraovarian Cyst
- Hydrosalpinx

Rare but Important
- Heterotopic Pregnancy

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Determine if mass is ovarian
  - Use TV probe to push ovary away from uterus or mass
    - Concurrent manual compression on low abdomen/pelvis can be helpful
  - Distinguish paraovarian or adnexal mass from ovarian mass by direct visualization during manipulation
- Look for secondary findings
  - Echogenic free fluid (hemorrhage)

Helpful Clues for Common Diagnoses
- **Corpus Luteum Cyst**
  - Very commonly identified in first trimester
  - Often complicated by hemorrhage of varying degrees
    - Look for typical reticular echoes seen in hemorrhagic cysts
    - Occasionally hemorrhage will appear homogeneous and simulate solid mass
  - "Ring of fire" appearance with Doppler imaging
  - Most resolve by early second trimester
- **Ovarian Teratoma**
  - Most common ovarian neoplasm in reproductive age group
  - Echogenic dermoid "plug" often present
    - Represents keratin
  - Appearance varies depending on content of teratoma
    - Cystic/hypoechoic fat or fat fluid level
    - Echogenic teeth/bones
    - Linear echogenic hair
- **Hyperstimulation Syndrome**
  - Bilateral enlarged, cystic ovaries
  - Ascites ± pleural effusions
    - Due to increased vascular permeability
  - History of in vitro fertility treatments
    - Usually in women undergoing ovulation induction
  - Usually suspected due to clinical presentation
    - Abdominal pain
    - Nausea/vomiting
    - Oliguria
    - Electrolyte imbalances
    - Hypotension
- **Theca Lutein Cysts**
  - Most often seen in clinical setting of infertility treatments
  - Rarely present with singleton pregnancy
  - Enlarged ovaries
    - Multiple simple cysts
    - Usually bilateral
  - Look for abnormal fetus
    - Gestational trophoblastic disease
    - Triploid fetus
    - Fetal hydrops
- **Endometrioma**
  - Diffuse, homogeneous, low-level echoes
  - Unilocular cyst
    - Occasionally multilocular, may mimic malignancy
  - Through transmission present
  - Can have fluid-fluid level if multiple ages of blood present
  - Look for echogenic foci with comet-tail artifact in the wall
    - Collections of cholesterol
  - Most often ovarian in location
    - Can be seen in broad ligament, cul-de-sac, adjacent to bowel
- **Ovarian Neoplasm**
  - Varying appearances depending on tissue of origin
    - Solid mass: Sex-chord stromal tumors
    - Complex cystic: Epithelial ovarian tumors
  - Suspicious ovarian masses can be further characterized with MR

Helpful Clues for Less Common Diagnoses
- **Pedunculated Fibroid**
  - Usually hypoechoic to myometrium

- Heterogeneous echotexture may be present
  - Hemorrhage with retracted clot
  - Cystic degeneration
  - Calcifications
- Use Doppler to look for vascular connection to underlying myometrium
- 50% of fibroids grow in size in the first 20 weeks of pregnancy
- May be painful
  - Degenerating fibroids
  - Twisting of pedunculated fibroid on stalk

- **Paraovarian Cyst**
  - Located in broad ligament
  - Round or oval mass medial to ovary
  - Almost always unilocular and anechoic
    - Infrequently may be multilocular or have minimal debris
  - Cyst moves separately from ovary with use of vaginal probe
- **Hydrosalpinx**
  - Thin-walled tubular structure
  - Anechoic fluid within
    - If internal debris present, consider hematosalpinx or pyosalpinx if patient is very ill
  - Look for thin endosalpingeal folds
    - "Beads on a string" or "cog wheel" appearance
    - Aids in confirmation that the "mass" is actually hydrosalpinx
    - Transvaginal ultrasound mandatory for visualization

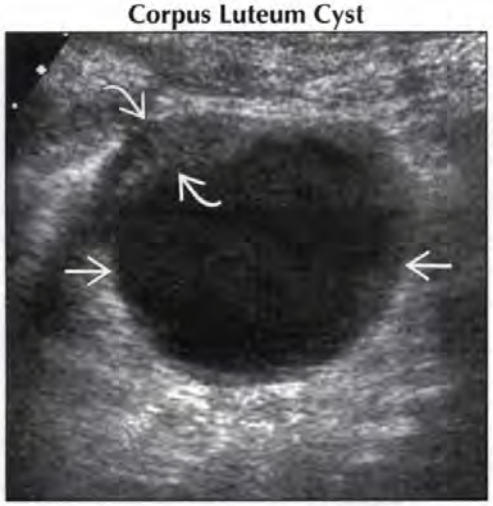
Helpful Clues for Rare Diagnoses

- **Heterotopic Pregnancy**
  - Suspect if adnexal mass with history of fertility treatments
    - 10-40% risk of ectopic pregnancy in fertility patients
    - 1:100-500 risk of heterotopic pregnancy in patients undergoing assisted reproductive technologies
  - Assess for intact gestational sac
    - ± Embryonic pole/heart beat
  - Beware of tubal or adnexal clot
    - Can be difficult to identify if isoechoic
    - Adjacent bowel loops should show peristalsis
  - Watch for secondary signs of ruptured ectopic
    - Echogenic free fluid
    - Fluid tracking into upper abdomen
    - Clinical peritoneal signs ± hypotension

Other Essential Information

- Radiographic suspicion of malignancy will guide clinical management of ovarian masses
  - Most benign-appearing masses will be observed through pregnancy
  - Monitor for signs of ovarian torsion if mass is large
  - Malignant-appearing masses should be managed surgically despite pregnancy
  - Surgery usually performed in the second trimester

Corpus Luteum Cyst



Transvaginal ultrasound at 13 weeks gestation shows a 9 cm anechoic left corpus luteum cyst. Note there is only a thin hypoechoic rim of visible ovarian tissue. This decreased to 3 cm at 17 weeks gestation.

Corpus Luteum Cyst



Color Doppler ultrasound at 5 weeks gestation shows a hemorrhagic corpus luteum cyst with a typical surrounding ring of vascularity.

ADNEXAL MASS IN FIRST TRIMESTER

Corpus Luteum Cyst



(Left) *Longitudinal transvaginal ultrasound shows a variant corpus luteum cyst, with fine reticular echoes ➔ indicating hemorrhage. It is surrounded by a thin echogenic rim ➔. The ovary is marked with calipers.* ***(Right)*** *Sagittal ultrasound shows an 8 cm ovarian mass with linear echoes (hair ➔) and an echogenic "plug" ➔. Dermoid cysts are the most common ovarian neoplasm discovered during pregnancy.*

Ovarian Teratoma



Hyperstimulation Syndrome

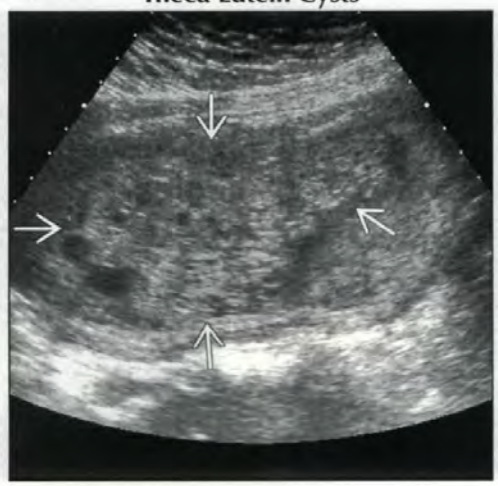


(Left) *Sagittal transvaginal ultrasound shows 2 gestational sacs in the uterus of an IVF patient with hyperstimulation syndrome. Anechoic free fluid ➔ is present in the pelvis.* ***(Right)*** *Axial ultrasound of the same IVF patient shows a hyperstimulated ovary measuring up to 19 cm (calipers). The patient also had severe shortness of breath due to large pleural effusions.*

Hyperstimulation Syndrome



Theca Lutein Cysts



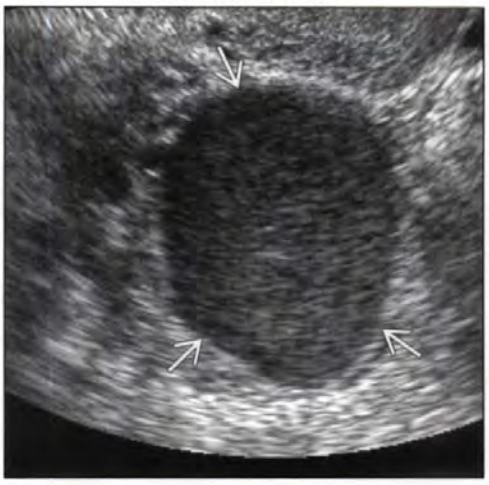
(Left) *Transverse transabdominal ultrasound shows a uterus filled with cystic material ➔ consistent with a complete hydatidiform mole. The patient presented with early hypertension at 15 weeks gestation by LMP.* ***(Right)*** *Longitudinal transvaginal ultrasound (same patient) shows enlarged ovaries (left ovary shown, calipers) measuring up to 5 x 8 cm. There are multiple associated theca lutein cysts, some of which are complicated by hemorrhage ➔.*

Theca Lutein Cysts

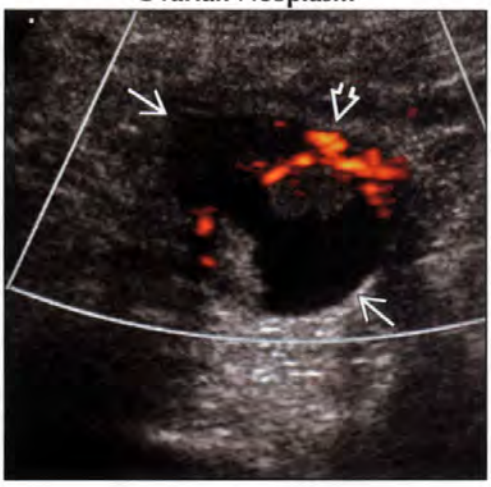


ADNEXAL MASS IN FIRST TRIMESTER

Endometrioma

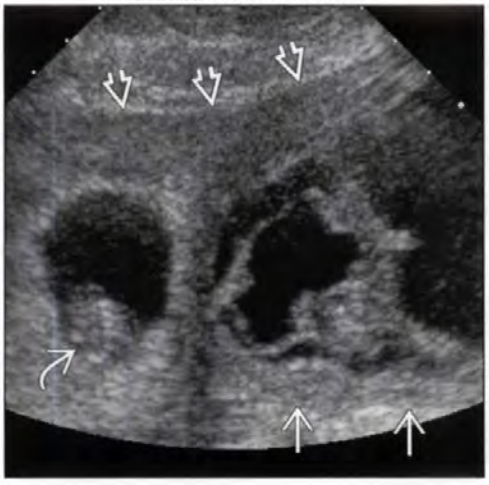


Ovarian Neoplasm

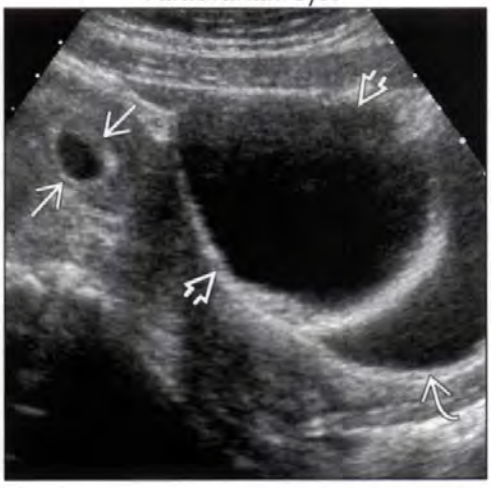


(Left) *Transvaginal ultrasound shows a hypoechoic ovarian endometrioma in a patient with history of endometriosis. Note the homogeneous, low-level internal echoes, which differ from the lace-like echoes of a hemorrhagic corpus luteum.* ***(Right)*** *Power Doppler ultrasound shows a solid and cystic 5 cm borderline ovarian tumor in a pregnant patient. Unlike a hemorrhagic corpus luteum, there is vascularity in the solid components.*

Pedunculated Fibroid



Paraovarian Cyst

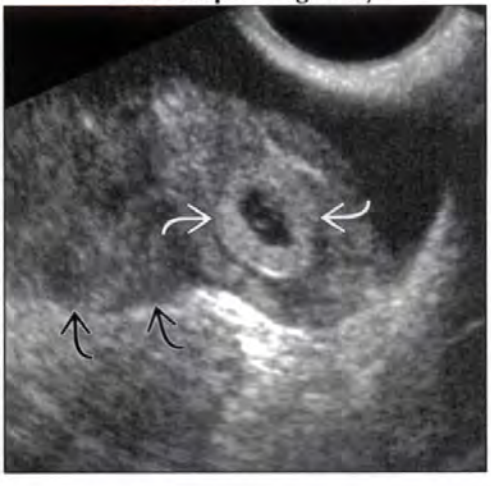


(Left) *Transverse transabdominal ultrasound shows an intrauterine embryo. Adjacent to the uterus in the left adnexa, there is a large cystic and solid degenerated fibroid with a bridge of myometrium confirming the site of origin.* ***(Right)*** *Sagittal transabdominal ultrasound shows an intrauterine gestational sac with a 9 cm anechoic paraovarian cyst displacing the bladder caudally. The ovaries were identified separately (not shown).*

Hydrosalpinx



Heterotopic Pregnancy



(Left) *Longitudinal transvaginal ultrasound shows a thin-walled, tubular cystic structure in the right adnexa, consistent with a hydrosalpinx.* ***(Right)*** *Sagittal transvaginal ultrasound of the adnexa shows a gestational sac with surrounding clot, consistent with a ruptured tubal ectopic in a patient with an early intrauterine pregnancy.*

SECTION 2
Central Nervous System

Brain

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ABSENT CAVUM SEPTI PELLUCIDI

DIFFERENTIAL DIAGNOSIS

Common
- Incorrect Scan Plane
- Agenesis of the Corpus Callosum
- Chiari II Malformation
- Severe Hydrocephalus

Less Common
- Holoprosencephaly Spectrum
  - Alobar Holoprosencephaly
  - Semilobar Holoprosencephaly
  - Lobar Holoprosencephaly

Rare but Important
- Septo-Optic Dysplasia
- Schizencephaly
- Syntelencephaly

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Scan technique is very important
  - Cavum septi pellucidi (CSP) should be the landmark for measurement of biparietal diameter and head circumference
  - Coronal images also helpful if fetal head position makes correct axial plane difficult
    - CSP seen between frontal horns inferior to corpus callosum
- Cavum septi et vergae is an anatomic variant that may cause confusion
  - Cavum vergae is posterior continuation of CSP, usually obliterates posterior to anterior so not seen in standard 18-20 week scan
  - If present → elongated, larger, fluid-filled space which may be confused with interhemispheric cyst
  - No other abnormalities present
- Normal CSP signifies normal midline brain development
  - If absent, significant malformation may be present though not immediately apparent

Helpful Clues for Common Diagnoses
- **Incorrect Scan Plane**
  - If scan plane too high or too low CSP not seen
  - Fornices are normal structures which create parallel echoes inferior to normal location of CSP
  - CSP appears as a fluid-filled "box": White line/black space/white line appearance
  - Fornices: Series of parallel black and white lines without intervening fluid-filled space
- **Agenesis of the Corpus Callosum**
  - Mild ventriculomegaly or colpocephaly
  - Lateral ventricles are parallel instead of divergent
  - Coronal images show "steer horn" appearance of frontal horns
  - Lack of normal anterior cerebral artery branch pattern into callosomarginal and pericallosal arteries on midline sagittal view
  - Stenogyria: Radiating "sunburst" pattern of medial cerebral gyri
  - MR very helpful to look for additional brain malformations
    - Heterotopia, lissencephaly, gyral abnormalities
- **Chiari II Malformation**
  - Ventriculomegaly with "boxy" appearance of frontal horns
  - "Banana" sign: Cerebellar prolapse into foramen magnum causes cerebellum to curve around brainstem
  - Obliteration of cisterna magna
  - "Lemon" sign: Bifrontal concavity
- **Severe Hydrocephalus**
  - Severe hydrocephalus leads to "blown out cavum"
    - Fenestrations appear in leaves of septum secondary to elevated cerebrospinal fluid pressure
    - Eventually leaves of cavum may be so thinned as to be invisible or torn
  - Most likely to occur with aqueductal stenosis

Helpful Clues for Less Common Diagnoses
- **Alobar Holoprosencephaly**
  - Supratentorial brain without division into cerebral hemispheres
  - "Rind", "cup", or "pancake" of cerebral tissue associated with monoventricle ± dorsal cyst
  - Head may be large if large dorsal cyst, or small and rounded in shape
  - Associated with severe facial malformation
  - Look for stigmata of trisomy 13
- **Semilobar Holoprosencephaly**

- Some division into cerebral hemispheres posteriorly but fusion anteriorly
- Head shape often round
- Look for stigmata of trisomy 13 or 18

- **Lobar Holoprosencephaly**
  - Two cerebral hemispheres are formed
  - May be single gyrus in continuity across midline
  - Midline fusion anomalies seen
    - Fused fornices create a round "mass" in the third ventricle

Helpful Clues for Rare Diagnoses

- **Septo-Optic Dysplasia**
  - Absent CSP associated with variable hypothalamic pituitary dysfunction and visual impairment
  - Mild dilatation of frontal horns
  - "Flat top" appearance to frontal horns
  - MR used to exclude additional malformations
    - Optic nerves and chiasm beyond resolution of fetal MR at present
  - Diagnosis is confirmed by clinical and ophthalmological evaluation of infant
- **Schizencephaly**
  - Cortical defect extending from ventricular surface to pia
  - May be "closed" or "open", unilateral or bilateral
  - Size of defect varies from very small to giant, involving most of frontoparietal cortex
  - Lateral ventricle on side of defect is distorted → tenting toward area of parenchymal loss
  - Bilateral giant open lip schizencephaly looks very similar to hydranencephaly
    - Key observation is that walls of a schizencephalic cleft are lined by grey matter
    - Important distinction as hydranencephaly is lethal
    - Giant open lip schizencephaly is associated with neurological impairment but is not necessarily lethal
- **Syntelencephaly**
  - Considered variant of holoprosencephaly spectrum by some
  - Two hemispheres are present but with some fusion of the dorsal aspect of cerebral hemispheres
    - Most common site of fusion is posterior frontal lobe

Other Essential Information

- Absent CSP is often the tip-off for underlying brain abnormality
- Agenesis of corpus callosum is associated with many other brain malformation as well as innumerable syndromes
  - Counsel parents that outcome highly variable
- Septooptic dysplasia associated with visual impairment/blindness as well as potential hypothalamic pituitary dysfunction

Incorrect Scan Plane



Axial oblique ultrasound of the fetal head shows the parallel lines produced by the fornices ⇒ which lie inferior to the CSP. This should not be confused with the cavum, which should appear as a fluid-filled "box".

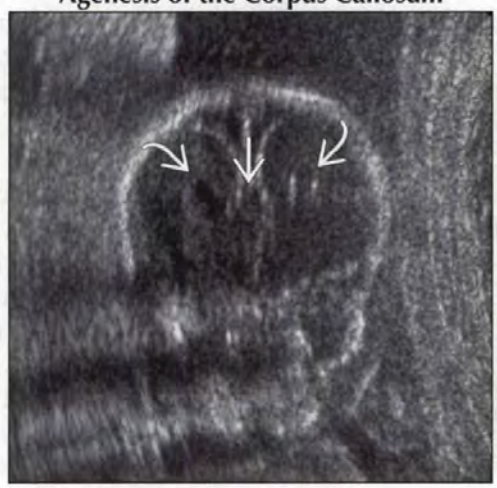
Incorrect Scan Plane



Axial oblique ultrasound shows the correct plane for demonstration of the line ⇒, space ⇒, line ⇒ or box-like structure of the normal cavum septi pellucidi.

ABSENT CAVUM SEPTI PELLUCIDI

Agenesis of the Corpus Callosum



Agenesis of the Corpus Callosum

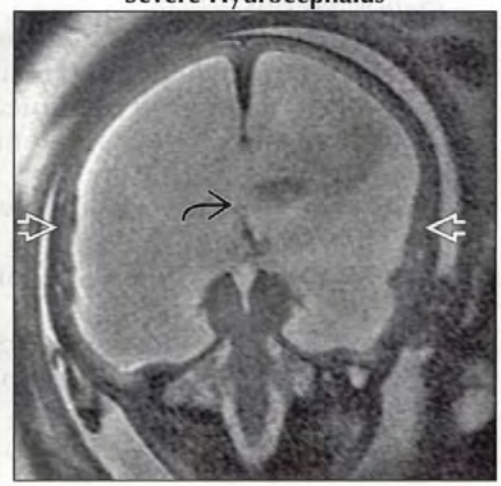


(Left) *Coronal ultrasound at 19 weeks shows no evidence of the cavum ➡ nor any evidence of a corpus callosum. There is a suggestion of steer horn configuration ➡ of the frontal horns.* ***(Right)*** *Sagittal color Doppler ultrasound in the same case confirms the suspicion. Normally the anterior cerebral artery ➡ gives rise to callosomarginal and pericallosal branches that run along the corpus callosum. The ➡ indicates where these vessels should be seen.*

Chiari II Malformation

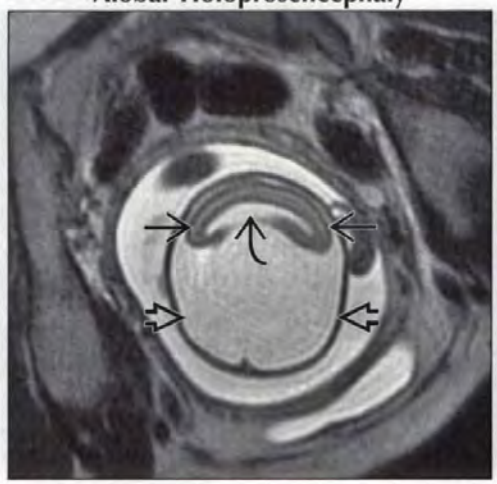


Severe Hydrocephalus

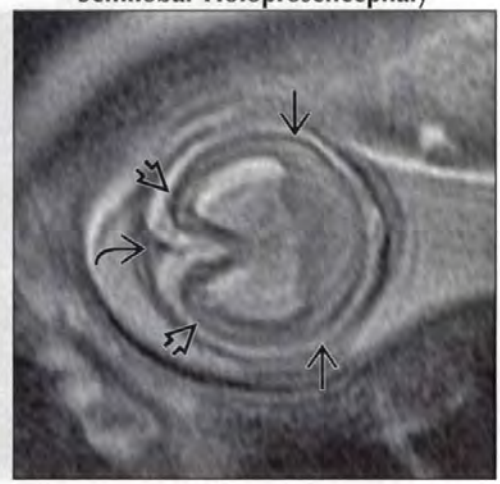


(Left) *Axial oblique ultrasound shows ventriculomegaly ➡, a "banana" cerebellum ➡, obliteration of the cisterna magna ➡ and absent cavum (➡ denotes expected location).* ***(Right)*** *Coronal T2WI MR shows lack of midline structures ➡ attributed to a "blown out" CSP secondary to severe hydrocephalus. This is a fetus with aqueductal stenosis. Note the compressed cortical mantle ➡.*

Alobar Holoprosencephaly



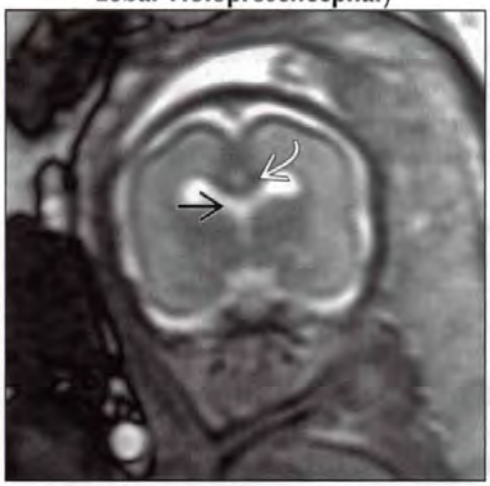
Semilobar Holoprosencephaly



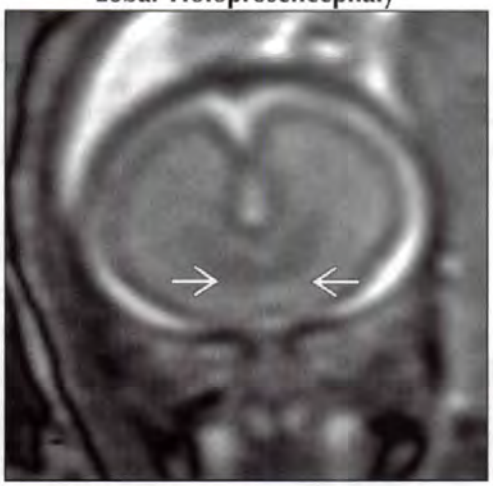
(Left) *Axial T2WI MR shows a "ball" ➡ of brain tissue with a monoventricle ➡ and a large dorsal cyst ➡. There is complete absence of midline structures including the CSP.* ***(Right)*** *Axial T2WI MR shows a round head shape with frontal lobe fusion ➡ but separate lobes ➡ posteriorly. Note the falx ➡ posteriorly.*

ABSENT CAVUM SEPTI PELLUCIDI

Lobar Holoprosencephaly



Lobar Holoprosencephaly

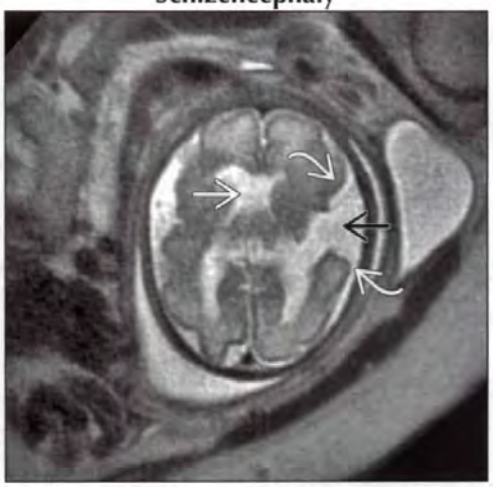


(Left) *Coronal T2WI MR shows an absent CSP ⇨ but intact corpus callosum ⇨.* ***(Right)*** *Coronal T2WI MR more anteriorly in the same case, shows gyral continuity ⇨ across the midline. The neuropathology literature defines lobar holoprosencephaly as having at least one gyrus in continuity. Fetal imaging literature is less stringent, including midline anomalies such as fused fornices as signs of lobar holoprosencephaly.*

Septo-Optic Dysplasia



Schizencephaly

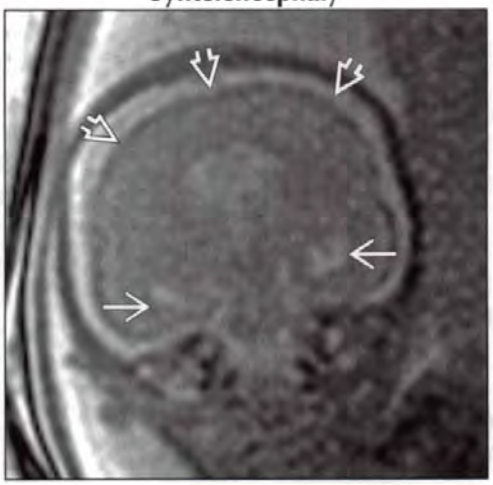


(Left) *Axial oblique ultrasound shows how subtle septo-optic dysplasia can be. The infant was diagnosed shortly after delivery. Review of OB scans showed no pictures of the cavum (⇨ denotes expected location). There were no other findings.* ***(Right)*** *Axial T2WI MR shows an absent CSP ⇨, and a parenchymal defect ⇨ with the edges lined by grey matter ⇨. Other images showed a similar defect on the other side making this bilateral open lip schizencephaly.*

Syntelencephaly



Syntelencephaly



(Left) *Axial T2WI MR shows an absent CSP ⇨ but formation of two cerebral hemispheres with frontal ⇨ and occipital ⇨ lobes.* ***(Right)*** *Coronal T2WI MR in the same case shows gyral continuity ⇨ in the midline at the level of the temporal horns ⇨. This is the typical appearance of syntelencephaly which has been described in association with 13q- deletion.*

MILD VENTRICULOMEGALY

DIFFERENTIAL DIAGNOSIS

Common
- Idiopathic
- Trisomy 21
- Agenesis of the Corpus Callosum
- Aqueductal Stenosis, Early
- Chiari II Malformation, Early

Less Common
- Trisomy 18
- Trisomy 13
- Encephalomalacia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Definition of mild ventriculomegaly is a lateral ventricle which measures 10-12 mm
- Lateral ventricle measurement
  - Part of routine anatomic survey
  - Measure atria at level of choroid plexus glomus
  - Normal measurement
    - < 10 mm from 14-40 wks
    - Mean diameter of 7.6 mm (± 0.6 mm)
    - Normal ventricular width is 70% of hemicalvarium at 18 wks and 30% of hemicalvarium at 28 wks

Helpful Clues for Common Diagnoses
- **Idiopathic**
  - 90% of isolated ventriculomegaly is idiopathic and has a normal outcome
  - 30% resolve in utero
  - More common in male fetuses, especially when presenting > 20 wks
    - Larger calvarial volume
- **Trisomy 21**
  - Look for other markers
    - Nuchal thickening (≥ 5 mm)
    - Short femur/humerus
    - Echogenic bowel
    - Intracardiac echogenic focus
    - Renal pelviectasis
    - Absent nasal bone
  - Associated major anomalies
    - Atrioventricular defect
    - Duodenal atresia
    - Esophageal atresia
- **Agenesis of the Corpus Callosum**
  - Agenesis may be complete or partial
    - Corpus callosum (CC) forms anterior to posterior
    - Posterior CC may not form
  - Colpocephaly
    - Teardrop-shaped lateral ventricle: Pointed frontal horns with posterior dilatation (in region of atria)
    - "Trident-shaped" frontal horns in coronal view
    - Frontal horns laterally displaced and parallel
  - Absent cavum septi pellucidi if complete
  - 50% with other brain anomalies
  - 10-20% with aneuploidy
  - 3% of all mild ventriculomegaly cases have agenesis of CC
- **Aqueductal Stenosis, Early**
  - Narrowing or occlusion of aqueduct of Sylvius
    - Distended lateral and 3rd ventricles
  - Obstructive hydrocephalus
    - Ventricles often normal or only mildly ↑ in 2nd trimester
    - Severe hydrocephalus common in 3rd trimester
    - Progressively ↑ head measurements
  - X-linked form
    - Bickers-Adams syndrome
    - < 5% of all aqueductal stenosis cases
    - 50% recurrence risk in male fetuses
    - Adducted thumbs
  - 90% with mental retardation
    - More severe if X-linked diagnosis
- **Chiari II Malformation, Early**
  - Hindbrain herniation
  - Almost always with spina bifida
  - Posterior fossa compression
    - Cisterna magna obliteration
    - Compressed cerebellum
    - "Banana sign" if cerebellum wraps around midbrain
  - Frontal calvarial findings are non-specific and transient
    - "Lemon-shaped" skull from narrow anterior-pointing frontal bones
    - Dolichocephaly
  - Variable amount of ventriculomegaly
    - 33% progress during pregnancy
    - 90% with ventriculomegaly at birth
  - Open spina bifida (no skin covering)
    - Bony dorsal arch defect

- Neural content exposure (± sac)
- 40% with additional anomalies
- 4% with aneuploidy

Helpful Clues for Less Common Diagnoses

- **Trisomy 18**
  - Hallmark is multiple major anomalies and intrauterine growth restriction
  - Brain anomalies in 30%
    - Ventriculomegaly
    - Chiari II malformation
    - Dandy-Walker continuum
    - Cerebellar hypoplasia
  - Second trimester markers
    - Choroid plexus cyst
    - "Strawberry-shaped" skull (brachycephaly)
    - Single umbilical artery
    - Umbilical cord cyst
- **Trisomy 13**
  - Multiple major anomalies in > 90%
  - Holoprosencephaly is hallmark anomaly
    - Variable severity
    - Monoventricle
    - Fused thalamus
    - Facial anomalies
  - Second trimester markers rarely isolated
    - Echogenic cardiac focus
    - Single umbilical artery
- **Encephalomalacia**
  - Brain parenchyma destruction
  - Early findings often subtle
    - Variable periventricular white matter heterogeneity
  - Cystic degeneration is late finding
  - Vascular causes
    - Hypoperfusion from any cause
    - Intracranial hemorrhage
  - Infectious and teratogen causes
    - Cytomegalovirus (most common)
    - Toxoplasmosis
    - Varicella/Zoster
    - Vitamin A

Other Essential Information

- Should we offer amniocentesis?
  - 4% with aneuploidy (likelihood ratio is 9)
  - ↑ Diagnosis of congenital infection
- Should we perform fetal MR?
  - Additional anomalies seen in 8%
  - Helpful for more subtle anomalies
  - Best after 28 wks
- Is isolated ventriculomegaly truly "isolated"?
  - 4% with subsequent brain anomaly
  - 9% with non CNS anomalies not diagnosed in utero

Alternative Differential Approaches

- Pay attention to head size
- Ventriculomegaly and small head
  - Trisomy 18
  - Trisomy 13
  - Encephalomalacia
- Ventriculomegaly and big head
  - Aqueductal stenosis
  - Trisomy 21 (brachycephaly is common)
  - Choroid plexus papilloma

Idiopathic



Axial ultrasound shows mild lateral ventriculomegaly (calipers). The cavum septi pellucidi ➡ and 3rd ventricle ➡ are seen well. The finding was isolated, and the outcome was normal.

Trisomy 21



Axial ultrasound shows mild ventriculomegaly in a fetus with nuchal fold thickening and mild pelviectasis (not shown). The presence of 3 markers is highly suggestive of aneuploidy.

MILD VENTRICULOMEGALY

Agenesis of the Corpus Callosum



Agenesis of the Corpus Callosum



(Left) *Axial ultrasound shows a "teardrop-shaped" lateral ventricle. The posterior portion is distended, and the anterior is pointed, typical of colpocephaly.* ***(Right)*** *T2WI MR images (axial left, coronal right) show colpocephaly, elevated third ventricle, and parallel frontal horns. On the coronal view, a "trident" appearance of the frontal horns is seen. Absence of the corpus callosum leads to the morphologic changes of the ventricles.*

Aqueductal Stenosis, Early



Aqueductal Stenosis, Early



(Left) *Axial transvaginal ultrasound shows bilateral mild ventriculomegaly at 20 weeks. This fetus was male with a sibling with X-linked aqueductal stenosis.* ***(Right)*** *Coronal ultrasound of the hand showed a fist. Neither hand ever opened, and the thumbs were adducted. The findings, along with amniocentesis results, were diagnostic of a second child affected with X-linked aqueductal stenosis.*

Chiari II Malformation, Early



Chiari II Malformation, Early



(Left) *Axial T2WI MR shows frontal bone concavity (lemon-shaped calvarium) and mild ventriculomegaly.* ***(Right)*** *Sagittal T2WI MR of the fetal brain and spine shows mild ventriculomegaly and hindbrain herniation into the foramen magnum, diagnostic of Chiari II malformation. A large spinal defect is seen involving the majority of the lumbo-sacral spine. The absence of a meningomyelocele sac is noted in this fetus with myeloschisis.*

MILD VENTRICULOMEGALY

Trisomy 18



Trisomy 18



(Left) *Axial ultrasound shows a "dangling choroid" in a fetus with mild ventriculomegaly. The fetus was small for its gestational age and had multiple other anomalies.* ***(Right)*** *Transabdominal ultrasound of the same fetus shows arthrogryposis. The legs were held in extension, and the left foot was clubbed. The fetus also had an absent stomach bubble. Multiple anomalies and fetal growth restriction are typical for trisomy 18.*

Trisomy 13



Trisomy 13

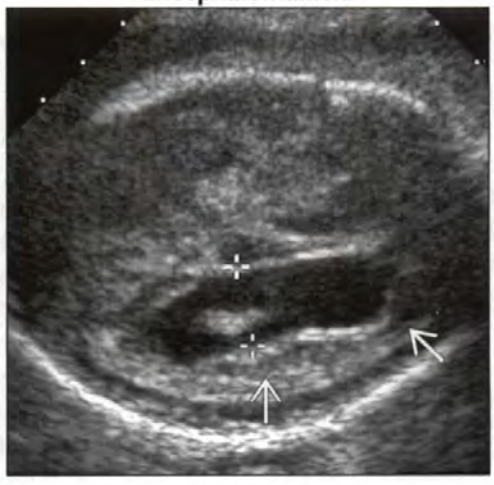


(Left) *Axial ultrasound shows mild ventriculomegaly (calipers).* ***(Right)*** *Axial ultrasound through the calvarium in the same fetus shows partial absence of the falx and fusion of the frontal lobes. This fetus has subtle semilobar holoprosencephaly. Severe facial anomalies were also present, and amniocentesis results yielded trisomy 13.*

Encephalomalacia



Encephalomalacia



(Left) *Axial ultrasound shows mild ventriculomegaly secondary to cystic periventricular degeneration. This fetus was a twin in a pregnancy complicated by twin/twin transfusion.* ***(Right)*** *Axial ultrasound shows mild ventriculomegaly (calipers) and subtle lucencies in the periventricular white matter in a patient with prior trauma and placental abruption. Periventricular leukomalacia was found at autopsy.*

HYDROCEPHALUS

DIFFERENTIAL DIAGNOSIS

Common
- Chiari II Malformation
- Aqueductal Stenosis, Late
- Dandy-Walker Continuum: Classic

Less Common
- Intracranial Hemorrhage
- Encephalomalacia

Rare but Important
- Obstructing Mass
- Choroid Plexus Papilloma

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Routine lateral ventricle measurement
  - Measure atria of lateral ventricle
    - At level of choroid plexus glomus
    - Measure inner to inner wall
  - Normal atrial measurements
    - < 10 mm between 14-40 wks
  - Ventriculomegaly
    - Mild: 10-12 mm
    - Moderate: 12-15 mm
    - Severe: > 15 mm
- Use transvaginal ultrasound
  - Cephalic presentation
    - Frontal fontanelle as acoustic window
  - Best for corpus callosum visualization
    - Color Doppler to show pericallosal artery

Helpful Clues for Common Diagnoses
- **Chiari II Malformation**
  - Hindbrain herniation
    - Causes obstructive hydrocephalus
  - Variable ventriculomegaly
    - 55% at time of diagnosis
    - 33% progress during pregnancy
  - Variable calvarial measurements
    - Normal or small in 2nd trimester
    - Macrocephaly if ↑ ventriculomegaly
  - Posterior fossa compression
    - Small or obliterated cisterna magna
    - Variable cerebellar compression
    - "Banana sign" if cerebellum wraps around midbrain
  - "Lemon-shaped" calvarium
    - Frontal bone narrowing and "tenting"
  - Almost always with spina bifida
    - 90% lumbosacral
    - 80% with overlying sac
- **Aqueductal Stenosis, Late**
  - Aqueduct of Sylvius narrowing/obstruction
    - ↑ 3rd and lateral ventricles
    - Normal 4th ventricle/posterior fossa
  - Progressive hydrocephalus
    - Initial normal or mild ventriculomegaly
    - Macrocephaly common near term
  - X-linked form
    - Bickers-Adams syndrome
    - < 5% of all cases
    - 50% recurrence risk in male fetuses
    - Adducted thumbs is additional finding
  - 90% with mental retardation
    - More severe retardation if X-linked
- **Dandy-Walker Continuum: Classic**
  - Most severe cystic posterior fossa malformation
    - Absent cerebellar vermis
    - Cystic dilatation of 4th ventricle
    - Variable ventriculomegaly
  - Ultrasound findings
    - Splayed cerebellar hemispheres
    - Small/absent cerebellum if severe
    - Posterior fossa cyst
  - Associated findings
    - Dysgenesis of corpus callosum
    - Encephalocele, neural tube defect
    - Cleft lip/palate
    - Cardiac defects
    - Polycystic kidneys

Helpful Clues for Less Common Diagnoses
- **Intracranial Hemorrhage**
  - Most common locations include: Subependymal, germinal matrix, intraventricular, intraparenchymal, subdural
  - Ventriculomegaly from 2 causes
    - Brain destruction ⇒ ventricular expansion
    - Intraventricular bleed ⇒ obstructive hydrocephalus
  - Ventricle findings
    - Hyperechoic intraventricular clot
    - Echogenic cerebrospinal fluid
    - Bulky irregular choroid plexus
    - Echogenic irregular ependyma
  - Causes
    - Maternal/fetal ↑ or ↓ blood pressure
    - Trauma
    - Thrombocytopenia/coagulation disorders

- Infection
- Fetal arteriovenous malformation

- **Encephalomalacia**
  - Brain parenchyma destruction
  - Ultrasound findings
    - Early: Periventricular heterogeneity
    - Late: Cystic change
  - Causes
    - Hypoperfusion from any cause
    - Infection
    - Teratogen exposure

Helpful Clues for Rare Diagnoses

- **Obstructing Mass**
  - Mass or mass-like lesions obstruct cerebrospinal flow
  - Cystic mass
    - Arachnoid cyst
    - Glioependymal cyst
    - Cystic teratoma
  - Solid mass
    - Solid teratoma
    - Astrocytoma or other rare tumor
    - Choroid plexus papilloma
  - Mass-like lesion
    - Vein of Galen malformation
    - Arteriovenous fistula
    - Intracranial hemorrhage
- **Choroid Plexus Papilloma**
  - Hydrocephalus from over production of cerebral spinal fluid (CSF)
    - Often rapid onset
  - Well-defined, lobular, hyperechoic mass
  - Occurs anywhere in ventricular system
    - Lateral ventricle most common
  - Mass may also obstruct ventricle causing asymmetric enlargement

Other Essential Information

- Normal hypoechoic brain can mimic ventriculomegaly
  - Use "dangling choroid" sign
    - Choroid "dangles" in a distended ventricle
    - Choroid lies almost parallel to falx in normal ventricle
- Confirm hydrocephalus with multiple views
  - 3D ultrasound helpful
- Asymmetric hydrocephalus is rare
  - Gravity dependent ventricle often slightly ↑ compared to other ventricle
- Role of MR
  - Additional brain anomalies seen in 8%
- Aneuploidy rates
  - Dandy-Walker continuum: 50%
  - Chiari II: 4%
  - Aqueductal stenosis: < 5% X-linked
  - Others: No ↑ risk

Alternative Differential Approaches

- Use posterior fossa view to help with differential
  - Small posterior fossa
    - Chiari II
  - Large/cystic posterior fossa
    - Dandy-Walker continuum
  - Normal posterior fossa
    - Aqueductal stenosis

Chiari II Malformation



Axial ultrasound of the fetal head shows a "dangling" choroid plexus in a distended lateral ventricle. The third ventricle and near field lateral ventricle are also dilated.

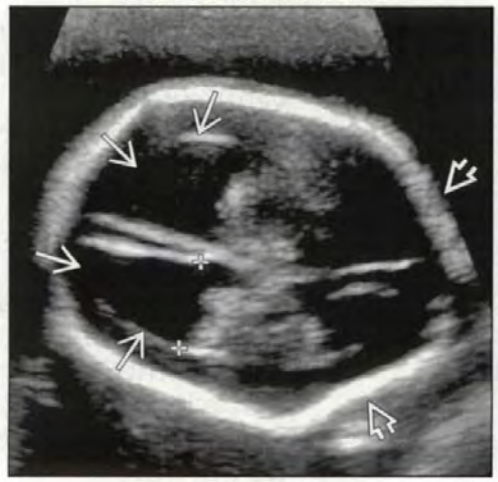
Chiari II Malformation



3D ultrasound of spina bifida in the same fetus shows a distal meningomyelocele sac, splaying of the dorsal lumbosacral spine ossification centers and a skin defect on the back.

HYDROCEPHALUS

Chiari II Malformation



Chiari II Malformation



(Left) *Axial ultrasound through the ventricles in a second trimester fetus shows severe ventriculomegaly* ➔ *and a "lemon-shaped" calvarium with frontal bone "tenting" and narrowing* ➔. ***(Right)*** *Axial ultrasound of the posterior fossa, shows a "banana-shaped" cerebellum* ➔. *The posterior fossa is compressed and the cerebellum wraps around the midbrain* ➔. *These findings are diagnostic of Chiari II. The spine must now be examined carefully to find the spina bifida.*

Aqueductal Stenosis, Late

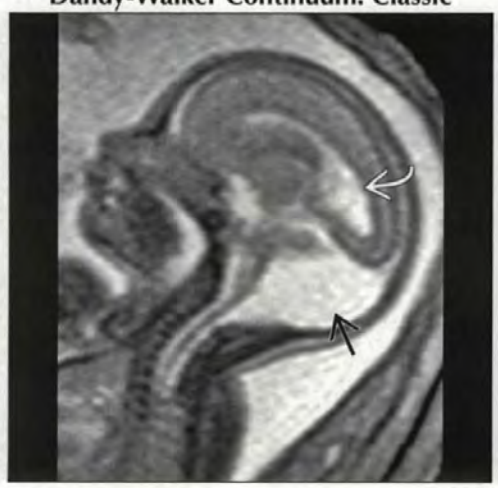


Aqueductal Stenosis, Late



(Left) *Axial ultrasound shows markedly distended lateral ventricles* ➔. ***(Right)*** *3D ultrasound of the fetal hands* ➔ *in the same case showed consistently clenched hands and adducted thumbs. The fetus was male and X-linked aqueductal stenosis was suspected. Chromosome analysis confirmed the diagnosis.*

Dandy-Walker Continuum: Classic



Dandy-Walker Continuum: Classic

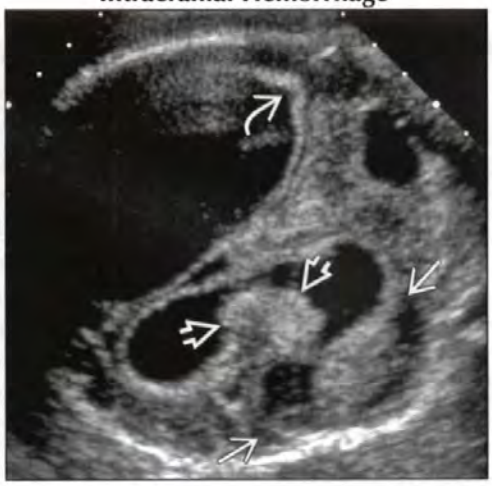


(Left) *Sagittal T2WI MR of a second trimester fetus shows a large posterior fossa cyst* ➔ *and ventriculomegaly* ➔. ***(Right)*** *Axial T2WI MR of the same fetus shows splaying of the cerebellar hemispheres* ➔ *and absence of the cerebellar vermis. The fourth ventricle appears to be contiguous with the cyst.*

Intracranial Hemorrhage

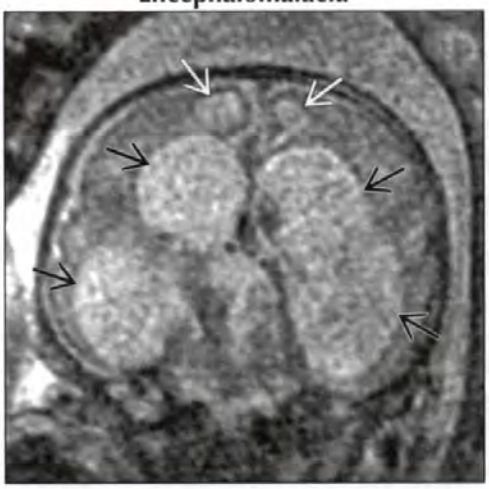


Intracranial Hemorrhage

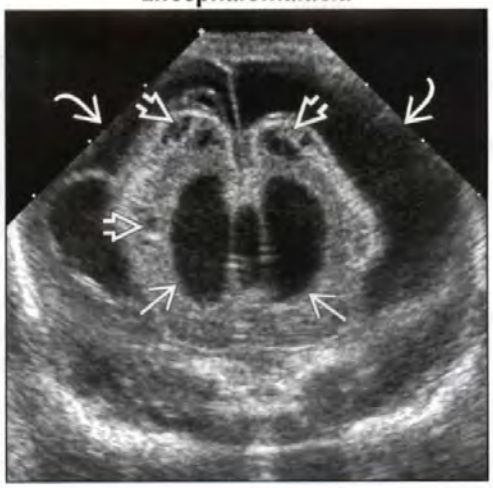


(Left) *Axial ultrasound in a third trimester pregnancy complicated by maternal thrombocytopenia shows a parenchymal and intraventricular hemorrhage. The lateral and third ventricles are also markedly distended.* ***(Right)*** *Coronal oblique ultrasound of the same fetus shows the parenchymal hemorrhage. In addition, the choroid plexus is nodular and the ventricular lining is echogenic, suggesting the presence of intraventricular blood.*

Encephalomalacia

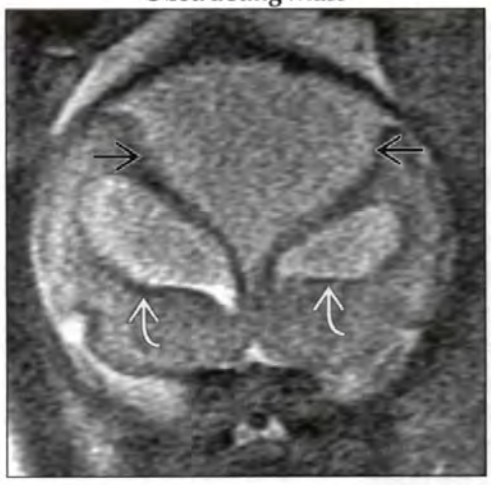


Encephalomalacia

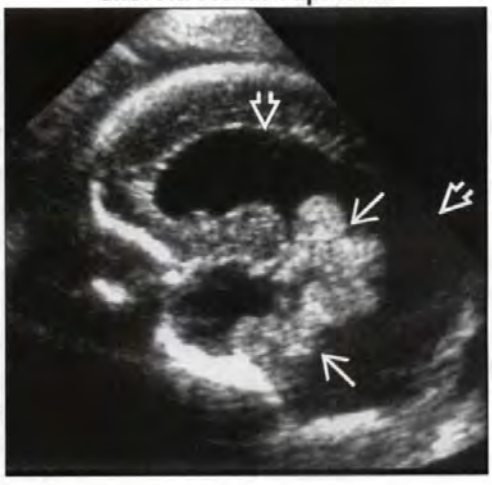


(Left) *Coronal T2WI MR in a fetus with prior cerebral hemorrhage and ischemia shows severe hydrocephalus and parenchymal brain cysts. Porencephaly is a late finding with encephalomalacia.* ***(Right)*** *Neonatal head ultrasound of the donor twin in a pregnancy with twin-twin transfusion shows cystic encephalomalacia from chronic hypoxia in utero. The ventricles and extra-cerebral spaces expand to fill the space left by destroyed brain.*

Obstructing Mass



Choroid Plexus Papilloma



(Left) *Coronal T2WI MR shows an interhemispheric glioependymal cyst. The frontal horns of the lateral ventricles are markedly distended secondary to obstruction of CSF flow at the foramen of Monro.* ***(Right)*** *Sagittal ultrasound shows a lobular, hyperechoic mass within the atrium of the dilated lateral ventricle. Hydrocephalus is the result of overproduction of CSF.*

FLUID-FILLED CALVARIUM

DIFFERENTIAL DIAGNOSIS

Common
- Severe Hydrocephalus
- Holoprosencephaly Spectrum
- Hydranencephaly

Less Common
- Dandy-Walker Continuum: Classic
- Severe Schizencephaly

Rare but Important
- Large Arachnoid Cyst
- Large Glioependymal Cyst

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Routine lateral ventricle measurement
  - Measure atria
    - At level of choroid plexus glomus
    - Inner-to-inner wall measurement
  - Normal is < 10 mm between 14-40 wks
- Routine brain anatomy assessment
  - Falx
  - Cavum septi pellucidi
  - Thalamus
  - Choroid plexus
  - Cerebellum
  - Cisterna magna
- Use vaginal ultrasound if fetus cephalic
- MR helpful for differential diagnosis

Helpful Clues for Common Diagnoses
- **Severe Hydrocephalus**
  - > 15 mm lateral ventricular (LV)
  - Thin cortical mantle
    - Compressed by ventricles
  - "Dangling" choroid
    - Choroid does not fill LV
    - Bilateral "dangling" if opposite choroid falls through foramen of Monro
  - Cavum septi pellucidi often absent
    - Fenestrations in wall from ↑ pressure
  - Macrocephaly if obstructive cause
  - Causes
    - Aqueductal stenosis
    - Dandy-Walker continuum
    - Chiari II malformation
- **Holoprosencephaly Spectrum**
  - Early arrest of brain cleavage and rotation
    - Variable severity
  - No clear division between different types
    - Alobar (most severe)
    - Semilobar (intermediate type)
    - Lobar (subtle)
  - Ventricle findings
    - Monoventricle
    - Dorsal sac
  - Variable absent midline structures
    - Cavum septi pellucidi
    - Falx
    - Corpus callosum
    - 3rd ventricle
    - Variable fused thalamus
  - Brain mantle appearance
    - "Pancake": Flat at skull base
    - "Cup": Partially surrounds monoventricle
    - "Ball": Mantle surrounds monoventricle
  - Associated facial anomalies
    - Hypotelorism ⇒ cyclopia
    - Abnormal nose ⇒ proboscis
    - Median or bilateral cleft lip/palate
  - Associations
    - Trisomy 13 (most common)
    - Trisomy 18
    - Maternal diabetes
- **Hydranencephaly**
  - Complete cerebral hemisphere destruction
  - Etiology
    - Vascular occlusion
    - Hemorrhage
    - Hypotension
    - Infection
  - Imaging features
    - Absent cerebral hemispheres
    - Present falx
    - Normal posterior fossa
    - Normal or splayed thalamus
    - Variable head size
    - No flow in middle or anterior cerebral arteries

Helpful Clues for Less Common Diagnoses
- **Dandy-Walker Continuum: Classic**
  - Dysgenesis of cerebellar vermis
    - Absent
    - Severely hypoplastic
  - Cystic dilatation of 4th ventricle
    - Ventricle communicates with cisterna magna
    - Posterior fossa cyst
  - Associated brain findings
    - Agenesis of corpus callosum
    - Hydrocephalus

- Encephalocele
- Neural tube defect
- Careful with diagnosis before 18 wks
  - Vermis not fully formed < 18 wks
  - Normal 1st trimester rhombencephalon is large and cyst-like

- **Severe Schizencephaly**
  - Brain cleft
    - Wedge-shaped defect
    - Filled with cerebrospinal fluid (CSF)
    - Extends from ventricle to brain surface
    - Lined with gray matter
  - Most common in cerebral hemispheres
    - Unilateral in 60%
    - Bilateral in 40%
  - Variable size
  - Associated brain anomalies
    - Absent cavum septi pellucidi
    - Septo-optic dysplasia
    - Heterotopia
    - Polymicrogyria
    - Pachygyria
  - Etiology
    - Neuronal migration anomaly

Helpful Clues for Rare Diagnoses

- **Large Arachnoid Cyst**
  - CSF cyst within layers of arachnoid
  - Location (fetal series)
    - Most common over cerebral convexities
    - ⅓ in posterior fossa
  - May grow rapidly in utero
    - Obstructive hydrocephalus
  - Agenesis of corpus callosum in 5%

- **Large Glioependymal Cyst**
  - Intracranial cyst with ependymal lining
  - Typically midline
    - Frontoparietal
  - Can cause obstructive hydrocephalus
  - Can look identical to arachnoid cyst

Other Essential Information

- Pitfalls
  - Shadowing from skull may cause calvarium to look fluid-filled
    - Use orthogonal planes
    - Use transvaginal ultrasound if vertex
  - Severe hydrocephalus can mimic hydranencephaly
    - Look for compressed brain mantle
    - MR is helpful
  - Structures may be present but compressed
    - Corpus callosum
    - Cavum septi pellucidi
- Use MR to better evaluate anatomy

Alternative Differential Approaches

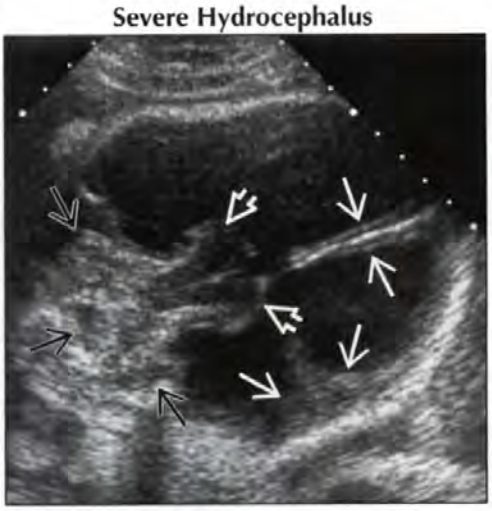
- What does the posterior fossa look like?
  - Compressed with Chiari II
  - Cystic with Dandy-Walker
  - Normal with aqueductal stenosis
- What does the brain mantle look like?
  - Thinned but normal with hydrocephalus
  - Absent with hydranencephaly
  - Small and abnormal with holoprosencephaly
  - Compressed and displaced with cysts
  - Partially absent with schizencephaly

Severe Hydrocephalus



Axial ultrasound shows severe hydrocephalus from aqueductal stenosis. The falx ➡ is present, and the calvarium is fluid-filled, secondary to severe ventriculomegaly.

Severe Hydrocephalus



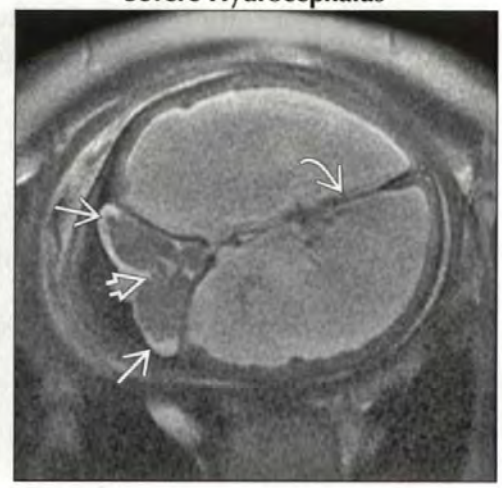
Coronal ultrasound of the same fetus shows compressed brain ➡ along the falx and calvarium. The presence of a normal thalamus ➡ and cerebellum ➡ helps with the differential diagnosis.

FLUID-FILLED CALVARIUM

Severe Hydrocephalus

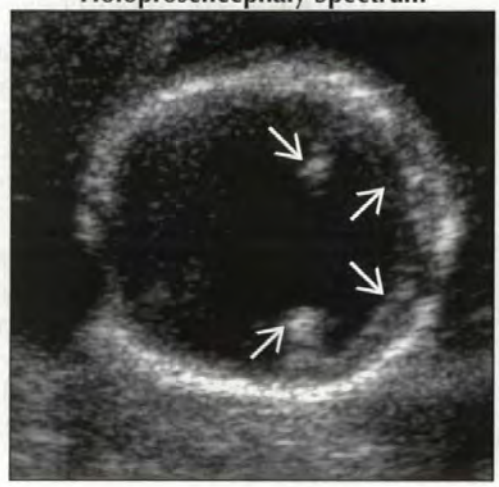


Severe Hydrocephalus

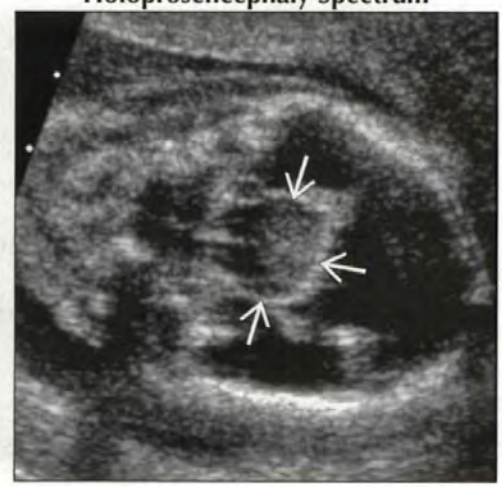


(Left) *Axial T2WI MR shows severe hydrocephalus. A thin but intact cortical mantle is seen.* ***(Right)*** *Coronal T2WI MR in the same fetus shows a normal posterior fossa with an intact cerebellar vermis and falx. These features help rule out hydranencephaly, holoprosencephaly, and Dandy-Walker continuum.*

Holoprosencephaly Spectrum



Holoprosencephaly Spectrum

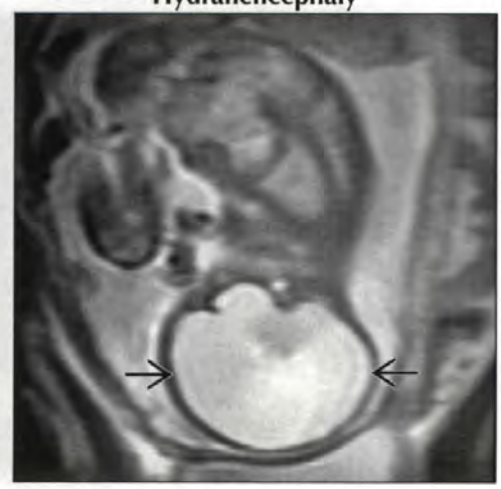


(Left) *Axial ultrasound shows a fluid-filled calvarium with a partial anterior brain mantle. There is no falx or significant lateral ventricle cleavage. Instead, there is a monoventricle.* ***(Right)*** *Coronal oblique ultrasound of the same fetus shows fused thalami. Multiple other anomalies were also seen in this case, and amniocentesis test results identified trisomy 13.*

Hydranencephaly



Hydranencephaly



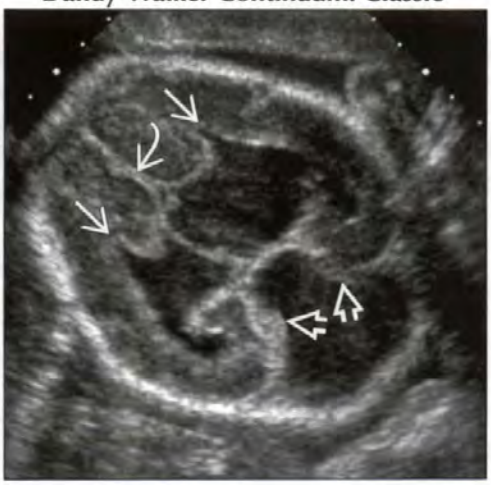
(Left) *Axial ultrasound at 14 weeks shows a fluid-filled calvarium with complete lack of cerebral tissue. The falx and brainstem are present. With hydranencephaly, the brainstem can herniate upwards and mimic fused thalami.* ***(Right)*** *Sagittal T2WI MR in another fetus shows a fluid-filled calvarium without any discernible cerebral tissue. This fetus had a normal brain ultrasound at 18 weeks, prior to a significant maternal traumatic event.*

FLUID-FILLED CALVARIUM

Dandy-Walker Continuum: Classic



Dandy-Walker Continuum: Classic



(Left) *Sagittal oblique ultrasound of the posterior fossa in a 3rd trimester fetus shows a large cyst. The cerebellum was compressed and not seen.* ***(Right)*** *Coronal oblique ultrasound in a 2nd trimester fetus shows a posterior fossa cyst, compressing the cerebellar hemispheres. This fetus also has agenesis of the corpus callosum (CC). Because the CC is absent, the falx extends directly to the ventricles, which have a pointed "trident shape".*

Severe Schizencephaly

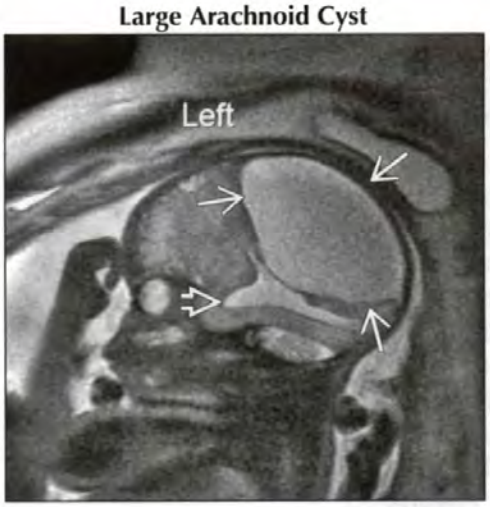


Severe Schizencephaly

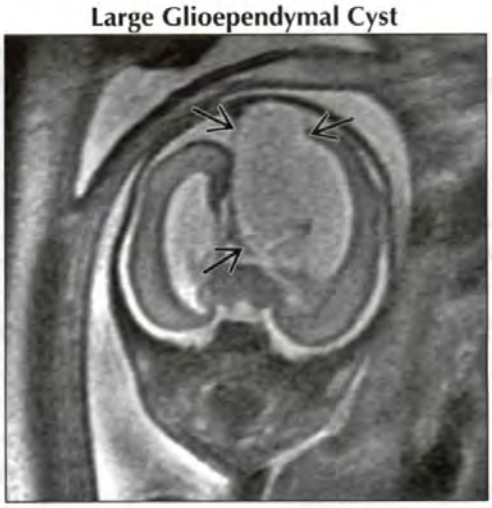


(Left) *Coronal ultrasound shows bilateral giant schizencephaly clefts extending from the ventricle to the undersurface of the calvarium. The thalami are not fused, distinguishing the diagnosis from holoprosencephaly.* ***(Right)*** *Coronal T2WI MR of the same fetus confirms the presence of bilateral large clefts and a normal cerebellum. In addition, the fetal MR demonstrates that the clefts are lined by gray matter.*

Large Arachnoid Cyst



Large Glioependymal Cyst



(Left) *Sagittal T2WI MR shows a large left supratentorial arachnoid cyst. This cyst progressively enlarged during pregnancy and caused ventriculomegaly.* ***(Right)*** *Coronal T2WI MR shows a large midline brain cyst associated with agenesis of the corpus callosum and obstructive hydrocephalus. The midline location suggests glioependymal cyst. Its appearance is otherwise identical to an arachnoid cyst.*

INTRACRANIAL CYSTS: MIDLINE

DIFFERENTIAL DIAGNOSIS

Common
- Cavum Vergae
- Agenesis of the Corpus Callosum
- Dandy-Walker Continuum: Classic
- Dandy-Walker Continuum: Variant
- Alobar Holoprosencephaly
- Semilobar/Lobar Holoprosencephaly
- Arachnoid Cyst

Less Common
- Glioependymal Cyst
- Cystic Teratoma
- Syntelencephaly

Rare but Important
- Vein of Galen Malformation
- Arteriovenous Fistula

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is it a vascular structure?
- Could it be a thrombosed arteriovenous fistula (AVF)?
- Where is it?
  - Supratentorial
    - Are there 2 separate cerebral hemispheres?
    - Is the corpus callosum (CC) intact?
    - Are there any solid components?
    - Is it simple or multiloculated?
  - Infratentorial
    - Is the cerebellar vermis normal?
    - Is the cyst in continuity with the 4th ventricle?
    - Is the torcular (confluence of sinuses) elevated?

Helpful Clues for Common Diagnoses
- **Cavum Vergae**
  - Anatomic variant
  - Cavum vergae is the posterior extension of the cavum septi pellucidi (CSP)
- **Agenesis of the Corpus Callosum**
  - Absence of CC and high-riding 3rd ventricle create prominent midline cerebrospinal fluid (CSF) space
  - Associated with midline cysts (usually glioependymal)
  - Absent CSP
  - Parallel lateral ventricles
  - Colpocephaly (tear-drop shaped ventricles)
  - "Steer horn" or "trident" configuration of frontal horns on coronal view
  - Abnormal branch pattern of anterior cerebral artery
  - Stenogyria: Gyri in radial "sunray" distribution in sagittal plane
- **Dandy-Walker Continuum: Classic**
  - Vermis severely hypoplastic or absent
  - Cystic dilatation of 4th ventricle
  - Large posterior fossa with big CSF cyst
  - 4th ventricle appears "open" and contiguous with cyst
  - Elevation of torcular
  - Ventriculomegaly may be present
- **Dandy-Walker Continuum: Variant**
  - Inferior vermis absent/dysplastic
  - Torcular position normal
  - Posterior fossa not enlarged
  - "Keyhole" appearance of 4th ventricle
- **Alobar Holoprosencephaly**
  - Monoventricle
  - Absent CSP
  - Absent falx
  - Often abnormal facies
    - Hypotelorism, cyclopia, facial cleft
    - Proboscis, absent nose, ethmocephaly, cebocephaly
  - Associated with aneuploidy, particularly trisomy 13
- **Semilobar/Lobar Holoprosencephaly**
  - Monoventricle anteriorly
  - Absent CSP
  - Separation into two lobes posteriorly
  - May be associated with abnormal facies
- **Arachnoid Cyst**
  - Extra-axial, avascular, simple
  - Majority over convexities but may be midline
    - ⅓ in posterior fossa in the fetus

Helpful Clues for Less Common Diagnoses
- **Glioependymal Cyst**
  - Avascular, may be multiloculated
  - More commonly midline and associated with agenesis of corpus callosum
- **Cystic Teratoma**
  - Part cystic, part solid mass
  - Gross distortion of cerebral architecture
  - Hydrocephalus
  - Polyhydramnios
- **Syntelencephaly**

- Separate ventricle anteriorly and posteriorly
- Parietal gyral continuity
- Fusion of ventricles in prefrontal area
- Associated with 13q-deletion
  - Syndactyly
  - Hypoplastic thumbs

Helpful Clues for Rare Diagnoses

- **Vein of Galen Malformation**
  - Elongated tubular midline vascular structure
    - Aneurysmal dilatation of median prosencephalic vein of Markowski
  - Located in cistern of velum interpositum and quadrigeminal plate cistern
  - Look for enlarged neck vessels
  - Look for cardiomegaly/hydrops from high output state
  - May cause hydrocephalus
  - May cause ischemic encephalomalacia
- **Arteriovenous Fistula**
  - Tubular structures in midline
  - Look for flow on Doppler
  - Thrombosed AVF looks hypoechoic, low level internal echoes, occasional echogenic clot within hypoechoic area
  - Associated with intracranial hemorrhage
    - Echogenic clot in ventricle
    - Echogenic ependyma
  - Associated with ischemic encephalomalacia
    - Loss of grey white matter differentiation
    - Ventriculomegaly
    - Porencephalic cyst
    - Microcephaly

Other Essential Information

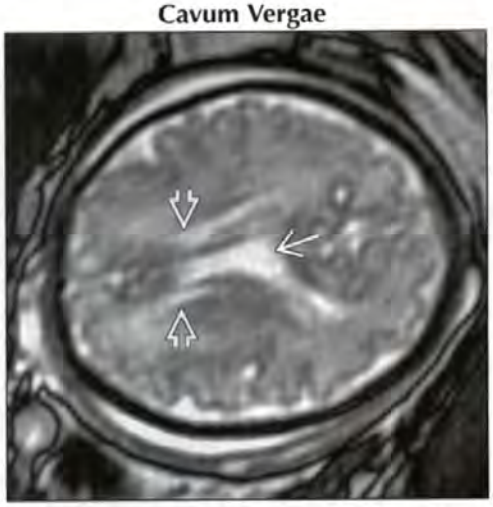
- Prognosis varies with etiology
  - Cavum vergae: No clinical significance
  - Agenesis of the corpus callosum
    - Prognosis depends on any associated syndromes/brain malformations
  - Dandy-Walker continuum
    - Outcome worse for classic malformation
    - Cognitive outcome depends upon associated syndromes/other brain malformations
  - Holoprosencephaly spectrum
    - Alobar: Many die in utero, rare survival beyond neonatal period reported
    - Semilobar/lobar outcome varies with type and severity of malformation
    - Developmental delay, seizure disorder, hypothalamic pituitary malfunction and visual impairment all reported
  - Arachnoid or glioependymal cyst
    - Prognosis depends on underlying brain malformation
    - Shunt placement required for obstructive hydrocephalus
  - Cystic teratoma
    - Dismal prognosis; 97% mortality if diagnosed before 30 weeks
  - Vascular malformations
    - Poor outcome if associated intracranial hemorrhage or ischemic encephalomalacia

Cavum Vergae



Axial oblique ultrasound shows an elongated, fluid-filled space ➡ in the midline. There was no flow, and the remaining brain was normal. This is typical of a cavum vergae.

Cavum Vergae



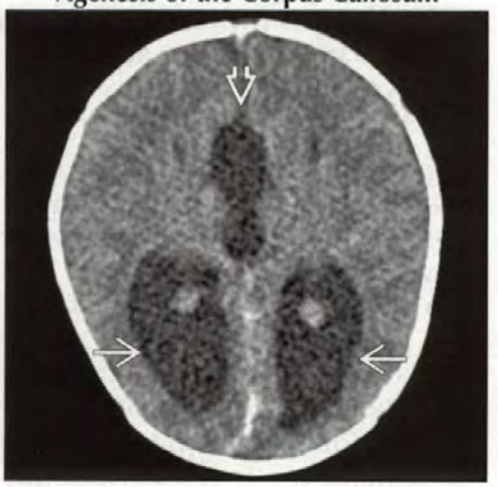
Axial oblique T2WI MR shows the elongated, fluid-filled cavum vergae ➡ lying between the lateral ventricles ➡. This is an anatomic variant of no clinical significance.

INTRACRANIAL CYSTS: MIDLINE

Agenesis of the Corpus Callosum



Agenesis of the Corpus Callosum

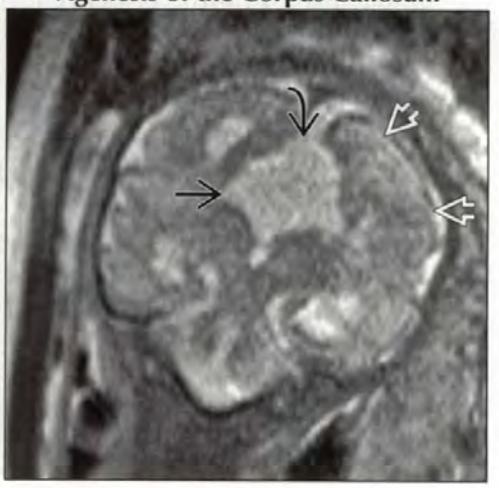


(Left) *Coronal T2WI MR shows a "steer horn" configuration of the anterior horns, absence of the corpus callosum, and increased CSF space between the hemispheres that can be mistaken for a midline cyst.* ***(Right)*** *Axial NECT in a newborn shows colpocephaly, a typical finding in agenesis of the corpus callosum. Note the interhemispheric collection of cerebrospinal fluid, which can mimic a a midline cyst.*

Agenesis of the Corpus Callosum



Agenesis of the Corpus Callosum

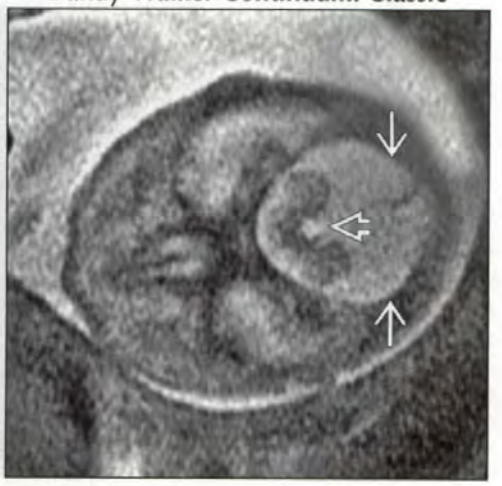


(Left) *Axial T2WI MR shows a large interhemispheric cyst in association with agenesis of the corpus callosum. This has mass effect, displacing the left cerebral hemisphere away from the midline.* ***(Right)*** *Coronal T2WI MR shows agenesis of the corpus callosum with an interhemispheric cyst and cortical dysplasia in a female fetus. The final diagnosis was Aicardi syndrome.*

Dandy-Walker Continuum: Classic



Dandy-Walker Continuum: Classic



(Left) *Axial oblique ultrasound shows a posterior fossa cyst in direct contact with the brainstem due to the absence of the vermis.* ***(Right)*** *Axial T2WI MR shows a posterior fossa cyst and absence of the vermis, which were associated with elevation of the torcular on sagittal views. These are the typical features of the Dandy-Walker malformation.*

Dandy-Walker Continuum: Variant



Dandy-Walker Continuum: Variant



(Left) *Axial oblique ultrasound shows a cleft in the inferior vermis in continuity with the 4th ventricle. The cisterna magna is not dramatically enlarged, nor was the torcular elevated.* ***(Right)*** *Coronal T2WI MR shows deficient inferior vermis but presence of the superior vermis. This fetus had multiple other anomalies including hypoplastic left heart. The infant did not survive, and autopsy was declined.*

Alobar Holoprosencephaly

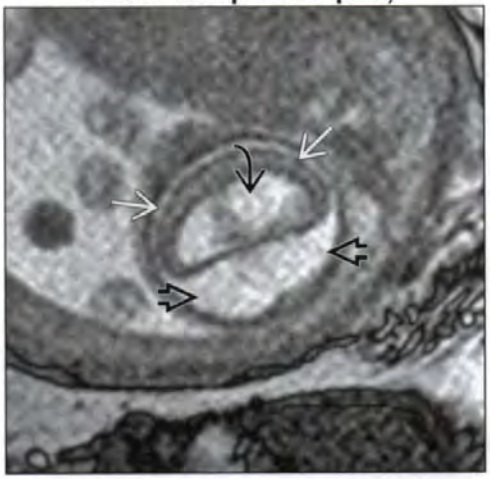


Alobar Holoprosencephaly



(Left) *Axial oblique ultrasound of a fetus shows fused thalami and a fluid-filled monoventricle, findings typical of alobar holoprosencephaly.* ***(Right)*** *Coronal oblique ultrasound of the neonatal head in the same case as previous image confirms thalamic fusion and monoventricle, typical for alobar holoprosencephaly.*

Alobar Holoprosencephaly



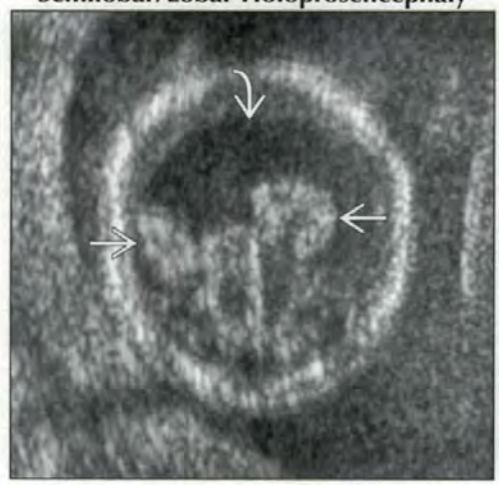
Alobar Holoprosencephaly



(Left) *Axial T2WI MR shows the small, abnormal brain and monoventricle displaced anteriorly by the large dorsal sac filled with cerebrospinal fluid.* ***(Right)*** *Gross pathology in a similar case shows a posterior view of the cranial cavity which has been "bivalved". The anteriorly placed, simple brain shows no evidence of cleavage into hemispheres and the wall of the deflated dorsal sac is seen adjacent to the reflected calvarium.*

INTRACRANIAL CYSTS: MIDLINE

Semilobar/Lobar Holoprosencephaly



(Left) *Axial oblique ultrasound shows an anterior monoventricle. The thalami are not fused and there are separate choroid plexus echoes indicating some attempt at formation of hemispheres, as seen in semilobar holoprosencephaly.* ***(Right)*** *Axial oblique ultrasound shows an extra-axial interhemispheric cyst lying posterior to the third ventricle. This stayed stable during the pregnancy and the infant was asymptomatic at birth.*

Arachnoid Cyst

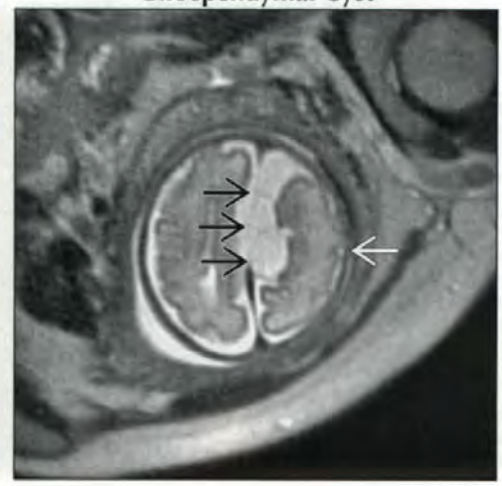


Arachnoid Cyst

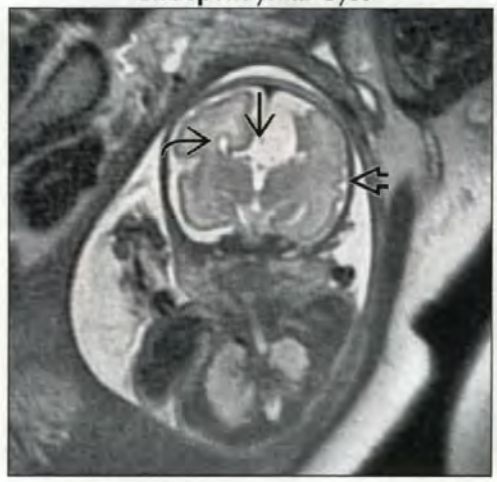


(Left) *Coronal color Doppler ultrasound in the same case as the previous image shows no internal vascularity. This is the typical appearance of an arachnoid cyst.* ***(Right)*** *Axial T2WI MR shows a multiloculated interhemispheric cyst associated with cortical dysplasia involving the left cerebral hemisphere in a fetus with agenesis of the corpus callosum. Glioependymal cysts are more likely to be multiloculated than arachnoid cysts.*

Glioependymal Cyst



Glioependymal Cyst



(Left) *Coronal T2WI MR in the same case as the prior image shows the cyst and associated absent corpus callosum. Note the "steer horn" appearance of the frontal horns and abnormal gyral pattern of the left cerebral hemisphere.* ***(Right)*** *Axial ultrasound shows a complex, midline echogenic mass with cystic and solid components causing obstructive hydrocephalus with dilatation of the occipital horn. Cystic teratoma was diagnosed on autopsy.*

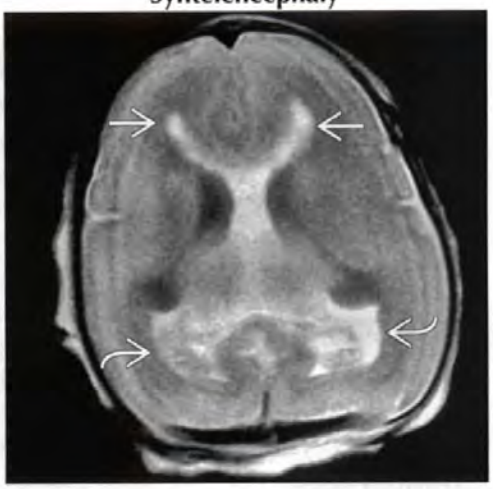
Cystic Teratoma



Syntelencephaly

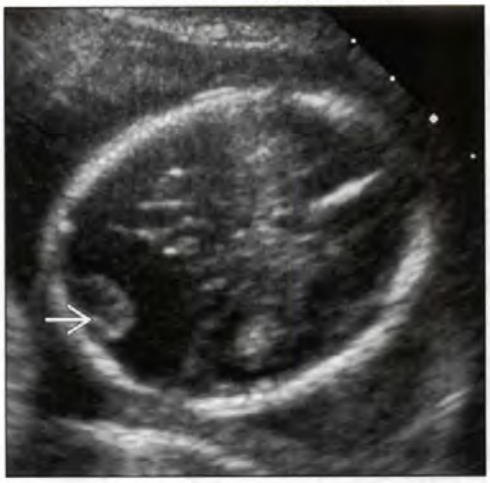


Syntelencephaly



(Left) *Coronal T2WI MR performed as part of an autopsy shows a monoventricle ➡ but separate germinal matrices ➡ and thalami ➡. **(Right)** Axial T2WI MR in the same case shows that there are separate frontal ➡ and occipital ➡ horns. Additional images showed gyral continuity in association with the monoventricle indicating syntelencephaly. In this case amniocentesis revealed a 13q-deletion.*

Vein of Galen Malformation

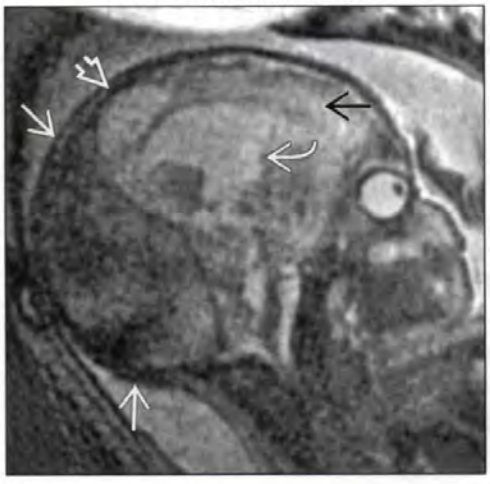


Vein of Galen Malformation



(Left) *Axial oblique ultrasound shows a posterior midline "cyst" containing an echogenic focus ➡. Doppler was negative, and the adjacent brain appeared normal. Working diagnosis was thrombosed vein of Galen malformation. No ventriculomegaly or signs of encephalomalacia developed. **(Right)** Sagittal T1WI MR in the neonate confirms thrombosed vein of Galen malformation ➡ with no evidence of bleeding into, or ischemia of, the adjacent brain.*

Arteriovenous Fistula



Arteriovenous Fistula



(Left) *Sagittal T2WI MR shows a large, thrombosed interhemispheric AVF ➡ with associated ventriculomegaly ➡, porencephalic cyst ➡, and encephalomalacia ➡. **(Right)** Coronal T2WI MR shows a dilated ➡ sagittal sinus (mistaken for a midline cyst on ultrasound). Doppler showed arterialized venous flow as this is the draining vein of an AVF. Note associated ischemic cortical atrophy ➡.*

INTRACRANIAL CYSTS: LATERAL

DIFFERENTIAL DIAGNOSIS

Common
- Choroid Plexus Cyst
- Arachnoid Cyst

Less Common
- Schizencephaly
- Arteriovenous Fistula

Rare but Important
- Porencephaly

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Assessment of fluid-filled intracranial structure
- Could it be a prominent normal structure such as lateral ventricle?
  - Mild ventriculomegaly
    - Choroid displaced from medial wall
    - Atria of lateral ventricle measures 10-12 mm
    - Look for signs of trisomy 21
    - Look for additional brain abnormalities
  - Colpocephaly: Tear drop shape to dilated occipital horns seen in agenesis of corpus callosum
    - Ventricles parallel
    - Absent cavum septi pellucidi
    - Loss of normal branch pattern of anterior cerebral artery
    - Stenogyria
- Once abnormal lateral ventricle excluded, determine location
  - Is it in the choroid plexus?
  - Is it in the brain substance (i.e., intra-axial)?
  - Is it outside the brain substance (i.e., extra-axial)?
- Is it replacing normal brain tissue?
  - Porencephaly
  - Schizencephaly
- Is it a space-occupying lesion displacing brain tissue?
  - Arachnoid cyst
  - Arteriovenous fistula
- Is it vascular?
- Could it be a thrombosed vascular structure?
  - Hypoechoic structure with increased through transmission
  - Often contains faint low level echoes
  - Usually tubular or round

Helpful Clues for Common Diagnoses
- **Choroid Plexus Cyst**
  - Cyst > 2 mm with defined wall, within choroid plexus
  - Variable size
  - Variable number
  - May be unilateral or bilateral
  - May be single or multiple
  - Look for associated signs of trisomy 18
    - Abnormal posturing
    - Facial cleft
    - Congenital heart disease
    - Neural tube defects
    - Omphalocele
- **Arachnoid Cyst**
  - Extra-axial cyst displaces brain parenchyma
  - Unilocular
  - Avascular
  - Most common over convexities
    - ⅓ fetal cases are in posterior fossa
  - May enlarge and cause hydrocephalus due to mass effect at foramen of Monro or aqueduct of Sylvius

Helpful Clues for Less Common Diagnoses
- **Schizencephaly**
  - Wedge-shaped defect in brain parenchyma
  - Extends from lateral ventricle to inner table of skull
  - Edges of cleft are lined by gray matter
  - Unilateral or bilateral
  - Open lip or closed lip
    - Open lip: Wedge-shaped defect, edges separated by cerebrospinal fluid-filled cleft
    - Closed lip: Edges of defect are in contact; unlikely to be detected on prenatal ultrasound
  - Size varies from small to "giant"
  - Consider MR to look for associated heterotopia, polymicrogyria, pachygyria, septo-optic dysplasia
- **Arteriovenous Fistula**
  - Hypoechoic structure on grayscale images
    - Blood flow on Doppler evaluation when patent
    - Thrombosed AVF is tubular/round structure with low level internal echoes but no flow on Doppler evaluation

- MR will show blood product signal even when flow is absent
- May be intra-axial, but dural (i.e., extra-axial) more common in fetus
- May be associated with high output cardiac failure
- May be associated with polyhydramnios
- Look for associated intraventricular or parenchymal hemorrhage
- Look for associated ischemic damage secondary to vascular "steal"
  - Cortical thinning, ventriculomegaly, microcephaly
  - MR more sensitive than ultrasound for detection of encephalomalacia

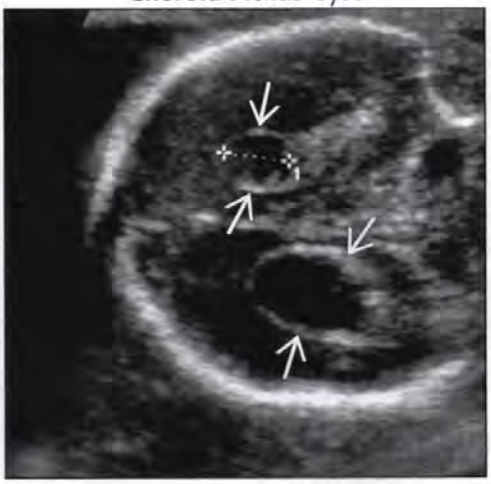
Helpful Clues for Rare Diagnoses

- **Porencephaly**
  - Porencephalic cyst replaces damaged brain
    - Intracerebral cavitation due to injury
    - Usually connected with lateral ventricle
  - Look for associated intracranial hemorrhage
  - Destructive process may present with mild ventriculomegaly and progress to encephalomalacia/porencephaly over time

Other Essential Information

- Prognosis is variable depending on underlying condition
  - Choroid plexus cysts
    - Seen in 1% of normal second trimester exams: No clinical significance if isolated finding
    - Seen in 50% of fetuses with trisomy 18: Usually associated with multiple other findings
  - Arachnoid cyst
    - Prognosis good if isolated
    - May require shunt or excision if large or causing obstructive hydrocephalus
  - Schizencephaly
    - Prognosis highly variable depending on size and bilaterality
    - Septo-optic dysplasia is common association: Visual impairment and hypothalamic/pituitary disturbance
    - Bilateral giant open lip schizencephaly → severe functional deficit ± epilepsy
  - Arteriovenous fistula
    - If brain is normal at birth, treatment recommended to prevent complications of hydrocephalus, progressive neurological deterioration, and acute bleed
    - Poor outcome if associated with ischemic encephalomalacia
    - Occasionally spontaneous thrombosis without associated brain injury
  - Porencephaly
    - Significant intellectual impairment, epilepsy, spasticity, blindness, depending on etiology and extent

Choroid Plexus Cyst



Coronal ultrasound shows bilateral choroid plexus cysts ➡. While most fetuses with choroid plexus cysts are normal, such cysts are a marker for trisomy 18 and a careful survey should be performed.

Choroid Plexus Cyst



3D ultrasound in the same case shows a consistently clenched fist with an overlapping second finger ➡ typical of trisomy 18. Multiple other anomalies were also present.

INTRACRANIAL CYSTS: LATERAL

Arachnoid Cyst

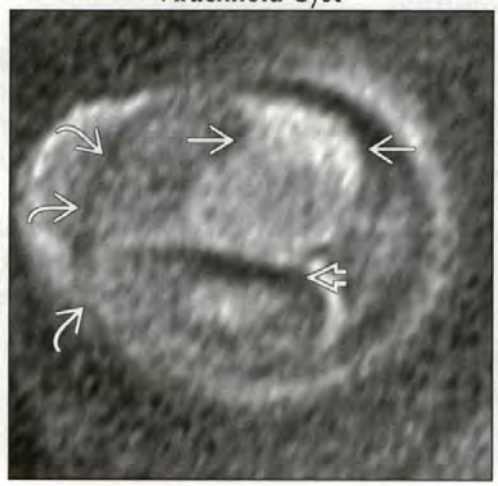


Arachnoid Cyst



(Left) *Coronal color Doppler ultrasound shows an extra-axial, simple, non-vascular cyst displacing otherwise normal brain parenchyma. This is typical of an arachnoid cyst.* ***(Right)*** *Coronal T2WI MR in the same case shows a CSF-intensity arachnoid cyst with mass effect causing elevation of the temporal lobe. Note the lesion is not destructive as there is no volume loss, and there is preservation of the hypointense cortical rim of gray matter.*

Arachnoid Cyst



Arachnoid Cyst

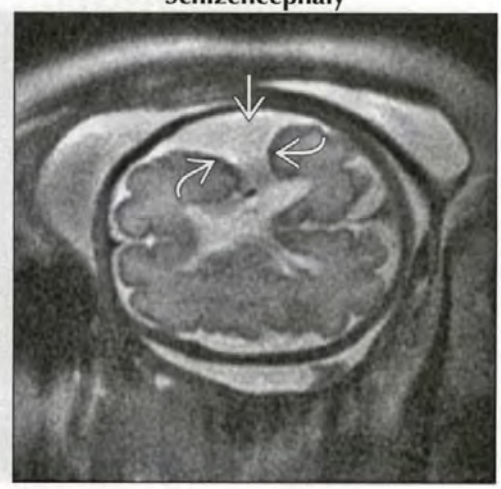


(Left) *Axial oblique T2WI MR shows a lateral arachnoid cyst in a fetus with semilobar holoprosencephaly and trisomy 13. The falx is partially present, but there is anterior fusion of the cerebral hemispheres.* ***(Right)*** *Axial ultrasound shows a posterior fossa arachnoid cyst, which could be confused with a mega cisterna magna. There was definite mass effect, which caused some remodeling of the cranium noted after delivery.*

Schizencephaly



Schizencephaly

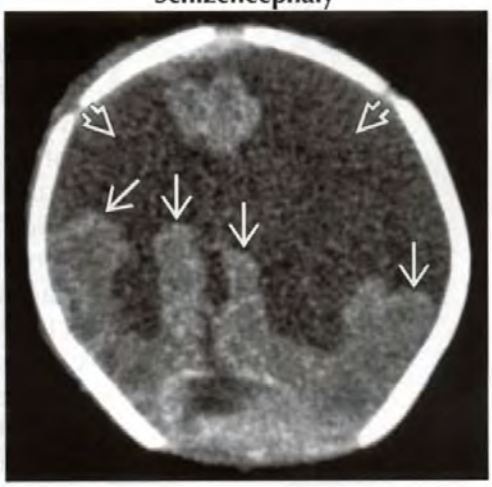


(Left) *Axial oblique ultrasound shows a wedge-shaped parenchymal defect in a fetus with an absent cavum septi pellucidi. This is typical of a small, open lip schizencephalic defect.* ***(Right)*** *Axial T2WI MR shows a small schizencephalic cleft on the right, with a CSF-filled space extending from the ventricle to the extra-axial space. Gray matter lines the cleft, differentiating this from porencephaly.*

Schizencephaly



Schizencephaly

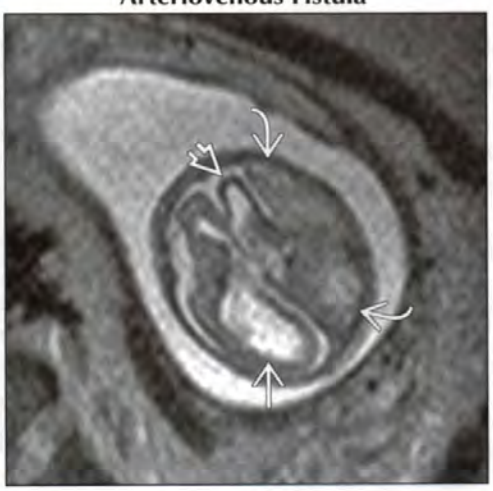


(Left) *Axial ultrasound shows a typical case of bilateral open lip schizencephaly with large bilateral clefts. The defects extend from the underlying ventricle to the inner table of the skull.* ***(Right)*** *Axial oblique NECT shows bilateral giant open lip schizencephaly with large clefts. Note intact gray matter lining the edges of the cleft.*

Arteriovenous Fistula



Arteriovenous Fistula

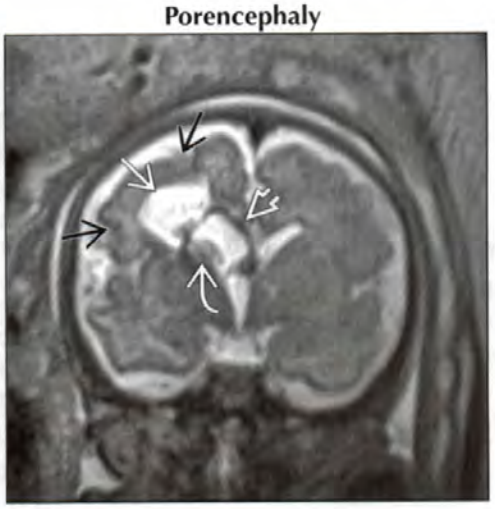


(Left) *Axial ultrasound shows ventriculomegaly and a hypoechoic structure with low-level internal echoes that was avascular on color Doppler.* ***(Right)*** *Axial T2WI MR in the same case confirms ventriculomegaly and shows mixed signal blood products within the thrombosed dural AVF. Note the extreme atrophy of the adjacent cerebral cortex. MR is superior to ultrasound for the detection of blood products.*

Porencephaly



Porencephaly



(Left) *Axial ultrasound shows an irregular porencephalic cyst extending from the lateral ventricle. Note the associated echogenic ependyma and intraventricular clot. This indicates a grade 4 intracranial hemorrhage.* ***(Right)*** *Coronal T2WI MR in the same patient shows a porencephalic cyst in communication with a dilated frontal horn. Note the associated intraventricular clot and cortical atrophy.*

ABNORMAL BRAIN PARENCHYMA

DIFFERENTIAL DIAGNOSIS

Common
- Exencephaly, Anencephaly
- Destructive Lesions
  - Intracranial Hemorrhage
  - Encephalomalacia
  - Cytomegalovirus
  - Toxoplasmosis

Less Common
- Schizencephaly
- Lissencephaly
- Gray Matter Heterotopia
- Pachygyria-Polymicrogyria
- Tuberous Sclerosis

Rare but Important
- CNS Tumors

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Define recognizable brain parenchymal structures
  - If no discernible brain structures or skull present, consider exencephaly or anencephaly
- Decide whether normal brain developed initially
  - Look for presence of basic structures: Gray/white matter, thalamus, ventricles/choroid, cerebellum/vermis, falx, corpus callosum
  - If suspect abnormal development, consider entities listed in less common diagnoses
  - If suspect normal development with later insult, consider destructive lesions
    - Characterize focal injury versus diffuse abnormality such as calcifications
- If intracranial mass present displacing brain parenchyma, consider CNS tumor
  - Use color Doppler to evaluate for vascularity

Helpful Clues for Common Diagnoses
- **Exencephaly, Anencephaly**
  - No calvarium and no recognizably normal neural tissue above orbits
  - Exencephaly
    - Dysmorphic neural tissue may be present initially
    - Usually "wears away" during gestation due to exposure to amniotic fluid and mechanical trauma
  - Anencephaly
    - No organized neural tissue remaining
  - Protuberant "frog-like" eyes due to shallow orbits
  - Polyhydramnios common due to impaired swallowing
  - Fluid often echogenic due to dissolved neural tissue
- **Intracranial Hemorrhage**
  - Echogenic intracranial "mass" without vascularity
  - Most commonly intraparenchymal
  - Use fetal MR to clarify anatomy and confirm location of clot(s)
    - If flow voids on T2WI, consider underlying vascular malformation
- **Encephalomalacia**
  - Result of various destructive brain processes
  - Sonographic findings often subtle
    - Look for associated ventriculomegaly as first clue
    - Periventricular lucencies due to cystic degeneration (later)
  - Porencephaly can occur due to parenchymal destruction
    - Intra-axial, avascular, round or irregular cavitary lesion without mass effect
- **Cytomegalovirus**
  - Widespread (non-shadowing) calcifications
  - Most commonly affects brain
    - Results in ventriculomegaly, microcephaly
  - Even in absence of sonographic findings, with known fetal infections, at least 20% have neurologic sequelae
- **Toxoplasmosis**
  - Non-shadowing intracranial and intrahepatic calcifications
    - Intracranial: Periventricular or random, may be very subtle and easily missed

Helpful Clues for Less Common Diagnoses
- **Schizencephaly**
  - CSF cleft extending from surface of brain to underlying ventricle
  - Open-lip is most common type identified in utero
  - Can be bilateral

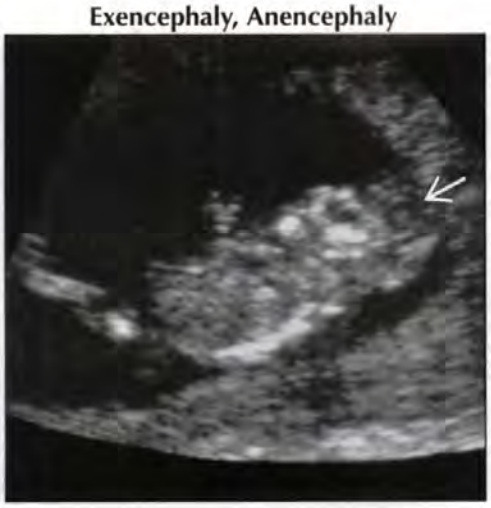
- Absent cavum septi pellucidi (CSP) in up to 70%
- Search for associated developmental brain anomalies

- **Lissencephaly**
  - Smooth contour to brain surface
    - Lack of normal gyral and sulcal development
  - Cannot assess until third trimester after sulcation begins
    - Sulcation normally starts around 23-24 weeks
    - Brain continues to appear "smooth" until approximately 26 weeks
  - Look specifically for shallow, poorly formed Sylvian fissures
  - May be linked to genetic defect or syndrome
    - Chromosome 17 mutation
    - X-linked lissencephaly - mothers have band heterotopia
    - Walker-Warburg syndrome
- **Gray Matter Heterotopia**
  - Gray matter cells in an abnormal location
    - Due to arrested neuronal migration to cortex
  - Subependymal: Nodules of gray matter along ventricular wall
  - Subcortical: Heterogeneous masses in subcortical location
  - Band: Homogeneous bands of gray matter between lateral ventricles and cortex
- **Pachygyria-Polymicrogyria**
  - Pachygyria
    - Much more common than complete lissencephaly
    - Can be focal or diffuse
    - Decreased sulcation (incomplete lissencephaly)
  - Polymicrogyria sometimes referred to as cortical dysplasia
    - Disorder of neuronal organization
    - Irregular cortical surface
    - Can appear flat and smooth due to innumerable microgyri
    - Seen with in utero ischemia, chromosomal abnormalities, congenital CMV
- **Tuberous Sclerosis**
  - Subependymal echogenic tubers can be identified along wall of ventricles
  - Subependymal giant cell astrocytoma near foramen of Monro
  - Cardiac rhabdomyoma(s) may be present
  - CNS findings often better visualized with fetal MR

Helpful Clues for Rare Diagnoses

- **CNS Tumors**
  - Solid intracranial mass with Doppler flow
    - Most are supratentorial
    - May extend through skull base into oral cavity
    - Size can be massive with rapid growth and hemorrhage
  - Significant overlap in appearance of tumors types

Exencephaly, Anencephaly



Sagittal transabdominal ultrasound at 12.5 weeks shows a lack of calvarium and exposed neural tissue ➡. No visible skull was identified.

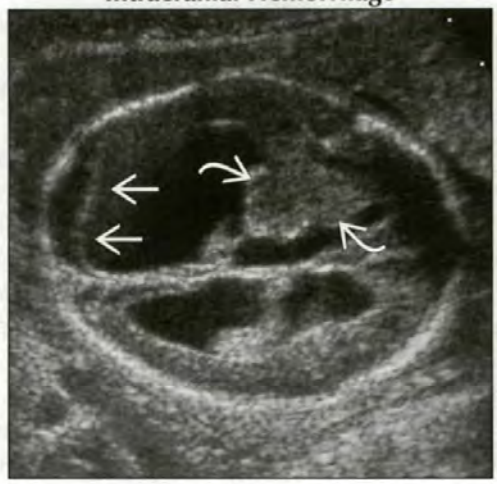
Exencephaly, Anencephaly



Coronal ultrasound shows some minimal residual neural tissue of a different fetus at 15 weeks ➡. Note the typical "frog-like" appearance of the face.

ABNORMAL BRAIN PARENCHYMA

Intracranial Hemorrhage



Encephalomalacia



(Left) *Axial transabdominal ultrasound of a fetus with trisomy 21 shows asymmetric dilatation of the ventricles with intraventricular clot and encephalomalacia. Etiology of the bleed was never identified.* ***(Right)*** *Axial transabdominal ultrasound shows asymmetric ventriculomegaly and porencephaly in an area of previously seen intracranial hemorrhage.*

Cytomegalovirus

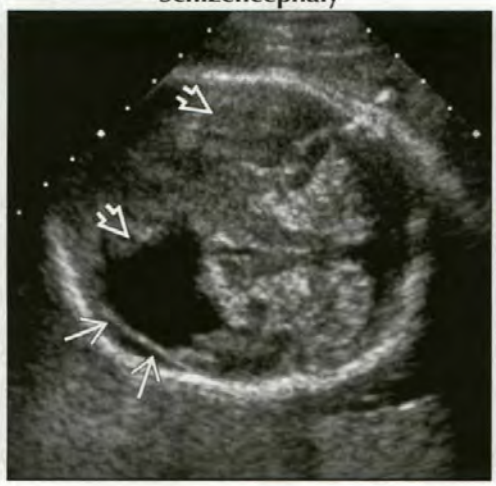


Toxoplasmosis



(Left) *Axial NECT in an infant with congenital cytomegalovirus shows periventricular calcifications. There is also cerebellar hypoplasia, creating an unusual appearance of the posterior fossa.* ***(Right)*** *Axial ultrasound shows periventricular and intraparenchymal calcifications. These findings are subtle and could be easily missed without clinical history of exposure. Findings were confirmed postnatally.*

Schizencephaly



Schizencephaly



(Left) *Axial ultrasound shows bilateral schizencephaly, with clefts extending from the underlying ventricle to the skull (up side more difficult to see secondary to reverberation artifact). A thin "roofing" membrane is also seen adjacent to the inner table of the skull.* ***(Right)*** *Coronal T2WI MR in the same case as previous image confirms bilateral clefts.*

Lissencephaly

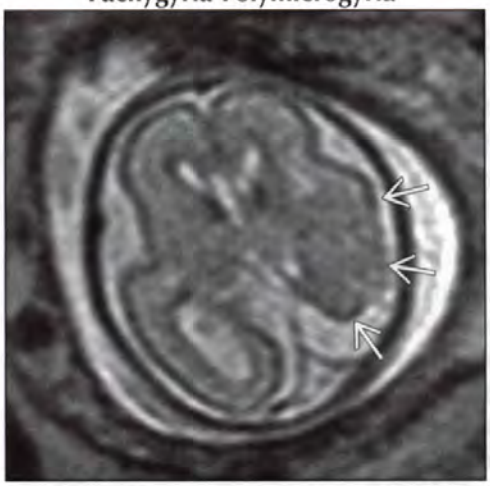


Gray Matter Heterotopia

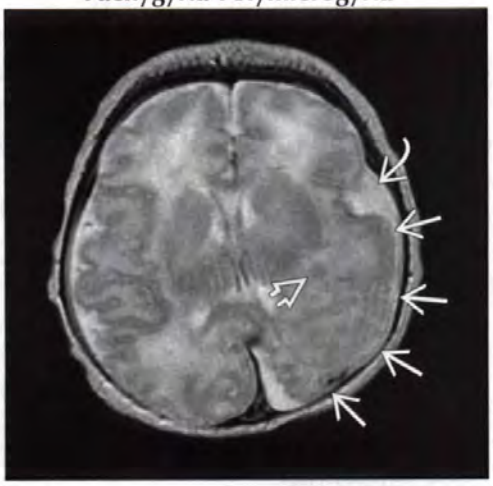


(Left) *Coronal T2WI MR in a third trimester fetus shows a smooth brain with lack of normal sulcation. Specifically note the shallow, poorly formed sylvian fissures.* ***(Right)*** *Coronal T2WI MR at 23.5 weeks shows asymmetric low signal gray matter adjacent to the left ventricle wall, consistent with subependymal gray matter heterotopia. Further posteriorly, a schizencephaly cleft was identified.*

Pachygyria-Polymicrogyria



Pachygyria-Polymicrogyria



(Left) *Axial T2WI MR in a fetus with an abnormal occipital lobe on ultrasound shows abnormal gray matter in the left occipital region even at 21 weeks.* ***(Right)*** *Axial T2WI MR postnatally shows a smooth cortical surface and irregular cortical organization on the left, most likely polymicrogyria. There is a focus of nodular heterotopia in the same hemisphere. Note the widened shallow sylvian fissure.*

Tuberous Sclerosis



CNS Tumors



(Left) *Axial T2WI MR shows two small hypointense subependymal nodules in a fetus with suspected tuberous sclerosis. Postnatal MR confirmed the diagnosis. These findings are subtle and often missed on prenatal ultrasound. Fetal MR is suggested in all suspected cases of tuberous sclerosis.* ***(Right)*** *Axial ultrasound shows a large, heterogeneously echogenic, midline tumor. Histology confirmed a teratoma.*

ABSENT CALVARIUM

DIFFERENTIAL DIAGNOSIS

Common
- Exencephaly, Anencephaly
- Acrania

Less Common
- Amniotic Band Syndrome
- Skeletal Dysplasia, Mimic
  - Osteogenesis Imperfecta
  - Hypophosphatasia
  - Achondrogenesis

Rare but Important
- Twin Reversed Arterial Perfusion (TRAP)

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is it a single or multiple gestation?
  - TRAP only possible in monochorionic twin or higher order multiple
- Is there a head?
  - If not, consider TRAP sequence in a twin pregnancy
  - Possible bands in singleton or a twin
- Is the cranial vault present?
  - If absent, differential diagnosis is between amniotic bands and anencephaly
  - If present, then differential diagnosis is between the mimics
    - Measure all long bones
    - Look for fractures
    - Assess compressibility of cranial vault
- Is there formed brain?
  - In anencephaly, there is a gelatinous mass of angiomatous stroma in the skull base
  - In exencephaly and acrania, a fully formed brain may be seen particularly early in pregnancy before repeated episodes of trauma destroy it
- Is the maternal serum alpha fetoprotein (msAFP) elevated?
  - Increased msAFP associated with open neural tube defect
- Is the fetus mobile within the uterine cavity?
  - If fetus seems tethered at one location, look carefully for amniotic bands
    - Change maternal position to try to float fetus away from uterine wall
    - Look for liner echoes attaching fetus or externalized organs to uterine wall

Helpful Clues for Common Diagnoses
- **Exencephaly, Anencephaly**
  - Brain present but not contained within skull vault
    - Brain contour lobulated ("Mickey Mouse") or spiked ("Bart Simpson")
  - In first trimester, may present with short crown rump length
  - Brain subject to trauma from fetal movement, abrasion against uterine wall, etc.
    - By third trimester may no longer see recognizable brain due to liquefaction
    - Look for echogenic debris in amniotic fluid
  - Orbits prominent → "frog eye" appearance
  - Often associated with cervical/lumbar spine defects
  - Polyhydramnios common
- **Acrania**
  - Absent calvarium above orbits
  - Occiput ossifies in cartilage; therefore, skull base intact
  - Meninges may be present
    - May provide protective barrier from erosion

Helpful Clues for Less Common Diagnoses
- **Amniotic Band Syndrome**
  - Multiple defects
  - Look for amputation/constriction defects in extremities
  - Look for asymmetric facial cleft
  - Look for missing orbit in association with "anencephalic" appearance
  - Key to establishing the diagnosis is demonstration of bands
    - Fine linear structures in amniotic fluid
    - Band often "tethers" fetus to uterine wall
    - Change maternal position to float fetus away from uterine wall
- **Skeletal Dysplasia, Mimic**
  - Calvarium is present but poorly ossified
  - Brain is seen "too easily"
  - Brain morphology is normal
    - Even though ossification is abnormal the brain is protected from erosion/trauma
  - Calvarium can be deformed by transducer pressure
  - **Osteogenesis Imperfecta**
    - Multiple fractures
    - Beaded ribs

- Short extremity bones
- "Thick" bones
- **Hypophosphatasia**
- Thin, bowed long bones
- **Achondrogenesis**
- Type I associated with poor mineralization of skull and completely unossified spine
- Fractures of ribs but not long bones seen in type Ia
- Spine ossification may be seen in pedicles in type Ib
- Type II has normal ossification of the skull but deficient ossification of spine

Helpful Clues for Rare Diagnoses

- **Twin Reversed Arterial Perfusion (TRAP)**
- Multiple gestation
- Monochorionic placentation
- Cranium often completely absent
- Upper extremities often poorly developed
- Even in well-developed TRAP fetus the brain is abnormal, skull vault may be present but surrounded by marked soft tissue edema
- Hallmark of this diagnosis is reversed flow in the umbilical artery
- Flow is away from placenta toward the TRAP fetus

Other Essential Information

- Prognosis is lethal if calvarium truly absent
- Anencephaly
- Most die within hours of birth, < 10% live to one week
- Prevention in future pregnancies possible by oral folate supplementation
- Amniotic band syndrome
- No significant recurrence risk
- Lethal if associated with acalvaria, major abdominoschisis
- Minor constriction defects are cosmetic only with normal life expectancy
- Skeletal dysplasias mimic absent calvarium due to poor mineralization
- Skeletal dysplasias with poor skull mineralization often carry poor prognosis
- Osteogenesis Imperfecta: Type II is perinatal lethal, other forms have variable prognosis
- Hypophosphatasia: Autosomal recessive in perinatal lethal form
- Achondrogenesis Ia/Ib: Autosomal recessive, type II is sporadic
- TRAP sequence
- TRAP fetus will not survive
- Pump twin at risk for high output failure
- Intervention indicated to prevent hydrops
- Radiofrequency ablation of TRAP twin cord now procedure of choice
- Use 3D ultrasound
- Useful in patient counseling
- Global view of extremity/head/chest/torso proportions

Exencephaly, Anencephaly



Coronal ultrasound image of the fetal face shows prominent, widely spaced orbits ➡. There is no neural tissue or cranium above the orbital ridge. This appearance has been described as "frog-like".

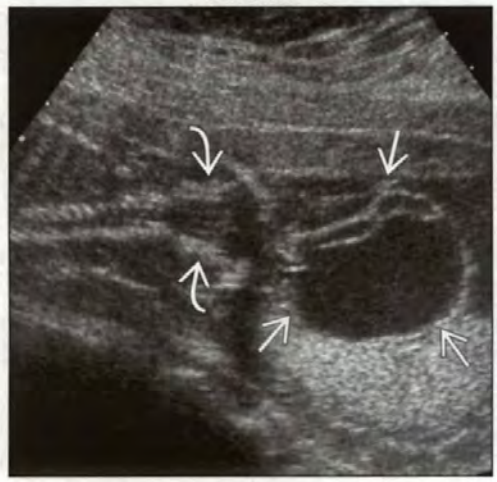
Exencephaly, Anencephaly

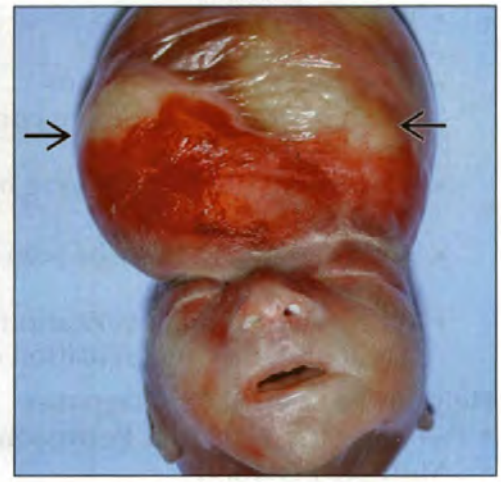


Clinical photograph taken at autopsy confirms the proptotic appearance of the eyes ➡ and the lack of a cranial vault and contents above the orbital ridge, typical of anencephaly.

ABSENT CALVARIUM

Acrania





(Left) *Coronal ultrasound shows brain tissue covered in a sac ➔ without an overlying skull. Note the splaying of the cervical spine ➔. Associated neural tube defects are common.* ***(Right)*** *Gross pathology in the same case of acrania shows some recognizable brain parenchyma with two hemispheres ➔ seen within the meningeal sac.*

Amniotic Band Syndrome





(Left) *Radiograph performed at autopsy in a case of amniotic band syndrome shows the ossified skull base ➔ and the amorphous mass of exteriorized brain ➔ with adherent amniotic band ➔.* ***(Right)*** *Gross pathology from a different case shows multiple anomalies caused by amniotic band syndrome. Note the large abdominal defect with herniation of bowel and liver ➔, exposed neural tissue ➔, large diagonal cleft lip ➔, and multiple extremity amputations ➔.*

Osteogenesis Imperfecta





(Left) *Axial ultrasound shows the malleable head shape ➔ of a fetus with osteogenesis imperfecta. The poorly ossified bone does not reflect much of the ultrasound beam; therefore, brain detail is remarkably well seen for the third trimester. The fact that the brain is normal indicates a mimic, rather than a truly absent calvarium.* ***(Right)*** *Lateral radiograph of the skull shows poorly ossified skull vault with wormian bones ➔ typical of osteogenesis imperfecta.*

ABSENT CALVARIUM

Hypophosphatasia



Hypophosphatasia



(Left) *Axial ultrasound shows excellent brain detail and poor reflection ➡ from the near field skull vault, due to under mineralization in a case of hypophosphatasia. The far field skull vault does produce an echo ➡, proving that it is present. Therefore, this is a mimic of absent calvarium.* ***(Right)*** *Frontal radiograph of the infant after birth confirms the presence of an intact, albeit poorly ossified, bony cranium ➡.*

Achondrogenesis



Achondrogenesis



(Left) *Axial ultrasound shows a thin skull vault due to achondrogenesis. Note that the far field echo is more convincing for an intact skull vault and the brain appears normal. With absence of the calvarium, repeated episodes of trauma from normal fetal movement cause loss of normal brain architecture.* ***(Right)*** *Gross pathology shows the extreme malleability of the skull. Deformity of the head ➡ was due to vaginal birth.*

Twin Reversed Arterial Perfusion (TRAP)



Twin Reversed Arterial Perfusion (TRAP)



(Left) *Ultrasound shows a relatively well-formed "acardiac" twin with a spine ➡, ribs, and lower extremities ➡ but no head. Note extensive soft tissue edema ➡ typical of the TRAP sequence.* ***(Right)*** *Gross pathology shows the defects ➡ left in the TRAP twin after radiofrequency ablation of the cord. Note the complete absence of a cranium ➡. The pump twin developed normally following intervention and was delivered at term.*

ABNORMAL CALVARIUM

DIFFERENTIAL DIAGNOSIS

Common

- Abnormal Shape
  - Poor Scan Technique
  - Dolichocephaly
  - Brachycephaly
  - "Lemon-Shaped"
  - "Strawberry-Shaped"
  - Round
  - Spaulding Sign
  - Craniosynostosis
- Calvarial Defect
  - Exencephaly, Anencephaly
  - Encephalocele
  - Amniotic Band Syndrome
- Abnormal Size
  - Macrocephaly
  - Microcephaly

Less Common

- Decreased Ossification
  - Osteogenesis Imperfecta
  - Achondrogenesis
  - Hypophosphatasia
- Scalp Masses

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Assess calvarial size, shape, and mineralization in all cases
  - Size
    - Is size concordant with gestational age and other biometric parameters?
  - Shape
    - Can you see standard scan plane anatomy?
    - If not, is it because of fetal position or maternal habitus?
    - Use transvaginal sonography for better resolution
    - 3D ultrasound allows volume acquisition
    - Data manipulation allows reproduction of true axial plane
  - Mineralization
    - Skull is formed after 10 weeks; use EV sonography from 10-14 weeks for better resolution if questions
    - If brain seen "too well" consider conditions with poor mineralization
    - Transducer pressure cannot deform a normally ossified cranium
- Is there a bony defect?
  - Essential to look at skull vault from several scan planes
  - Refraction of beam may create an apparent defect where none exists
    - Cystic hygroma may be mistaken for an occipital encephalocele
  - Must know normal anatomy: Do not mistake metopic suture for frontal encephalocele

Helpful Clues for Common Diagnoses

- **Poor Scan Technique**
  - Make sure thalami and cavum septi pellucidi are visible
- **Dolichocephaly**
  - Boat shaped: Long back-to-front, narrow side-to-side
    - Seen with breech presentation, oligohydramnios, myelomeningocele
- **Brachycephaly**
  - Short back-to-front, wide side-to-side
  - Described in trisomy 21
- **"Lemon-Shaped"**
  - Bifrontal concavity seen with Chiari II malformation
    - Resolves in third trimester in all cases
  - Occurs in various other conditions and 1% of normal fetuses
- **"Strawberry-Shaped"**
  - Triangular configuration described in trisomy 18
  - Most fetuses with trisomy 18 have multiple other anomalies
- **Round**
  - May be technical if measurement obtained in wrong scan plane
  - If normal anatomic markers are not identified and head shape appears round from multiple acoustic windows, underlying brain is usually abnormal
    - Look carefully for signs of aprosencephaly/holoprosencephaly spectrum
- **Spaulding Sign**
  - Bones of skull vault overlap as brain collapses following demise
- **Craniosynostosis**
  - Abnormal head shape secondary to premature closure of sutures

- Look for features of associated conditions (e.g., Crouzon, Pfeiffer, Apert, skeletal dysplasia)

- **Exencephaly, Anencephaly**
  - Exencephaly: Lack of cranial vault but brain tissue present
  - Anencephaly: Cranial vault absent, no brain tissue, skull base contains gelatinous angiomatous stroma
- **Encephalocele**
  - Occipital: Herniation of intracranial structures through an occipital defect
    - Look for other anomalies/signs of aneuploidy
  - Frontal: Herniation of intracranial structures through an anterior skull defect
    - Look for associated hypertelorism, callosal dysgenesis, midline lipoma
- **Amniotic Band Syndrome**
  - Look for linear echoes from bands in amniotic fluid
  - Look for associated extremity amputation or constriction defects
  - "Anencephaly" with asymmetric orbits or facial cleft ⇒ bands highly likely
- **Macrocephaly**
  - Enlarged head: Biparietal diameter (± head circumference) > 2 SD above mean
  - Look for underlying abnormalities (e.g., hydrocephalus, tumor, megalencephaly)
- **Microcephaly**
  - Small head: Biparietal diameter (± head circumference) > 2 SD below mean
  - Seen with infection, ischemia, syndromes, malformations

Helpful Clues for Less Common Diagnoses

- **Osteogenesis Imperfecta**
  - Associated with fractures in long bones, beaded ribs
- **Achondrogenesis**
  - Hallmark is lack of vertebral ossification
- **Hypophosphatasia**
  - Associated with micromelia and thin, bowed bones in perinatal lethal form
- **Scalp Masses**
  - Calvarium normal
  - Mass (e.g., lymphangioma, hemangioma) arises from scalp

Other Essential Information

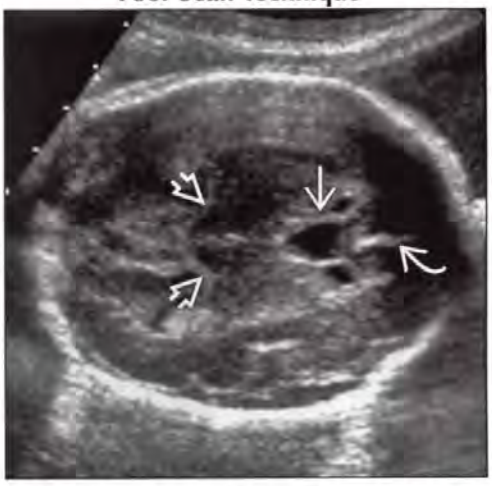
- Technique very important in head measurement and evaluation of calvarial contour
- Biparietal diameter (BPD)
  - Measure at level of thalami and cavum septi pellucidi
  - Cerebellar hemispheres should not be visible
  - Midline echoes in center of oval-shaped cross-section
  - Measure outer edge proximal skull to inner edge distal skull
- Head circumference: Measure at outer edge of skull in same plane as BPD

Poor Scan Technique



Axial ultrasound shows an odd calvarial contour because the orbits ➡ are included in the scan plane, which is incorrect. The thalami and cavum are not well seen.

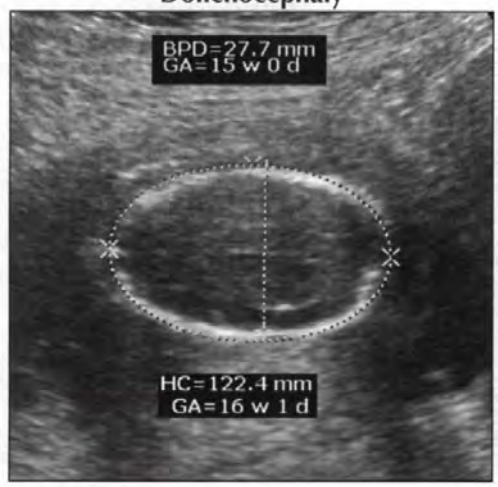
Poor Scan Technique



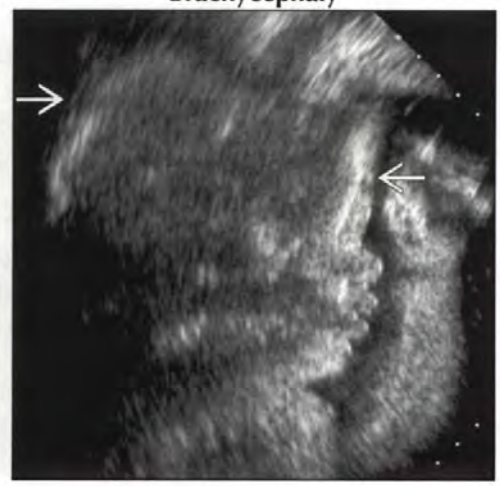
Axial oblique ultrasound for comparison shows the correct scan plane with the cavum ➡, thalami ➡, and central midline echo ➡ all well seen.

ABNORMAL CALVARIUM

Dolichocephaly

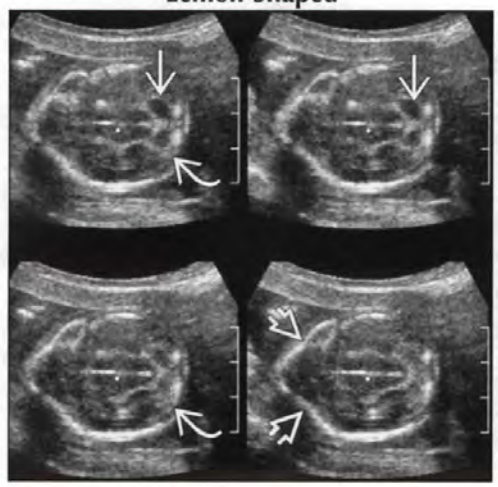


Brachycephaly



(Left) *Axial ultrasound shows a dolichocephalic head shape in an extremely obese patient. Note the elongated shape; the BPD, therefore, measures less than expected. The head circumference measurement is more accurate for dating if the head has a dolichocephalic shape.* ***(Right)*** *Sagittal ultrasound shows brachycephaly (i.e., shortening in the AP plane ➡) in a fetus with trisomy 21.*

"Lemon-Shaped"



"Lemon-Shaped"

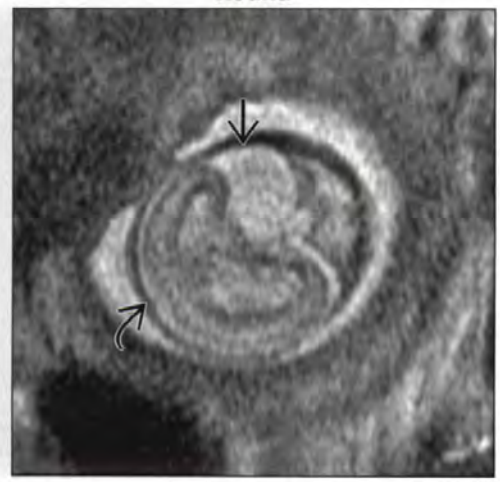


(Left) *3D ultrasound, shown as a series of axial "slices" akin to CT or MR, illustrates the "lemon" head ➡ and the typical associated "banana" cerebellum ➡, both of which are signs of Chiari II malformation. The cisterna magna ➡ is obliterated.* ***(Right)*** *Axial ultrasound shows bifrontal concavity ➡ in this normal fetus. When seen, this sign should prompt careful evaluation; however, it is not pathognomic for the Chiari malformation.*

"Strawberry-Shaped"



Round



(Left) *Axial ultrasound shows the more triangular "strawberry" shape with short AP ➡ diameter and loss of the expected oval shape. Multiple anomalies prompted amniocentesis which revealed trisomy 18.* ***(Right)*** *Axial T2WI MR shows an extra-axial cyst ➡ displacing brain tissue. The frontal lobes are fused across the midline ➡. This fetus had semilobar holoprosencephaly with an arachnoid cyst. The head shape was round in all scan planes.*

ABNORMAL CALVARIUM

Spaulding Sign



Craniosynostosis



(Left) *Axial ultrasound shows discontinuity of the vault and overlap ➡ of the skull bones in a case of intrauterine fetal demise.* ***(Right)*** *Axial ultrasound shows an abnormal calvarial shape with a narrow frontal area ➡ and wider parietal area ➡ in a fetus ultimately diagnosed with Pfeiffer syndrome.*

Exencephaly, Anencephaly



Encephalocele



(Left) *Coronal ultrasound shows the typical "frog-eyed" appearance ➡ in anencephaly due to absence of the skull vault above the orbital ridge.* ***(Right)*** *Sagittal T2WI MR shows a calvarial defect ➡ and large occipital encephalocele ➡. Note the small head size ➡. This is caused by the large amount of brain parenchyma in the encephalocele sac.*

Encephalocele



Amniotic Band Syndrome



(Left) *Coronal ultrasound shows a soft tissue mass ➡ between the orbits (calipers), a typical location for a frontal encephalocele. There was associated hypertelorism.* ***(Right)*** *Lateral radiograph as part of an autopsy shows the facial bones ➡ and occiput ➡ with a mass of amorphous exteriorized brain ➡ to which bands were adherent.*

ABNORMAL CALVARIUM

Macrocephaly



Macrocephaly

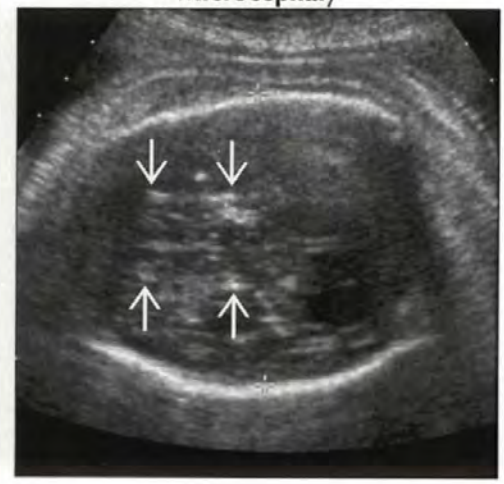


(Left) *Sagittal T2WI MR shows head enlargement secondary to agenesis of the corpus callosum and a large interhemispheric cyst. Note how disproportionately large the cranium appears. The fetus also had hemifacial microsomia and left anophthalmia* ⇨. ***(Right)*** *Axial ultrasound shows an enlarged biparietal diameter (calipers) of 11.5 cm in a fetus with aqueductal stenosis. Cesarean delivery was required.*

Microcephaly

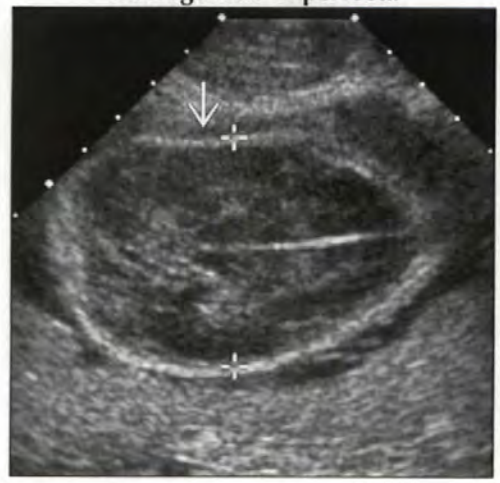


Microcephaly

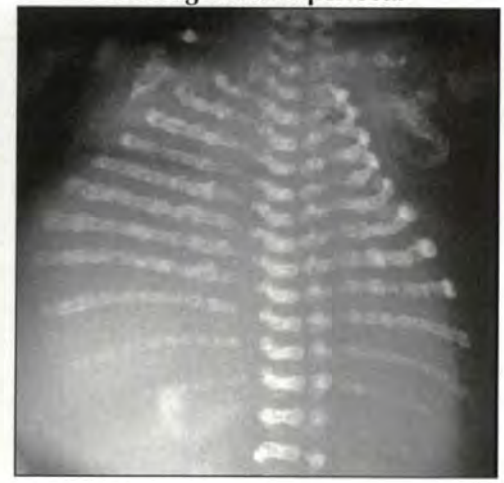


(Left) *Table shows a severely diminished head size in a fetus with an early scan that confirmed menstrual dating. Note that the head measurements lag behind the femur length and abdominal circumference.* ***(Right)*** *Axial ultrasound in the same case shows loss of normal grey white matter differentiation and multiple punctate calcifications* ➡. *The infant died within hours of delivery, and the family declined autopsy. Infection was considered the most likely cause.*

Osteogenesis Imperfecta



Osteogenesis Imperfecta



(Left) *Axial ultrasound shows deformation of the skull* ➡ *by transducer pressure. The abnormal ossification accounts for the compressibility.* ***(Right)*** *Radiograph of a stillborn shows multiple rib fractures as seen in osteogenesis imperfecta.*

Achondrogenesis

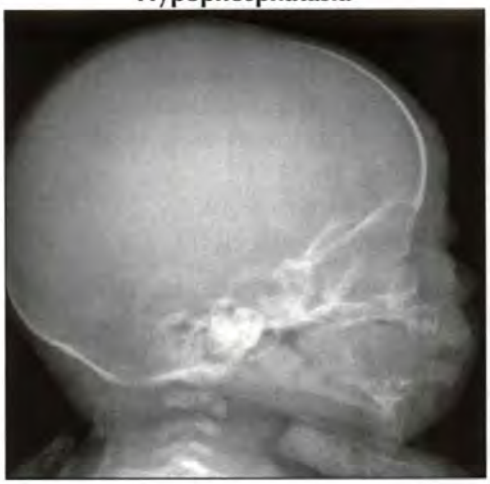


Achondrogenesis



(Left) *Axial oblique ultrasound shows that the brain detail is remarkably well seen and that the skull vault ➡ can be compressed by transducer pressure.* ***(Right)*** *Coronal ultrasound in the same case shows that the chest ➡ is small in relation to the abdomen ➡. There was poor vertebral body ossification. Autopsy confirmed achondrogenesis type 1a.*

Hypophosphatasia



Hypophosphatasia

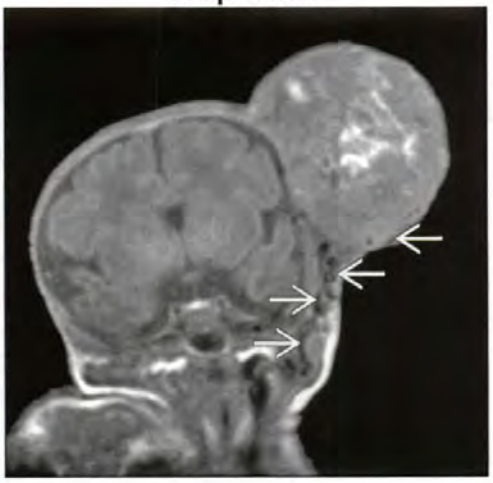


(Left) *Radiograph shows marked reduction in mineralization in this neonate with hypophosphatasia.* ***(Right)*** *Ultrasound shows a short curved femur in a fetus with hypophosphatasia. A poorly ossified skull should prompt careful evaluation of the long bones.*

Scalp Masses



Scalp Masses



(Left) *Axial transabdominal ultrasound shows a solid scalp mass ➡ with a few small cystic spaces ➡. There was no calvarial defect or underlying brain abnormality. The mass was resected after delivery and shown to be a hemangioma.* ***(Right)*** *Coronal T1WI MR shows another scalp hemangioma in a newborn. There are large vessels seen as flow voids ➡, feeding this scalp mass. The brain anatomy is normal.*

MICROCEPHALY

DIFFERENTIAL DIAGNOSIS

Common
- Idiopathic
- Symmetric IUGR
- Exencephaly, Anencephaly

Less Common
- Encephalocele
- Atelencephaly, Aprosencephaly
- Destructive Processes

Rare but Important
- Syndromes
  - Cornelia de Lange Syndrome
  - Neu Laxová Syndrome
- Teratogens
  - Fetal Alcohol Syndrome
  - Hydantoin Syndrome
  - Valproic Acid

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Ensure that measurements are obtained correctly
- Repeat measurement for confirmation
- Look at parents and consider measuring parental head circumference
- Is the whole fetus small or just the head?
  - If all measurements are small consider incorrect dates versus early onset growth restriction
  - If head measurements smaller than other parameters, true microcephaly more likely
- Is there a cranial vault?
- Is there a defect in the vault?
- Is the face normal?
  - Atelencephaly/aprosencephaly strongly associated with abnormal facies
- Are there calcifications?
  - Good predictor of infection but may be subtle: Small, non-shadowing
  - Use transvaginal scans for improved resolution if fetus in cephalic presentation
- Is there evidence of bleeding?
  - Look for echogenic clot in ventricles, nodular ependymal thickening, porencephaly
- Is the maternal serum alpha fetoprotein elevated?
  - Encephalocele, anencephaly, exencephaly most likely

Helpful Clues for Common Diagnoses
- **Idiopathic**
  - Small head size with normal interval growth
  - Structurally normal fetus
  - Structurally normal brain
- **Symmetric IUGR**
  - Size less than expected for dates
  - All biometric parameters affected
  - Often early onset
  - More likely due to intrinsic fetal abnormality than placental insufficiency
  - Look for signs of aneuploidy/syndromes
- **Exencephaly, Anencephaly**
  - Cranial vault absent
  - Variable amounts of brain tissue present
  - Short crown rump length in first trimester
  - In exencephaly the externalized brain may confer spiky or lobulated contour to head
  - "Frog eye" appearance of large, shallow orbits and absent vault above orbital ridge
  - Look for amniotic bands as cause
    - Linear echoes in amniotic fluid
    - Constriction/amputation defects in extremities
    - "Slash" defects elsewhere (e.g., abdominoschisis, facial cleft)

Helpful Clues for Less Common Diagnoses
- **Encephalocele**
  - If large amount of brain parenchyma in encephalocele, skull vault is small
    - Microcephaly in 25% of occipital encephaloceles
  - Diverse appearance of herniated tissue
    - Gyral pattern may be identified
  - Look for osseous defect
  - Look for associated anomalies for syndromic diagnosis
    - Meckel Gruber syndrome associated with abnormal kidneys/polydactyly
- **Atelencephaly, Aprosencephaly**
  - Developmental arrest of formation of telencephalon &/or prosencephalon
  - No normal cerebral structures
  - Cerebellum often hypoplastic
  - Facial anomalies, often severe
    - Absent eyes/nasal structures, midline oculofacial defects including cyclopia
  - Radial ray anomalies including absent thumbs

- Oligodactyly, camptodactyly, clinodactyly, clubfoot
- **Destructive Processes**
  - Look for calcifications in TORCH infections
  - Infarction/hemorrhage/ischemic "steal" phenomena in arteriovenous shunts
    - Use Doppler to evaluate all "cystic" structures for flow
  - MR helpful to show blood products
  - MR helpful to demonstrate encephalomalacia, porencephalic cysts

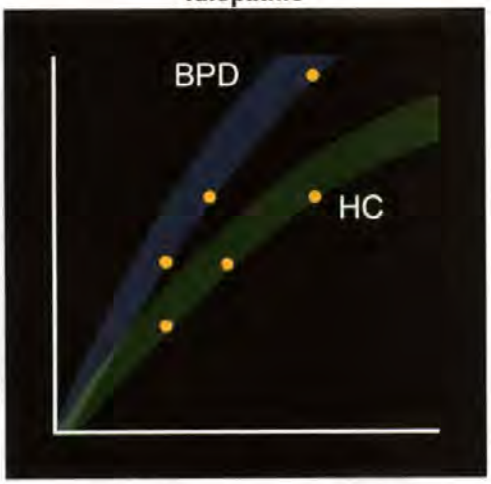
Helpful Clues for Rare Diagnoses

- **Cornelia de Lange Syndrome**
  - Microcephaly
  - Micrognathia with protruding upper lip
  - Upper limb reduction defects
  - Intrauterine growth restriction (IUGR)
  - Diaphragmatic hernia
  - Cardiac defects: Pulmonary stenosis, ventricular septal defect
- **Neu Laxová Syndrome**
  - Microcephaly
  - Receding forehead
  - Proptosis
  - IUGR
  - Central nervous system malformations
  - Limb contractures
  - Abnormal genitalia
- **Fetal Alcohol Syndrome**
  - IUGR
  - Cardiac defects
  - Short palpebral fissures, smooth philtrum, thin upper lip
- **Hydantoin Syndrome**
  - IUGR
  - Cardiac defects
- **Valproic Acid**
  - IUGR
  - Neural tube defects
  - Radial ray malformation

Other Essential Information

- Prognosis for true microcephaly is poor
  - 67% fetuses with microcephaly have other brain anomalies
  - Non-CNS anomalies also common
  - Infant HC 2-3 SD below mean: 33% moderate to severe intellectual impairment
  - Infant HC > 3 SD below mean: 62% moderate to severe intellectual impairment
- Exencephaly, anencephaly, aprosencephaly, atelencephaly are lethal
  - Folate supplementation decreases anencephaly risk in future pregnancies
  - Amniotic band syndrome is sporadic, unlikely to recur
- Prognosis in encephalocele varies with amount of herniated brain and associated abnormalities
  - Operative approach is resection of extruded brain and closure of defect

Idiopathic



Graphic shows appropriate interval growth with BPD and HC measurements at the 5th percentile for GA. The infant was normal at birth.

Symmetric IUGR

LMP =	MA = 32w3d
MA = 28w5d±13d	
	MA ±SD
BPD	29w1d±15d
HC	29w6d±14d
AC	27w1d±15d
FL	29w2d±15d
LMP%	<10%
AFI	5.66cm

Table shows growth restriction and oligohydramnios in a fetus with gastroschisis and confirmed menstrual dating. Poor biophysical scores precipitated delivery, and the infant had a stormy post-operative course.

MICROCEPHALY

Exencephaly, Anencephaly



Encephalocele



(Left) *Sagittal ultrasound ultrasound image shows lack of a bony calvarium superior to the skull base. The skull base ossifies in cartilage independent of the skull vault, which ossifies in membrane.* ***(Right)*** *Sagittal transvaginal ultrasound shows a huge encephalocele containing most of the cerebral hemispheres. The skull vault is very small. The infant did not survive.*

Encephalocele

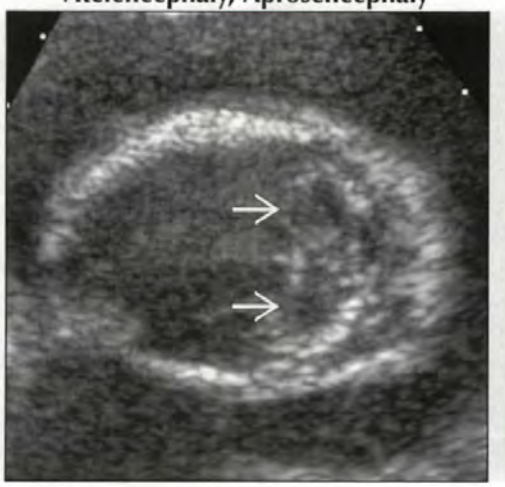


Encephalocele

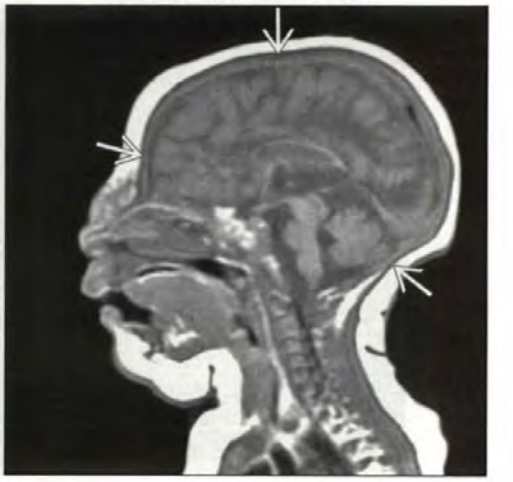


(Left) *Sagittal T2WI MR shows a large amount of brain in the encephalocele sac. The head is small, and the forehead shows the typical sloped configuration seen in microcephaly.* ***(Right)*** *Gross pathology shows the same case at autopsy. Note the small head, sloped forehead, and large encephalocele.*

Atelencephaly, Aprosencephaly



Destructive Processes



(Left) *Axial oblique ultrasound shows a normal bilobed cerebellum but no identifiable structures above the tentorium. Autopsy diagnosis was atelencephaly. There were no orbits, nasal structures, mouth, or cerebral hemispheres.* ***(Right)*** *Sagittal T1WI MR in a severely impaired survivor of twin-twin transfusion and co-twin demise shows a small calvarium due to ischemic periventricular leukomalacia.*

Destructive Processes

LMP = MA = 36w1d

MA = 30w0d±17d

	MA	±SD
BPD	26w2d	±15d
HC	27w0d	±14d
AC	33w0d	±21d
FL	34w3d	±21d

Destructive Processes



(Left) *Table shows biometric data in a fetus referred for evaluation of IUGR. Clearly head measurements lag far behind the AC and FL, concerning for microcephaly.* ***(Right)*** *Sagittal ultrasound of the same case shows loss of normal brain architecture and multiple intracranial calcifications. Note skin thickening secondary to hydrops. The infant died within hours of birth, and the family declined autopsy. Infection is considered the most likely diagnosis.*

Cornelia de Lange Syndrome



Cornelia de Lange Syndrome

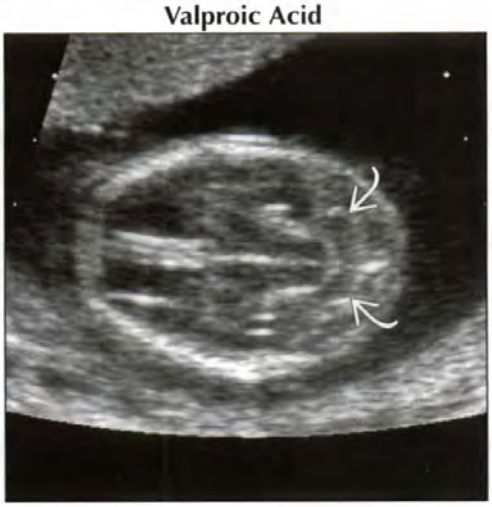


(Left) *Clinical photograph shows the typical appearance with microcephaly, hirsutism with low set hairline, long eyelashes, finely penciled eyebrows, and micrognathia. Bilateral limb reduction defects are also present.* ***(Right)*** *Transabdominal ultrasound shows the typical upper extremity limb reduction defect seen in Cornelia de Lange syndrome.*

Neu Laxová Syndrome



Valproic Acid



(Left) *Sagittal ultrasound of the chest and head shows the typical sloping forehead seen in severe microcephaly. At autopsy this fetus was found to have features of Neu Laxová syndrome.* ***(Right)*** *Axial ultrasound in a fetus exposed to valproate shows the "banana" cerebellum of the Chiari II malformation. The head size was small for gestational age. Myelomeningocele, radial club hands, and absent thumbs were also attributed to valproate embryopathy.*

MACROCEPHALY

DIFFERENTIAL DIAGNOSIS

Common
- Benign Familial
- Macrosomia
- Aqueductal Stenosis
- Intracranial Cysts

Less Common
- Holoprosencephaly Spectrum
- CNS Tumors
- Beckwith-Wiedemann Syndrome

Rare but Important
- Hydranencephaly
- Chiari II Malformation

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Ensure that measurements are obtained correctly
  - Biparietal diameter is measured at level of thalami and cavum septi pellucidi
    - Outer edge of near field skull to inner edge of far field
  - Head circumference in same axial plane,
    - Bone circumference only, does not include soft tissues
- Repeat measurements for confirmation
- Look at parents and consider measuring parental head circumference
- Does the brain look normal?
  - Megalencephaly associated with abnormal gyral pattern and loss of architecture
- Are the ventricles enlarged?
  - Where is the level of obstruction?
  - Look for mass or cyst obstructing flow of cerebrospinal fluid (CSF)
- Is the posterior fossa normal?
  - Look for signs of Chiari malformation, Dandy-Walker continuum, posterior fossa cyst/mass
- Is the falx present?
  - If absent, holoprosencephaly spectrum is most likely diagnosis
  - If present must differentiate hydranencephaly from severe hydrocephalus
- Is there a cortical mantle?
  - MR may be necessary to make this determination

Helpful Clues for Common Diagnoses
- **Benign Familial**
  - Measure parental head size
  - Underlying brain normal
  - Interval growth normal (i.e., no progressive enlargement)
- **Macrosomia**
  - Estimated fetal weight > 90th percentile
  - Abdominal circumference may be larger than other parameters
  - Increased subcutaneous fat visible in third trimester
  - Often associated with polyhydramnios
  - Associated with overgrowth syndromes
  - Seen in infant(s) of diabetic mother(s)
- **Aqueductal Stenosis**
  - Obstructive hydrocephalus
    - Moderate to severe lateral ventricular dilatation (> 15 mm)
    - 3rd ventricle dilated
    - "Dangling" choroid
    - Cavum septi pellucidi may be absent
  - Posterior fossa structures look normal
    - No evidence of Chiari II or Dandy-Walker continuum as etiology of hydrocephalus
  - X-linked form: Adduction-flexion deformity of thumbs in male fetus
- **Intracranial Cysts**
  - Arachnoid
    - Extra-axial
    - Simple
    - Typically stable in size but may enlarge and cause obstruction of CSF flow
  - Glioependymal
    - Extra-axial
    - Midline
    - Extend anterior to posterior
    - More likely to be multiloculated
    - High protein concentration: May show increased signal on T1WI MR

Helpful Clues for Less Common Diagnoses
- **Holoprosencephaly Spectrum**
  - Lobar form more likely to be associated with significant ventriculomegaly and macrocephaly
    - Look for midline fused fornices
    - Look for gyral continuity across midline
    - Look for asymmetric falx placement

- Small head size more likely with alobar/semilobar forms unless very large associated dorsal cyst

- **CNS Tumors**
  - Solid intracranial mass with Doppler flow
  - Supratentorial but precise point of origin often hard to determine
  - May present with hydrocephalus before tumor visible
  - Exhibit rapid growth
- **Beckwith-Wiedemann Syndrome**
  - Fetal overgrowth syndrome
  - Look for associated findings
    - Omphalocele
    - Macroglossia
    - Nephromegaly, hepatosplenomegaly, hemihyperplasia

Helpful Clues for Rare Diagnoses

- **Hydranencephaly**
  - Head size usually normal but macrocephaly may be seen
  - Fluid-filled supratentorial space
  - Falx present
  - Normal posterior fossa
  - Medial temporal or inner occipital lobes also may be present
    - Perfused by posterior circulation therefore preserved
- **Chiari II Malformation**
  - Although associated with ventriculomegaly, head size often normal
  - Look for associated neural tube defect (NTD)
  - Brain findings
    - Posterior fossa "banana sign"
    - Calvarial "lemon sign"
    - Obliteration of cisterna magna

Other Essential Information

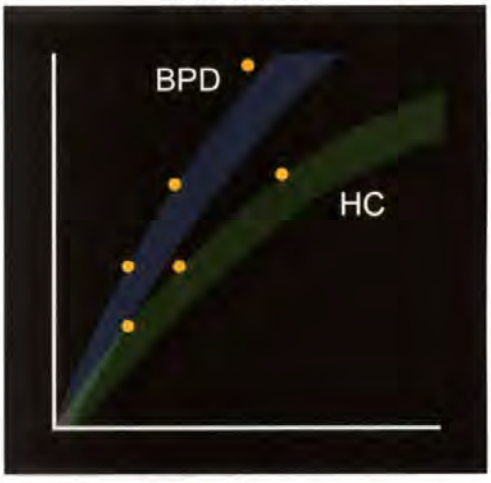
- Prognosis varies depending on underlying condition
  - Aqueductal stenosis
    - May have relatively good outcome after shunt placement
    - X-linked form is associated with profound mental impairment
  - Holoprosencephaly spectrum
    - Alobar holoprosencephaly, atelencephaly and aprosencephaly are lethal
    - Semilobar holoprosencephaly often lethal, profound intellectual impairment
    - Lobar holoprosencephaly has variable outcome, usually not lethal
  - CNS tumors
    - Dismal prognosis: 97% mortality if diagnosed before 30 weeks
  - Hydranencephaly
    - Dismal prognosis: 50% liveborn infants die in first month
    - 85% mortality by end of first year
  - Chiari II
    - Folate supplementation dramatically decreases recurrence risk
    - Prognosis depends on level of NTD ± any additional anomalies

Benign Familial



Axial ultrasound shows a normal brain in a fetus with BPD and HC approaching 2SD above the mean for gestational age. The infant was normal at birth; the mother also had a large head circumference.

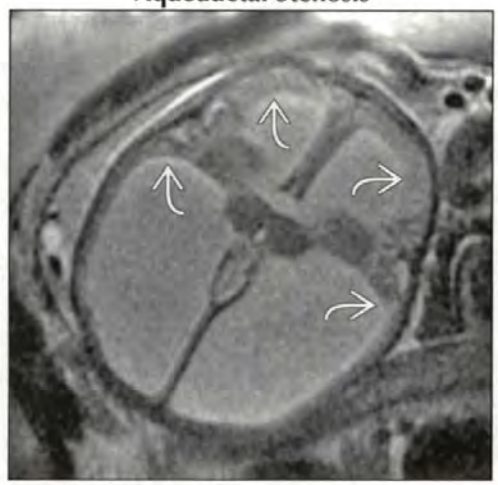
Macrosomia



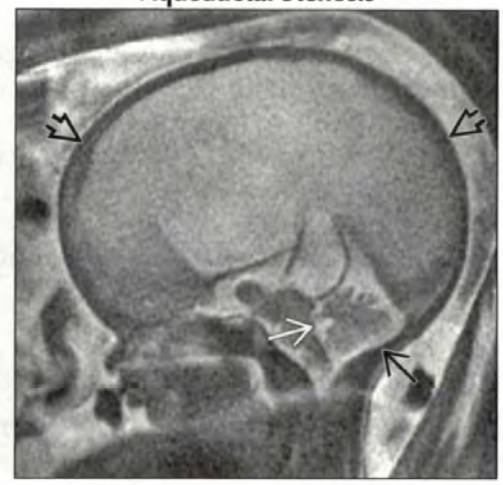
Graphic shows both HC and BPD exceeding the 95th percentile in a generally large fetus. The infant was normal at birth and did not exhibit any features of a pathological overgrowth syndrome.

MACROCEPHALY

Aqueductal Stenosis



Aqueductal Stenosis

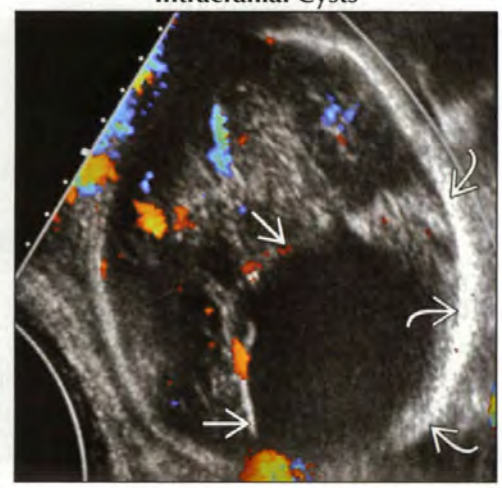


(Left) *Axial T2WI MR shows severe hydrocephalus and a compressed, thin cortical mantle. This excludes hydranencephaly.* ***(Right)*** *Sagittal T2WI MR in the same fetus shows a normal cerebellum and posterior fossa, excluding Dandy-Walker continuum and Chiari malformation as a cause of hydrocephalus. The fourth ventricle is normal as the obstruction is higher, at the aqueduct of Sylvius. Note marked cranial enlargement.*

Aqueductal Stenosis



Intracranial Cysts



(Left) *Coronal oblique ultrasound shows intact midline, thin rind of cerebral cortex, and severe hydrocephalus. The thalami are normal as is the cerebellum. Doppler flow was seen, confirming severe hydrocephalus rather than hydranencephaly.* ***(Right)*** *Axial ultrasound shows a large arachnoid cyst exerting mass effect on the cranial vault. This cyst had enlarged significantly in the third trimester and was causing hydrocephalus.*

Holoprosencephaly Spectrum



Holoprosencephaly Spectrum



(Left) *Sagittal T2WI MR shows an enlarged cranium, which is primarily fluid-filled. The profile is abnormal with a flattened nose and a rudimentary globe. Abnormal facies is common, but not universally seen, in holoprosencephaly (final diagnosis aprosencephaly).* ***(Right)*** *Clinical photograph shows macrocephaly in an infant with alobar holoprosencephaly. Facial features are normal as were chromosomes. Not all holoprosencephaly is associated with aneuploidy.*

CNS Tumors



CNS Tumors



(Left) *Sagittal ultrasound shows a large complex mass with both cystic ⇨ and solid ⇨ components completely replacing most of the normal intracranial contents. The head size was dramatically enlarged.* ***(Right)*** *Gross pathology in the same case shows massive cranial enlargement secondary to an intracranial teratoma which extended into the oral cavity.*

Beckwith-Wiedemann Syndrome

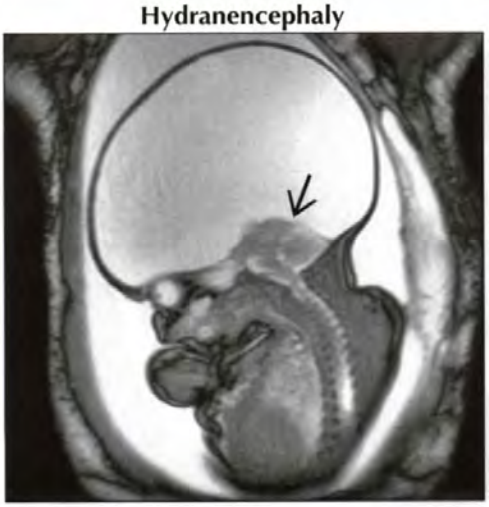


Beckwith-Wiedemann Syndrome

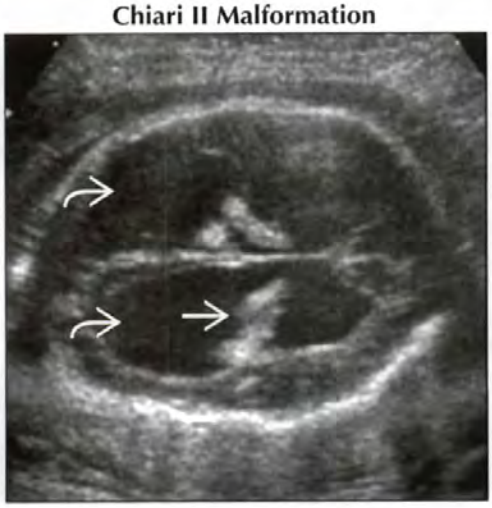


(Left) *Coronal 3D ultrasound in a fetus with an overgrowth pattern shows macroglossia ⇨. Hepatosplenomegaly was also present, both features of Beckwith-Wiedemann syndrome.* ***(Right)*** *Coronal transabdominal ultrasound in a different case shows renal enlargement. At 30 weeks, the right kidney measured 4.8 cm in length and the left measured 6.9 cm (calipers). Beckwith-Wiedemann syndrome was diagnosed after clinical evaluation of the newborn.*

Hydranencephaly



Chiari II Malformation



(Left) *Sagittal T2WI MR shows complete lack of cerebral tissue in a fetus with macrocephaly. Biparietal diameter and head circumference were > 99th percentile. The posterior fossa ⇨ was normal.* ***(Right)*** *Axial ultrasound shows dangling choroid ⇨, large ventricles ⇨, and enlarged head size in a fetus with a myelomeningocele. Head size was such that Cesarean delivery was required. It is more usual for the head size to remain normal in Chiari malformation.*

INTRACRANIAL CALCIFICATIONS

DIFFERENTIAL DIAGNOSIS

Common

- Maternal Infection
  - Cytomegalovirus
  - Toxoplasmosis
  - Varicella

Rare but Important

- Teratoma

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Significant overlap in imaging findings of in utero infections
  - Intrahepatic and intracranial calcifications most common findings
  - Intracranial calcifications may be non-shadowing and subtle
- Requires maternal/fetal serologies to make definitive diagnosis

Helpful Clues for Common Diagnoses

- **Cytomegalovirus**
  - Most common congenital infection
  - Main reservoir is children under < 2 years
  - Brain most commonly affected area
    - Calcifications (predominately periventricular), ventriculomegaly, microcephaly
  - Other findings include intrauterine growth restriction (IUGR), hepatosplenomegaly, cardiomyopathy, echogenic bowel and hydrops
- **Toxoplasmosis**
  - Cats are definitive hosts: Oocyst shed in feces
  - Human infection from contaminated soil, water, undercooked meats
  - Non-shadowing intracranial and intrahepatic calcifications
  - Intracranial calcifications may be periventricular or random in distribution
  - Other findings include ventriculomegaly, IUGR and echogenic bowel
- **Varicella**
  - Transplacental infection of fetus following maternal chickenpox infection
  - Intrahepatic and intracranial calcifications
    - May also see liver, heart, renal calcifications
  - Polyhydramnios due to neurologic impairment of swallowing
  - Limb hypoplasia and contractures
  - Paradoxical diaphragmatic motion on real time sonography due to unilateral paralysis
  - Cutaneous lesions in dermatomal distribution seen in neonate

Helpful Clues for Rare Diagnoses

- **Teratoma**
  - Most common brain tumor in fetus
  - Obvious, large, destructive mass with cystic and solid components
  - Calcification most specific feature but not always present

Cytomegalovirus



Coronal ultrasound focused on the frontal horns shows periventricular calcifications ➡. Only minimal shadowing is seen, which is typical.

Cytomegalovirus



Axial NECT in an infant shows extensive periventricular ➡ and basal ganglia ➡ calcification. The patient had microcephaly and ventriculomegaly, all features of congenital CMV infection.

INTRACRANIAL CALCIFICATIONS

Toxoplasmosis



Toxoplasmosis



(Left) *Axial ultrasound shows periventricular and intraparenchymal calcifications. These findings are subtle and could be easily missed. Findings confirmed postnatally.* ***(Right)*** *Coronal ultrasound shows echogenic bowel. Both intracranial calcifications and echogenic bowel are non-specific and can be seen in many congenital infections. Patient history is key, and maternal/fetal serologies are necessary to make a definitive diagnosis.*

Varicella



Varicella



(Left) *Ultrasound of the arm shows a terminal transverse defect. A proximal radius and ulna are present but no wrist or hand is visualized.* ***(Right)*** *Clinical photograph after delivery shows very rudimentary fingers in this case of congenital varicella infection. Other neonatal findings of fetal varicella syndrome include cutaneous lesions in a dermatomal distribution, chorioretinitis and varying degrees of neurologic dysfunction.*

Teratoma



Teratoma



(Left) *Coronal ultrasound of a 17 week fetus shows macrocephaly (compare to the normal chest). There is a large, solid intraparenchymal mass with areas of calcification and shadowing. Calcifications are the most specific finding of a teratoma but are not always present.* ***(Right)*** *Axial posterior fossa ultrasound shows a predominately cystic teratoma with several focal calcifications. The mass is bulging the calvarium and distorting the cerebellum.*

INTRACRANIAL MASS

DIFFERENTIAL DIAGNOSIS

Common
- Intracranial Hemorrhage

Rare but Important
- CNS Tumors
  - Teratoma
  - Astrocytoma
  - Craniopharyngioma
  - Choroid Plexus Papilloma
  - Lipoma
  - Primitive Neuroectodermal Tumor

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Doppler critically important in evaluating any intracranial mass
  - Intracranial hemorrhage (ICH) will show no flow
  - Tumors will have vascular flow
    - Tumors may have areas of hemorrhage; important to sample entire mass
- Follow-up studies helpful
  - ICH will evolve over time with developing areas of encephalomalacia and porencephaly
  - Many tumors will show rapid growth rates
- MR very helpful in characterizing intracranial masses
  - Sensitive for blood products
  - Better anatomic evaluation for extent of hemorrhage or mass
- Considerable overlap in appearance of many tumor types (teratoma, astrocytoma, craniopharyngioma, primitive neuroectodermal tumor)
  - Differentiation often not possible or even necessary
    - Universally dismal prognosis
  - Most are supratentorial
  - Precise point of origin can often not be determined
  - May extend through skull base into oral cavity
  - May cause gross distortion of cerebral architecture
  - Hydrocephalus common
  - Polyhydramnios secondary to decreased swallowing from hypothalamic dysfunction
- Important to recognize lipomas and choroid plexus papillomas as prognosis is substantially better

Helpful Clues for Common Diagnoses
- **Intracranial Hemorrhage**
  - Bleeds may be intraparenchymal, subependymal, intraventricular, or subdural
    - Always use Doppler to confirm no flow in "mass"
    - Look for vascular malformation as potential cause of bleed
  - Typically present as a non-perfused, echogenic, intracranial "mass"
  - Most are supratentorial
  - Posterior fossa uncommon
  - Intraventricular hemorrhages may appear as an irregular, bulky choroid plexus
    - Also look for echogenic, irregular ependyma as residual sign of earlier bleed
  - Hemorrhage usually extensive when diagnosed in utero
    - Normal intracranial landmarks often obscured
  - Initially clot is hyperechoic
  - Becomes hypoechoic with time
    - Look for evolving areas of encephalomalacia/porencephaly
    - Hydrocephalus commonly develops
  - MR excellent for evaluating for blood products
    - T1WI high signal (methemoglobin)
    - T2WI low signal

Helpful Clues for Less Common Diagnoses
- **Teratoma**
  - Most common tumor (approximately 50% of all fetal CNS tumors)
  - Complex masses with cystic and solid components
  - Typically midline location
  - Calcifications most specific finding but not always present
  - May fill entire cranial vault and extend through skull base into mouth
- **Astrocytoma**
  - Solid tumors
  - Arise in cerebral hemispheres
  - Can be seen in the setting of tuberous sclerosis
    - Most common at foramen of Monro

- **Craniopharyngioma**
  - Arise from Rathke pouch, an ectodermal diverticulum from roof of mouth
  - Suprasellar midline mass
  - Frequently calcify
  - Indistinguishable from teratoma
- **Choroid Plexus Papilloma**
  - May occur anywhere in ventricular system
    - Lateral ventricle most common
  - Well-defined, lobular, hyperechoic mass
  - Hydrocephalus from over production of cerebral spinal fluid (CSF)
    - Often rapid onset
  - Mass may also obstruct ventricle causing asymmetric enlargement
- **Lipoma**
  - Well-defined, echogenic mass
  - Midline or lateral ventricles
  - Up to 50% of midline lipomas associated with agenesis of the corpus callosum
  - May be multiple
- **Primitive Neuroectodermal Tumor**
  - Highly malignant small-cell tumor
  - Derive from neural crest
  - Extremely rapid growth
  - Indistinguishable from other intracranial tumors

Other Essential Information

- Findings in agenesis of the corpus callosum
  - Absent cavum septi pellucidi
  - Elevation of 3rd ventricle creating "trident" shape in coronal plane
  - Colpocephaly (teardrop-shaped ventricles)
  - Midline lipoma or cyst

Alternative Differential Approaches

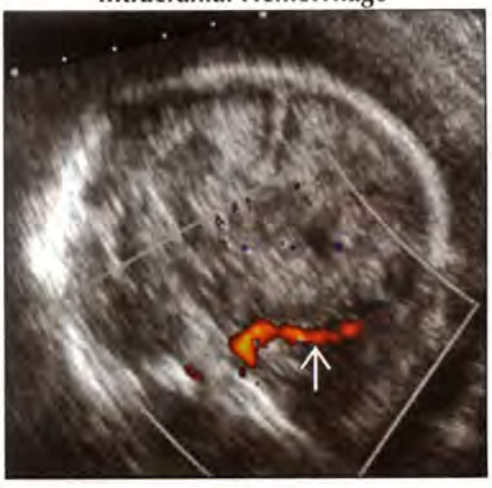
- Intraventricular masses
  - Choroid plexus cyst
    - Very commonly seen in normals but does have an association with trisomy 18
    - Large ones potentially confused with choroid plexus papilloma
    - Cystic, not solid
    - Resolve on follow-up examination
    - No hydrocephalus
  - Choroid plexus papilloma
    - Hydrocephalus usually present
  - Lipoma
    - No hydrocephalus
  - Intracranial hemorrhage
    - Evolves over time
    - Develops areas of porencephaly/encephalomalacia
    - Hydrocephalus worsens as encephalomalacia progresses
- Intracranial mass with calcifications
  - Teratoma
    - More common
    - Located anywhere in brain
  - Craniopharyngioma
    - Suprasellar but often so large cannot tell point of origin
  - In utero infection causes intracranial calcifications but not masses

Intracranial Hemorrhage



Coronal ultrasound shows a large, echogenic intraparenchymal bleed ➡, which is distorting the normal landmarks.

Intracranial Hemorrhage



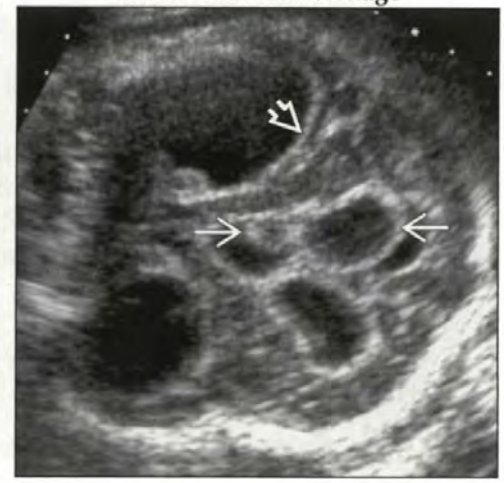
Coronal power Doppler ultrasound shows flow within the middle cerebral artery ➡ but not within the "mass". Always evaluate with color Doppler to rule out an intracranial tumor.

INTRACRANIAL MASS

Intracranial Hemorrhage



Intracranial Hemorrhage



(Left) *Axial oblique ultrasound shows intraventricular hemorrhage associated with co-twin demise in a monochorionic twin pregnancy. Note the presence of hydrocephalus. **(Right)** Axial oblique transabdominal ultrasound shows mixed echogenicity intraventricular clot. The ependyma is markedly echogenic.*

Teratoma

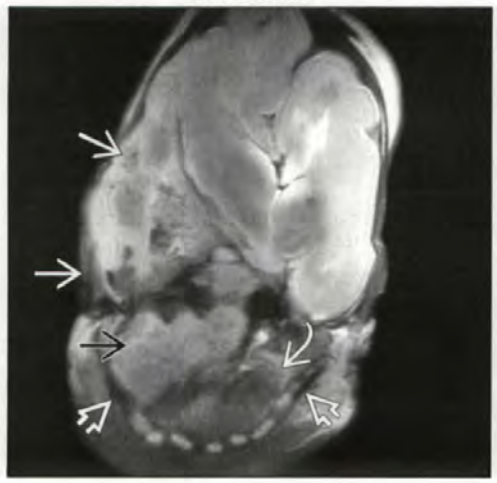


Teratoma

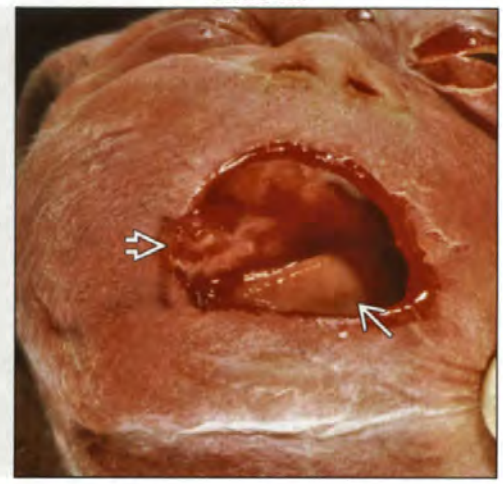


(Left) *Axial ultrasound of the head shows a massive teratoma with complete replacement of normal intracranial contents. **(Right)** Sagittal ultrasound in the same case shows the mass has grown into the oral cavity. It has both solid and cystic components. The cystic component is preventing the mouth from closing. Note the protrusion of the lower lip and mandible.*

Teratoma



Teratoma

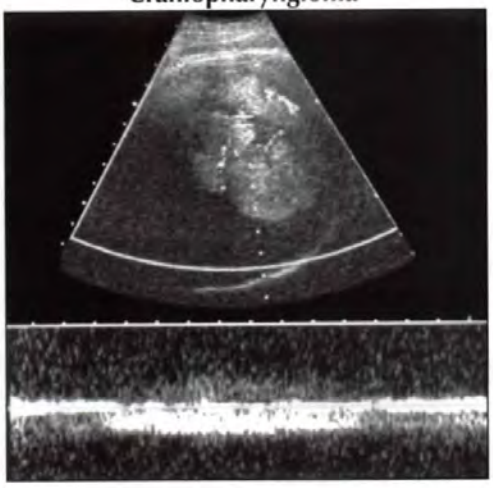


(Left) *Coronal T2WI MR postmortem scan shows a large extra-axial mass which has grown through the skull and into the oropharynx. It is splaying the mandible and displacing the tongue. **(Right)** Gross photograph in the same case shows the distortion of the right side of the face by the tumor, which can be seen within the oral cavity, displacing the tongue.*

Craniopharyngioma



Craniopharyngioma

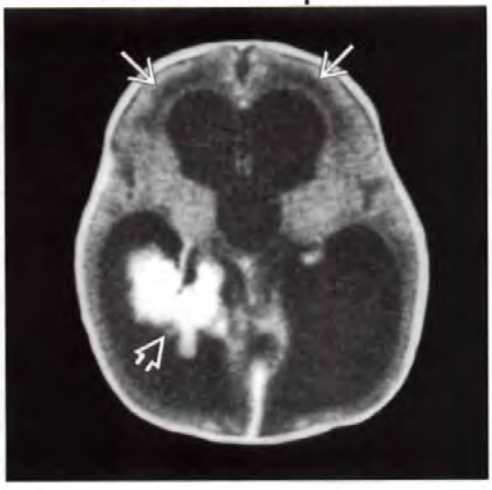


(Left) *Axial ultrasound shows a large, echogenic, well-defined, midline mass ➡ within the fetal brain. This appearance is non-specific and could easily have been an intraparenchymal bleed.* ***(Right)*** *Pulsed Doppler ultrasound shows flow within the mass, confirming this is a tumor rather than an area of hemorrhage.*

Choroid Plexus Papilloma



Choroid Plexus Papilloma

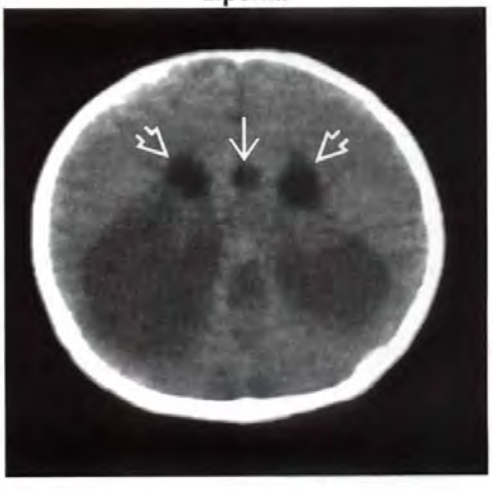


(Left) *Axial ultrasound shows a well-defined, lobular, echogenic mass ➡ within the atrium of the right lateral ventricle. There is also marked hydrocephalus.* ***(Right)*** *Axial CECT after delivery shows brisk enhancement of this mass ➡. There is severe hydrocephalus with transependymal edema ➡. Hydrocephalus is caused by the overproduction of CSF.*

Lipoma



Lipoma



(Left) *Axial ultrasound shows three distinct echogenic masses, one in the midline ➡ and one in each lateral ventricle ➡.* ***(Right)*** *Axial NECT confirms both an interhemispheric lipoma ➡ and two intraventricular lipomas ➡. Note the "teardrop-shaped" ventricles (colpocephaly). This fetus also had agenesis of the corpus callosum, a common associated finding with midline lipomas.*

POSTERIOR FOSSA CYST/FLUID COLLECTION

DIFFERENTIAL DIAGNOSIS

Common

- Incorrect Scan Plane
- Mega Cisterna Magna
- Dandy-Walker Continuum: Classic
- Dandy-Walker Continuum: Variant
- Arachnoid Cyst

Less Common

- Cerebellar Hypoplasia
- Vein of Galen Malformation
- Arteriovenous Fistula

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Review normal anatomy of posterior fossa structures
  - Cisterna magna should measure ≤ 10mm
  - Assess cerebellar size
    - Diameter should measure ≈ gestational weeks in mm in second trimester
    - Normative charts exist for more accurate third trimester measurements
  - Cerebellum should be bilobed and symmetric
- Identify if vermis hypoplastic/absent
  - Hypoplasia/absent in Dandy-Walker continuum
  - Beware of oblique scan plane
    - May simulate vermian defect
  - Later in gestation check sagittal plane
    - Vermis is midline echogenic posterior fossa structure between cerebellar hemispheres
- Doppler is critical
  - "Cyst" may actually be a vascular malformation
  - May not have significant flow if thrombosis present
    - May see clot
  - MR will show typical flow void from high velocity flow
    - Thrombosis will have variable signal based on evolution of blood products within clot

Helpful Clues for Common Diagnoses

- **Incorrect Scan Plane**
  - An overly coronal image gives false impression of mega cisterna magna or even Dandy-Walker malformation
- **Mega Cisterna Magna**
  - Measure space between cerebellum and inner skull
    - Normal linear echogenic dural folds traverse space
  - Mega cisterna magna if > 10 mm
  - Cerebellum and vermis normally formed
  - Complete detailed anatomy scan
    - Carefully evaluate brain parenchyma
    - Can be associated with chromosomal abnormalities (trisomy 18)
  - May not have clinical significance in isolation
- **Dandy-Walker Continuum: Classic**
  - Agenesis or severe hypoplasia of vermis
  - Communication of 4th ventricle with large posterior fossa cyst/cisterna magna
  - Elevated torcular herophili (confluence of sinuses)
  - Do not diagnose in first trimester
    - Normal rhombencephalon is cystic at that time
  - 70-90% have additional anatomic abnormalities
    - Chromosomal abnormalities in 50%
    - Other CNS findings include callosal dysgenesis, encephalocele, polymicrogyria, heterotopias, holoprosencephaly
  - May develop hydrocephalus antenatal
    - More common to have postnatal hydrocephalus
- **Dandy-Walker Continuum: Variant**
  - Partial agenesis/hypoplasia of inferior vermis
    - Vermis not always completely formed until 17 weeks gestation
  - Torcular herophili in normal location, unlike Classic DW
  - Avoid over-diagnosis
    - Beware of oblique scan plane
    - If early in gestation, follow until 18 weeks to allow for complete development of vermis
  - May be associated with other CNS and extra-cranial abnormalities
    - Similar to Classic DW in frequency
- **Arachnoid Cyst**
  - Anechoic extra-axial cyst
    - Contains cerebral spinal fluid

- Most common over cerebral convexities in fetus
- 1/3 in posterior fossa
- If in posterior fossa, normal dural folds will be displaced

◦ Size is variable
- Need to monitor growth
- If rapid growth → obstructive hydrocephalus

◦ Can have scalloping of inner skull
◦ If supratentorial and midline look for agenesis of corpus callosum
◦ Mostly sporadic but occasionally seen with syndromes
- Consider amniocentesis

Helpful Clues for Less Common Diagnoses

• **Cerebellar Hypoplasia**
◦ Cerebellum present but small for gestational age creating enlarged cisterna magna
◦ Differentiate from atrophy of cerebellar hemispheres
- Cerebellum small and cerebellar fissures enlarged if atrophy
- Hypoplasia implies small cerebellum but fissures normal in proportion with folia

◦ Can be isolated or associated with chromosomal or structural abnormalities
- Trisomy 18, 13
- Pontocerebellar hypoplasia (rare)
- X-linked cerebellar hypoplasia (rare)

• **Vein of Galen Malformation**
◦ Elongated midline cystic structure
- In cistern of velum interpositum and quadrigeminal plate cistern
- Extends posteriorly to occipital region
- Actually the medial prosencephalic vein of Markowski

◦ Doppler shows turbulent flow
- High velocity, low resistance arterial flow

◦ May present with hydrops in third trimester, due to high output heart failure
◦ Rarely may thrombose
- Look for hypoechoic intraluminal clot
- Can also see calcifications in clot

• **Arteriovenous Fistula**
◦ Focal area of enlarged vessels
- May appear as tubular cystic structures on grayscale
- Color Doppler shows alternating directions of flow
- Enlarged associated meningeal arteries and draining veins present

◦ Look for secondary effects
- Hydrocephalus
- Ischemia manifested by ventriculomegaly, periventricular leukomalacia, porencephaly
- Intracranial hemorrhage
- Cortical atrophy
- Premature closure of cranial sutures

◦ Hydrops may result due to shunt volume
- Check for cardiomegaly as early sign
- AVF may enlarge and shunt can worsen in utero

Mega Cisterna Magna



Axial ultrasound shows the cisterna magna measuring more than 10 mm (calipers). This was consistently reproducible. No other anatomic abnormalities were found.

Mega Cisterna Magna



Sagittal T2WI MR of the same fetus shows an intact vermis ⇨ and a normal fourth ventricle ⇨. Therefore, this is not a Dandy-Walker variant but an isolated mega cisterna magna.

POSTERIOR FOSSA CYST/FLUID COLLECTION

Incorrect Scan Plane



(Left) *Coronal oblique ultrasound shows an apparent large cisterna magna and suggests an absent inferior vermis (DW variant). Axial images showed a completely normal posterior fossa.* ***(Right)*** *Sagittal T2WI MR shows marked elevation of the torcular Herophili and a large posterior fossa cyst. There is no vermis covering the fourth ventricle. The fetus had multiple other anomalies including agenesis of the corpus callosum and a large interhemispheric cyst.*

Dandy-Walker Continuum: Classic

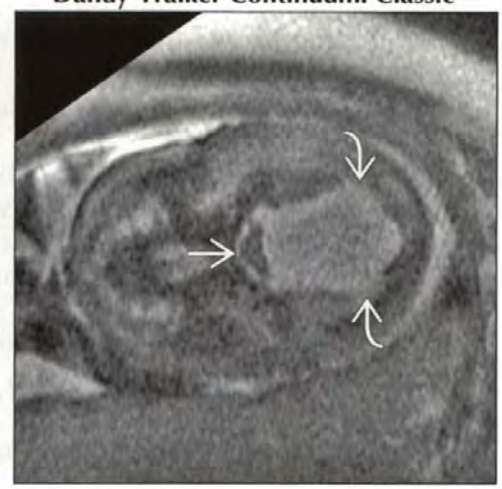


Dandy-Walker Continuum: Classic

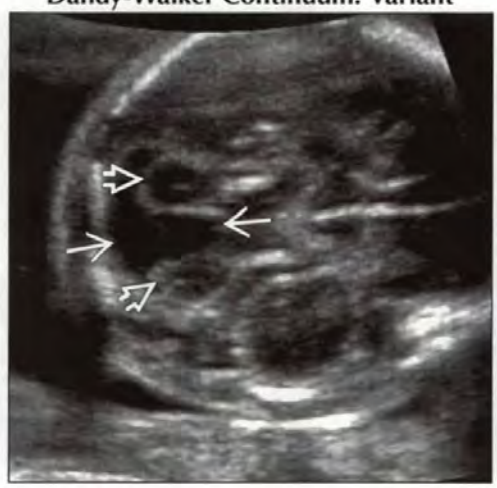


(Left) *Coronal ultrasound shows a large cystic space in the posterior fossa. The cerebellar hemispheres were absent. Diagnosis was confirmed postnatally as well.* ***(Right)*** *Axial T2WI MR shows a huge posterior fossa cyst in contact with the brainstem, which was flattened. The vermis was absent, and the cerebellar hemispheres were hypoplastic.*

Dandy-Walker Continuum: Classic

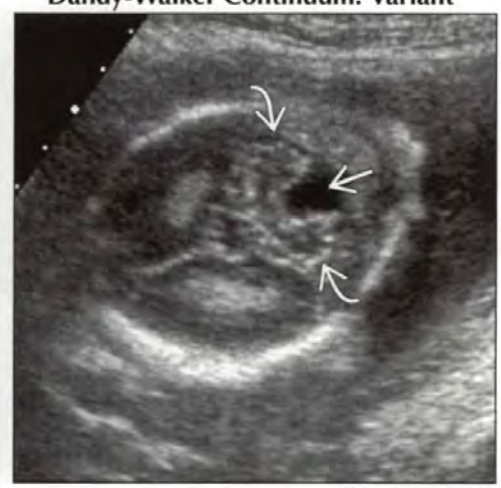


Dandy-Walker Continuum: Variant



(Left) *Axial ultrasound shows splaying of the inferior cerebellar hemispheres and communication between the cisterna magna and the fourth ventricle, secondary to an absent inferior vermis.* ***(Right)*** *Coronal ultrasound shows open communication of the fourth ventricle with the cisterna magna. The cerebellar lobes are only mildly hypoplastic. This finding was confirmed on axial planes.*

Dandy-Walker Continuum: Variant

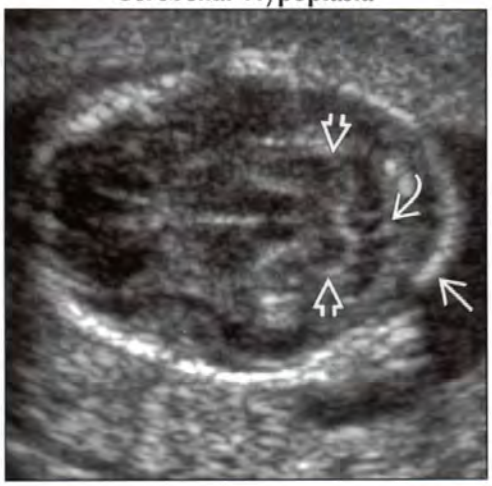


POSTERIOR FOSSA CYST/FLUID COLLECTION

Arachnoid Cyst

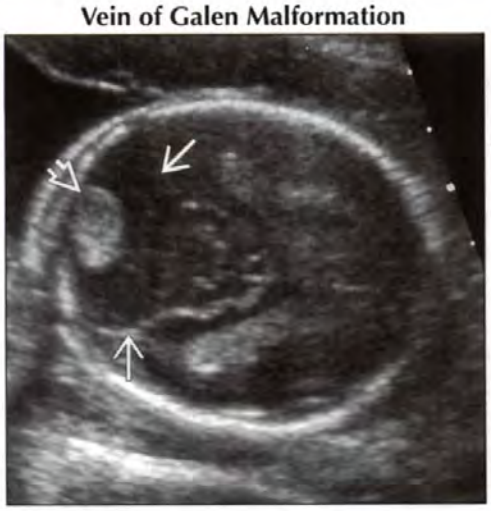


Cerebellar Hypoplasia



(Left) *Coronal T2WI MR shows an infratentorial arachnoid cyst ➡ that displaces the cerebellum ➡. On other images the vermis was shown to be intact. These features distinguish the cyst from a Dandy-Walker malformation.* ***(Right)*** *Axial ultrasound shows a small bilobed cerebellum ➡, resulting in an enlarged cisterna magna ➡. There is also nuchal fold thickening ➡. This fetus also had bilateral enlarged echogenic kidneys. Chromosomes showed trisomy 13.*

Vein of Galen Malformation

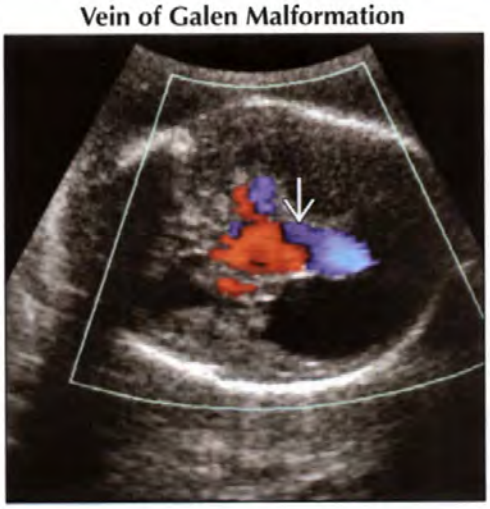


Vein of Galen Malformation

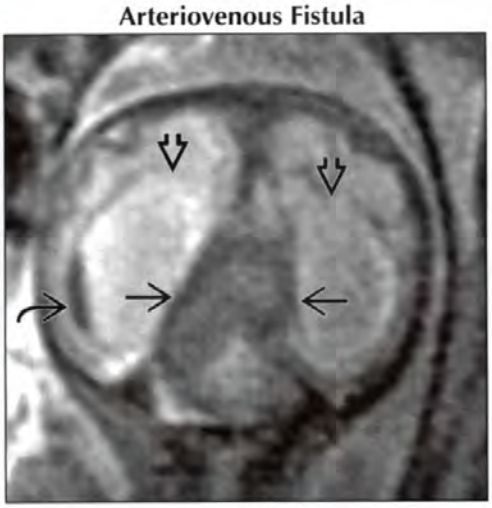


(Left) *Axial oblique ultrasound shows a large "cyst" ➡ in the posterior fossa, with an echogenic nodule ➡. No flow was seen with Doppler.* ***(Right)*** *Sagittal T2WI MR in the same patient as previous image shows a large, intermediate signal intensity extra-axial mass in the posterior cranium ➡. There is a low-signal clot ➡ corresponding to the hyperechoic area on ultrasound. This is a thrombosed vein of Galen malformation.*

Vein of Galen Malformation



Arteriovenous Fistula



(Left) *Axial oblique color Doppler ultrasound shows flow ➡ within a large, midline, elongated "cyst". It is important to put Doppler on cystic appearing lesions to rule out a vascular malformation.* ***(Right)*** *Coronal T2WI MR shows clot extending between the cerebral hemispheres ➡. Ventriculomegaly ➡, and intraventricular hemorrhage ➡ are present. Thrombosed dural AVF was confirmed at birth.*

ABNORMAL CEREBELLUM

DIFFERENTIAL DIAGNOSIS

Common
- Chiari II Malformation
- Dandy-Walker Continuum: Classic
- Dandy-Walker Continuum: Variant
- Arachnoid Cyst
- Mega Cisterna Magna
- Mega Cisterna Magna (Mimic)

Less Common
- Cerebellar Hypoplasia
- Rhombencephalosynapsis
- Joubert Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Assess cerebellar size and shape
  - Measure transcerebellar diameter
    - Gestational age ~ transcerebellar diameter until about 3rd trimester
    - Use normative tables for exact percentiles
- Absent or partially absent cerebellum/vermis
  - Consider Dandy-Walker spectrum if vermian defect present
    - May also be associated with posterior fossa cyst
  - If vermis not actually absent but distorted/compressed, consider arachnoid cyst in posterior fossa
- Cerebellum appears small
  - Confirm small cerebellum versus relatively large posterior fossa
  - If cerebellum actually normal, could be mega cisterna magna or posterior fossa arachnoid cyst
- Cerebellum is flattened and "banana-shaped"
  - Consider Chiari II malformation
  - Look for associated neural tube defect

Helpful Clues for Common Diagnoses
- **Chiari II Malformation**
  - "Banana sign" in posterior fossa
    - Flattened cerebellum
    - Curved around midbrain
  - "Lemon sign"
    - Refers to appearance of calvarium secondary to flattened frontal bones and dolichocephaly
    - Typical appearance often resolves by 3rd trimester
    - Nonspecific finding, can even be seen in normals
  - Very small or absent cisterna magna
    - Due to caudal displacement of posterior fossa structures (tonsillar herniation) through foramen magnum
  - Ventriculomegaly
    - Usually mild but can progress during pregnancy
  - If typical intracranial findings identified, search carefully for the associated open neural tube defect (ONTD)
    - Can be subtle at times despite dramatic intracranial abnormalities
    - May be skin covered
    - Posterior ossification sites of vertebral bodies are splayed
    - Check sagittal view to visualize overlying sac (seen in 80%)
  - May have associated scoliosis/kyphosis at level of ONTD
  - Look for clubfoot, additional anomalies
- **Dandy-Walker Continuum: Classic**
  - Absent vermis
  - Large posterior fossa cyst
  - 4th ventricle open to posterior fossa cyst
  - Elevated torcular Herophili (confluence of superior sagittal, straight, & occipital sinuses)
  - Look for other associated anomalies (seen in 70-90%)
    - Callosal dysgenesis
    - ONTD
    - Heterotopias
    - Chromosomal abnormalities
    - Cardiac anomalies
- **Dandy-Walker Continuum: Variant**
  - Milder form of Dandy-Walker malformation
  - Absent or hypoplastic inferior vermis
  - 4th ventricle open to cisterna magna
    - "Keyhole" appearance of 4th ventricle on axial ultrasound
  - Posterior fossa should be normal size
  - Avoid over diagnosis
    - Vermis not always completely formed until 17 weeks
    - Overly coronal oblique scan plane can simulate vermian defect
- **Arachnoid Cyst**
  - One third of cases occur in posterior fossa

- Vermis is intact
- Cyst does not communicate with 4th ventricle
- Cerebellum often appears distorted by mass effect from cyst

- **Mega Cisterna Magna**
  - Enlarged cisterna magna measuring > 10 mm
    - Make sure this is a true measure
    - Should see cavum septi pellucidi
  - Can give overall impression of relatively small cerebellum
  - Actual cerebellar diameter normal for gestational age
  - Usually an incidental finding of no clinical consequence
  - May be part of multiple findings seen with trisomy 18
- **Mega Cisterna Magna (Mimic)**
  - An overly coronal image gives false impression of mega cisterna magna

Helpful Clues for Less Common Diagnoses

- **Cerebellar Hypoplasia**
  - Cerebellar axial diameter less than expected for gestational age
  - Normally formed cerebellar hemispheres present
    - Folia appear normal (prominent sulci seen with atrophy, not hypoplasia)
  - Look for other anatomic abnormalities
    - Associated with chromosomal anomalies
- **Rhombencephalosynapsis**
  - Congenital fusion of cerebellar lobes
  - Agenesis of vermis
  - Cerebellum appears small and "ball-like"
    - Typical bilobed appearance is absent
    - Abnormal 4th ventricle shape ("pointed" appearance on postnatal MR)
  - Look for other associated intracranial abnormalities
    - Consider fetal MR for optimal visualization of intracranial structures
- **Joubert Syndrome**
  - Cerebellar cleft at midline between hemispheres
    - Represents prominent interpeduncular fossa
    - May be seen in conjunction with ventriculomegaly, encephalocele, polydactyly, micropenis
    - Can be confused with Dandy-Walker
  - "Molar tooth" sign on fetal MR
    - Deepened interpeduncular fossa
    - Thick, straight superior cerebellar peduncles
    - Hypoplastic vermis
    - Abnormal midbrain (↓ anteroposterior diameter)
  - Can be seen with fetal hyperpnea up to 140-160 breaths/min
  - Autosomal recessive disorder
    - Clarify family history to assess
    - Genetic counseling useful for future pregnancy planning

Chiari II Malformation



Axial ultrasound shows the typical appearance of a "banana" cerebellum ➡ wrapped around the brainstem and a "lemon" skull ➡. These findings should prompt a careful search for associated ONTD.

Chiari II Malformation



Sagittal ultrasound of the spine in the same fetus shows a subtle sacral neural tube defect ➡, which could easily be missed without the associated intracranial findings.

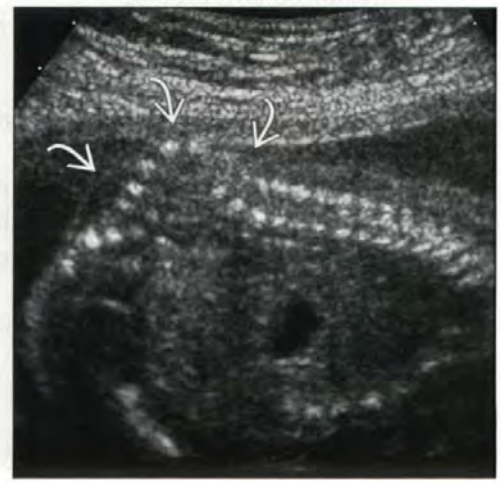
ABNORMAL CEREBELLUM

Chiari II Malformation



(Left) *Axial ultrasound of a fetus seen for screening ultrasound shows cerebellar compression with a resultant "banana" cerebellum. A "lemon" skull is also seen.* ***(Right)*** *Sagittal ultrasound of the same fetus as previous image shows an extensive ONTD of the lumbar spine with marked kyphosis at the level of the defect.*

Chiari II Malformation

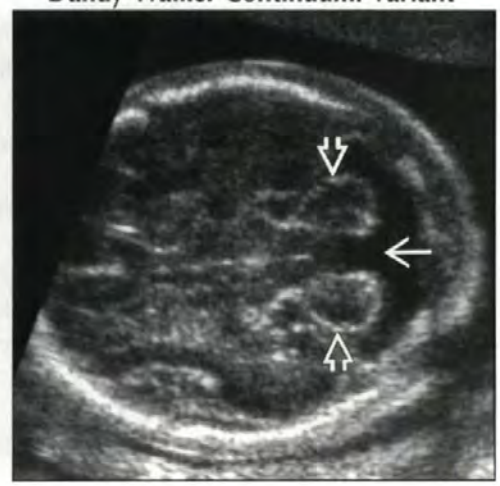


Dandy-Walker Continuum: Classic



(Left) *Axial ultrasound shows absence of the vermis and direct communication of the 4th ventricle & a posterior fossa cyst (calipers). The cerebellar hemispheres are small and splayed. In addition, the cavum septi pellucidi is absent.* ***(Right)*** *Axial oblique ultrasound shows a bilobed cerebellum with a vermian defect identified on routine screening at 22 weeks gestation.*

Dandy-Walker Continuum: Variant



Dandy-Walker Continuum: Variant



(Left) *Axial ultrasound of a fetus at 33 weeks gestation shows open communication of the inferior 4th ventricle with the cisterna magna ("keyhole" defect). This defect is relatively small. Dandy-Walker malformation forms a true continuum from severe to mild.* ***(Right)*** *Sagittal T2WI MR shows a posterior fossa arachnoid cyst. The vermis is intact, torcular position is normal, and there is no communication with the 4th ventricle.*

Arachnoid Cyst



Mega Cisterna Magna (Mimic)

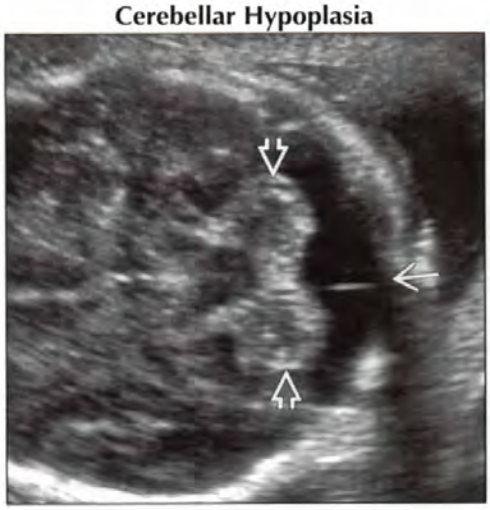


Mega Cisterna Magna (Mimic)

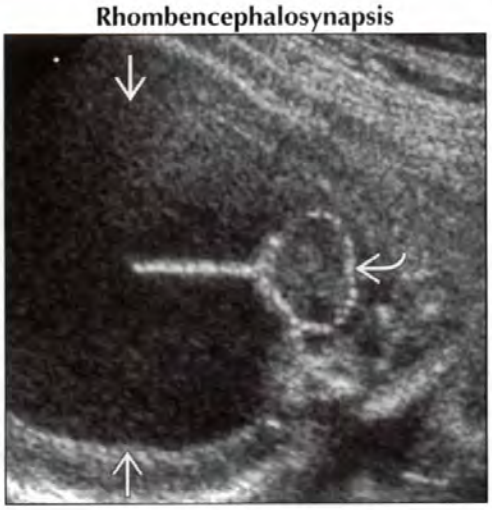


(Left) *Coronal oblique ultrasound shows an apparently enlarged cisterna magna, measuring 13 mm (calipers).* ***(Right)*** *Axial ultrasound which includes the cavum septi pellucidi in the same fetus as previous image, shows the cisterna magna is actually 9 mm and therefore within normal limits. The vermis is also noted at midline. Scanning at too steep an angle can give the false appearance of a mega cisterna magna.*

Cerebellar Hypoplasia

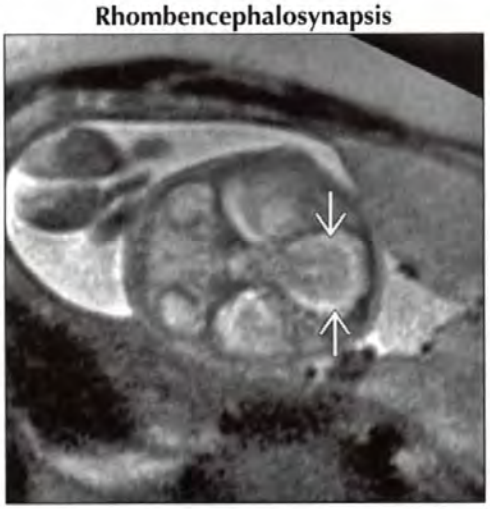


Rhombencephalosynapsis

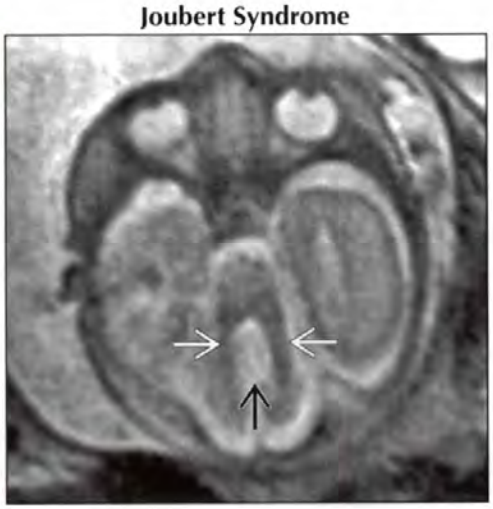


(Left) *Axial ultrasound of a fetus with trisomy 18 shows a small cerebellum, measuring at the 5th percentile for 30 weeks. The cisterna magna is enlarged, measuring 1.3 cm. The fetus died in utero near term.* ***(Right)*** *Coronal oblique ultrasound shows a small, globular cerebellum. There is no normal hemispheric differentiation or vermian structures, findings typical of rhombencephalosynapsis. Lobar holoprosencephaly was also present.*

Rhombencephalosynapsis



Joubert Syndrome



(Left) *Axial T2WI MR shows a small cerebellum with lack of normal hemispheric differentiation. The cerebellum looks like a homogeneous soft tissue mass ("ball-like") without an identifiable vermis.* ***(Right)*** *Axial oblique T2WI MR shows the "molar tooth" described in Joubert syndrome, created by the elongated superior cerebellar peduncles on either side of a cleft between the cerebellar hemispheres.*

SPINAL MASS

DIFFERENTIAL DIAGNOSIS

Common
- Myelomeningocele
- Myeloschisis
- Sacrococcygeal Teratoma

Less Common
- Lipoma/Lipomyelomeningocele
- Lymphangioma
- Myelocystocele

Rare but Important
- Iniencephaly
- Chiari III Malformation

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Multicystic mass in region of distal spine
  - Differentiate myelomeningocele from sacrococcygeal teratoma due to prognostic implications
  - Myelomeningocele
    - Prognosis depends on associated malformations and level of defect
    - Amniocentesis offered as associated with aneuploidy
    - Not associated with high output state
  - Sacrococcygeal teratoma (SCGT)
    - Prognosis depends on size, intrapelvic extension, growth rate and vascularity
    - Solid rapidly growing SCGT → high output state → hydrops
    - Fetal therapy indicated for impending hydrops
    - Multicystic lesions have good prognosis
- Maternal history important
  - Valproate embryopathy
  - Diabetic embryopathy
- Differential diagnosis of patient with elevated maternal serum alpha fetoprotein (msAFP)
  - "Open anything"
  - Myelomeningocele important diagnosis as potential to decrease recurrence risk in future pregnancies

Helpful Clues for Common Diagnoses
- **Myelomeningocele**
  - Elevated msAFP
  - Associated with Chiari II brain malformation in 99% of cases
    - Obliteration of cisterna magna
    - "Banana" shaped cerebellum
    - Ventriculomegaly/hydrocephalus
    - "Lemon-shaped" head (bifrontal concavity which resolves in third trimester)
  - Open neural tube defect
    - Posterior elements splayed or divergent: Lack of normal "teepee" shape on axial view
    - Parallel posterior elements abnormal if reproducible from different scan planes
- **Myeloschisis**
  - Open defect with no covering membrane
  - Neural placode exposed to amniotic fluid
  - Everted edges may appear as spinal mass
- **Sacrococcygeal Teratoma**
  - Cystic, solid, or mixed echogenicity mass arising from sacrum
    - Purely cystic in 15%
    - May contain calcifications
  - Posterior elements intact
  - Sacrum present
  - Solid lesions may be hypervascular putting fetus at risk for developing hydrops
  - Grading system developed by American Academy of Pediatric Surgery Section
    - Type 1: Completely external or minimal presacral component
    - Type 2: External and internal component extending into presacral space
    - Type 3: External and internal component extending into abdomen
    - Type 4: Completely internal, no external component
  - Brain is structurally normal

Helpful Clues for Less Common Diagnoses
- **Lipoma/Lipomyelomeningocele**
  - Skin covered defect
  - Not associated with elevated msAFP
  - Subcutaneous mass of variable echogenicity
  - Lipoma: No spinal canal enlargement
  - Lipomyelomeningocele: Tethered low-lying spinal cord inserts into lipoma through dysraphic defect
- **Lymphangioma**
  - Multicystic mass
  - May be anywhere along torso
  - Skin-covered

- May mimic myelomeningocele or cystic sacrococcygeal teratoma
  - Myelomeningocele associated with open neural tube defect; sac extends out of spine defect
  - Sacrococcygeal teratoma arises from sacrum
- **Myelocystocele**
  - Skin-covered lumbosacral dysraphism
  - Hydromyelic cord traverses meningocele, expands into large contiguous terminal cyst
  - Abnormal cystic dilatation of thecal sac (i.e., intradural)
    - All other cystic spinal masses arise in soft tissues
  - May be associated with other abnormalities
    - Caudal regression or OEIS syndrome (omphalocele, exstrophy of the bladder, imperforate anus, and spinal anomalies)

Helpful Clues for Rare Diagnoses

- **Iniencephaly**
  - Lethal open neural tube defect
  - High cervical location, spinal defect may extend to thoracic/lumbar area (rachischisis)
  - Cervical vertebrae are missing or fused
  - Associated occipital encephalocele
  - Associated with fixed hyperextension of neck → "stargazer" fetus
  - Mandibular skin contiguous with chest
- **Chiari III Malformation**
  - Intracranial findings of Chiari II
  - High cervical ± low occipital osseous defect
  - Herniation of posterior fossa contents (i.e., cerebellum, brain stem, fourth ventricle ± upper cervical spinal cord) through osseous defect

Other Essential Information

- Essential to scan fetal spine in three planes
  - Axial for neural tube closure
  - Sagittal for global alignment
  - Coronal for vertebral body evaluation/assessment of posterior elements
- Myelomeningocele may be extremely subtle when small in second trimester
  - Maternal habitus ± fetal position may compromise assessment of distal spine
  - Look at brain for Chiari malformation
- Lateral recumbent fetus pitfall
  - Posterior elements may appear parallel in normal fetus when scanned in lateral recumbent position
    - Scan from different acoustic windows
    - Orthogonal views at suspicious level
    - Correlate with msAFP
- Myelomeningocele may be associated with other malformations
  - If multiple anomalies, risk of aneuploidy is increased
- Spinal lipoma
  - May be associated with tethered cord
  - Important to counsel parents about potential for neurological impairment

Myelomeningocele



3D ultrasound displayed as serial axial slices shows the splayed posterior elements ➡ and myelomeningocele sac ➡.

Myelomeningocele



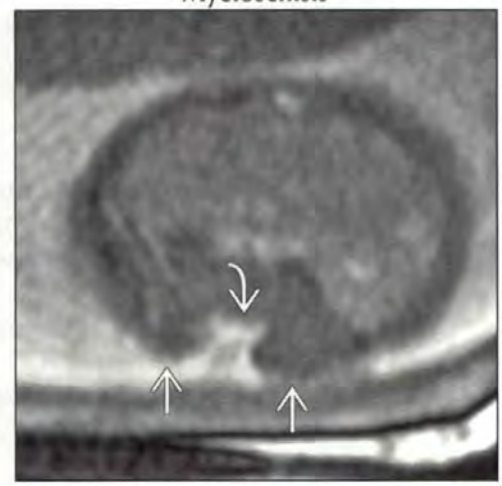
Sagittal ultrasound shows a myelomeningocele ➡ at the level of the sacral spine (➡ = L5). This was associated with the typical brain findings of Chiari II malformation.

SPINAL MASS

Myelomeningocele

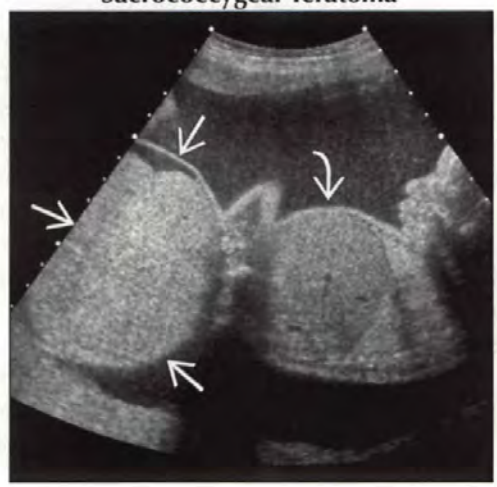


Myeloschisis



(Left) *Sagittal T2WI MR shows a small myelomeningocele in the lumbosacral area. Maternal habitus precluded adequate sonographic assessment, although the intracranial findings of Chiari II malformation were seen.* ***(Right)*** *Axial T2WI MR shows a myeloschisis defect. In this lesion, there is no encasing dural membrane, and the neural placode is open to amniotic fluid. The everted edges of the defect may suggest a spinal mass.*

Sacrococcygeal Teratoma

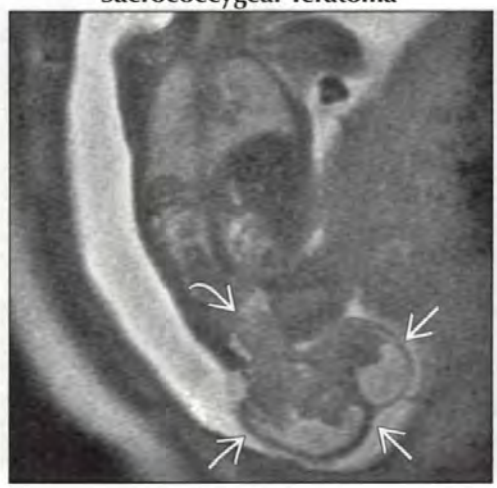


Sacrococcygeal Teratoma



(Left) *Sagittal ultrasound shows a large, solid mass that is larger than the fetal torso. Solid teratomas are at risk for high output cardiac failure and hydrops.* ***(Right)*** *Clinical photograph shows the bilobed appearance of a large, solid sacrococcygeal teratoma involving the perineum and buttocks of the neonate.*

Sacrococcygeal Teratoma



Sacrococcygeal Teratoma



(Left) *Coronal T2WI MR shows a mixed cystic and solid mass extending into the pelvis. This elevated the bladder, but there was no associated urinary tract obstruction.* ***(Right)*** *Coronal T2WI MR shows a sacrococcygeal teratoma with primarily cystic external component but an unexpectedly large solid intrapelvic extension. The mass remained stable, and surgical excision was successful.*

Lipoma/Lipomyelomeningocele

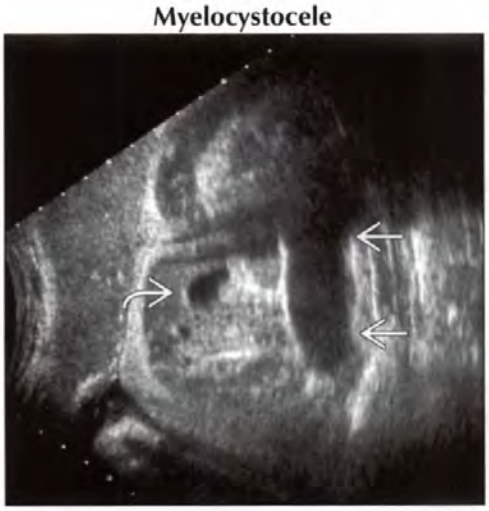


Lymphangioma

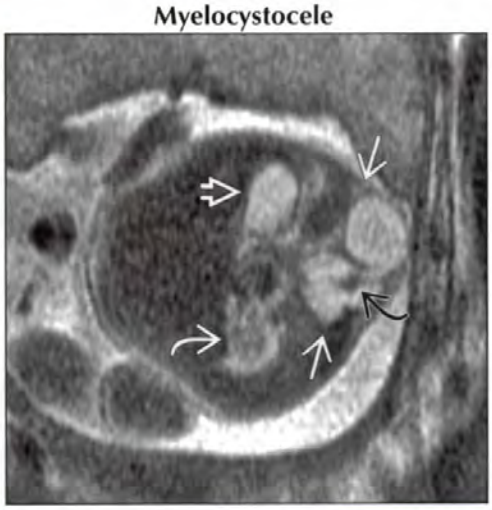


(Left) *Sagittal ultrasound shows intact skin covering* ➡ *over a hypoechoic mass* ➡ *associated with dysraphism in the sacral region. Maternal serum screening and views of the brain were normal. Postnatal imaging showed a lipomyelomeningocele and tethered cord.* ***(Right)*** *Axial ultrasound shows a septated cystic mass* ➡ *arising from the soft tissues of the neck not involving the bony spinal column. Note normal converging posterior elements* ➡.

Myelocystocele

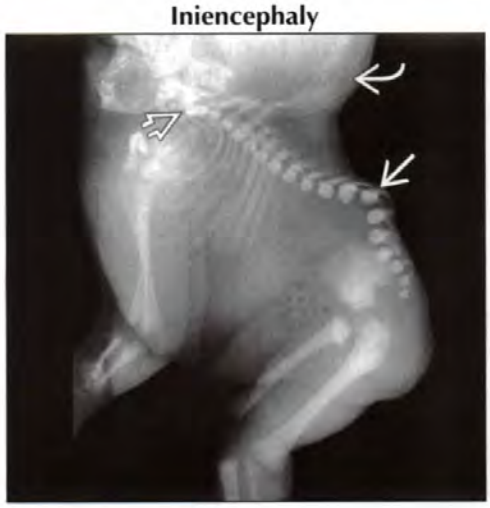


Myelocystocele



(Left) *Sagittal oblique ultrasound shows an unusual posterior tubular fluid collection* ➡. *Note gallbladder* ➡. *Spinal bony elements were not seen.* ***(Right)*** *Axial T2WI MR confirms posterior multicystic mass* ➡ *surrounding the spinal cord* ➡. *The stomach* ➡ *and the right adrenal* ➡ *are seen at this level. This is a terminal myelocystocele associated with severe caudal regression. The infant died within minutes of birth.*

Iniencephaly



Chiari III Malformation



(Left) *Lateral radiograph shows an acute lumbar kyphosis* ➡. *There is marked head extension* ➡ *due to shortening of the cervical spine, which is the result of missing and fused vertebrae* ➡. ***(Right)*** *Sagittal ultrasound shows cervical dysraphism and an occipital bony defect* ➡ *with an associated encephalomeningocele* ➡. *This defect, combined with the typical intracranial finding seen in Chiari II, constitutes a Chiari III malformation.*

ABNORMAL SPINE POSITION

DIFFERENTIAL DIAGNOSIS

Common
- Positional
- Spina Bifida
- Body Stalk Anomaly
- VACTERL Association
- Vertebral Anomaly

Less Common
- Amniotic Band Syndrome
- Thanatophoric Dysplasia
- Achondroplasia
- Asphyxiating Thoracic Dysplasia

Rare but Important
- Conjoined Twins
- Iniencephaly

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is fetal position fixed or variable?
  - If variable, less likely to be significant problem
  - If fixed, evaluate global fetal movement
    - Any evidence of arthrogryposis/akinesia sequence
    - Serial evaluation important as akinesis sequences may be progressive
  - Look at abdominal wall and cord insertion site to exclude body stalk anomaly
  - Scan through amniotic fluid looking for bands
- Are bones of spine normal?
  - Use high frequency transducer for better resolution
  - Assess vertebral body height, mineralization, presence of 3 ossification centers at all levels
- Is spine length normal?
  - Caudal regression sequence may cause spine position to look unusual as loss of normal sacral curvature
    - Make sure vertebra are present between the iliac wings
  - Scan through cervical, thoracic, lumbar, and sacral spine systematically

Helpful Clues for Common Diagnoses
- **Positional**
  - Most commonly idiopathic due to fetal stretching movements
    - Fetus seen to move extremities
    - Normal amniotic fluid
    - Change in position on follow-up scans
  - May be due to fetal crowding
    - Multiple gestations, late pregnancy
  - Associated with oligohydramnios
    - Renal agenesis/other bilateral renal anomalies
    - Twin-twin transfusion syndrome donor
  - Look for synechiae
    - Fetal parts oriented around large synechia
  - Large uterine fibroids may distort cavity → unusual fetal position
  - Müllerian duct anomalies may result in decreased cavity size/unusual fetal lie
- **Spina Bifida**
  - Depending on level of neural tube defect (NTD) may see sharp kyphosis
    - Thoracic NTD most likely to result in abnormal spine position
    - Iniencephaly is high cervical NTD causing neck hyperextension due to shortening of cervical spine
  - Look for associated brain findings of Chiari II malformation
    - Ventriculomegaly, "banana" cerebellum, "lemon-shaped" head
  - Look for splayed/absent posterior elements
  - Look for associated myelomeningocele/myeloschisis
- **Body Stalk Anomaly**
  - Result of embryologic maldevelopment with interruption of normal embryonic folding process
    - Open abdominal wall defect
    - Peritoneum in continuity with amnion therefore fetus fixed to placenta
    - Body stalk/yolk stalk fusion fails: Short or absent umbilical cord
  - Normal cord development allows fetus to move freely within amniotic sac
  - If no cord, fetus tethered to uterine wall
    - Extremities and cranium move
    - Spine growth/elongation → hyperextension about fixed point where abdominal cavity is open and adherent to placenta
- **VACTERL Association**
  - Non-random association of seven core anomalies

- Vertebral anomalies
- Anal atresia
- Cardiac anomalies
- Tracheoesophageal atresia
- Renal anomalies
- Limb defects (radial ray)

- **Vertebral Anomaly**
  - Look for hemivertebra/fused vertebrae at apex of scoliosis/kyphosis
  - Ask for history of maternal diabetes, alcohol use, drug exposure (e.g., valproate)

Helpful Clues for Less Common Diagnoses

- **Thanatophoric Dysplasia**
  - Lethal skeletal dysplasia with severe micromelia
  - "Telephone receiver" femur in type 1
  - Kleeblattschädel (cloverleaf) skull type 2
  - Distinctive spine appearance
    - Marked platyspondyly with intervertebral disc height > vertebral body height (normally equal)
    - Lumbar kyphosis
- **Achondroplasia**
  - Thoracolumbar kyphosis
  - Narrowed interpedicular distance
  - Progressive rhizomelic limb shortening
  - Macrocephaly with frontal bossing
  - Trident hands
- **Asphyxiating Thoracic Dysplasia**
  - Thoracic lordosis, lumbar kyphosis
  - Short, horizontal ribs with small thorax

Helpful Clues for Rare Diagnoses

- **Conjoined Twins**
  - Contiguous skin covering between twins
  - Spines hyperextended due to fixed anterior point of union
  - Bridging tissue may be pliable → orientation of twins can vary from scan to scan
- **Iniencephaly**
  - Lethal extensive open neural tube defect
    - Defect in occiput and inion
    - Occipital encephalocele + spinal dysraphism
    - Fixed cervical hyperextension → "stargazer" fetus

Other Essential Information

- Twin gestation
  - Determine chorionicity
  - Conjoined twins are monochorionic
  - Differential diagnosis for fixed scoliosis in one twin of dichorionic twins is as for singleton
- Apparently isolated hemivertebra causing abnormal spine position may be "tip of the iceberg"
  - Perform formal fetal echocardiogram at 18-22 weeks
  - Look carefully for other stigmata of VACTERL association
  - Assess spinal cord with high resolution transducer if possible
    - Tethered cord
    - Lipoma
    - Diastematomyelia

Positional



Sagittal ultrasound shows prominent but transient thoracic kyphosis. This was a transient finding and is seen with normal fetal movement.

Positional

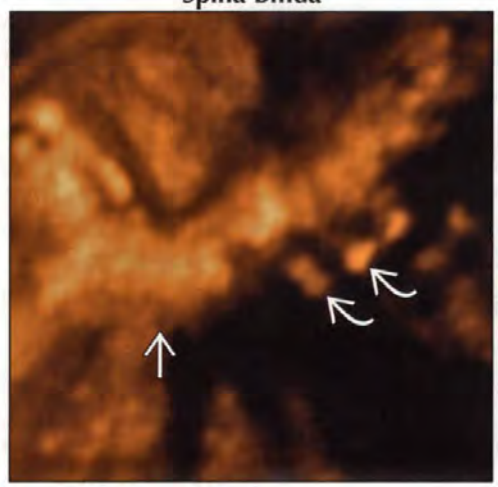


Sagittal ultrasound shows a 6 cm fibroid ➡ compressing the fetal spine ➡. This was exacerbated by transducer pressure. The fetus moved normally.

ABNORMAL SPINE POSITION

(Left) *3D ultrasound shows acute angulation ➡ of the thoracic spine in a fetus with a high thoracic neural tube defect. Origins of some of the upper ribs ➡ are seen; the rest of the torso was twisted out of plane.* ***(Right)*** *Axial oblique ultrasound in the same case as previous image shows the myelomeningocele sac ➡ involving the thoracic spine. Several ribs ➡ are imaged due to the degree of twisting of the torso.*

Spina Bifida



Spina Bifida



(Left) *Radiograph in the infant at birth shows a hemivertebra ➡ and several "jumbled" vertebral bodies inferiorly ➡. Increased tissue density in the midline is due to the mass of the associated myelomeningocele sac.* ***(Right)*** *Ultrasound shows two points of acute angulation of the spine ➡ in a fetus with the body stalk anomaly. Fixed spine position is a hallmark of body stalk anomaly.*

Spina Bifida

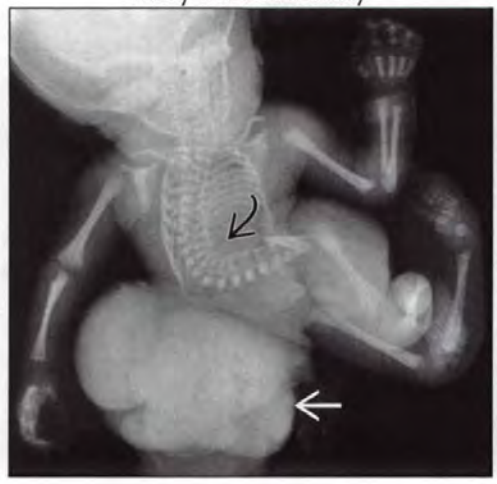


Body Stalk Anomaly

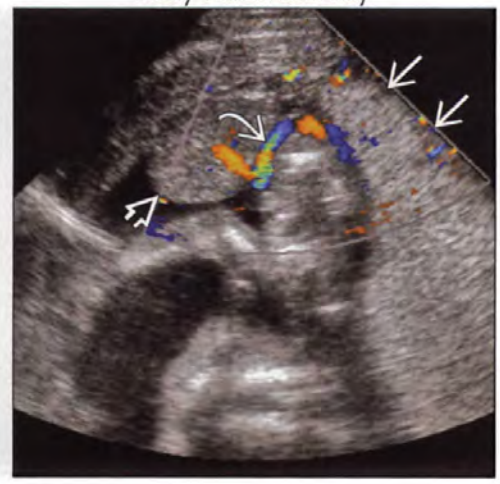


(Left) *Radiograph shows marked scoliosis ➡ associated with evisceration of the thoraco-abdominal contents ➡.* ***(Right)*** *Transvaginal ultrasound at 16 weeks gestation shows the fetus "stuck" to the placenta ➡. The liver ➡ is exteriorized. Umbilical vessels ➡ are seen to traverse the defect but there is no normal umbilical cord. Lack of normal cord development is another key feature of the body stalk anomaly.*

Body Stalk Anomaly



Body Stalk Anomaly



ABNORMAL SPINE POSITION

VACTERL Association

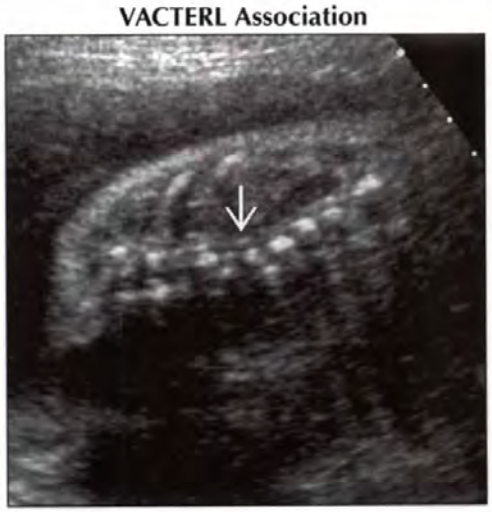


VACTERL Association

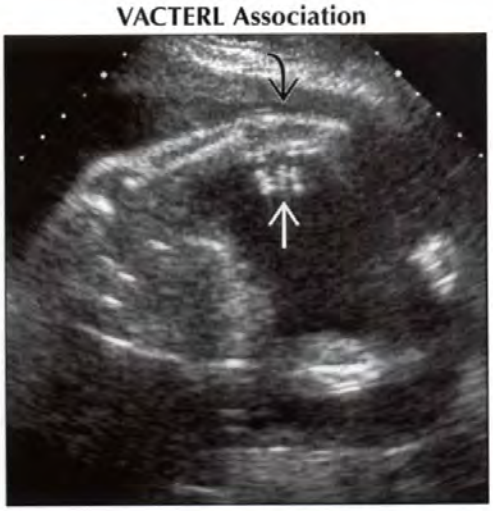


(Left) *Coronal ultrasound after amnioinfusion in a fetus with severe oligohydramnios showed persistent thoracolumbar scoliosis* ➡ *with at least one vertebral body anomaly* ➡. ***(Right)*** *Coronal color Doppler ultrasound in the same case as previous image confirms absent renal arteries* ➡. *Bilateral radial ray anomaly and rockerbottom feet were also present, a constellation of findings typical of the VACTERL association.*

VACTERL Association



VACTERL Association

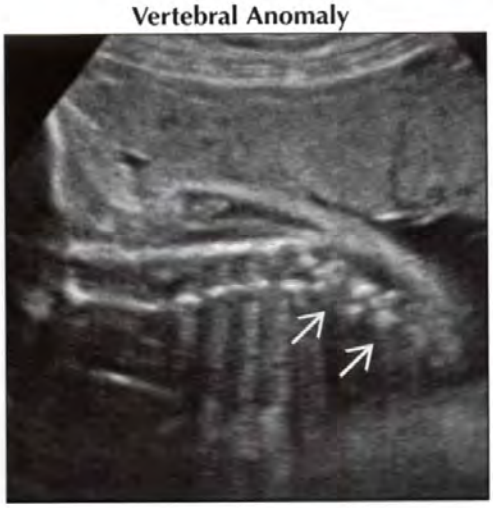


(Left) *Ultrasound shows persistent scoliosis* ➡ *in a fetus with multiple anomalies including complex congenital heart disease.* ***(Right)*** *Ultrasound in the same case as previous image shows a short forearm* ➡ *and an abnormally positioned hand* ➡ *with only 3 digits. Amniocentesis performed for suspicion of trisomy 18 showed normal chromosomes. VACTERL association provides a unifying diagnosis for this group of anomalies.*

Vertebral Anomaly



Vertebral Anomaly



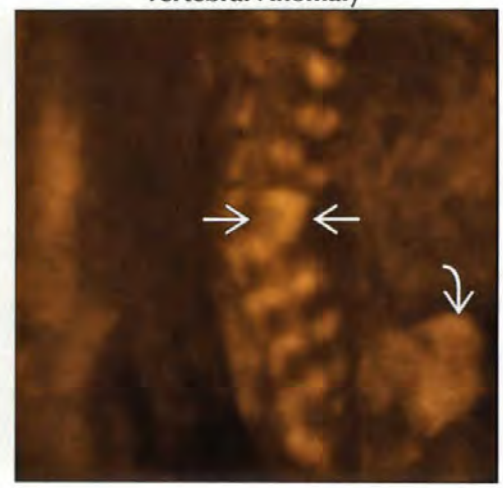
(Left) *Lateral radiograph shows complete loss of the lower lumbar vertebral bodies (12th rib denoted by* ➡*), with only rudimentary vertebrae at L1 and L2* ➡. *Five sacral segments* ➡ *are present. The infant was born to a poorly controlled diabetic who received little prenatal care.* ***(Right)*** *Sagittal ultrasound shows "jumbled" thoracic vertebrae* ➡ *in a fetus with diabetic embryopathy.*

ABNORMAL SPINE POSITION

Vertebral Anomaly



Vertebral Anomaly



(Left) *Sagittal ultrasound shows typical appearance of isolated focal kyphosis of the lumbar spine seen with 2D ultrasound.* ***(Right)*** *Coronal 3D ultrasound shows fusion of vertebral bodies and scoliosis (left iliac crest). The cisterna magna and cerebellum were normal, essentially ruling out an open neural tube defect.*

Amniotic Band Syndrome



Amniotic Band Syndrome

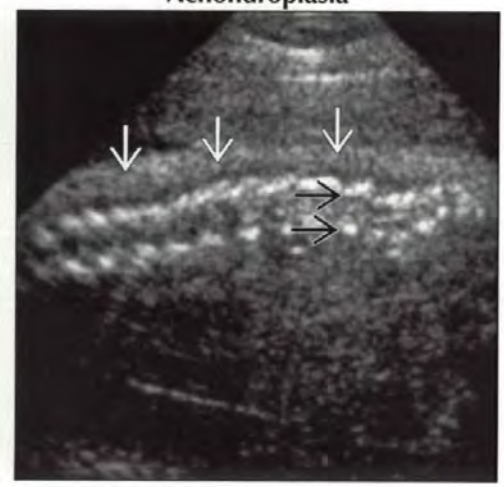


(Left) *Sagittal ultrasound shows a persistent thoracic kyphosis that was associated with a large abdominal wall defect, normal umbilical cord, and linear bands in the amniotic fluid, consistent with amniotic band syndrome.* ***(Right)*** *Coronal 3D ultrasound shows the fetus apparently "resting" on its liver. Note the apparent band.*

Thanatophoric Dysplasia



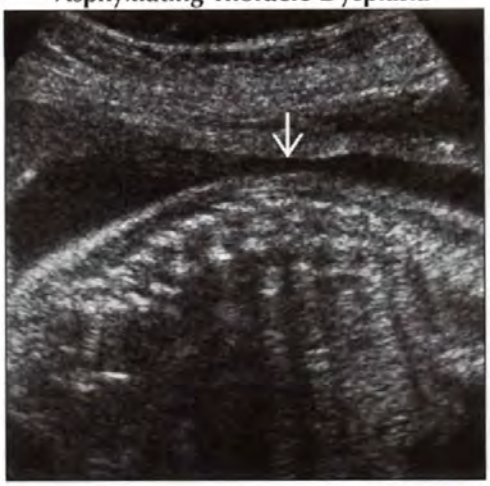
Achondroplasia



(Left) *Sagittal ultrasound shows the typical appearance of the spine in thanatophoric dysplasia in the third trimester. Note lumbar kyphosis, severe platyspondyly, and polyhydramnios. Trident hand and metaphyseal flaring were also seen.* ***(Right)*** *Coronal ultrasound of the spine shows thoracolumbar kyphosis with narrow interpedicular distance in a fetus with heterozygous achondroplasia.*

ABNORMAL SPINE POSITION

Asphyxiating Thoracic Dysplasia



Asphyxiating Thoracic Dysplasia



(Left) *Sagittal ultrasound shows a persistent lumbar kyphosis. Initial images showed possible cardiomegaly, which was, in fact, due to small thoracic circumference. The ribs were short and horizontal without fractures.* ***(Right)*** *Lateral radiograph of the thoracolumbar spine in the same case after birth confirms short horizontal ribs with a long narrow thorax. Note also the persistent lumbar kyphosis and thoracic lordosis.*

Conjoined Twins



Conjoined Twins

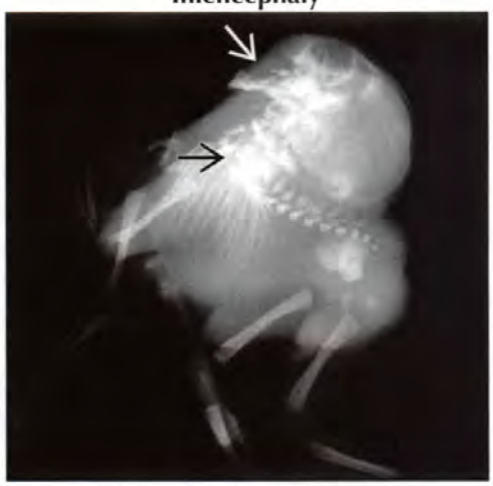


(Left) *Gross pathology shows thoracoomphalopagus twins joined at the chest and abdomen. The left twin has marked lumbar lordosis as well as cervical spine hyperextension.* ***(Right)*** *Radiograph in another set of thoracoomphalopagus twins shows cervical spine hyperextension in one of the twins. In conjoined twins with ventral fusion, the spines are often arched away from the site of fusion.*

Iniencephaly



Iniencephaly



(Left) *Sagittal ultrasound at 12 weeks gestation shows marked hyperextension of the head. The body appears small compared to the head, secondary to absent cervical and upper thoracic vertebrae. These are first trimester features of iniencephaly. Dilated ventricles were also present.* ***(Right)*** *Lateral radiograph shows the typical stargazer head position. There are abnormal/missing vertebrae with extreme extension of the cervical spine.*

ABNORMAL VERTEBRAL COLUMN

DIFFERENTIAL DIAGNOSIS

Common
- Spina Bifida
- Segmentation and Fusion Anomalies

Less Common
- Caudal Regression Sequence
- VACTERL Association
- Skeletal Dysplasias
  - Achondrogenesis
  - Thanatophoric Dysplasia

Rare but Important
- Iniencephaly
- Chiari III Malformation

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Are all vertebral bodies present?
  - Take care to assess all segments systematically
  - Caudal regression may be quite subtle when mild
  - Coronal views useful to see tapering sacral segments, look for hemivertebrae
- Are all posterior elements closed?
  - Axial views essential to avoid missing spinal dysraphism
  - Look for intracranial findings of Chiari II malformation if suspect neural tube defect
- Is spine orientation normal?
  - Does orientation change with fetal movement?
  - Is odd position/alignment due to crowding/amniotic cavity distortion?
- Is fluid volume normal?
  - Polyhydramnios: Skeletal dysplasias, tracheoesophageal fistula, maternal diabetes (macrosomia)
  - Oligohydramnios: Significant renal anomalies, maternal diabetes (microvascular disease)

Helpful Clues for Common Diagnoses
- **Spina Bifida**
  - Associated with elevated maternal serum alpha fetoprotein
  - Associated with brain findings of Chiari II malformation
  - Spinal laminae parallel or divergent
    - Normally converge to form a "teepee"
  - Look for associated myelomeningocele
    - Interrupted skin echo
    - Fine membranous sac ± contained neural elements
  - Look for associated myeloschisis
    - Interrupted skin echo with rolled edges, no sac
- **Segmentation and Fusion Anomalies**
  - Result from aberrant vertebral column formation
    - Block vertebra: Segmentation failure of two or more vertebral somites
    - Posterior neural arch anomalies: Dysraphism ± unilateral pedicle aplasia/hypoplasia
    - Partial or complete failure of formation: Vertebral aplasia, hemivertebra, butterfly vertebra
    - Partial duplication: Supernumerary hemivertebra
  - Often cause kyphosis/scoliosis
  - May be isolated but if found look for other malformations
    - Gastrointestinal (20%),
    - Congenital cardiac disease (10%)
    - Renal anomalies also described
  - Evaluate spinal cord carefully for associated tethering or diastematomyelia (split cord)
  - Check for maternal alcohol abuse/medications history (valproate)

Helpful Clues for Less Common Diagnoses
- **Caudal Regression Sequence**
  - Strong association with poorly controlled maternal diabetes
  - Due to missing vertebra rather than abnormal vertebra
  - Spine looks "short"
  - Tapers abnormally high, not at sacral tip
    - Assess cervical, thoracic, lumbar, & sacral segments systematically in every fetus
  - Look for vertebral bodies between iliac wings on axial images
  - "Shield sign" describes the appearance of apposed iliac wings when sacral vertebra are absent
  - Lower extremities often held in fixed "Buddha" or "cross-legged tailor" position
- **VACTERL Association**

- Non-random association of anomalies involving multiple organ systems except brain
  - Vertebral anomalies
  - Anal atresia
  - Cardiac anomalies
  - Tracheoesophageal fistula
  - Renal anomalies
  - Limb (radial ray)
- Three or more associated malformations required for diagnosis

- **Achondrogenesis**
  - Hallmark is lack of vertebral ossification
    - Type Ia: Completely unossified, associated with rib but not long bone fractures
    - Type Ib: Pedicles may be ossified
    - Type II: Deficient spine mineralization
  - Severe micromelia
  - Disproportionately large head
    - Ossification may be normal (type II) or deficient (type I)
  - Small thorax with protuberant abdomen
- **Thanatophoric Dysplasia**
  - Severe platyspondyly with prominent lumbar kyphosis
  - Associated with severe micromelia with prominent bowing but normal ossification
  - Telephone receiver femur in type I
  - Kleeblattschädel ("cloverleaf") skull deformity in type II
  - Small, narrow thorax

Helpful Clues for Rare Diagnoses

- **Iniencephaly**
  - Lethal open neural tube defect
  - High cervical location with severe shortening of cervical spine
  - Defect may extend to thoracic/lumbar spine (rachischisis)
  - Cervical vertebrae missing or fused
  - Associated occipital encephalocele
  - Fixed neck hyperextension → "stargazer" head position
- **Chiari III Malformation**
  - Intracranial findings of Chiari II
  - High cervical ± low occipital osseous defect
  - Herniation of posterior fossa contents
    - Cerebellum, fourth ventricle ± upper cervical cord through defect

Other Essential Information

- Use 3D ultrasound
  - Volume acquisition allows manipulation of data and may reveal findings not appreciated on 2D scans
  - Serial slices comparable to CT/MR allows assessment of level
  - Surface rendered reconstructions useful for global view and to counsel parents
- Think of the VACTERL association when vertebral anomalies are seen in a fetus with other known malformations and normal chromosomes
  - 28% neonatal mortality
  - 48% mortality in first year

Spina Bifida



Gross pathology shows a large thoracolumbar neural tube defect. The myelomeningocele membrane ➔ was torn during delivery. Note the open spinal column ➔ in the center of the soft tissue defect.

Spina Bifida



Sagittal oblique ultrasound shows a sacral neural tube defect ➔ with associated tethering of the cord. The conus ➔ is visible to the level of the defect; it should not be lower than L2/3.

ABNORMAL VERTEBRAL COLUMN

Segmentation and Fusion Anomalies



Segmentation and Fusion Anomalies



(Left) *Sagittal ultrasound shows a persistent "kink" at the thoracolumbar junction. This was an isolated finding in this fetus, but always look for other signs of VACTERL association.* ***(Right)*** *Axial ultrasound in the same case shows incomplete ossification of the left lateral half of the vertebral body at the level of the "kink". This is most consistent with an isolated hemivertebra. The parents elected no further prenatal evaluation.*

Caudal Regression Sequence

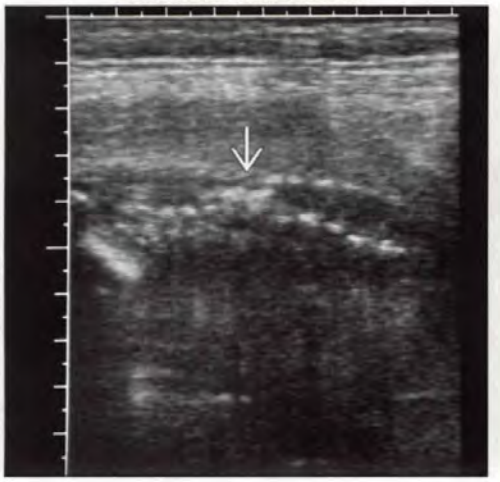


Caudal Regression Sequence

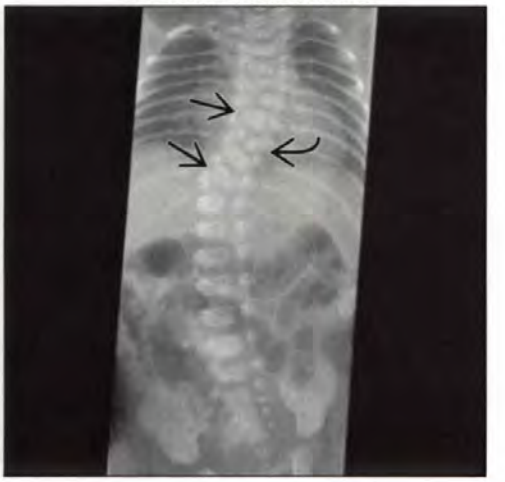


(Left) *Sagittal T2WI MR shows abrupt early termination of the spine associated with a small pelvis and multiple other malformations. The mother was not diabetic.* ***(Right)*** *Frontal radiograph in the same case as previous image shows additional vertebral anomalies in the cervical and thoracic spine, as well as mediastinal shift due to unilateral left pulmonary agenesis.*

VACTERL Association



VACTERL Association



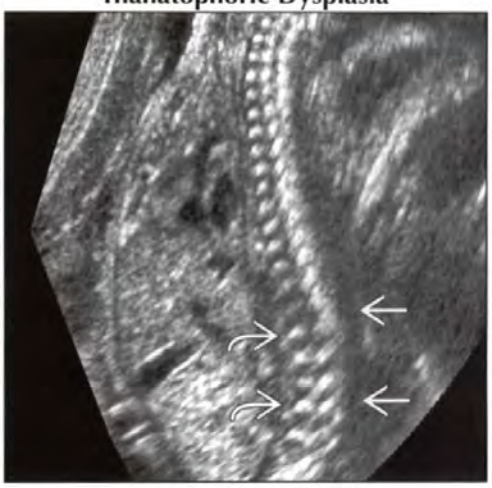
(Left) *Ultrasound shows the use of a high frequency transducer to improve bone detail. There was anhydramnios in this pregnancy, but multiple fused vertebrae are seen at the thoracolumbar junction. VACTERL sequence was confirmed at autopsy.* ***(Right)*** *Radiograph in a different, liveborn infant with multiple anomalies in keeping with VACTERL association shows hemivertebra, butterfly vertebra, and double curve scoliosis.*

ABNORMAL VERTEBRAL COLUMN

Achondrogenesis



Thanatophoric Dysplasia



(Left) *Sagittal ultrasound shows absent/decreased ossification in the thoracic* ➡ *and lumbar spine* ➡. *In association with severe limb shortening, this is typical of achondrogenesis.* ***(Right)*** *Sagittal ultrasound shows prominent lumbar kyphosis* ➡ *and platyspondyly* ➡, *typical for thanatophoric dysplasia.*

Thanatophoric Dysplasia



Iniencephaly



(Left) *Sagittal radiograph performed as part of an autopsy shows the same features of prominent lumbar kyphosis* ➡ *and platyspondyly* ➡ *in this lethal skeletal dysplasia.* ***(Right)*** *Sagittal oblique transvaginal ultrasound at 12 weeks gestation shows a short and small appearing torso* ➡ *and large occipital encephalocele* ➡, *allowing confident diagnosis of iniencephaly.*

Chiari III Malformation



Chiari III Malformation



(Left) *Axial ultrasound shows a large occipital encephalocele* ➡ *in a patient who presented late in pregnancy.* ***(Right)*** *Sagittal ultrasound in the same case as previous image shows associated cervical dysraphism* ➡ *with herniation of the posterior fossa contents and fourth ventricle* ➡. *Given the poor prognosis, the couple elected nonintervention during labor.*

SECTION 3
Face/Neck

Facial Cleft	3-2
Micrognathia	3-6
Abnormal Midface	3-12
Abnormal Eyes	3-16
Abnormal Ears	3-22
Macroglossia	3-24
Facial Mass	3-26
Neck Mass	3-30

FACIAL CLEFT

DIFFERENTIAL DIAGNOSIS

Common
- Isolated Cleft Lip, Palate
- Trisomy 18
- Trisomy 13

Less Common
- Holoprosencephaly
- Amniotic Band Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Routine face views
  - Profile
  - Coronal nose and lip
- 3D ultrasound technique for fetal face
  - Obtain good 2D profile view
    - Midsagittal
    - Fluid in front of face
  - Perform 3D sweep or 4D acquisition
    - 4D is real time 3D
    - 4D resolution may not be as good as 3D sweep acquisition
  - Soft tissue rendered view
    - Look for intact lip
    - Look at nares and eyes
  - Bone rendered view
    - Look for intact palate
  - Reverse face 3D technique
    - Flip acquired profile view so baby is looking "down"
    - Rendered 3D view looks from "behind the palate"
    - Shadow artifact is decreased
    - May improve detection of soft palate defects and small bony defects
- "Face predicts the brain"
  - Look carefully at fetal brain if any facial anomaly identified
  - Consider brain MR
    - Midline facial defects associated with midline brain defects
    - Better than ultrasound for subtle abnormalities, such as gray matter heterotopia
- Isolated cleft palate is often missed
  - Posterior soft tissue defect
  - Fetal MR helpful
  - Sagittal view may be best
    - Fluid in nasal cavity
    - Back of tongue in defect

Helpful Clues for Common Diagnoses
- **Isolated Cleft Lip, Palate**
  - Type 1 cleft lip (CL)
    - Unilateral CL
    - No palate defect
    - ± Flattened nares
  - Type 2 CL, cleft palate (CP)
    - 80% of all CL, CP
    - Unilateral CL and CP
    - Associated flat nares
  - Type 3 CL, CP
    - Bilateral CL and CP
    - Premaxillary protrusion on profile view
    - Dysplastic medial anterior palate is mass-like and protrudes anteriorly
    - Severe nose deformity often seen
  - Type 4 CL, CP
    - Midline CL and CP
    - Anterior/mid palate defect
    - Midface hypoplasia
    - Flat dysplastic nose
  - Isolated CP
    - Rare
    - Often involves only posterior soft palate
    - Fetal MR may be best diagnostic test
- **Trisomy 18**
  - 15% with CL/CP (type 2)
    - Rarely type 1
  - Other facial anomalies
    - Micrognathia
    - Low set ears
  - Other associated markers/anomalies
    - Cardiac defects (90%)
    - Choroid plexus cysts (50%)
    - Clenched hand & overlapping index finger (50%)
    - Intrauterine growth restriction (50%)
    - Brain anomaly (30%)
    - Omphalocele (20%)
    - Spina bifida (12%)
    - Clubfoot
    - Rockerbottom foot
    - Diaphragmatic hernia
    - Bladder outlet obstruction
- **Trisomy 13**
  - 50% with facial anomalies
    - Type 3 or 4 CL/CP
    - Cyclopia
    - Hypotelorism
    - Proboscis

- Other associated markers/anomalies
  - Holoprosencephaly (40%)
  - Postaxial polydactyly (75%)
  - Cardiac defects (80%)
  - Intracardiac echogenic focus
  - Cystic kidneys (50%)
  - Omphalocele
  - Intrauterine growth restriction (50%)

Helpful Clues for Less Common Diagnoses

- **Holoprosencephaly**
  - Arrest in brain cleavage and rotation
  - Alobar holoprosencephaly
    - Most severe
    - Fused thalamus
    - Single ventricle
    - Brain mantle
    - Dorsal sac
  - Semilobar holoprosencephaly
    - Less severe than alobar
    - Partially fused thalamus
    - Monoventricle anterior & occipital horns
    - Callosal dysgenesis
  - Lobar holoprosencephaly
    - Most mild form
    - Absent cavum septi pellucidi
    - Fused fornices
    - Hypoplastic frontal lobes
    - ± Callosal dysgenesis
  - Associated facial anomalies
    - Cyclopia
    - Hypotelorism
    - Proboscis
    - Type 3 or 4 CL/CP
- **Amniotic Band Syndrome**
  - Entrapment of fetal parts by disrupted amnion
  - Bizarre facial clefts
    - Do not conform to pattern of developmental clefts
    - Often oblique through face
  - Other anomalies
    - Bizarre abdominal wall defects
    - Chest wall defects
    - Scoliosis
    - Atypical cephaloceles
    - Acrania/acalvaria-like defect
    - Amputations
    - Limb constrictions with lymphedema
  - Bands may or may not be visible

Other Essential Information

- Teratogens associated with CL/CP
  - Alcohol
  - Valproic acid
  - Hydantoin
  - Retinoic acid
- Aneuploidy rates with CL/CP
  - Type 1, rare
  - Type 2, 20%
  - Type 3, 30%
  - Type 4, 50%
- Ethnicity demographics for CL/CP
  - 1:600 Asian
  - 1:1,000 Caucasian
  - 1:2,500 African-American

Isolated Cleft Lip, Palate



Coronal 3D ultrasound shows a left-sided cleft lip with associated mild left nares flattening. No other fetal anomalies were seen and the family opted not to have an amniocentesis.

Isolated Cleft Lip, Palate



Clinical photograph of the same case after delivery shows the left cleft lip and nasal flattening. The baby was otherwise normal and did well after surgery.

FACIAL CLEFT

Isolated Cleft Lip, Palate





(Left) *Coronal 3D ultrasound bone rendered view shows a cleft palate defect in association with a cleft lip. This fetus has isolated type 2 cleft lip/palate.* ***(Right)*** *Axial ultrasound at the level of the fetal tongue in another fetus shows a midline cleft lip/palate. The tip of the nose is flattened and lies along the anterior edge of the defect. This fetus has a type 4 cleft lip and palate defect.*

Isolated Cleft Lip, Palate

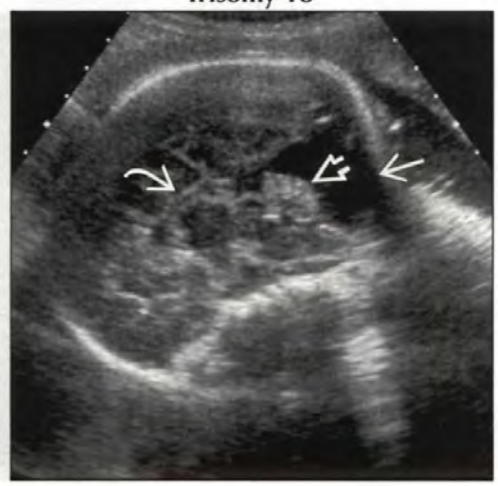




(Left) *Coronal 3D ultrasound shows a mild type 1 cleft lip. The cleft barely deforms the upper lip and does not extend to the nasal ala.* ***(Right)*** *Clinical photograph of the same fetus after delivery shows the minimal cleft in the lip. Note the nose is not flattened. There was no associated cleft palate.*

Trisomy 18





(Left) *Coronal ultrasound shows a bilateral cleft lip in a third trimester fetus.* ***(Right)*** *Sagittal ultrasound of the same fetus shows mega cisterna magna and a small cerebellum. In addition, there is partial absence of the corpus callosum with the anterior portion present and the posterior part missing. The fetus also had clenched hands. Upon delivery, trisomy 18 was diagnosed.*

FACIAL CLEFT

Trisomy 13

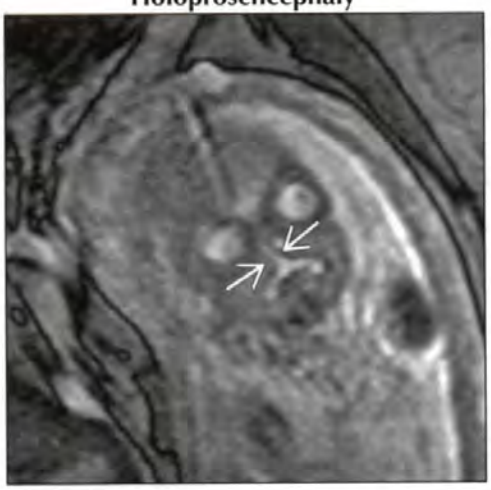


Trisomy 13

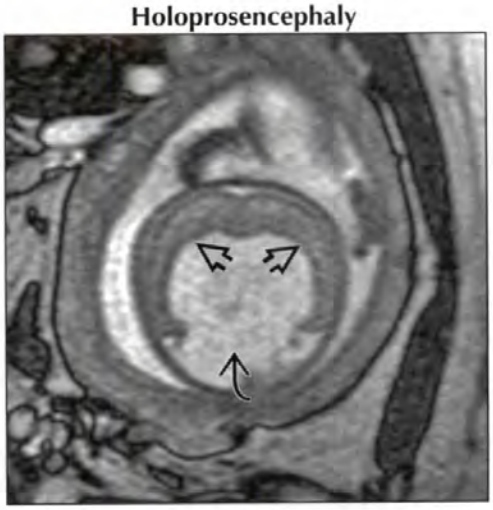


(Left) *Sagittal ultrasound of the fetal face shows a premaxillary protuberance. The appearance is caused by bilateral cleft lip and palate. The central bony maxillary tissue is dysplastic and mass-like.* ***(Right)*** *Axial ultrasound through the fetal heart in the same fetus shows an echogenic cardiac focus, a ventricular septal defect and a single atrium. Chromosome analysis was performed and revealed trisomy 13.*

Holoprosencephaly



Holoprosencephaly

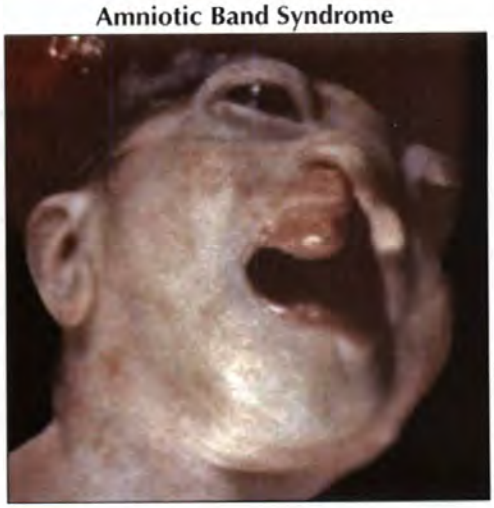


(Left) *Coronal T2WI MR of the fetal face shows a cleft palate. High signal fluid is seen within the defect, extending from the oral cavity to the nasal cavity.* ***(Right)*** *Axial T2WI MR through the calvarium of the same fetus shows an anterior brain mantle, lack of an interhemispheric falx and a monoventricle, consistent with holoprosencephaly.*

Amniotic Band Syndrome



Amniotic Band Syndrome



(Left) *Coronal ultrasound through the upper lip shows an oblique lip defect that extends to the nares. In addition, the fetus had a bizarre abdominal wall defect and acrania.* ***(Right)*** *Clinical photograph of the same fetus after delivery shows the large oblique facial cleft. Proptosis and acrania are also present. Oblique clefts, which do not conform to a normal developmental pattern, should always raise the suspicion of amniotic bands.*

MICROGNATHIA

DIFFERENTIAL DIAGNOSIS

Common
- Technical
- Idiopathic
- Oligohydramnios
- Trisomy 18

Less Common
- Amniotic Band Syndrome

Rare but Important
- Pierre Robin Syndrome
- Diabetic Embryopathy
- Treacher Collins Syndrome
- Cornelia de Lange Syndrome
- Otocephaly
- Other Syndromes/Conditions

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is micrognathia real or technical due to incorrect scan plane?
  - Reproducible finding if real
- Use 3D ultrasound if available
  - Helpful to assess additional dysmorphic features (e.g., ear malposition, ear malformation, eye orientation)
  - Volume acquisition increases likelihood that true midline sagittal view of profile is being analyzed
  - Surface rendering → way to qualitatively evaluate chin from different perspectives
    - Help parents understand appearance and consulting services plan treatment
- Mandibular measurements
  - Plethora of measurement described with some nomograms available, most are technically challenging & not widely used
    - Jaw index
    - Mandibular area
    - Inferior facial angle, mandibular angle
    - Mandible width/maxilla width ratio

Helpful Clues for Common Diagnoses
- **Technical**
  - Incorrect scan plane
- **Idiopathic**
  - No other abnormalities identified
  - May be familial; look at both parents
- **Oligohydramnios**
  - Part of Potter sequence
    - Beaked nose, redundant skin, low set ears, club feet
- **Trisomy 18**
  - Facial features include micrognathia and clefting
  - Usually associated with growth restriction and multiple anomalies
    - Omphalocele, congential heart disease, abnormal finger positioning, arthrogryposis/radial ray malformation, central nervous system anomalies, congenital diaphragmatic hernia

Helpful Clues for Less Common Diagnoses
- **Amniotic Band Syndrome**
  - Random constriction/amputation defects; "slash" defects
  - Careful search for bands mandatory as no significant recurrence risk
    - Linear echoes in amniotic fluid
    - Extend from fetal parts to uterine wall
    - Fetus appears tethered or in fixed position
  - Occasionally, inspection of placenta after delivery may be only way to confirm diagnosis

Helpful Clues for Rare Diagnoses
- **Pierre Robin Syndrome**
  - Micrognathia often severe
  - U-shaped palatal cleft hard to see sonographically as lip intact but may be evident on MR
  - Glossoptosis (posterior displacement of tongue), also easier to see on MR
- **Diabetic Embryopathy**
  - Caudal regression sequence ± extremity malformations
  - Brain malformations including holoprosencephaly
  - Congenital heart disease, especially transposition and double outlet right ventricle
  - Long standing diabetes → IUGR, oligohydramnios
  - Gastrointestinal malformations (e.g., anorectal atresia)
  - Genitourinary malformations (e.g., renal agnesis)
- **Treacher Collins Syndrome**
  - Genetic disorder characterized by craniofacial deformities
  - Down sloping palpebral fissures

- Malar hypoplasia
- Microtia

- **Cornelia de Lange Syndrome**
  - Typical facies: Prominent upper lip, crescent-shaped mouth, micrognathia, fine arched eyebrows, long eyelashes
  - Upper extremity limb reduction defects
  - Congenital diaphragmatic hernia, occasionally bilateral
  - Intrauterine growth restriction (IUGR)
- **Otocephaly**
  - Extremely rare but lethal anomaly
    - Microstomia
    - Aglossia or oroglossal hypoplasia
    - Agnathia or mandibular hypoplasia
    - Synotia: Low set medially rotated ears
  - Fetus cannot swallow → marked polyhydramnios
- **Other Syndromes/Conditions**
  - Online Mendelian Inheritance in Man (OMIM) database includes 211 genetic conditions featuring micrognathia
  - Many other multiple anomaly complexes without unifying diagnosis also feature micrognathia
  - Micrognathia is rarely isolated
  - Presence mandates careful search for other anomalies and consideration of of formal fetal echocardiography
    - 22q11 deletion associated with micrognathia and conotruncal malformations
  - Families should be evaluated by clinical genetics service
- Important to recognize autosomal recessive conditions as 25% recurrence risk
  - Neu-Laxová syndrome
    - Lethal syndrome with IUGR
    - Microcephaly
    - Exophthalmos, absent eyelids
  - Nager syndrome
    - Severe micrognathia and malar hypoplasia
    - Spectrum of radial ray malformations
    - 28% survival reported
    - Some dispute as to nature of inheritance, some cases appear dominant

Other Essential Information

- Micrognathia may be associated with skeletal dysplasia
  - Assess bone density
  - Measure long bone lengths
  - Look at vertebral contours
  - Use 3D ultrasound
- If associated with ear anomalies may also be associated with renal malformations
  - Neonatal renal ultrasound worthwhile
- Other potential complications
  - Polyhydramnios → increased risk of preterm labor
  - Respiratory distress ± feeding difficulties and typically requires one or more surgical procedures for repair
  - Increased risk of genetic/syndromic condition

Technical



Sagittal ultrasound shows possible micrognathia ➡ in the second trimester. No other abnormal findings. Because fetal position precluded a better profile view, follow-up was suggested.

Technical



Sagittal transabdominal ultrasound in the same fetus as previous image, shows normal profile in the third trimester. The apparent micrognathia was due to incorrect scan plane.

MICROGNATHIA

Oligohydramnios



Oligohydramnios



(Left) *Clinical photograph shows typical Potter syndrome facies with low set ears and flattened nose. Also note micrognathia.* ***(Right)*** *Transabdominal ultrasound is extremely compromised by maternal obesity and oligohydramnios. The lack of fluid causes compression of the fetus resulting in the "squashed" appearance seen in Potter syndrome, which was due to renal agenesis in this case.*

Trisomy 18

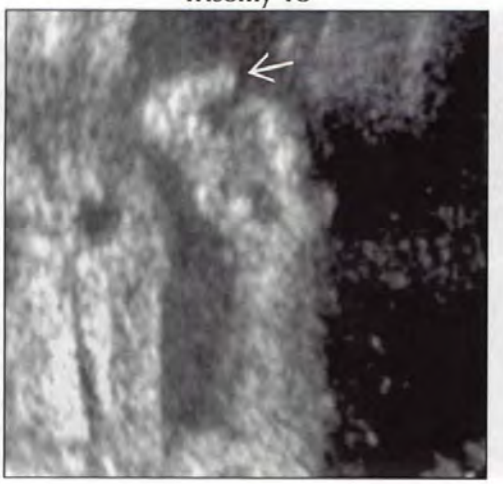


Trisomy 18



(Left) *Sagittal ultrasound shows a small mandible in a fetus with multiple other anomalies and polyhydramnios. Amniocentesis showed trisomy 18.* ***(Right)*** *Clinical photograph in the same patient, after near term intrauterine fetal demise, confirms micrognathia.*

Trisomy 18



Trisomy 18



(Left) *Transabdominal ultrasound shows abnormal posturing of the hand with an over-riding index finger, typical of that seen in trisomy 18.* ***(Right)*** *Axial transabdominal ultrasound shows a strawberry-shaped head. This is associated with trisomy 18, especially if seen in association with multiple anomalies.*

MICROGNATHIA

Amniotic Band Syndrome



Amniotic Band Syndrome

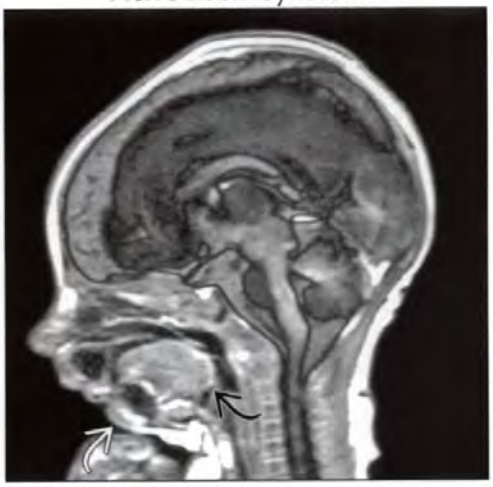


(Left) *Coronal transabdominal ultrasound shows hypertelorism, externalized brain and a small mandible.* ***(Right)*** *Transabdominal ultrasound in the same case as previous image, shows a band in the amniotic fluid anchoring the externalized brain to the uterine side wall.*

Pierre Robin Syndrome

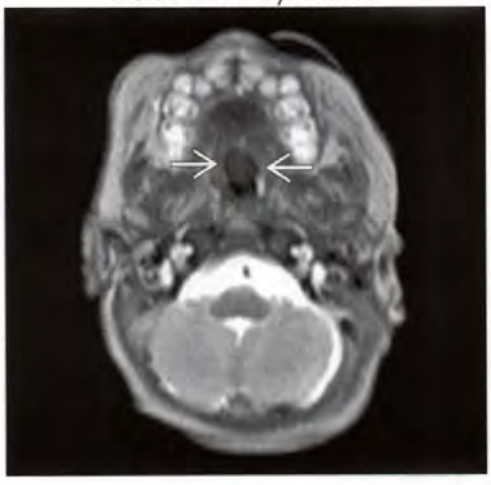


Pierre Robin Syndrome



(Left) *Sagittal ultrasound shows polyhydramnios and micrognathia. Lissencephaly was also present. Polyhydramnios may have been caused by neurological impairment and/or mechanical problems with swallowing due to micrognathia.* ***(Right)*** *Sagittal T1WI MR after delivery shows micrognathia and glossoptosis (posterior displacement of tongue), typical of Pierre Robin syndrome. Lissencephaly and heterotopia were also confirmed.*

Pierre Robin Syndrome



Pierre Robin Syndrome



(Left) *Axial T2WI MR in the same infant as previous image, shows the typical U-shaped palatal cleft.* ***(Right)*** *Sagittal ultrasound in a different case shows micrognathia with marked protuberance of the upper lip. The cavum septi pellucidi was not seen and fetal MR was performed. This showed callosal dysgenesis as well as a palatal cleft and glossoptosis. Pierre Robin syndrome was confirmed at birth.*

MICROGNATHIA

Diabetic Embryopathy



Diabetic Embryopathy

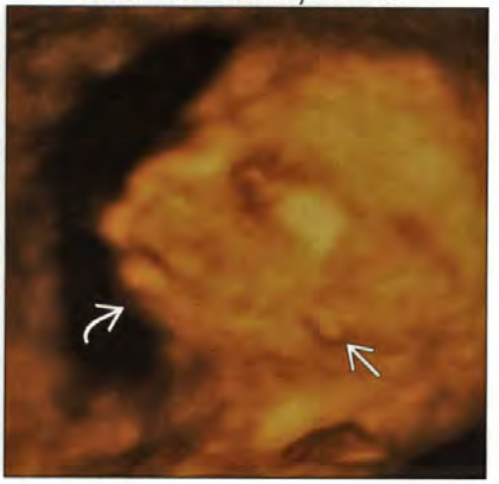


(Left) *Coronal ultrasound through the midface of a 15 week fetus shows a palate ⇒ but no mandible ⇒. This fetus also had severe caudal regression. The mother was a poorly controlled insulin dependent diabetic.* ***(Right)*** *Clinical photograph after termination shows severe micrognathia, confirming the ultrasound findings. Also note the small low-set ears ⇒.*

Treacher Collins Syndrome



Treacher Collins Syndrome

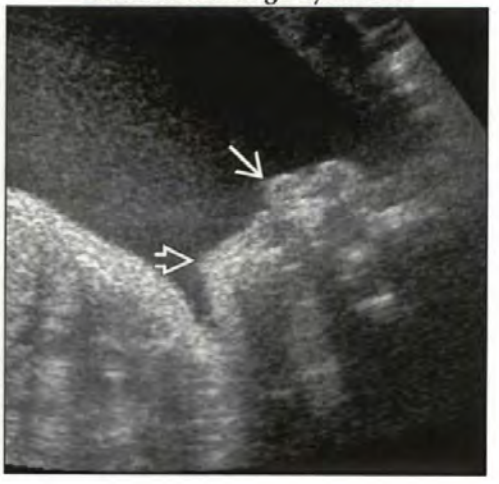


(Left) *Sagittal ultrasound shows severe retrognathia and micrognathia ⇒ typical for this condition.* ***(Right)*** *Sagittal oblique 3D ultrasound in the same fetus as previous image, shows microtia ⇒ as well as the severe micrognathia ⇒.*

Cornelia de Lange Syndrome



Cornelia de Lange Syndrome



(Left) *Clinical photograph shows the typical facial appearance including hirsutism, low set hairline and high arched brows. Micrognathia ⇒, upper lip protuberance ⇒ and limb reduction defects ⇒ are characteristic findings on prenatal ultrasound.* ***(Right)*** *Sagittal transabdominal ultrasound shows the fetal sonographic correlation in this case. The micrognathia ⇒ and upper lip prominence ⇒ are characteristic of this syndrome.*

MICROGNATHIA

Cornelia de Lange Syndrome



Cornelia de Lange Syndrome



(Left) *Transabdominal ultrasound in the same case as previous image, shows bilateral limb reduction defects, one of which is seen here. The limbs are well-seen even in the third trimester due to polyhydramnios.* ***(Right)*** *Coronal transabdominal ultrasound through the chest shows a large right-sided diaphragmatic hernia with liver and gallbladder in the chest. Note that the stomach has been pulled over to a midline location.*

Otocephaly



Otocephaly

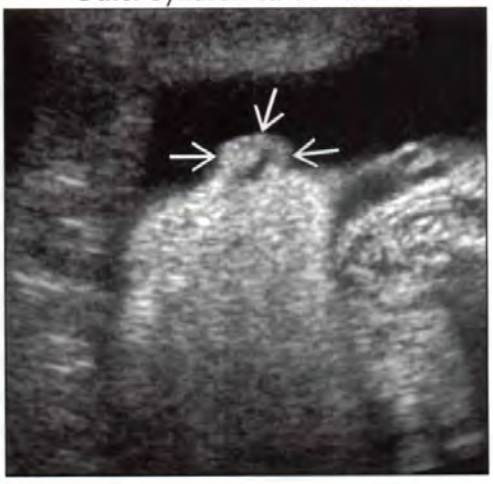


(Left) *Sagittal transabdominal ultrasound shows polyhydramnios and severe micrognathia.* ***(Right)*** *Clinical photograph of the infant shows the typical features of otocephaly including microstomia, synotia (low-set medially rotated ears), and agnathia.*

Other Syndromes/Conditions



Other Syndromes/Conditions



(Left) *Sagittal ultrasound shows micrognathia. There was also polyhydramnios, which is a typical associated finding secondary to impaired swallowing.* ***(Right)*** *Coronal ultrasound in the same case as previous image shows low-set, small ears. Ear anomalies are often seen with micrognathia. There was no definite unifying diagnosis in this case.*

ABNORMAL MIDFACE

DIFFERENTIAL DIAGNOSIS

Common
- Midface Hypoplasia
- Absent/Small Nasal Bone (NB)
- Trisomy 21
- Trisomy 13
- Cleft Lip, Palate
- Holoprosencephaly Spectrum

Less Common
- Skeletal Dysplasia

Rare but Important
- Warfarin (Coumadin) Exposure

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Obtain profile view routinely
  - Assess nasal bone
    - Should be present in first trimester
    - > 2.5 mm in second trimester
  - Note relationship of maxilla to mandible
    - Mandible slightly posterior to maxilla
  - Use 3D to reconstruct profile if not obtained by 2D
- Look for intact lip and palate routinely
  - Coronal nose/lip view
    - Axial palate view if suspect defect
  - 3D ultrasound helpful
- Cleft lip/palate (CL/CP) classification
  - Type 1: CL only, no CP
  - Type 2: Unilateral CL and CP
  - Type 3: Bilateral CL and CP
  - Type 4: Midline CL and CP
  - CP only is rare and difficult to diagnose
    - Posterior soft tissue defect
    - MR may be a better test

Helpful Clues for Common Diagnoses
- **Midface Hypoplasia**
  - Flat profile
    - Depressed nasal bridge
  - Maxillary hypoplasia
    - Maxilla lines up with mandible
    - Reverse overbite, if severe
  - Often associated with other craniofacial anomalies
    - Craniosynostosis
    - Hypotelorism
    - Hypertelorism
    - Cleft lip and palate
- **Absent/Small Nasal Bone (NB)**
  - Absent NB in first trimester
    - Assess for NB at time of nuchal translucency screening
  - Small NB in second trimester
    - < 2.5 mm at 15-20 wks
  - Marker for trisomy 21, but most often seen in normals, especially in Asian population
  - Likelihood ratios (LR) of trisomy 21
    - LR of 35 if absent NB in 1st trimester
    - LR of 9 if small NB in 2nd trimester
- **Trisomy 21**
  - Flat face
    - Small nose
    - Midface hypoplasia
  - Other markers
    - Increased nuchal translucency/fold
    - Mild ventriculomegaly
    - Short humerus/femur
    - Echogenic bowel
    - Echogenic cardiac focus
    - Renal pelviectasis
    - Clinodactyly
  - Major anomalies
    - Atrioventricular septal defect
    - Duodenal atresia
- **Trisomy 13**
  - Holoprosencephaly (40%)
    - + Associated facial anomalies
    - Type 3 or 4 cleft lip/palate
  - Polydactyly (75%)
  - Cardiac defects (80%)
  - Echogenic cystic kidneys (50%)
  - Intrauterine growth restriction (50%)
- **Cleft Lip, Palate**
  - Nose is affected by CL/CP
    - Cleft extends to nares
    - Flat nares with types 1, 2
  - Flat midface
    - Most common with type 4
    - Nose invaginates towards defect
  - Premaxillary protuberance
    - Mass-like area just below nose
    - 2° to dysplastic anterior palate
    - Most often seen with type 3 CL/CP
    - Large type 2 with abnormal profile also
  - CL/CP associations
    - Trisomy 18, 13
    - Holoprosencephaly
  - Aneuploidy and type of CL /CP
    - Type 1: Rare

- Type 2: 20%
- Type 3: 30%
- Type 4: 50%

- **Holoprosencephaly Spectrum**
  - Alobar, semilobar, lobar
  - Cyclopia with proboscis
  - Ethmocephaly
    - Proboscis with hypotelorism
  - Cebocephaly
    - Flat nose with single nostril
  - Median cleft lip/palate

Helpful Clues for Less Common Diagnoses

- **Skeletal Dysplasia**
  - Thanatophoric dysplasia
    - Cloverleaf skull (Kleeblattschädel)
    - Frontal bossing
    - Short upturned nose
    - Depressed nasal bridge
    - Micromelia
    - "Telephone receiver" femur
    - Platyspondyly
  - Achondroplasia (heterozygous)
    - Frontal bossing
    - Depressed nasal bridge
    - Progressive rhizomelia
    - Trident hands
  - Achondroplasia (homozygous)
    - Severe and early bone shortening
    - Lethal

Helpful Clues for Rare Diagnoses

- **Warfarin (Coumadin) Exposure**
  - Severe nasal hypoplasia
  - Rhizomelia (proximal limb shortening)
  - Stippled epiphyses

Other Essential Information

- Craniosynostosis syndromes associated with midface hypoplasia
  - Apert syndrome
    - Acrocephalosyndactyly type 1
    - Coronal suture fusion
    - Conical skull shape
    - "Mitten" hand and feet syndactyly
    - Extensive, often boney, fusion
  - Carpenter syndrome
    - Acrocephalosyndactyly type 2
    - Multiple suture craniosynostosis
    - Preaxial polydactyly
    - Partial syndactyly
    - Cardiac defects
    - Omphalocele, hernia
- Other syndromes associated with midface hypoplasia
  - Cornelia de Lange syndrome
    - Protruding upper lip with micrognathia
    - Upper limb reduction
    - Diaphragmatic hernia
    - Short 1st metacarpal
  - Fryns syndrome
    - Depressed nasal bridge
    - Orofacial cleft
    - Cardiac defect
    - Diaphragmatic hernia
    - Digital hypoplasia

Midface Hypoplasia



Sagittal ultrasound shows a flat midface. The nose and maxilla are small. The mandible protrudes anterior to the maxilla. This fetus had other anomalies as well.

Absent/Small Nasal Bone



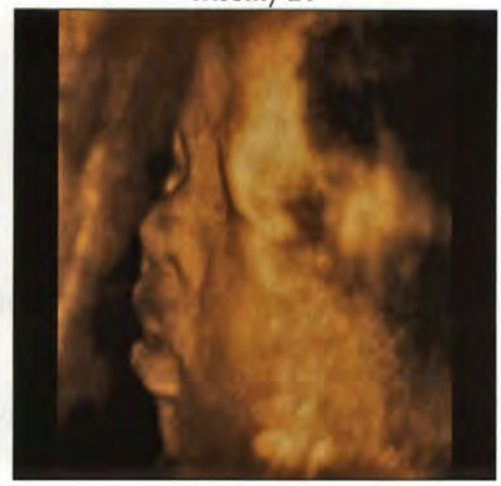
Sagittal ultrasound at 18 weeks shows a small nasal bone. While most fetuses with this finding are normal, it is associated with trisomy 21 and a careful targeted exam should be performed.

ABNORMAL MIDFACE

Trisomy 21



Trisomy 21



(Left) *Sagittal ultrasound in the second trimester shows a depressed nasal bridge with a hypoplastic nasal bone and a flat midface. No other anomalies were appreciated and genetic testing was declined.* ***(Right)*** *Sagittal 3D ultrasound of the same fetus as previous image, in the third trimester shows a flat, small midface, typical for trisomy 21. The diagnosis was made after delivery.*

Trisomy 13

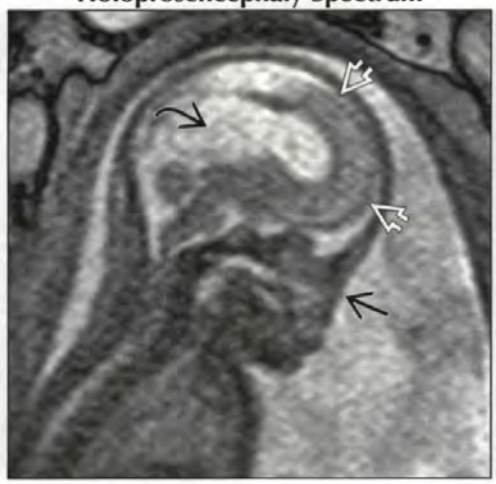


Cleft Lip, Palate



(Left) *Sagittal transvaginal ultrasound through the fetal head at 14 weeks shows a premaxillary mass and nuchal edema. The premaxillary mass suggests the presence of bilateral CL/CP. Chorionic villus sampling was performed on the same day and yielded trisomy 13.* ***(Right)*** *Sagittal ultrasound of another fetus shows a flat face secondary to a large palate defect. Fluid is seen extending into the nasal cavity. The nose is flat and deformed.*

Holoprosencephaly Spectrum



Holoprosencephaly Spectrum



(Left) *Sagittal T2WI MR shows an absent nose and a flat midface in a fetus with holoprosencephaly. An anterior brain mantle and monoventricle are also seen in this view.* ***(Right)*** *3D ultrasound shows a flat, small midface and a tiny proboscis in another fetus with holoprosencephaly, hypotelorism, and normal chromosomes.*

ABNORMAL MIDFACE

Holoprosencephaly Spectrum

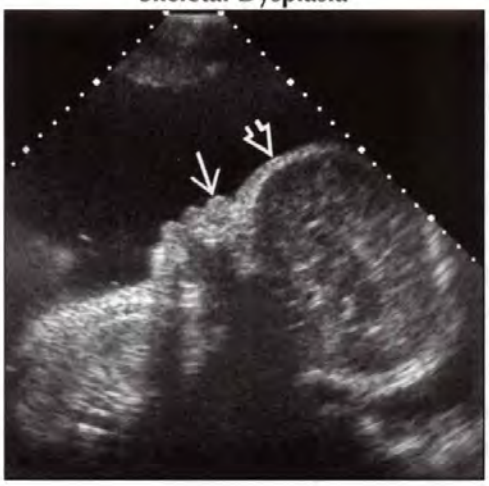


Holoprosencephaly Spectrum



(Left) *Sagittal ultrasound shows a midline orbit, above which is a proboscis in a fetus with cyclopia and holoprosencephaly.* ***(Right)*** *Sagittal ultrasound of another fetus with holoprosencephaly and trisomy 13 shows a proboscis adjacent to the fetal forehead. The midface is small and flat. A two vessel cord is also seen.*

Skeletal Dysplasia



Skeletal Dysplasia



(Left) *Sagittal ultrasound of a fetus with thanatophoric dysplasia shows a short upturned nose, frontal bossing, and a flat midface. The limbs were extremely short, and there was also chest hypoplasia and severe polyhydramnios.* ***(Right)*** *Sagittal radiograph of the same fetus as previous image, after delivery, shows an abnormal head shape with craniosynostosis and midface hypoplasia.*

Warfarin (Coumadin) Exposure



Warfarin (Coumadin) Exposure



(Left) *Sagittal ultrasound of a third trimester fetus exposed to warfarin shows severe nasal and midface hypoplasia.* ***(Right)*** *Clinical photograph of the same fetus as previous image, after delivery, confirms the ultrasound findings.*

ABNORMAL EYES

DIFFERENTIAL DIAGNOSIS

Common
- Hypotelorism
- Hypertelorism

Less Common
- Proptosis
- Dacrocystocele

Rare but Important
- Orbital Tumors
- Anophthalmia/Microphthalmia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Consider routine imaging of fetal orbits
  - Axial view at level of eyes
    - Evaluate bony orbit and globes
  - Coronal face view with 3D ultrasound
- Orbital biometry
  - Binocular diameter (BOD)
    - Outer to outer margin of both orbits
  - Interocular diameter (IOD)
    - Inner to inner margin between orbits
  - Ocular diameter (OD)
    - Single bony orbit diameter
- Rule of "thirds" for normal biometry
  - Normal IOD = OD
    - A "third eye" should fit between orbits
- Orbital nomograms are available
  - Gestational age vs. OD, IOD, BOD values
  - IOD/BOD percentiles
- "Face predicts the brain"
  - Look carefully for brain anomalies
  - Look for other face anomalies, including subtle findings such as lack of vermillion border

Helpful Clues for Common Diagnoses
- **Hypotelorism**
  - Eyes too close together
    - ↓ IOD, ↓ BOD
  - Hypotelorism is rarely an isolated finding
    - Holoprosencephaly is a major association
  - Associated nose anomalies
    - Proboscis (tube-like nose)
    - Nose with single nostril
    - Superior displacement of nose or proboscis is a common finding
  - Cyclopia is most severe type
    - Single bony orbit
    - Variable amount of globe doubling
    - Dysplastic tissue may cover orbit
    - Proboscis often above eyes
  - Ethmocephaly
    - Proboscis separates close set eyes
  - Cebocephaly
    - Infraorbital flat nose with single nostril
  - Associated cleft lip and palate
    - Most common is median cleft lip/palate
- **Hypertelorism**
  - Eyes too far apart
    - ↑ IOD
    - ↑ BOD is less prominent feature
  - Associated brain anomalies may be subtle
    - Agenesis of corpus callosum
  - Other craniofacial defects common
    - Bilateral cleft lip and palate
    - Large unilateral cleft lip and palate
    - Craniosynostosis

Helpful Clues for Less Common Diagnoses
- **Proptosis**
  - Exophytic eyes
  - Forward displacement of globes
  - Often associated with boney cranial anomalies
- **Dacrocystocele**
  - Dilatation of lacrimal drainage system
    - Imperforate valves of Hasner
    - Can be large and cause nasal obstruction
  - "Cyst" medial to orbit
    - Unilateral or bilateral
  - Most resolve on own
    - In utero or during first year of life

Helpful Clues for Rare Diagnoses
- **Orbital Tumors**
  - Solid mass in orbit
    - Often large and vascular
  - Globe may be displaced or destroyed
    - Unilateral proptosis most common finding
  - Boney orbit distorted or destroyed
  - Look for calcifications in mass (teratoma)
- **Anophthalmia/Microphthalmia**
  - Absent or small globe
    - Optic vesicle fails to form appropriately
  - Unilateral or bilateral

Other Essential Information
- 3D ultrasound extremely helpful
  - Soft tissue detailed anatomy
  - Bone-rendered images

- Facial clefts
- Fused sutures (craniosynostosis)
- Helps non-imagers see the anomalies
- Parents, genetic counselors, surgeons

- Fetal MR
  - Helps identify subtle and additional brain anomalies
  - Better delineate extent of tumors
- Look for signs of holoprosencephaly if hypotelorism present
  - Alobar holoprosencephaly
    - Severe facial anomalies
    - Single ventricle
    - No falx
    - Fused thalami
    - Dorsal sac
  - Semilobar holoprosencephaly
    - Less severe than alobar
    - Face with less severe anomalies or normal
    - Monoventricle anteriorly with separate occipital horns
    - Incomplete falx
    - Partially cleaved thalamus
  - Lobar holoprosencephaly
    - Often with normal face
    - Least severe form
    - Absent cavum septi pellucidi
    - Fused fornices

Alternative Differential Approaches

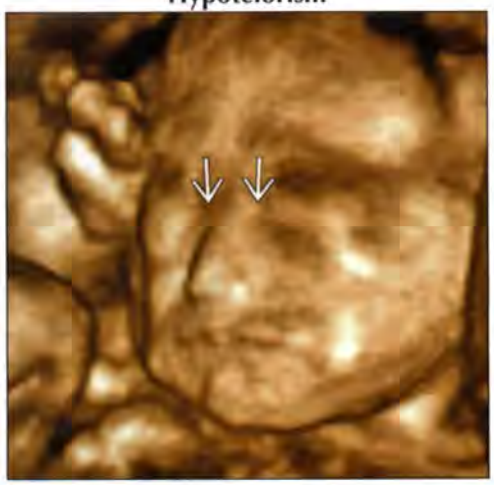
- Hypotelorism Associations
  - Trisomy 13
  - Trisomy 18
  - Microcephaly
  - Craniosynostosis
    - Trigonocephaly
  - Smith-Lemli-Opitz syndrome
  - Meckel-Gruber syndrome
  - Myotonic dystrophy
- Hypertelorism Associations
  - Agenesis of corpus callosum
  - Craniosynostosis
    - Apert syndrome
    - Carpenter syndrome
    - Crouzon syndrome
    - Thanatophoric dysplasia
  - Anterior encephalocele
  - Midline facial mass or cleft
  - Turner syndrome
  - Trisomy 13
  - Antiepileptic drug use
- Proptosis Associations
  - Anencephaly
  - Apert syndrome
  - Treacher-Collins syndrome
  - Pfeiffer syndrome
  - Neu-Laxová syndrome
- Anophthalmus/Micropthalmus Causes
  - Single gene mutation
  - Infection
  - X-ray exposure
- Orbital Tumors Types
  - Teratoma
  - Retinoblastoma
  - Soft tissue sarcoma
  - Rhabdoid tumor
  - Neurofibroma

Hypotelorism



Axial ultrasound shows the two orbits ➡ are very close to each other. A small interocular distance ➡ is seen. The fetus also had semilobar holoprosencephaly and normal chromosomes.

Hypotelorism



Coronal 3D ultrasound of the same fetus shows the decreased interocular distance ➡. In addition, the fetus has a small nose and a flat midface.

ABNORMAL EYES

Hypotelorism

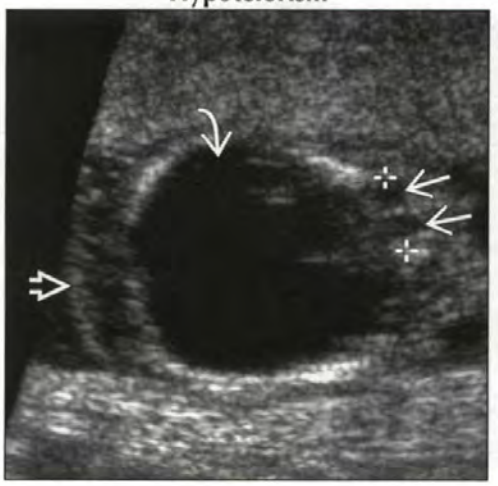


Hypotelorism



(Left) *Coronal ultrasound shows two small orbits with no significant intraorbital distance. The nose is missing and the fetal mouth is small without a normal upper lip vermillion border.* ***(Right)*** *Coronal ultrasound in another case shows a single orbit without a discernible globe. In addition, a proboscis is seen en face and superior to the orbit.*

Hypotelorism



Hypotelorism



(Left) *Axial ultrasound shows two globes in one bony orbit (calipers). Note the nuchal skin thickening and the fluid-filled calvarium.* ***(Right)*** *Coronal ultrasound through the head in the same fetus shows fused thalami. This fetus had alobar holoprosencephaly and trisomy 13.*

Hypertelorism



Hypertelorism



(Left) *Axial T2WI MR through the orbits shows that eyes are far apart and slightly proptotic. The calvarium is also misshapen and brachycephalic.* ***(Right)*** *Clinical photograph of the same case shows hypertelorism as well as a markedly misshapen head from craniosynostosis.*

ABNORMAL EYES

Hypertelorism



Hypertelorism



(Left) *Coronal ultrasound shows increased interocular distance (calipers).* ***(Right)*** *Coronal ultrasound through the upper lip ➡ shows a lack of a vermillion border. The fetus also had a flat midface, echogenic kidneys and a single umbilical artery.*

Hypertelorism



Hypertelorism



(Left) *Coronal transabdominal ultrasound shows an increased interocular distance ➡ and a consistently clenched hand ➡ from syndactyly in this fetus with Apert syndrome.* ***(Right)*** *Clinical photograph shows hypertelorism in the same fetus with Apert syndrome.*

Hypertelorism



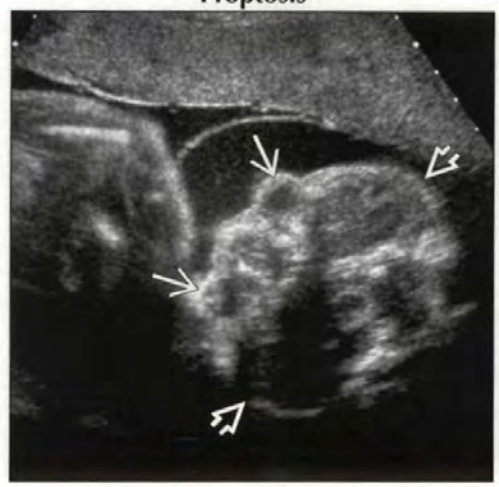
Hypertelorism



(Left) *Axial ultrasound shows that the distance between the eyes (calipers) is approximately twice what is expected.* ***(Right)*** *Frontal 3D ultrasound confirms widely spaced eyes ➡. The fetus also had an inferior vermian defect and normal chromosomes.*

ABNORMAL EYES

Proptosis

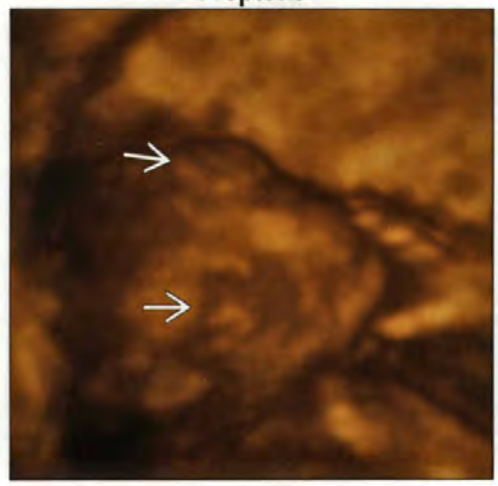


Proptosis

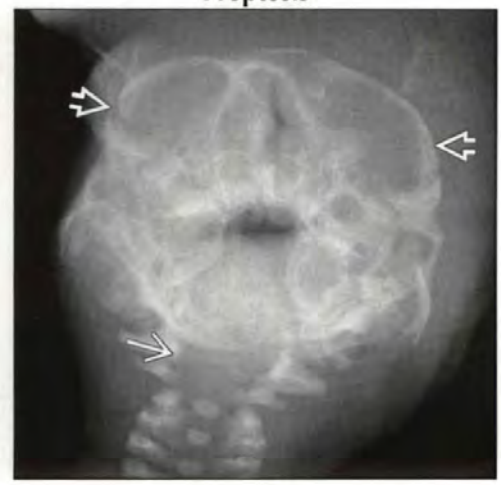


(Left) *Axial ultrasound through the orbits of a twin shows proptosis ➡ and hypertelorism. The fetal head shape is abnormal and there is bulging of the temporal lobes ➡. **(Right)** Coronal ultrasound through the calvarium in the same case shows a cloverleaf configuration of the skull ➡ caused by craniosynostosis. This is a case of Pfeiffer syndrome.*

Proptosis



Proptosis



(Left) *Coronal oblique 3D ultrasound shows proptosis ➡ in a fetus with anencephaly. The skull base is malformed with shallow orbits creating this appearance. **(Right)** Anteroposterior radiograph shows abnormally shaped orbits ➡ in another fetus with exencephaly and proptosis. Note the splaying of the superior cervical vertebral bodies ➡.*

Dacrocystocele



Dacrocystocele



(Left) *Axial ultrasound shows two small cysts ➡ located medial to the globes ➡, adjacent to the nose. These cysts are in the expected region of the lacrimal drainage ducts. **(Right)** Coronal 3D ultrasound shows the mass-like dacrocystocele ➡ between the eye ➡ and the nose.*

ABNORMAL EYES

Dacrocystocele



Dacrocystocele

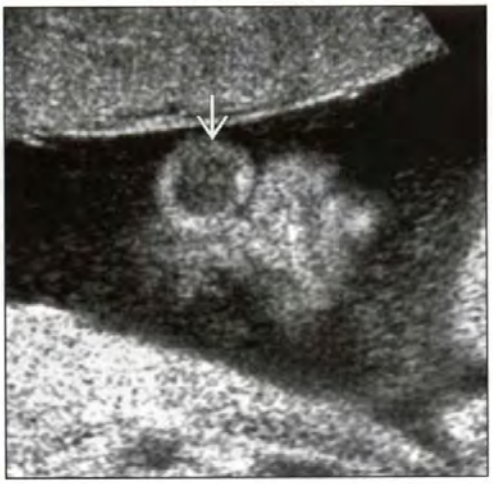


(Left) *Sagittal oblique 3D ultrasound from the prior case show the dacrocystocele in a different projection.* ***(Right)*** *Clinical photograph of the newborn shows a small slightly discolored medial eye mass. Most dacrocystoceles resolve in utero or during the first year of life.*

Orbital Tumors



Orbital Tumors

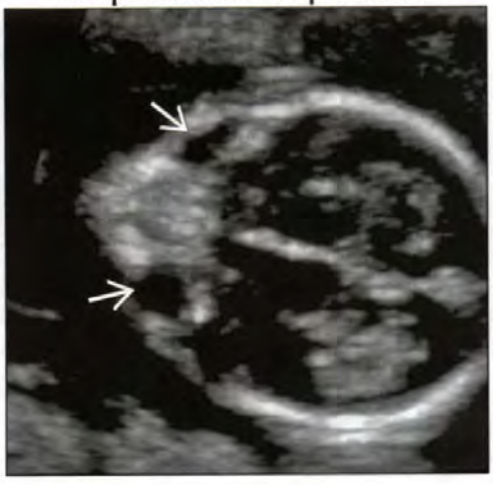


(Left) *Axial ultrasound through the orbits shows unilateral proptosis secondary to an enlarged echogenic globe. The normal eye is partially seen.* ***(Right)*** *Coronal ultrasound of the face confirms the diagnosis of a globe enlarged by tumor. Unilateral proptosis is the most common finding of an orbital tumor.*

Anophthalmia/Microphthalmia



Anophthalmia/Microphthalmia



(Left) *Coronal ultrasound shows no identifiable facial features in a fetus with atelencephaly (lethal malformation with developmental arrest of telencephalon). On autopsy, there were no orbits, nasal structures or mouth.* ***(Right)*** *Axial ultrasound through the orbits shows another case with hypertelorism, very small orbits, and microphthalmia.*

ABNORMAL EARS

DIFFERENTIAL DIAGNOSIS

Common
- Low Set Ears
- Deficient or Absent Ear

Less Common
- Lop Ear
- Protruding Ear

ESSENTIAL INFORMATION

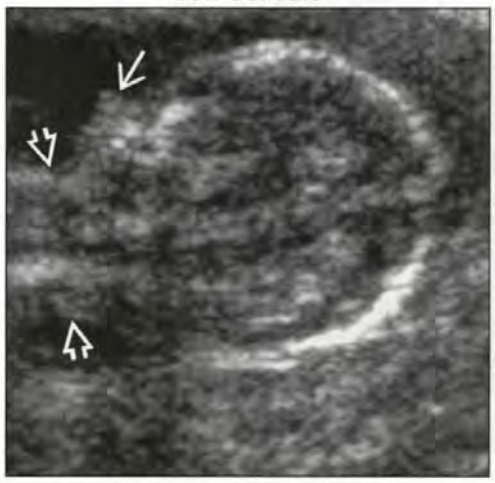
Key Differential Diagnosis Issues
- Normal ear position
  - Top of helix is at the inner canthi eye level
- Ear size evaluation
  - Ear length = 1/3 biparietal diameter
  - Ear width is more variable
- Use 3D to evaluate morphology
  - Helix is most external curve of auricle
  - Antihelix is Y-shaped internal auricle
  - Tragus lies over external meatus
  - Antitragus faces tragus

Helpful Clues for Common Diagnoses
- **Low Set Ears**
  - Top of helix is lower than inner canthi line
  - Associated with micrognathia
    - Treacher Collins syndrome
    - Nager syndrome
    - Pierre Robin syndrome
    - Aneuploidy
- **Deficient or Absent Ear**
  - Microtia = small ears
    - With or without absent auricular components
  - Anotia = no external ears

Helpful Clues for Less Common Diagnoses
- **Lop Ear**
  - Deformed ear cartilage
    - Top of ear curls downward
  - Often isolated
    - Autosomal dominant inheritance described
  - Associated with other anomalies
    - Anencephaly
    - Other syndromes with cartilage defects
- **Protruding Ear**
  - Ear protrudes > 25° from head
  - Most often idiopathic
    - Show ears are not low set

Other Essential Information
- Association with aneuploidy
  - Trisomy 21
  - Trisomy 18
  - Trisomy 13
  - Turner syndrome
- Ear anomalies are highly associated with micrognathia
- Low set and deficient ears commonly seen in oligohydramnios
- Supernumerary parts can mimic facial masses
  - Auricular tags
  - Auricular duplication

Low Set Ears



Coronal ultrasound shows the ear ➡ located near the top of the neck ⇔ in this fetus with Treacher Collins syndrome.

Low Set Ears



Sagittal ultrasound in the same fetus with Treacher Collins syndrome shows micrognathia. The mandible is small and recessed ➡.

ABNORMAL EARS

Deficient or Absent Ear

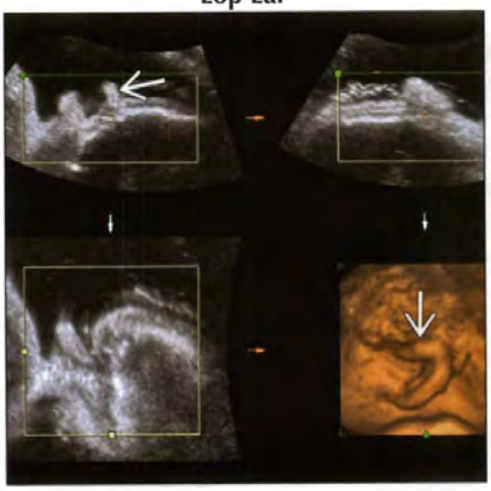


Deficient or Absent Ear



(Left) *Coronal oblique ultrasound shows a small, low-set ear ➡ in a fetus with multiple other anomalies.* ***(Right)*** *Clinical photograph shows a low set deficient ear ➡. The top of the ear should be at the level of the inner canthus line ⇒. The fetus also had a cleft lip and micrognathia ⇒. Chromosomes were normal.*

Lop Ear

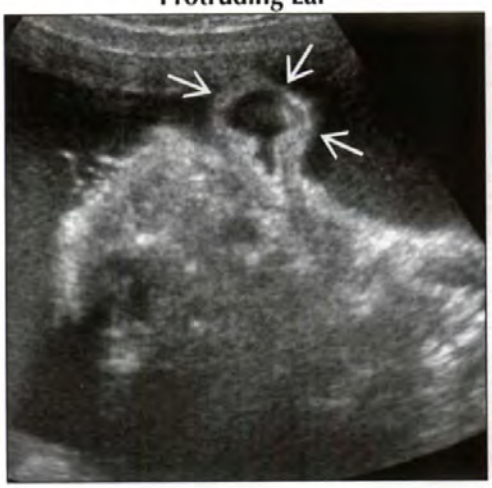


Lop Ear



(Left) *3D ultrasound shows the top of the ear bent away from the head and curled downward ➡.* ***(Right)*** *Clinical photograph of the neonate confirms the diagnosis ➡. The baby had multiple other anomalies as well.*

Protruding Ear



Protruding Ear



(Left) *Coronal ultrasound shows the ear protruding away from the fetal head ➡. This fetus also had a unilateral cleft lip and palate.* ***(Right)*** *3D ultrasound shows normal ear morphology and positioning in the same patient. Protruding ears are usually idiopathic and may be hereditary.*

MACROGLOSSIA

DIFFERENTIAL DIAGNOSIS

Common
- Idiopathic
- Trisomy 21

Less Common
- Beckwith-Wiedemann Syndrome
- Oral Mass (Mimic)

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Normal fetal movements include swallowing, thumb sucking and tongue motion
- Macroglossia implies that the tongue is too large to fit in oral cavity
- Down syndrome fetuses may exhibit "tongue thrusting" movements in third trimester
  - Tongue protrudes intermittently due to lax muscle tone
- If tongue seems "too easy to see" look carefully for facial cleft
  - Coronal view of nose/lips
  - Axial view of tooth buds
- Lymphangioma may cause tongue enlargement
  - Rare amongst head and neck lymphangiomas
  - More likely to present in childhood than in fetus
- Oral masses can be confusing
  - Sometimes hard to tell if a mass originates in tongue or palate
  - Epignathus typically very large with cystic/solid/calcified components

Helpful Clues for Common Diagnoses
- **Idiopathic**
  - Structurally normal fetus
    - No signs of aneuploidy particularly trisomy 21
    - Size appropriate for dates
- **Trisomy 21**
  - Correlate with a priori risk and look for sonographic markers
    - Absent nasal bone/thick nuchal fold/mild ventriculomegaly
    - Congenital heart disease, particularly atrioventricular septal defect
    - Duodenal atresia/echogenic bowel/pyelectasis
    - Short humerus/femur
    - Clinodactyly/sandal gap toe

Helpful Clues for Less Common Diagnoses
- **Beckwith-Wiedemann Syndrome**
  - Macroglossia in 97%
  - Fetal overgrowth in 88%
  - Omphalocele/umbilical hernia in 80%
  - Organomegaly
  - At increased risk for severe neonatal hypoglycemia sufficient to cause brain injury
  - Childhood tumors develop in up to 10% of cases (Wilms tumor most common)

Idiopathic



Sagittal transabdominal ultrasound during the course of a growth assessment for poor maternal weight gain showed this fetus "licking" the placenta. The tongue ➡ fit in the mouth, and the infant was normal at birth.

Idiopathic



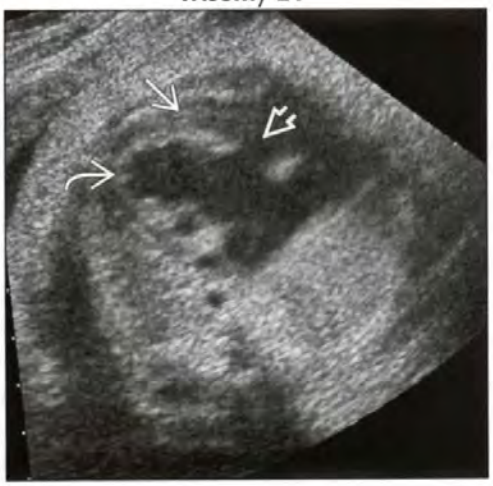
Sagittal transabdominal ultrasound shows another fetus with its tongue protruding ➡. This was a transient phenomenon, and the infant was normal at birth.

MACROGLOSSIA

Trisomy 21



Trisomy 21



(Left) *Coronal transabdominal ultrasound of the face shows a protruding tongue and broad flat nose typical of the Down syndrome facies.* ***(Right)*** *Four chamber view of the heart in the same fetus shows an unbalanced atrioventricular septal defect with a small right ventricle, larger left ventricle, and ventricular septal defect. Amniocentesis confirmed trisomy 21.*

Trisomy 21

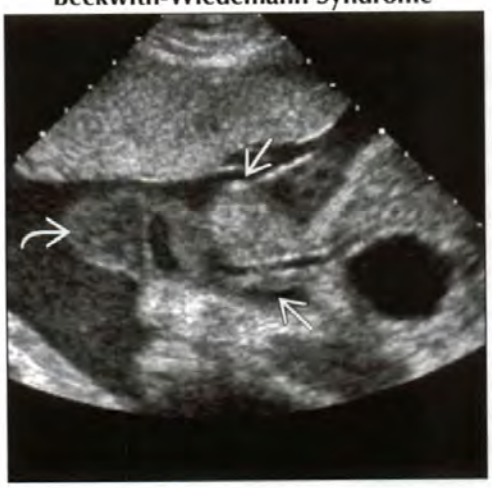


Beckwith-Wiedemann Syndrome



(Left) *Sagittal ultrasound in the same fetus as two prior images shows persistent protrusion of the tongue. Persistent protrusion is the hallmark of macroglossia, as the tongue is too large to be contained within the oral cavity.* ***(Right)*** *Coronal ultrasound shows a more subtle appearance of tongue protrusion in a fetus with Beckwith-Wiedemann syndrome. This was not appreciated prospectively.*

Beckwith-Wiedemann Syndrome



Beckwith-Wiedemann Syndrome



(Left) *Axial ultrasound in the same fetus as the prior image shows an omphalocele and cystic degeneration of Wharton jelly. Omphalocele is a common finding in Beckwith-Wiedemann syndrome.* ***(Right)*** *Axial ultrasound in a different case shows enlargement of the left kidney noted during a biophysical profile. There were no other abnormalities, but the fetus was large for dates. Beckwith-Wiedemann syndrome was diagnosed postnatally.*

FACIAL MASS

DIFFERENTIAL DIAGNOSIS

Common
- Premaxillary Protrusion
  - Cleft Lip, Palate
- Proboscis

Less Common
- Epignathus
- Frontal Encephalocele

Rare but Important
- Preauricular Tags
- Orbital Tumors

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Routine views of fetal face
  - Profile
    - Nasal bone assessment
    - Rule out micrognathia
  - Orbit
    - Axial and coronal views
    - Note distance between eyes
  - Nose and lip assessment
    - Coronal soft tissue view
- 3D ultrasound technique
  - Acquisition best from profile view
  - Need fluid in front of face
  - Soft tissue rendered views
    - Assess eyes, nose and lips on one image
  - Bone rendered views
    - Bony palate
    - Sutures
- 4D ultrasound
  - 3D in real time = 4D
  - Static 3D has better resolution
- MR
  - Better delineation of extension of masses
  - Best for associated subtle brain anomalies

Helpful Clues for Common Diagnoses
- **Premaxillary Protrusion**
  - From bilateral cleft lip, palate
    - Bilateral cleft palate leaves midline "island" of dysplastic maxillary tissue
    - Axial palate view shows defects best
    - Profile view shows mass-like structure
    - 3D shows mass under nose
  - May be seen with unilateral cleft lip/palate
    - Large cleft palate defect
    - Alveolar ridge anteriorly displaced
    - Palate protuberance on profile view
    - 3D shows morphology best
  - Cleft lip, palate (CL/CP) classification
    - Type 1: Unilateral CL only
    - Type 2: Unilateral CL/CP
    - Type 3: Bilateral CL/CP
    - Type 4: Midline CL/CP
- **Proboscis**
  - Tube-like soft tissue midface structure
    - Proboscis instead of normal nose
    - Superior to eyes or between eyes
    - May lie flush with forehead
  - Orbit anomalies with proboscis
    - Cyclopia
    - Hypotelorism
  - Holoprosencephaly association
    - Alobar
    - Semilobar
  - Aneuploidy
    - Trisomy 13 most common
    - Trisomy 18

Helpful Clues for Less Common Diagnoses
- **Epignathus**
  - Teratoma
    - Arises from oral or nasal cavity
    - Calcification in 50% (diagnostic)
  - Large, fungating oral mass
    - Fills oral cavity then emanates from mouth and/or nose
  - Can have intracranial extension
    - Transsphenoidal extension
    - Extra-axial mass
  - Polyhydramnios from impaired swallowing
  - MR
    - Helpful in determining extent of mass
    - Can identify fat within lesion
- **Frontal Encephalocele**
  - Skull defect in frontoethmoidal region with herniation of intracranial structures
    - Skin-covered
  - Nasofrontal
    - Between frontal and nasal bones
  - Nasoethmoidal
    - Between nasal bones and nasal cartilage
  - Nasoorbital
    - Through medial orbital defect
  - Associated with other midline anomalies
    - Hypertelorism
    - Corpus callosum anomalies
    - Heterotopia
    - Interhemispheric lipoma

- Most common in Southeast Asia
  - 1:1,000
- Better prognosis than occipital or parietal encephaloceles

Helpful Clues for Rare Diagnoses

- **Preauricular Tags**
  - Supernumerary ear parts
    - Duplication of embryologic components
  - Skin "tags" with variable differentiation
    - Found at embryologic junctions
  - Preauricular tag
    - Anterior to ear
    - Looks like a cheek mass
  - Associated with other facial and ear anomalies
    - Micrognathia most common
- **Orbital Tumors**
  - Group of rare tumors
    - Retinoblastoma
    - Teratoma
    - Rhabdomyosarcoma
    - Rhabdoid tumor
    - Neurofibroma
  - Often large solid mass
    - Involved globe may or may not be identified
    - Bony anatomy may be distorted
    - Mass is vascular
  - Small masses
    - May present with proptosis only

Other Essential Information

- Frontal encephaloceles easily missed
  - Hypertelorism may be the only finding
    - Small bony defect
    - Subtle brain herniation
  - Consider MR for unexplained hypertelorism
- Facial anomalies with holoprosencephaly
  - Cyclopia is most severe type
    - Single bony orbit
    - Variable amount of globe present
    - Proboscis often above orbit
  - Ethmocephaly
    - Proboscis separates close set eyes
  - Cebocephaly
    - Infraorbital flat nose with single nostril
- Orbital tumors may present with proptosis (2 etiologies)
  - Retroorbital tumor
    - Pushes globe forward
  - Tumor involves globe
    - Enlarged globe
- Look for fat or calcification in mass
  - Teratoma most likely diagnosis if either present

Alternative Differential Approaches

- "Mass-like" face lesions
  - Premaxillary protuberance
  - Proboscis
  - Frontal encephalocele
  - Preauricular tags
- True face masses
  - Epignathus
  - Orbital tumors

Premaxillary Protrusion



Sagittal ultrasound shows an abnormal fetal profile with a "mass-like" bony protrusion ➡ beneath the nose ➡. This appearance is typical for type 3 cleft lip/palate.

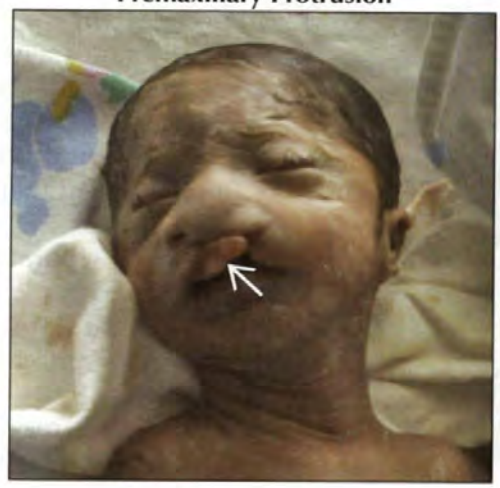
Premaxillary Protrusion



Frontal 3D ultrasound of the same fetus shows the bilateral cleft lip defects ➡. The anterior maxillary "mass" ➡ beneath the nose is seen well with 3D ultrasound.

FACIAL MASS

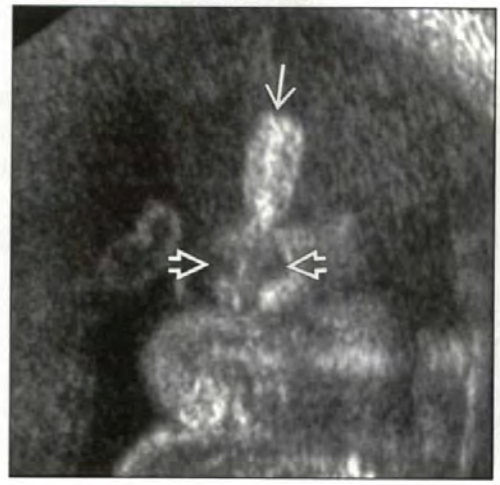
Premaxillary Protrusion





(Left) *Clinical photograph of a large unilateral cleft lip, palate shows anterior displacement of the maxilla. On a profile view, this tissue will look mass-like.* ***(Right)*** *Axial ultrasound through the alveolar ridge of a fetus with a large cleft lip, palate defect shows the palate defect and the anteriorly displaced alveolar ridge, as compared to the contralateral side.*

Proboscis





(Left) *Coronal ultrasound shows a tube-like proboscis extending superiorly above the eyes. There is severe hypotelorism and holoprosencephaly (not shown).* ***(Right)*** *Clinical photograph shows a proboscis in another fetus with holoprosencephaly and cyclopia.*

Epignathus



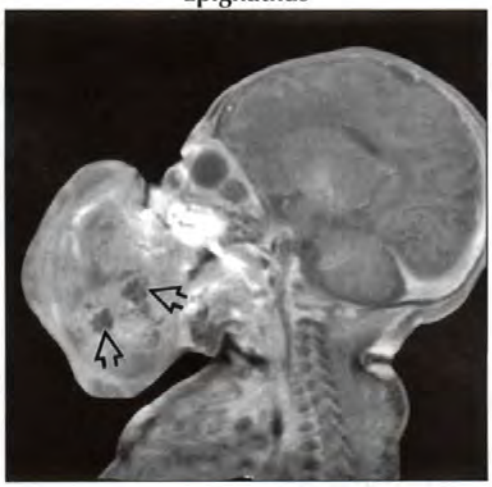


(Left) *Sagittal ultrasound of the fetal face shows a large mixed cystic and solid mass growing out of the mouth. The mandible is held open.* ***(Right)*** *Clinical photograph shows the fetus during ex utero intrapartum treatment (EXIT). The mass is controlled while the intubation is performed. The teratoma was subsequently resected.*

Epignathus



Epignathus

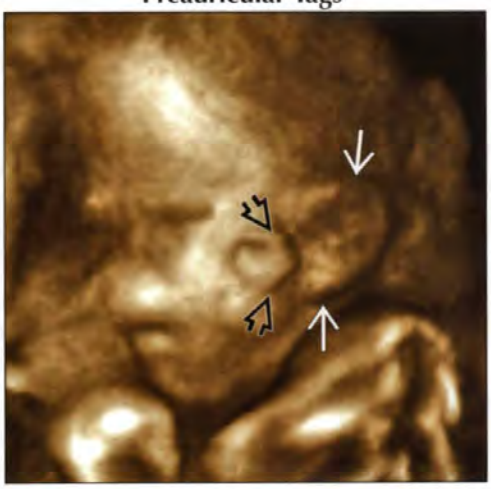


(Left) *Sagittal T2WI MR shows a mostly solid mass ⇨ extending from the fetal mouth.* ***(Right)*** *Sagittal T1WI MR after delivery shows the mass had grown during fetal life. Focal areas of low signal ⇨ may be calcification or fluid.*

Frontal Encephalocele

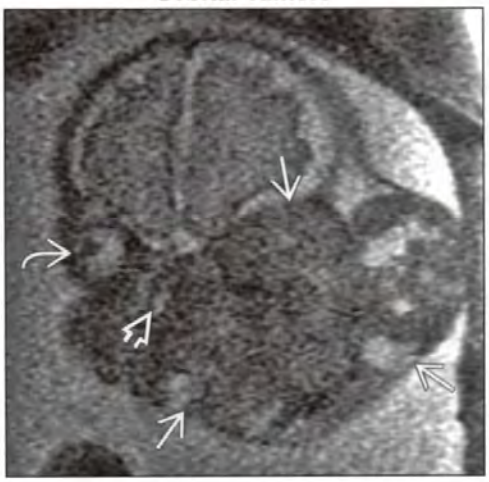


Preauricular Tags



(Left) *Sagittal ultrasound shows an anterior calvarial bony defect ➡ with herniating brain ⇨. This fetus also had hypertelorism.* ***(Right)*** *Sagittal oblique 3D ultrasound in another fetus shows a normal ear ➡ and a curvilinear preauricular skin tag ⇨. The fetus also had multiple other anomalies.*

Orbital Tumors



Orbital Tumors



(Left) *Coronal T2WI MR shows a large solid and cystic orbital mass ➡. The involved globe is not identified and the nasal septum ⇨ is deviated towards the normal eye ⇨.* ***(Right)*** *Sagittal color Doppler ultrasound focused on the mass shows it is very vascular. The final diagnosis was rhabdomyosarcoma.*

NECK MASS

DIFFERENTIAL DIAGNOSIS

Common
- Cystic Hygroma
- Nuchal Cord, Mimic
- Occipital Encephalocele

Less Common
- Cervical Teratoma
- Goiter
- Truncal Lymphangioma

Rare but Important
- Iniencephaly

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is the mass predominately cystic or solid?
  - Cystic favors cystic hygroma
  - Solid favors teratoma, goiter or encephalocele
- Where is it located?
  - Posterior masses favor cystic hygroma and cephaloceles
  - Anterior masses favor teratoma or goiter
- Always check the calvarium and spine
  - If intact, encephalocele and spinal defect excluded
- Always use color Doppler
  - Rule out nuchal cord
  - Look for flow within mass

Helpful Clues for Common Diagnoses
- **Cystic Hygroma**
  - Result of failed/delayed jugular venous-lymphatic connection
    - Creates a multiseptated nuchal fluid collection
  - Located in posterior subcutaneous tissues, frequently wrapping around laterally
    - May involve only one side of neck causing postural abnormality
  - May be massive and mimic amniotic fluid
    - Concern for airway obstruction at birth
  - Internal septations
    - Multiple, fine linear septations
    - Thick, midline septation is nuchal ligament
  - Nonimmune hydrops common with large hygromas
  - Small cystic hygromas can evolve into thick nuchal fold
  - Aneuploidy in 2/3 of fetuses with 2nd trimester cystic hygroma
    - Turner syndrome most common, especially for larger septated hygromas
    - Trisomy 21 next most common; smaller less complicated hygromas and nuchal skin thickening
  - Cardiovascular anomalies often present
    - Aortic arch defects most frequent, especially in Turner syndrome
- **Nuchal Cord, Mimic**
  - Nuchal cord can be confused with cystic hygroma on grayscale images
  - Obvious flow with color Doppler
  - Cords with less vascular coiling are more pliable and more likely to wrap around neck
  - Longer cords also have increased incidence of nuchal wrapping
- **Occipital Encephalocele**
  - Osseous defect should be demonstrated
    - Usually midline: Occipital
    - Lateral: Parietal, inferior temporal
    - May be difficult to see with small defect
  - Cephalocele
    - More generic term for herniation of intracranial contents
  - Encephalocele
    - Meninges & brain
    - Most common
  - Cranial meningocele
    - Meninges only
  - Chiari III
    - Hindbrain malformation
    - Cerebellum herniated into cephalocele
  - Diverse appearance of herniated neural tissue
    - Gyral pattern may be identified
    - Mixed cystic/solid mass
    - "Cyst within a cyst" or "target" sign suggests prolapsed 4th ventricle
    - In first trimester head may look small or irregular
  - Microcephaly in 25%
  - Other central nervous system anomalies common
  - Large occipital cephaloceles may also involve cervical spine

Helpful Clues for Less Common Diagnoses
- **Cervical Teratoma**

- Mixed cystic and solid mass involving anterior aspect of neck
  - Frequently extends to involve surrounding structures
- Calcifications are virtually pathognomonic of teratoma
- Head is often held in hyperextension or deviated to one side
- Polyhydramnios from upper esophageal obstruction
- Solid portions of mass often very vascular
  - Arteriovenous shunting may be present
  - Vascular shunting may result in hydrops

• Goiter

- Fetal goiter may result from overtreatment of maternal hyperthyroidism, transplacental passage of anti-thyroid antibodies or congenital hypothyroidism
- Solid, homogeneous anterior neck
- Maintains normal thyroid contour and echogenicity
- Use color Doppler in coronal view to evaluate carotid arteries and jugular veins
  - Large goiter will cause lateral displacement
- May obstruct swallowing causing polyhydramnios
- May prevent normal fetal "chin tuck" → extended neck → obstructed labor
  - If fetus is able to flex the chin to chest, a goiter is unlikely to affect mode of delivery or airway at birth

• Truncal Lymphangioma

- May extend into deep tissues of neck but primarily involves axilla, chest and arm
- Morphologically identical to cystic hygroma

Helpful Clues for Rare Diagnoses

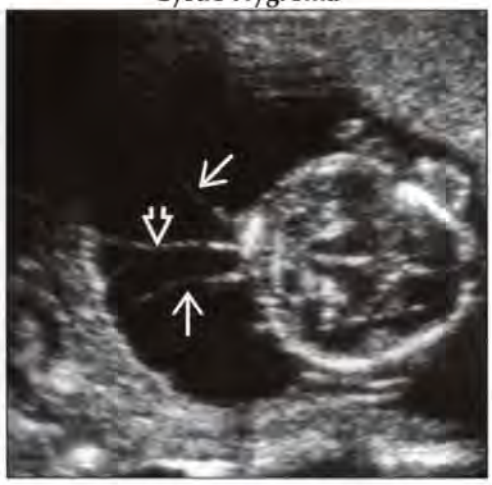
• Iniencephaly

- Extensive open neural tube defect characterized by
  - Defect in occipital bone and inion
  - Occipital encephalocele
  - Spinal dysraphism which often extends into thoracic and even lumbar area
  - Fixed hyperextension of head ("stargazer" position)
  - Missing or fused cervical vertebrae

Other Essential Information

- Postural abnormality of neck may be progressive as mass grows over gestation
  - May require cesarean section
- Large neck masses may compromise airway at birth leading to asphyxia
  - Preplanning essential
  - May require EXIT procedure (ex-utero intrapartum treatment)
    - Fetus is partially delivered by cesarean section section while placenta and umbilical cord remain intact
    - Uteroplacental gas exchange maintained
    - Fetus remains hemodynamically stable while airway is established
    - Avoids "crash" attempt at achieving airway at birth

Cystic Hygroma



Axial ultrasound shows a large cystic hygroma with multiple thin septations ⇨ emanating away from the fetal head. The midline septation is the nuchal ligament ⇨. This fetus had Turner syndrome.

Cystic Hygroma



Radiograph shows a giant hygroma ⇨ engulfing the fetal head. Also note the soft tissue edema from generalized hydrops, a common complication. Large hygromas are often associated with Turner syndrome.

NECK MASS

Cystic Hygroma



Cystic Hygroma

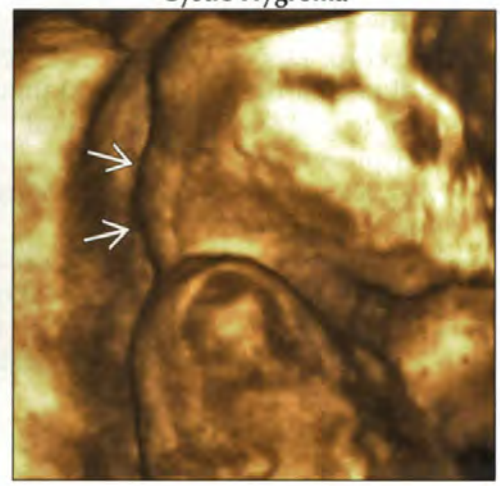


(Left) *Coronal ultrasound of the fetal neck shows a large, multiseptated cystic mass ➡ along the right side of the neck.* ***(Right)*** *Coronal T2WI MR in the same case as previous image, shows the hygroma involving the deep tissues of the neck ➡. It surrounds the neck vessels, which appear as flow voids ➡. Postnatal imaging confirmed the infiltrative nature of the mass. This fetus had normal chromosomes.*

Cystic Hygroma



Cystic Hygroma

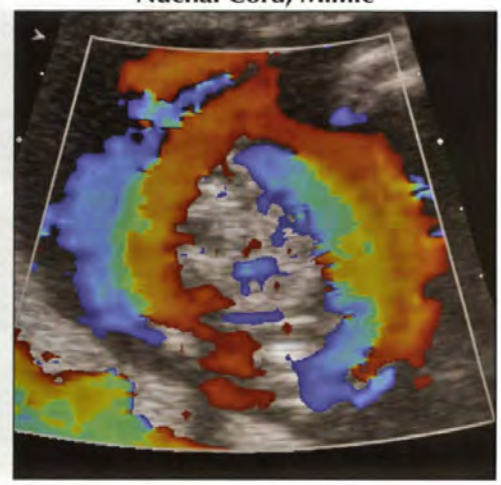


(Left) *Axial ultrasound shows nuchal fold thickening (calipers) with fluid, consistent with a relatively small cystic hygroma.* ***(Right)*** *Sagittal 3D ultrasound also shows the increased bulge behind the neck ➡. In the third trimester, the cystic hygroma fluid resolved, and the fetus was left with residual increased skin thickness behind the neck. This appearance of a cystic hygroma is more commonly seen in trisomy 21, which was confirmed after delivery.*

Nuchal Cord, Mimic



Nuchal Cord, Mimic



(Left) *Axial ultrasound shows a cystic "mass" around the neck ➡.* ***(Right)*** *Axial color Doppler ultrasound quickly reveals that the mass is actually a nuchal cord with two loops wrapped around the neck. Note the cord does not show the normal coiling, which is felt to be a risk factor for developing a nuchal cord.*

Occipital Encephalocele



Occipital Encephalocele

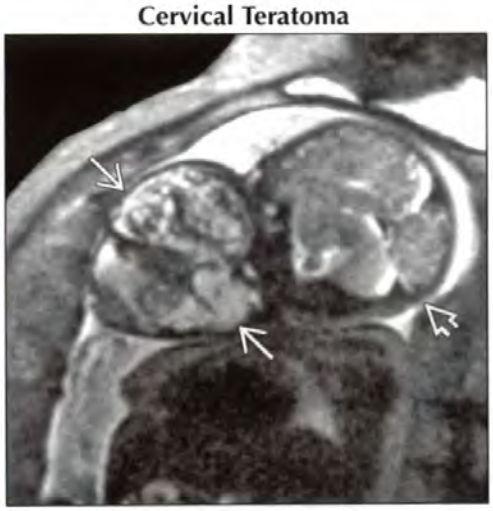


(Left) *Sagittal ultrasound shows a large septated nuchal mass, which could be confused with a cystic hygroma. Note that spinous processes of the cervical spine are not seen.* ***(Right)*** *Axial oblique ultrasound in the same case as previous image, shows a large occipital skull defect, with herniation of intracranial contents. There is a "cyst within a cyst" or "target" sign, which is created when the 4th ventricle herniates into the cephalocele.*

Occipital Encephalocele

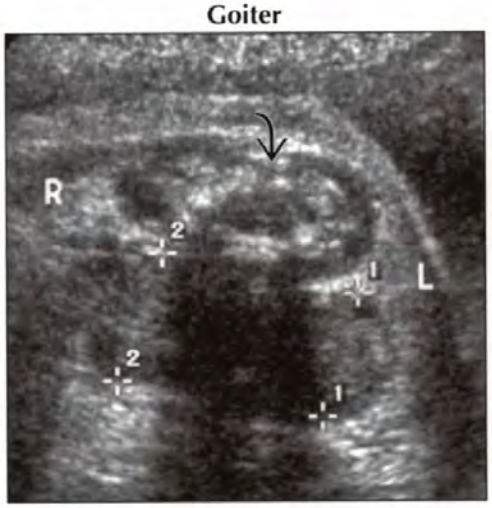


Cervical Teratoma

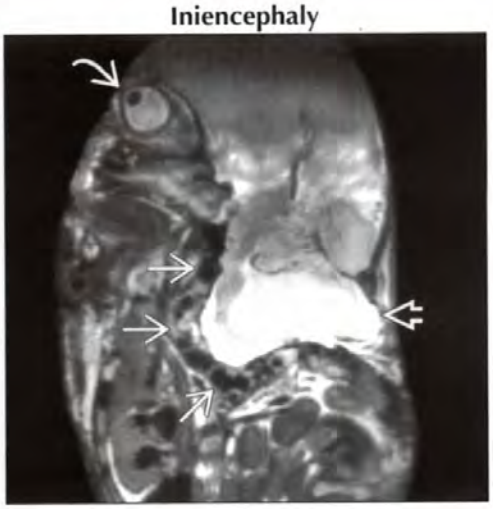


(Left) *Clinical photograph from the same case as previous image, shows the large occipital/high cervical meningoencephalocele, a Chiari III malformation.* ***(Right)*** *Coronal T2WI MR shows a typical example of a cervical teratoma. There is a complex neck mass, with both cystic and solid components. The head is displaced to the side. The mass is approximately the same size as the fetal head. The fetus was delivered using the EXIT procedure and the mass resected.*

Goiter



Iniencephaly



(Left) *Axial ultrasound of the fetal neck (spinous process of cervical vertebra) shows a markedly enlarged thyroid gland (calipers). It has the same morphology and homogeneous echogenicity as an adult thyroid.* ***(Right)*** *Sagittal T2WI MR postmortem shows typical features of iniencephaly. There are missing and fused vertebrae and a large cranial and spinal defect. The neck is retroflexed, and the eyes are held in the "stargazer" position.*

SECTION 4
Chest

Thoracic Fluid Collection	4-2
Solid/Echogenic Lung Mass	4-6
Cystic Lung Mass	4-10
Small Chest	4-14

THORACIC FLUID COLLECTION

DIFFERENTIAL DIAGNOSIS

Common

- Bilateral Pleural Effusion
  - Hydrothorax
- Unilateral Pleural Effusion
  - Chylothorax
- Pericardial Effusion

Less Common

- Effusions Associated with Lung Masses
  - Bronchopulmonary Sequestration
  - Congential Cystic Adenomatoid Malformation (CCAM)
- Masses That Can Mimic Simple Fluid Collection
  - Unilocular CCAM
  - Congenital Diaphragmatic Hernia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- First determine where the fluid is located
  - Pleural space, pericardial space or within a mass
- Pleural effusion
  - Routine four chamber heart view
    - Curvilinear, anechoic fluid
    - Echogenic lung displaced medially
  - Coronal chest view
    - Lung displaced superiorly and medially
    - "Wing-like" lungs float in fluid
- Pericardial effusion
  - Fluid collection surrounds fetal heart
  - If large, heart is seen beating in a "bag of water"
  - Lungs will be compressed posteriorly, not free floating
- Large, unilocular cystic masses may simulate pleural effusion
  - Masses are rounded or ovoid
  - Have mass effect on lungs and mediastinal structures
    - Lung shifted away from fluid, not surrounded by fluid as in a pleural effusion

Helpful Clues for Common Diagnoses

- **Hydrothorax**
  - Serous fluid collection
  - Effusions are bilateral and symmetric
  - Hallmark finding in hydrops fetalis (both immune and nonimmune)
    - Hydrops defined as fluid accumulation in 2 or more body cavities
      - Pleural effusion
      - Ascites
      - Skin edema
      - Pericardial fluid
    - Polyhydramnios and placentomegaly also often present if there is hydrops
    - Anomalies commonly associated with effusions/hydrops
      - Cystic hygroma (Turner syndrome) most common
      - Trisomy 21 (other markers usually seen); trisomy 18, 13 less likely
      - Cardiac defects
      - Cardiac arrhythmia
      - Infection
      - Cystic adenomatoid malformation (< 10% have hydrops)
    - Fetal masses causing high output failure (and possible hydrops)
      - Sacrococcygeal teratoma
      - Goiter
      - Vascular shunting: Vein of Galen malformation, arteriovenous fistulas, hepatic hemangioendothelioma, placental chorioangioma
    - First trimester pleural effusion
      - Can be seen as early as 7 weeks
      - Associated with increased nuchal translucency
      - Poor prognosis when present before 15 wks
      - Aneuploidy common: Turner syndrome most likely
- **Chylothorax**
  - Chylous fluid collection
  - Effusion is unilateral
  - Primary congenital lymphatic defect
    - Atresia, fistula, or absence of thoracic duct
  - Thoracic duct crosses from right to left at 5th thoracic level
    - Level of obstruction determines right versus left-sided effusion
    - Equal incidence of right and left-sided effusions
  - Fluid is anechoic
    - Only after neonatal feeding will chylous fluid appear "milky"
  - Mass effect common

- Mediastinal shift
- Flattened diaphragm
- May lead to hydrops when large
  - Consider performing fetal thoracentesis if evidence of fetal compromise
- 5% associated with aneuploidy
  - Turner syndrome
  - Trisomy 21
  - Noonan syndrome
- Amniocentesis warranted
- 15% resolve in fetal life
- Near 100% survival without hydrops and normal chromosomes

- **Pericardial Effusion**
  - Seen best on standard four chamber view
  - Lenticular or oval collection of fluid adjacent to, or surrounding heart
  - Trace of fluid along one ventricular wall is normal
    - Can be up to 2 mm
    - Majority of fetuses (50-80%) have trace fluid if careful search done
    - Generally transient
    - If fetus not at increased risk, follow-up not necessary
  - Significant effusion if fluid surrounds atria as well as ventricles
  - Pericardial effusion seen in many conditions
    - Evaluate heart for structural abnormality, arrhythmia or mass
    - Look for other signs of hydrops
    - Look for signs of congenital infection
    - Look for anemia

Helpful Clues for Less Common Diagnoses

- **Bronchopulmonary Sequestration**
  - 6-10% may develop unilateral pleural effusion
  - May cause tension hydrothorax requiring fetal thoracentesis to decompress
  - 90% left-sided
  - Look for triangular, solid mass surrounded by pleural fluid
- **Congenital Cystic Adenomatoid Malformation (CCAM)**
  - May see effusions if fetus develops hydrops
    - Occurs in < 10% of cases
  - Unilocular CCAM can appear as a simple fluid collection
    - Look for stomach below diaphragm to rule out congenital diaphragmatic hernia
- **Congenital Diaphragmatic Hernia**
  - Appears as simple fluid collection if only stomach is herniated
    - In isolated gastric herniation, stomach generally located posteriorly in chest (herniation through foramen of Bochdalek)
  - Look carefully for small bowel and liver, which are often present but difficult to visualize
    - Use color Doppler and consider fetal MR

Hydrothorax



Axial ultrasound in a fetus with immune hydrops shows bilateral effusions ➡ and marked skin edema ➡. The lungs are displaced medially, "hugging" the heart.

Hydrothorax



Coronal oblique ultrasound in a fetus with non-immune hydrops (fetus had Smith-Lemli-Opitz syndrome) shows typical "winged" appearance of floating lungs ➡.

THORACIC FLUID COLLECTION

(Left) *Axial ultrasound shows typical appearance of Turner syndrome presenting with a large cystic hygroma and hydrops. A large nuchal cystic hygroma ➡ mimics amniotic fluid and contains septations ➡.* ***(Right)*** *Coronal ultrasound in the same case shows body wall edema ➡, bilateral pleural effusions ➡, and ascites ➡, diagnostic of associated hydrops fetalis.*

Hydrothorax



Hydrothorax

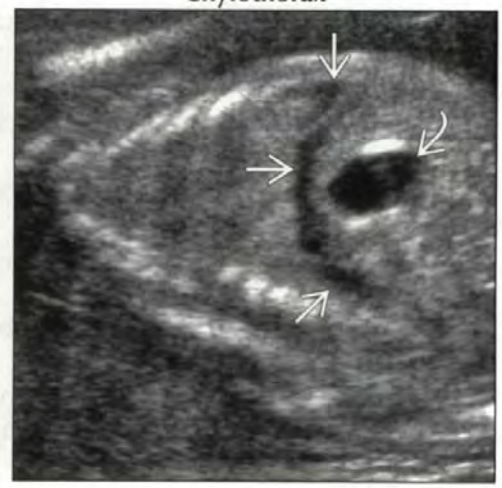


(Left) *Sagittal ultrasound shows the right hemithorax in a fetus with supraventricular tachycardia and pleural effusions. Fluid can actually be seen tracking in the major fissure ➡.* ***(Right)*** *Sagittal ultrasound shows a small isolated pleural effusion ➡ in an otherwise normal fetus (stomach ➡). A follow-up study performed 2 weeks later showed complete resolution of the effusion.*

Hydrothorax

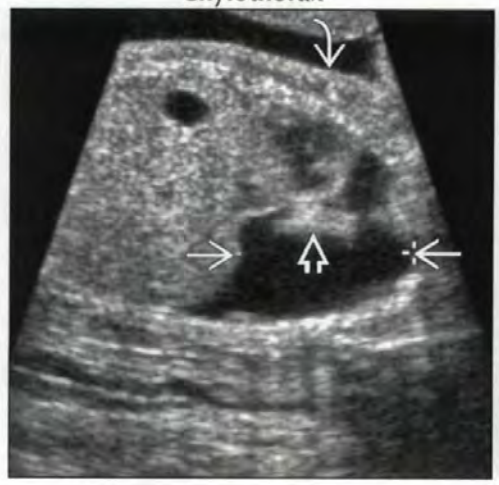


Chylothorax



(Left) *Coronal ultrasound shows typical features of a large, unilateral chylothorax ➡. It is compressing the right lung ➡ and deviating the heart. Skin edema is also present ➡.* ***(Right)*** *Four chamber view of the heart shows a small, localized pericardial effusion (calipers), which resolved on follow-up studies.*

Chylothorax



Pericardial Effusion

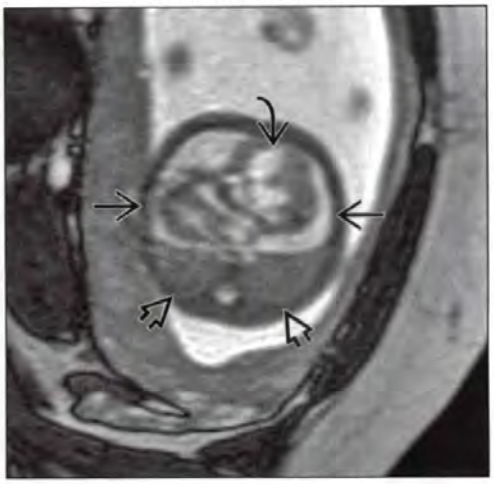


THORACIC FLUID COLLECTION

Pericardial Effusion

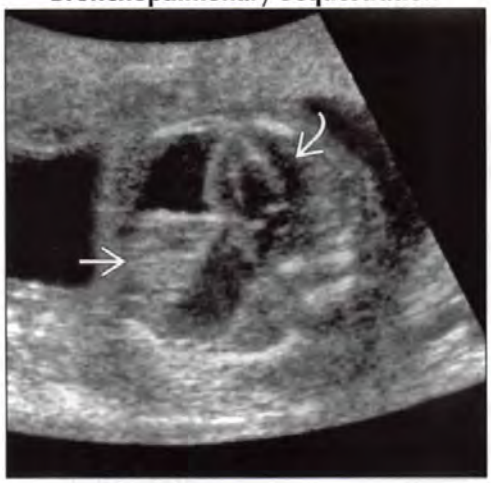


Pericardial Effusion



(Left) *Four chamber view in a fetus with a cytomegalovirus infection shows cardiomegaly with a pericardial effusion. Note the lungs are compressed posteriorly and are not floating in the fluid, as would be seen with pleural effusions.* ***(Right)*** *Axial T2WI MR shows a large intrapericardial teratoma and pericardial effusion. Note how the lungs are compressed posteriorly by this large effusion.*

Bronchopulmonary Sequestration



Congential Cystic Adenomatoid Malformation (CCAM)



(Left) *Axial ultrasound shows a unilateral pleural effusion surrounding a triangular mass at the left lung base. The heart is shifted to the right. Sequestrations can cause a tension hydrothorax requiring in utero drainage.* ***(Right)*** *Axial ultrasound shows small bilateral pleural effusions in a fetus with a CCAM who was developing hydrops. Effusions are not common in CCAM, unlike in sequestrations, unless they are part of generalized hydrops.*

Unilocular CCAM



Congenital Diaphragmatic Hernia



(Left) *Axial ultrasound shows a large, unilocular cyst in the fetal chest deviating the heart to the right. The stomach was in the normal position, making a unilocular CCAM the most likely diagnosis.* ***(Right)*** *Axial ultrasound shows a posteriorly located, unilocular cyst in the chest, displacing the heart to the right. No stomach bubble was seen in the abdomen. The stomach had herniated through the foramen of Bochdalek.*

SOLID/ECHOGENIC LUNG MASS

DIFFERENTIAL DIAGNOSIS

Common
- Congenital Cystic Adenomatoid Malformation, Microcystic
- Bronchopulmonary Sequestration
- Congenital Diaphragmatic Hernia

Less Common
- Teratoma, Pericardial
- Teratoma, Chest
- Congenital Lobar Emphysema

Rare but Important
- Tracheal Atresia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Doppler is key for diagnosing solid chest mass
  - Congenital cystic adenomatoid malformation (CCAM) has vascular supply (both arterial and venous) from pulmonary circulation
  - Sequestration has prominent feeding vessel from aorta
    - Generally drains into systemic circulation (inferior vena cava, azygous)
  - Diaphragmatic hernia containing liver will show portal/hepatic veins
  - Other masses may show flow but usually no dominant feeding vessel
- Where is the lesion?
  - Right side favors CCAM
  - Left side could be either CCAM or sequestration
  - Bilateral
    - Tracheal atresia: Look for massive chest enlargement, fluid-filled trachea/bronchi
    - Bilateral CCAM
    - Bilateral congenital diaphragmatic hernia: Stomach with cause cystic mass on left side
- Is the mass surrounded by fluid?
  - Pericardial vs. pleural effusion
    - May be confusing if large
    - Pericardial effusion: Lungs compressed posteriorly
    - Pleural effusion: Lungs float in fluid and appear "wing-like"
  - Pericardial effusion common with pericardial teratomas
  - Unilateral pleural effusion suggests sequestration
  - Bilateral effusions as part of generalized hydrops
    - Most common with CCAM
- "Disappearing" lung mass
  - Common in both CCAM and sequestration

Helpful Clues for Common Diagnoses
- **Congenital Cystic Adenomatoid Malformation, Microcystic**
  - Morphology varies from solid appearing (microcystic) to complex cystic mass (macrocystic) or even unilocular
  - Microcystic CCAM appear as solid lesions
    - Cysts < 5 mm
    - Uniformly echogenic
    - Well-defined masses
    - 95% are unilateral and affect only 1 lobe
    - No side predilection
    - May see small, scattered macroscopic cysts
  - Color Doppler
    - Vascular supply from pulmonary artery
    - Venous drainage to pulmonary vein (more difficult to see)
  - Greatest growth 20-26 weeks
    - May regress and even "disappear" later in pregnancy
  - May be complicated by hydrops (< 10%)
    - Near 100% mortality with hydrops if untreated
- **Bronchopulmonary Sequestration**
  - Uniformly echogenic, well-marginated, triangular-shaped mass
  - 90% left-sided, 90% supradiaphragmatic
  - Color Doppler
    - Prominent feeding vessel from aorta (may have more than one)
    - Venous drainage to inferior vena cava or azygous (often difficult to see)
  - Unilateral pleural effusion in 6-10%
    - May cause tension hydrothorax
- **Congenital Diaphragmatic Hernia**
  - Right-sided hernia more likely to present as solid mass because stomach remains below diaphragm
    - Stomach may be more medially located than usual
  - Contents of hernia vary in echogenicity

- Liver more hypoechoic, bowel more hyperechoic
- Liver may be difficult to differentiate from lung
  - Use Doppler to look for hepatic/portal veins
  - Fetal MR best tool to evaluate contents of hernia
  - "Liver up" has worse prognosis
- Bilateral hernias may be difficult to diagnose
  - Abnormal cardiac axis may be only clue; apex will be more midline
  - Abdominal circumference will measure less than expected
- Pulmonary hypoplasia worse for CDH than other chest masses of comparable size
- Up to 50% have an associated abnormality, including chromosomal

Helpful Clues for Less Common Diagnoses

- **Teratoma, Pericardial**
  - May be either intrapericardial or extrapericardial
  - Intrapericardial masses invariably will have pericardial effusion
    - May be massive and mistaken for pleural effusion
    - At risk for cardiac tamponade
- **Teratoma, Chest**
  - Typically originate from anterior mediastinum and can cross midline
  - May contain both solid and cystic components
  - Calcifications most specific feature but not always present
  - Can grow extremely rapidly
- **Congenital Lobar Emphysema**
  - Uniformly echogenic
  - More commonly upper lobe (L > R)
  - Rare to diagnosis in utero
  - Generally present in neonatal period with air trapping

Helpful Clues for Rare Diagnoses

- **Tracheal Atresia**
  - Symmetric, bilateral lung enlargement
  - Chest circumference enlarged
  - Inversion of diaphragm
  - Fluid-filled trachea and bronchi
  - Heart appears midline and compressed

Other Essential Information

- Postnatal workup should be done in all cases even if mass has "disappeared" in utero
  - Mass not truly gone, just regressed to a point that it is not discernible by routine scanning
  - Perform postnatal contrast-enhanced CT or MR
    - Chest X-ray may not show lesion
  - Postnatal resection somewhat controversial in asymptomatic individuals
    - Most feel risk of infection and malignancy warrants resection in all cases

Congenital Cystic Adenomatoid Malformation, Microcystic



Axial transabdominal ultrasound shows a typical example of a large microcystic CCAM ➡ at 22 weeks gestation. Several small macrocysts ➡ are seen in the periphery. There were no signs of hydrops.

Congenital Cystic Adenomatoid Malformation, Microcystic



Coronal ultrasound shows this lesion is causing significant mediastinal shift ➡. This mass gradually decreased in size. The newborn was stable at delivery and had an elective resection at 2 months of age.

SOLID/ECHOGENIC LUNG MASS

Congenital Cystic Adenomatoid Malformation, Microcystic



Bronchopulmonary Sequestration



(Left) *Coronal view of the fetal chest shows a large mass in the left hemithorax. Color Doppler shows vascular connections with the pulmonary circulation (Doppler cursor). This is typical for a CCAM.* ***(Right)*** *Coronal power Doppler ultrasound of the fetal chest shows a large feeding artery arising from the aorta supplying a wedge-shaped, echogenic lung mass. This is classic for a sequestration. Doppler is essential for differentiating CCAMs from sequestrations.*

Bronchopulmonary Sequestration



Bronchopulmonary Sequestration



(Left) *Coronal ultrasound shows a well-defined, triangular, echogenic mass at the left lung base. The heart was displaced to the right.* ***(Right)*** *Axial color Doppler ultrasound in the same case shows a large feeding vessel arising from the aorta. The fetus did well, and the sequestration was resected after delivery.*

Congenital Diaphragmatic Hernia



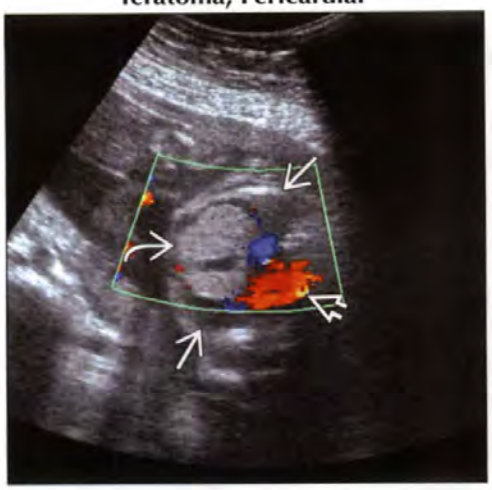
Congenital Diaphragmatic Hernia



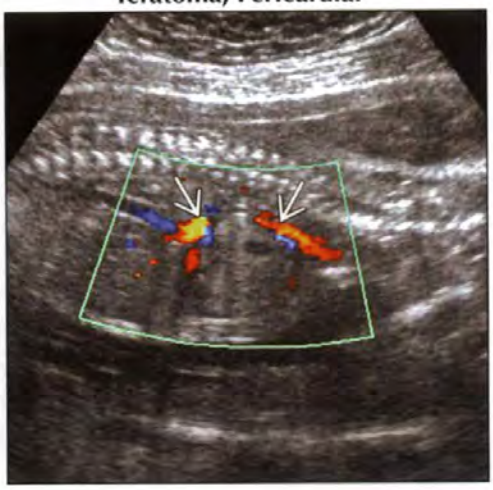
(Left) *Axial ultrasound through the fetal chest shows a large, solid, right-sided chest mass. The heart is being shifted to the left.* ***(Right)*** *Axial color Doppler ultrasound in the same case shows flow within a hepatic vein in this right-sided CDH. Liver is more hypoechoic than would be typical for either a CCAM or sequestration.*

SOLID/ECHOGENIC LUNG MASS

Teratoma, Pericardial

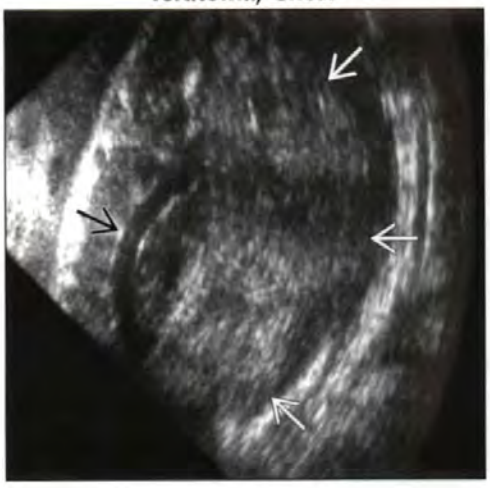


Teratoma, Pericardial

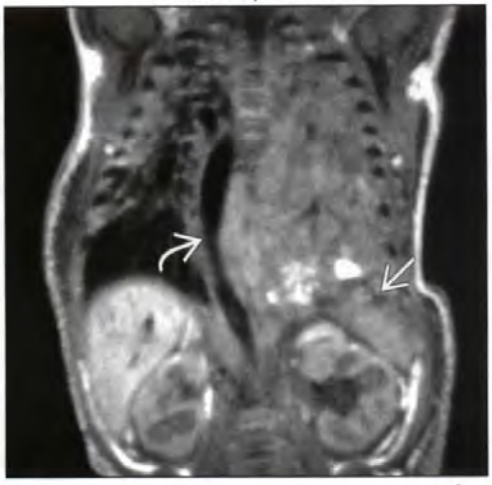


(Left) *Axial color Doppler ultrasound shows a large intrapericardial teratoma adjacent to the heart. It is surrounded by a massive pericardial effusion.* ***(Right)*** *Sagittal color Doppler ultrasound shows deviation of the aorta, but the mass itself had little flow and no feeding vessel could be identified. This helps differentiate a teratoma from a sequestration or CCAM.*

Teratoma, Chest

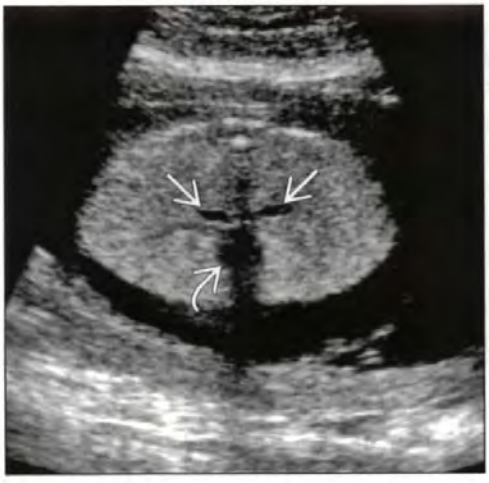


Teratoma, Chest

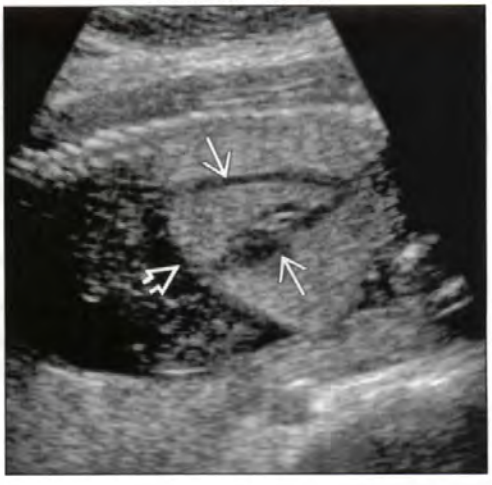


(Left) *Coronal ultrasound shows a large, solid mass within the left hemithorax displacing the aorta to the right. Calcifications were seen on other views. This fetus had a normal scan 6 weeks earlier.* ***(Right)*** *Coronal T1WI MR after delivery likewise shows displacement of the aorta by the mass. The mass has grown through the diaphragm into the upper abdomen. An immature teratoma was found at autopsy. Teratomas can have extremely rapid growth rates.*

Tracheal Atresia



Tracheal Atresia



(Left) *Axial ultrasound through the fetal chest shows markedly enlarged, hyperechoic lungs. The bronchi are fluid-filled. The heart is shifted to the midline and appears small, almost completely obscured by shadowing from the vertebral body. Massive bilateral lung enlargement is typical for tracheal atresia.* ***(Right)*** *Sagittal ultrasound shows fluid-filled bronchi well out into the periphery of the lung. Also note that the diaphragm is inverted.*

CYSTIC LUNG MASS

DIFFERENTIAL DIAGNOSIS

Common

- Congenital Diaphragmatic Hernia
- Congenital Cystic Adenomatoid Malformation, Macrocystic

Less Common

- Lymphangioma
- Bronchogenic Cyst
- Neurenteric Cyst

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Where is the stomach?
  - If it is below the diaphragm, a congenital diaphragmatic hernia (CDH) is less likely
    - CDH without gastric herniation (small bowel, liver) generally appears as solid or echogenic mass
- Simple cyst vs. complex cystic mass
  - Bronchogenic cyst and neurenteric cyst more likely to be single simple cysts
  - CDH, macrocystic congenital cystic adenomatoid malformation (CCAM), and lymphangioma are generally large complex masses
- Does the mass extend beyond the chest wall?
  - Lymphangioma has bulk of mass in subcutaneous tissues, not in chest cavity
- Always evaluate spine
  - Neurenteric cysts often associated with thoracic boney abnormality

Helpful Clues for Common Diagnoses

- **Congenital Diaphragmatic Hernia**
  - Left-sided hernias most common (80-90%)
  - Stomach causes cystic mass in chest
  - Four classic findings of left-sided hernia
    - Cystic mass in left side of chest
    - Absence of fluid-filled stomach in abdomen
    - Deviation of heart toward right
    - Polyhydramnios
  - Small bowel and liver are often also herniated but harder to see
    - Very important to determine contents of hernia
    - Those with "liver up" have poorer prognosis
  - Abdominal circumference less than expected
  - Most hernias are posterior through foramen of Bochdalek
    - Must evaluate diaphragm in sagittal plane
    - Anterior coronal plane can completely miss diaphragmatic defect
  - Look carefully for other anomalies including chromosomal
    - Up to 50% will have an associate abnormality
  - Color Doppler helpful to look for portal/hepatic veins in herniated liver
    - Even with Doppler, it may be difficult to see liver if only a small portion of the left lobe is herniated
  - MR extremely valuable for determining contents of hernia
    - Meconium-filled bowel is bright on T1WI
    - Fluid-filled bowel is bright on T2WI
    - Liver is dark on T2WI
    - Normal lung intermediate signal on T2WI
- **Congenital Cystic Adenomatoid Malformation, Macrocystic**
  - Lung hamartoma with proliferation of terminal bronchioles and lack of normal alveoli
  - Morphology varies from solid appearing (microcystic) to complex cystic mass (macrocystic) or even unilocular
  - Macrocystic congenital cystic adenomatoid malformation (CCAM)
    - 1 or more cysts > 5 mm
    - Often multiple cysts of varying sizes
    - May have single large cyst
    - Borders poorly defined
  - Both arterial supply and venous drainage from pulmonary circulation
  - Stomach is below diaphragm
  - Abdominal circumference is normal
  - 95% unilateral and affect only 1 lobe
  - No predilection for side (R = L)

Helpful Clues for Less Common Diagnoses

- **Lymphangioma**
  - Complex cystic body wall mass
    - Sonolucent cysts
    - Septa of variable thicknesses
    - No solid components

- May enlarge during pregnancy
- Associated anomalies rare

- Mediastinal involvement common but bulk of the mass will be outside thoracic cavity
- Can be located anywhere in soft tissues
  - 70% are axillary
  - 30% involve trunk or limbs
- Axillary masses typically between arm and chest wall
  - Arm held away from fetal trunk
  - May extend down arm
  - Secondary lymphedema common
  - Rib deformity common
  - Associated pleural effusion rare

- **Bronchogenic Cyst**
  - Smooth, unilocular cyst
  - Most commonly mediastinal but may be in lung parenchyma
  - Mediastinal cysts
    - Majority in middle mediastinum typically paratracheal, carinal, or hilar
  - Pulmonary
    - Majority in medial third of lungs
    - More frequent in lower lobes
    - Equal incidence in both lungs (R = L)
- **Neurenteric Cyst**
  - Single, smooth, unilocular cyst
  - Round to dumbbell shape
    - Dumbbell shape suggests extension into spinal canal
  - Located midline by spine
    - Thoracic spine most common site, followed by cervical spine
  - Vertebral anomalies (both segmentation and fusion) in up to 50%
    - Butterfly vertebrae
    - Hemivertebrae
    - Vertebral clefts
    - Diastematomyelia
  - Etiology thought to be incomplete separation of notochord layer from endoderm (primitive foregut)
    - Small piece of primitive gut becomes trapped in developing spinal canal

Other Essential Information

- Consider fetal MR
  - Especially important for evaluating contents of CDH
    - MR excellent to look for herniated liver
  - Evaluate mediastinal invasion of lymphangioma
  - Look for intraspinal extension of neurenteric cyst

Alternative Differential Approaches

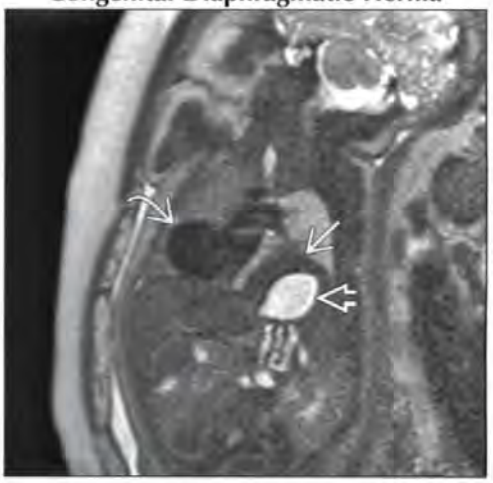
- Simple cyst
  - Bronchogenic cyst
  - Neurenteric cyst
  - CDH with only stomach herniated
  - CCAM with only one cyst
- Complex cystic mass
  - Congenital diaphragmatic hernia
  - Cystic adenomatoid malformation
  - Lymphangioma

Congenital Diaphragmatic Hernia



Axial ultrasound shows the stomach, bowel, and liver in the fetal chest. The heart is shifted into the right hemithorax.

Congenital Diaphragmatic Hernia



Coronal T2WI MR shows the heart shifted to the right. MR clearly shows herniation of the left lobe of the liver (stomach). This was not seen at US and is important as it confers a worse prognosis.

CYSTIC LUNG MASS

Congenital Diaphragmatic Hernia



Congenital Diaphragmatic Hernia



(Left) *Axial ultrasound shows the heart shifted into the right hemithorax. The stomach is in the chest.* ***(Right)*** *Axial ultrasound of the abdomen shows no stomach bubble, confirming the cystic structure within the chest, was indeed, the stomach. The abdominal circumference measured less than expected, which is typical, especially when the CDH is large.*

Congenital Cystic Adenomatoid Malformation, Macrocystic



Congenital Cystic Adenomatoid Malformation, Macrocystic



(Left) *Axial ultrasound shows the heart deviated to the right by a complex echogenic mass with scattered cysts. The right lung is compressed. Doppler interrogation of the mass showed pulmonary arterial supply and venous drainage.* ***(Right)*** *Sagittal ultrasound in the same case shows the mass filling the left hemithorax with larger cysts in the upper lobe. It is common to have both macrocystic and microcystic areas within a CCAM.*

Congenital Cystic Adenomatoid Malformation, Macrocystic



Congenital Cystic Adenomatoid Malformation, Macrocystic



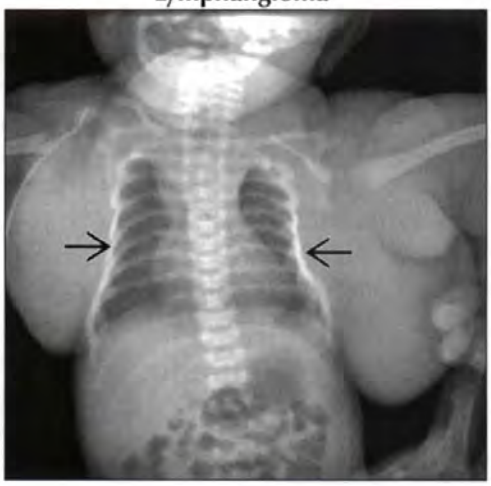
(Left) *Sagittal ultrasound shows the stomach clearly below the diaphragm, which is intact. There are two large cysts in the left hemithorax.* ***(Right)*** *Radiograph after delivery shows these cysts are now filled with air. There is mass-effect with shift of the heart to the right. Note the stomach is below the diaphragm.*

CYSTIC LUNG MASS

Lymphangioma

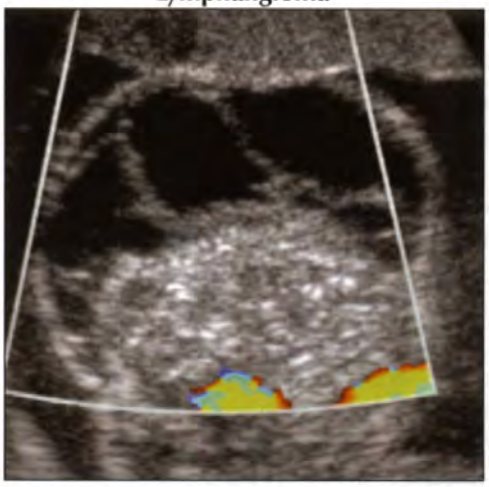


Lymphangioma

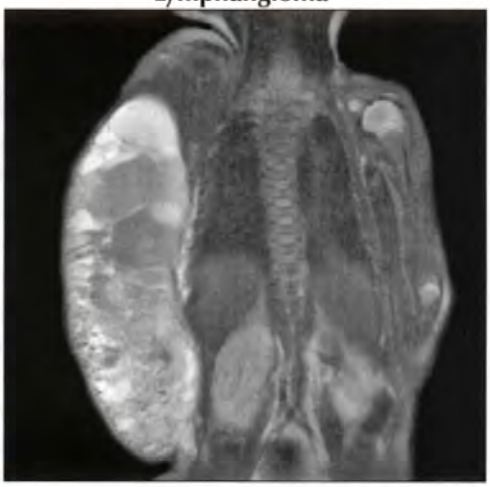


(Left) *Axial ultrasound shows bilateral, large, complex, multiloculated chest wall masses. Thin and thick septations are seen throughout.* ***(Right)*** *Postnatal chest radiograph shows the arms extending away from the trunk and lateral rib deformity secondary to in utero mass effect.*

Lymphangioma

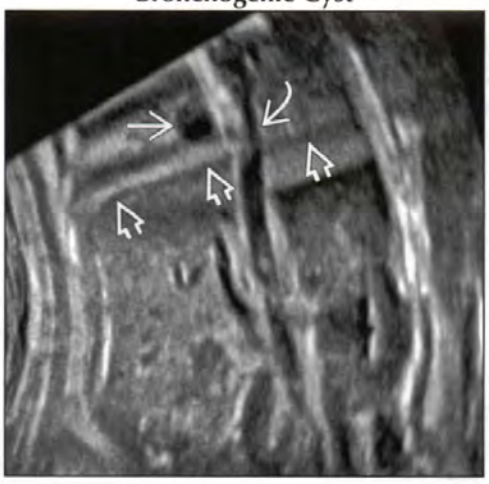


Lymphangioma



(Left) *Axial color Doppler ultrasound shows an avascular, cystic, chest wall mass.* ***(Right)*** *Coronal T2WI MR after delivery shows multiple locules of varying signal intensity. The mass extended from the axilla to the pelvis. Lymphangiomas may extend into the pleural space, mediastinum or abdomen but the bulk of the mass is generally subcutaneous.*

Bronchogenic Cyst



Bronchogenic Cyst



(Left) *Coronal ultrasound shows a small, simple, midline cyst near the right lung base. Note that the diaphragm is intact (aorta).* ***(Right)*** *Axial CECT after delivery shows a simple, fluid-density cyst. Note the adjacent vertebral body is normal making neurenteric cyst less likely. Resection confirmed a bronchogenic cyst.*

SMALL CHEST

DIFFERENTIAL DIAGNOSIS

Common

- Severe Oligohydramnios
  - Renal Agenesis
  - Autosomal Recessive Polycystic Kidney Disease, Severe
  - Bilateral Multicystic Dysplastic Kidneys
  - Posterior Urethral Valves, Complete
  - Severe Intrauterine Growth Restriction (IUGR)

Less Common

- Skeletal Dysplasias
  - Thanatophoric Dysplasia (TD)
  - Osteogenesis Imperfecta (OI)
  - Achondroplasia
  - Achondrogenesis
  - Asphyxiating Thoracic Dysplasia (Jeune Syndrome)
  - Short Rib-Polydactyly

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Is the chest truly small?
  - Large heart or distended abdomen may create appearance of a small chest
- Anything that causes severe oligohydramnios can result in a small chest and pulmonary hypoplasia
  - Fetal compression causes decreased space for lung growth and restriction of breathing movements
  - Efflux of lung fluid into amniotic space
    - Fetal lung fluid functions as a stent keeping developing air spaces distended
    - Vital for normal lung development
- Many skeletal dysplasias have a small thoracic cavity
  - Must look at multiple factors in order to make appropriate diagnosis
    - Which long bones are effected and how severely?
    - Is ossification normal or decreased?
    - Are there rib/long bone fractures or bowing?

Helpful Clues for Common Diagnoses

- **Renal Agenesis**
  - Absent kidneys and bladder
  - Anhydramnios
- **Autosomal Recessive Polycystic Kidney Disease, Severe**
  - Large, echogenic kidneys
  - Abdominal circumference often enlarged
  - Amount of amniotic fluid varies according to severity of renal disease
- **Bilateral Multicystic Dysplastic Kidneys**
  - Non-functioning kidneys filled with multiple cysts of varying size
  - Absent bladder
- **Posterior Urethral Valves, Complete**
  - Grossly distended bladder in male fetus
  - Look for "keyhole" appearance of dilated posterior urethra
  - May have associated hydronephrosis
- **Severe Intrauterine Growth Restriction (IUGR)**
  - Asymmetric IUGR: Abdomen and chest are small, with "sparing" of head growth
  - Often associated with oligohydramnios
  - Early onset asymmetric IUGR very concerning for triploidy

Helpful Clues for Less Common Diagnoses

- **Thanatophoric Dysplasia (TD)**
  - Divided into 2 subtypes based on morphologic findings
  - Chest findings are similar in both types
    - Thorax is small and narrow with short horizontal ribs
    - Ossification is normal
    - No rib fractures
    - Pulmonary hypoplasia
  - TD type I
    - "Telephone receiver" femur
    - All long bones severely affected (micromelia) with bowing
    - Platyspondyly
    - Prominent lumbar kyphosis
    - Trident-shaped hands (short fingers, appear same length; gap between 3rd and 4th fingers)
  - TD type II
    - Kleeblattschädel ("cloverleaf") skull
    - Femurs longer, less curved
    - Platyspondyly less marked
- **Osteogenesis Imperfecta (OI)**
  - Presence of fractures distinguishes OI from other skeletal dysplasias (except achondrogenesis type 1A)
  - Multiple different types based on phenotype

- Type II is most severe form and most likely to be diagnosed in utero
- Type II is perinatal lethal
- Chest is small with "beaded" ribs (result of multiple rib fractures)
- Long bone shortening/angulation secondary to fractures
- Callus formation gives bones a "crumpled" appearance
- Decreased mineralization
- Brain "too well seen"
- Skull deformation from transducer pressure ("soft" bones)

- **Achondroplasia**
  - Most common heritable, non-lethal skeletal dysplasia
  - Chest usually has mild "bell-shaped" appearance
    - Much less severe than other dysplasias
  - Early scans usually normal
  - Rhizomelia (proximal limb shortening) noted after 22 weeks
  - Normal ossification without fractures
  - No bowing or angulation seen prenatally
  - Progressive macrocephaly with frontal bossing
  - Prominent thoracolumbar kyphosis
  - Trident hands
- **Achondrogenesis**
  - Group of lethal disorders with severe micromelia, unossified spine, short trunk and disproportionately large head
  - Chest findings vary with type
  - Type 1A
    - Multiple rib fractures
    - Poorly ossified skull
  - Type 1B
    - No rib fractures
    - Poorly ossified skull
  - Type II
    - No rib fractures
    - Normal skull ossification
- **Asphyxiating Thoracic Dysplasia (Jeune Syndrome)**
  - Characterized by a severely constricted, long, narrow thorax
  - Cystic kidneys
  - Normal ossification
  - Polydactyly in 15%
  - Limbs shortened but may not be as profound as in other conditions
- **Short Rib-Polydactyly**
  - Characterized by severe micromelia, short horizontal ribs, polydactyly, visceral anomalies
  - Visceral anomalies include cardiac, urogenital and central nervous system

Alternative Differential Approaches

- Decreased ossification with beaded ribs
  - Osteogenesis imperfecta
  - Achondroplasia type Ia
- Absent spine ossification
  - Achondrogenesis (all types)
- Small chest and polydactyly
  - Short rib-polydactyly
  - Asphyxiating thoracic dysplasia

Renal Agenesis



Coronal ultrasound of a fetus with renal agenesis shows a very small bell-shaped chest. There is complete anhydramnios.

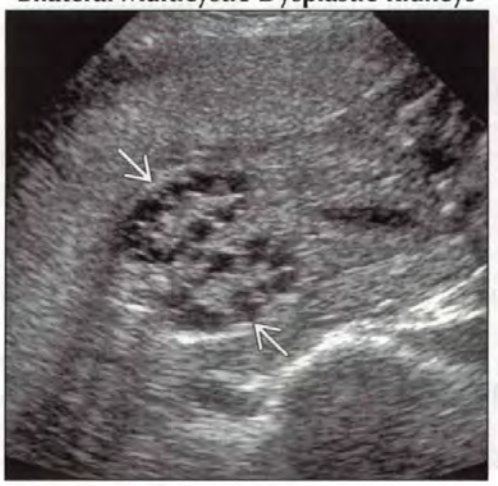
Autosomal Recessive Polycystic Kidney Disease, Severe



Coronal postmortem T2WI MR shows a tiny thoracic cavity. The kidneys are massively enlarged with no normal remaining parenchyma.

SMALL CHEST

Bilateral Multicystic Dysplastic Kidneys



(Left) *Coronal ultrasound of fetal abdomen shows bilateral, enlarged, cystic kidneys. As multicystic dysplastic kidneys are nonfunctioning, there is no amniotic fluid & consequently normal lung development cannot occur.* ***(Right)*** *Coronal ultrasound shows a dramatically distended bladder in a 15 week male fetus. Note the small chest and severe oligohydramnios.*

Posterior Urethral Valves, Complete



Severe Intrauterine Growth Restriction (IUGR)

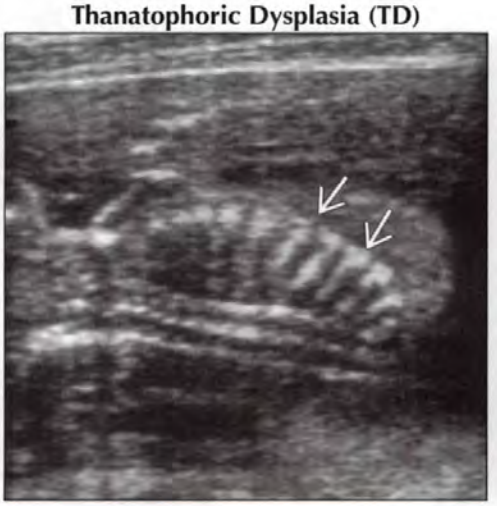


(Left) *Coronal ultrasound of a fetus from a dichorionic twin gestation. The fetal chest and abdomen are significantly smaller than the head. Amniocentesis showed triploidy. The other twin was normal.* ***(Right)*** *Postmortem radiograph shows normal ossification with dramatically shortened and bowed humeri. Note the platyspondyly with "H-shaped" vertebrae. Platyspondyly can be difficult to appreciate on ultrasound.*

Thanatophoric Dysplasia (TD)

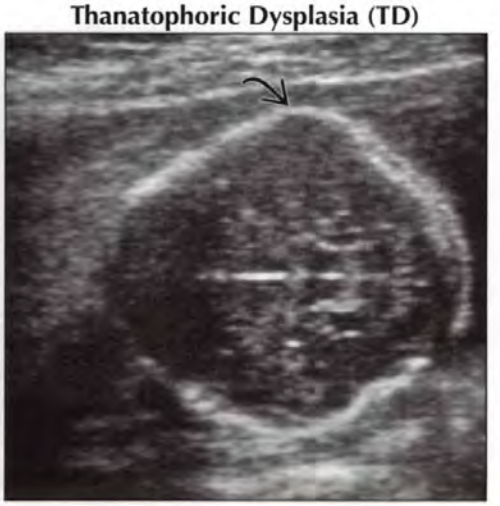


Thanatophoric Dysplasia (TD)



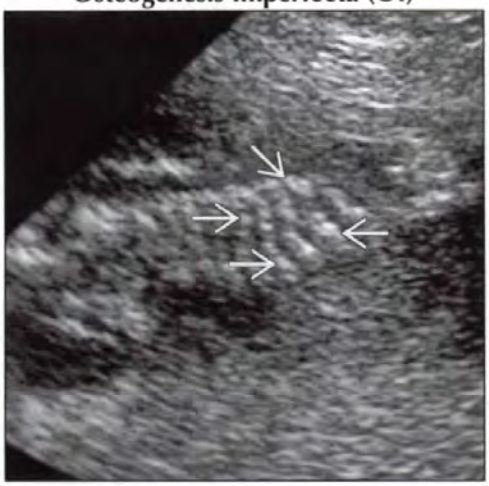
(Left) *Coronal oblique ultrasound of the fetal thorax shows short straight ribs, which do not go around the chest. The ossification is normal, and there are no rib fractures.* ***(Right)*** *Axial ultrasound of the fetal head in the same case shows the classic kleeblattschädel or "cloverleaf" skull configuration typical for TD type II.*

Thanatophoric Dysplasia (TD)



SMALL CHEST

Osteogenesis Imperfecta (OI)



Osteogenesis Imperfecta (OI)

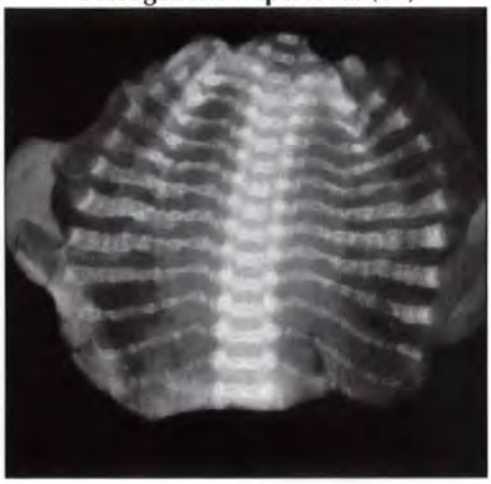


(Left) *Sagittal oblique ultrasound focused on the ribs shows they have a wavy, "beaded" appearance ➡. This is the result of fractures with callus formation.* ***(Right)*** *Axial ultrasound of the fetal head in the same case shows flattening of skull ➡ from the transducer pressure. Because the skull is poorly mineralized, there is little reverberation artifact, resulting in the brain being "too well seen".*

Osteogenesis Imperfecta (OI)



Osteogenesis Imperfecta (OI)



(Left) *Frontal radiograph shows a typical case of OI type 2 in a fetus with innumerable rib, vertebral, & long bone fractures. Arms & legs are short due to angulation & deformity resulting from the fractures. These fractures give the bones a "crumpled" appearance.* ***(Right)*** *Specimen radiograph of a fetal thoracic cavity, in a different case, shows innumerable rib fractures creating a "beaded" appearance.*

Achondroplasia



Achondroplasia



(Left) *Sagittal ultrasound shows a slightly small chest ➡, with a protuberant abdomen ➡. This is much less pronounced than in other skeletal dysplasias.* ***(Right)*** *Axial head ultrasound in the same fetus shows normal ossification of the skull, with reverberation artifact in the near field. The fetus was 33.2 weeks, but the head size measured 36.5 weeks. Macrocephaly with frontal bossing is a common 3rd trimester finding in achondroplasia.*

SMALL CHEST

Achondroplasia



(Left) *Lateral radiograph shows the calvarial shape of an infant with achondroplasia. The bone is well ossified. Note prominent frontal bossing* ➔ *and depressed nasal bridge* ➔. ***(Right)*** *Sagittal ultrasound shows a small chest in relation to the abdomen, but the key finding is the complete lack of vertebral body ossification* ➔.

Achondrogenesis



Achondrogenesis



(Left) *Sagittal ultrasound focused on the ribs* ➔ *of the same fetus shows they are very short and "beaded", the result of rib fractures.* ***(Right)*** *Axial oblique ultrasound shows the calvarium is deformed by transducer pressure* ➔. *These findings are characteristic of achondrogenesis type 1A, which was confirmed at autopsy.*

Achondrogenesis

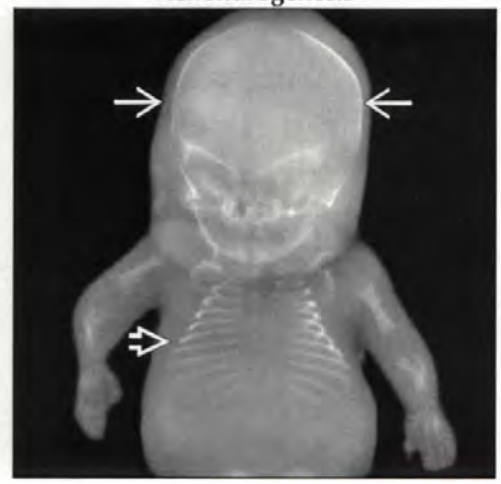


Achondrogenesis



(Left) *Radiograph of a fetus with achondrogenesis type IA shows poor skull ossification* ➔ *and thin, wavy ribs secondary to multiple fractures* ➔. *Note the lack of vertebral body ossification.* ***(Right)*** *Radiograph shows a well-ossified calvarium* ➔ *with lack of spine ossification in type II achondrogenesis. Note the absence of rib fractures* ➔.

Achondrogenesis



Asphyxiating Thoracic Dysplasia (Jeune Syndrome)

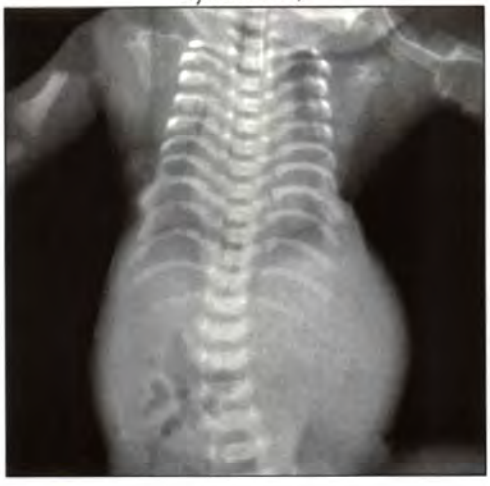


Asphyxiating Thoracic Dysplasia (Jeune Syndrome)



(Left) *Sagittal ultrasound shows a markedly small chest ➡ when compared to the fetal abdomen ➡. Note the chest has a more elongated shape than typically seen in other dysplasias.* ***(Right)*** *Axial ultrasound through the chest shows the ribs ➡ are very short and horizontal.*

Asphyxiating Thoracic Dysplasia (Jeune Syndrome)



Asphyxiating Thoracic Dysplasia (Jeune Syndrome)



(Left) *Frontal radiograph from the prior case shows a very long and narrow thoracic configuration typical of Jeune syndrome. Note that ossification is normal.* ***(Right)*** *Lateral radiograph shows how "sunken" the chest appears in relation to the abdomen. Note the extreme rib shortening ⇒.*

Short Rib-Polydactyly



Short Rib-Polydactyly



(Left) *Ultrasound of the hand shows postaxial polydactyly, with six fingers ➡ and a thumb ➡. Both pre- and postaxial polydactyly have been described with short rib-polydactyly syndrome.* ***(Right)*** *Lateral radiograph shows severely shortened ribs ⇒ but normal ossification. Multiple other anomalies may be present including cardiac, urogenital, and central nervous system.*

SECTION 5
Cardiac

Abnormal Cardiac Axis	5-2
Chamber Asymmetry	5-8
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Septal Defect	5-16
Abnormal Outflow Tracts	5-20
Abnormal Rate or Rhythm	5-24
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ABNORMAL CARDIAC AXIS

DIFFERENTIAL DIAGNOSIS

Common

- Chest Mass
  - Congenital Diaphragmatic Hernia
  - Cystic Adenomatoid Malformation
  - Bronchopulmonary Sequestration
  - Pleural Effusion
  - Teratoma
- Cardiac
  - Chamber Asymmetry
  - Conotruncal Malformation
  - Heterotaxy, Cardiosplenic Syndromes

Less Common

- Pulmonary Agenesis

Rare but Important

- Ectopia Cordis

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Important to have systematic approach
- In all OB scans check fetal orientation
  - Which is the fetal anatomic left and right?
- Check position of stomach
- Check position of cardiac apex
- Stomach and cardiac apex should both be on the left
  - If both on right, likely complete situs inversus with good prognosis
  - If opposite sides, likely heterotaxy syndrome
    - Strong association with complex congenital heart disease
- Normal four chamber view is seen on an axial image of the chest
  - Ribs should be symmetric and C-shaped
- Normal cardiac axis is 35° to 45°
  - Draw a line from spine to sternum
  - Draw a line along axis of intraventricular septum
- If axis is abnormal
  - Does the heart appear displaced within the thorax?
    - May be "pushed" to one side by a mass
    - May be "pulled" to one side if lung small or absent
  - Ectopia cordis implies heart situated outside thorax
    - Intra-abdominal
    - Extrathoracic
  - Is the internal cardiac structure normal?
    - Normal right and left atria
    - Normal right and left ventricles
    - Normal outflow tracts crossing as they exit the heart
    - Atrioventricular concordance
    - Ventriculoarterial concordance

Helpful Clues for Common Diagnoses

- **Congenital Diaphragmatic Hernia**
  - Stomach/intestine ± liver in chest
  - Heart displaced away from side of hernia
    - In bilateral hernias, there may be minimal cardiac shift
  - Look for peristalsis within chest
  - Look for "bucket handle" motion of diaphragm on coronal view
  - Strong association with aneuploidy
- **Cystic Adenomatoid Malformation**
  - Chest mass with perfusion from pulmonary artery branches
  - May be uniformly echogenic to multicystic depending on type
  - Heart displaced away from mass
- **Bronchopulmonary Sequestration**
  - Echogenic mass with perfusion from aorta
  - Usually on left, with cardiac shift to the right side
- **Pleural Effusion**
  - Large solitary effusion may displace heart
  - Look for floating lung
  - Differentiate from pericardial effusion
    - Surrounds heart, displaces lung posteriorly
- **Teratoma**
  - Complex cystic/solid mass ± calcifications
- **Chamber Asymmetry**
  - Which chamber is abnormal? Or is it a single ventricle heart?
  - Right heart enlargement
    - Shunt lesions with increased venous return
    - Incipient hydrops
    - Severe placental insufficiency
    - Left heart outflow obstruction
  - Small right ventricle (RV)
    - Pulmonary atresia/stenosis (RV can also be normal)
    - Left dominant unbalanced atrioventricular septal defect (AVSD)
  - Small left ventricle (LV)

- Hypoplastic left heart syndrome (may have poorly functioning echogenic LV in aortic stenosis with endocardial fibroelastosis)
- Right dominant unbalanced AVSD
- Large right atrium
- Ebstein anomaly/tricuspid dysplasia
- Pulmonary stenosis/atresia

- **Conotruncal Malformation**
  - Four chamber view often shows normal chambers
  - Look at outflow tracts in every case
    - Single outflow: Truncus most likely if normal sized ventricles and VSD present
    - Parallel outflow tracts: Transposition of the great arteries or double outlet right ventricle
    - Large aorta overriding VSD with separate, small PA: Tetralogy of Fallot
- **Heterotaxy, Cardiosplenic Syndromes**
  - Check situs in every OB scan: Cardiac apex and stomach should be on the left
  - Look for interrupted inferior vena cava with azygous continuation to the superior vena cava
    - Vessel located posterior to the aorta at the level of the diaphragm
  - Look for transverse, midline liver
  - Complex congenital heart disease
    - Often AV septal defect
    - Often single ventricle
    - Often abnormal outflow tracts
    - Systemic and pulmonary venous abnormalities

Helpful Clues for Less Common Diagnoses

- **Pulmonary Agenesis**
  - Heart displaced to chest wall on side of missing lung
  - Diaphragm elevated but present on side of missing lung
  - No evidence of diaphragmatic hernia/lung mass "pushing" heart
  - Look for associated vertebral anomalies or congenital heart disease
  - Look for other features of VACTERL association

Helpful Clues for Rare Diagnoses

- **Ectopia Cordis**
  - Heart in abnormal location
  - Look for amniotic bands if exterior to thorax
  - Pentalogy of Cantrell
    - Anterior diaphragmatic hernia
    - Midline abdominal wall defect
    - Cardiac anomalies
    - Defect of diaphragmatic pericardium
    - Low sternal defect

Other Essential Information

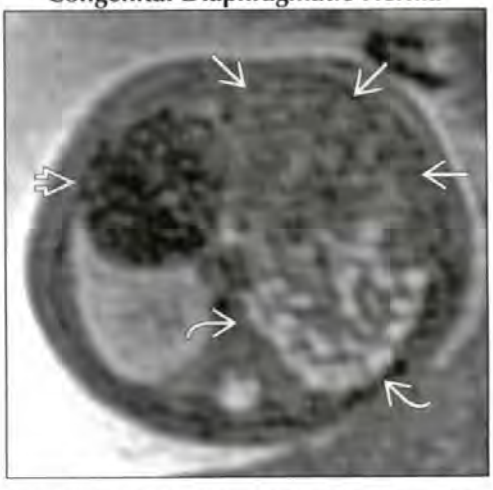
- Prognosis in heterotaxy syndromes depends on complexity of cardiac disease
  - Association with complete heart block almost uniformly fatal
- Prognosis in diaphragmatic hernia depends on liver position and presence of cardiac defects
  - "Liver up" or complex cardiac anomaly confers worse prognosis

Congenital Diaphragmatic Hernia



Axial ultrasound shows the stomach ➡ behind the heart ➡, which is displaced to the right. Posterior displacement of the stomach suggests "liver up" CDH, which confers poor prognosis.

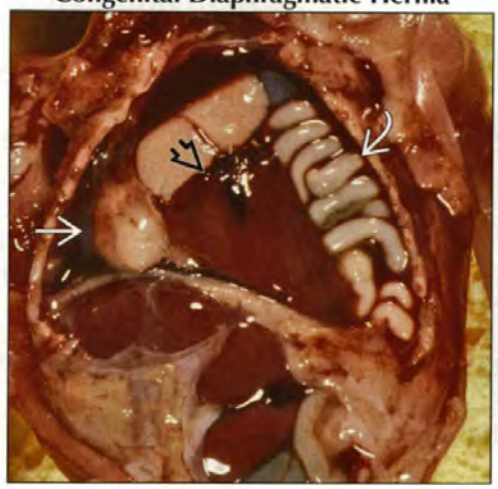
Congenital Diaphragmatic Hernia



Axial T2WI MR shows bowel ➡ and compressed lung ➡ (which could be mistaken for liver) in the left chest with rightward heart ➡ displacement. Coronal views proved that the liver was not in the chest.

ABNORMAL CARDIAC AXIS

Congenital Diaphragmatic Hernia

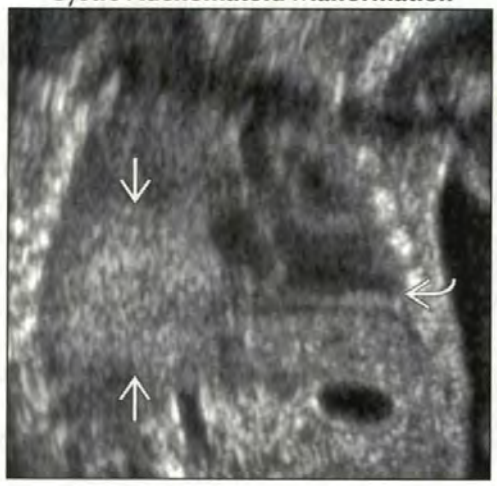


Cystic Adenomatoid Malformation



(Left) *Gross pathology shows both small bowel and liver in the chest. The heart is displaced to the right.* ***(Right)*** *Axial ultrasound shows a large, echogenic mass with small, scattered cysts consistent with a congenital cystic adenomatoid malformation. There is marked displacement of the heart and compression of the contralateral lung.*

Cystic Adenomatoid Malformation

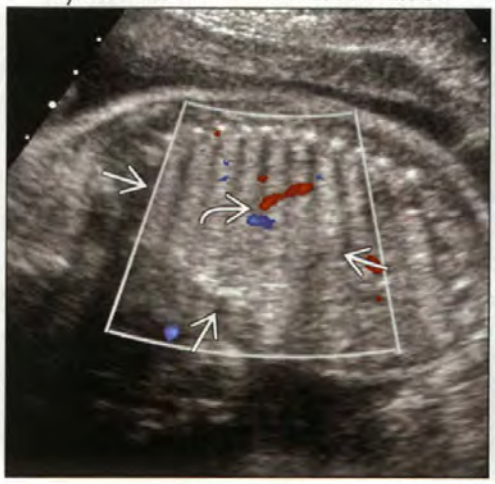


Cystic Adenomatoid Malformation

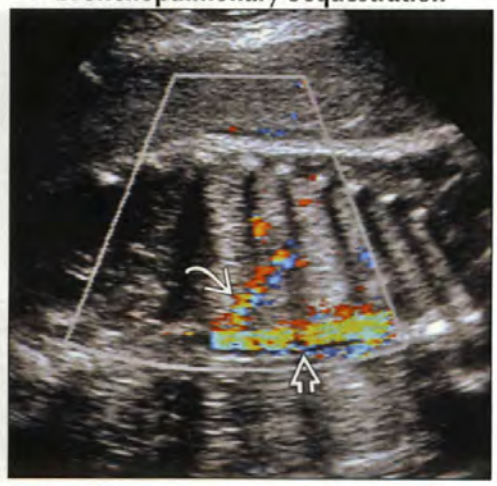


(Left) *Coronal ultrasound shows an echogenic CCAM displacing the heart to the left chest wall.* ***(Right)*** *Coronal T2WI MR shows a CCAM occupying the right upper lobe with minimal mediastinal shift and cardiac displacement to the left.*

Cystic Adenomatoid Malformation



Bronchopulmonary Sequestration



(Left) *Sagittal oblique color Doppler ultrasound shows a large congenital cystic adenomatoid malformation perfused by branches of the pulmonary artery.* ***(Right)*** *Coronal color Doppler ultrasound shows the blood supply to an echogenic, non-cystic chest mass arising from the aorta, typical for sequestration.*

ABNORMAL CARDIAC AXIS

Bronchopulmonary Sequestration

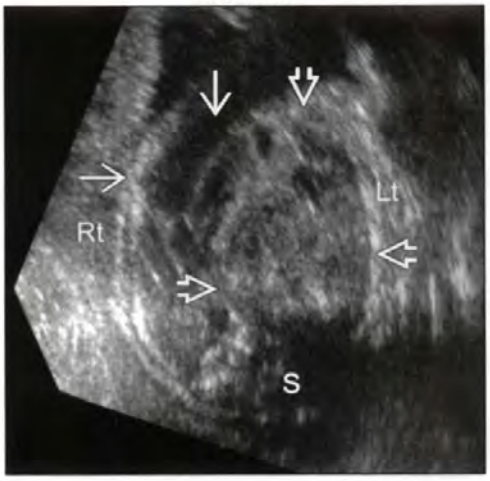


Pleural Effusion



(Left) *Axial ultrasound in the same case as the previous image shows a well-defined, echogenic mass ➡ at the left lung base. The heart ➡ is displaced to the right.* ***(Right)*** *Axial ultrasound shows a huge, right-sided chylothorax ➡ compressing the right lung ➡ and deviating the heart ➡. Skin edema is also present. Fetal thoracentesis was performed, but hydrops worsened despite intervention.*

Teratoma

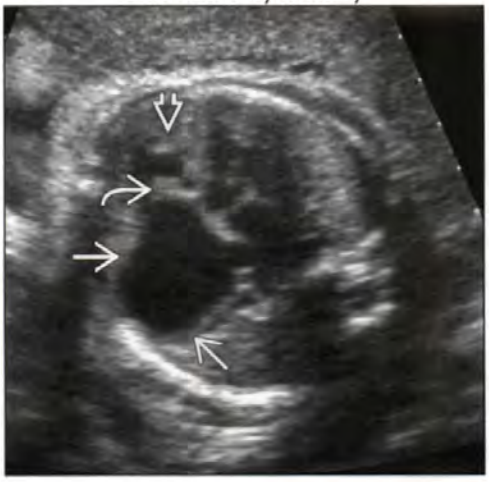


Chamber Asymmetry

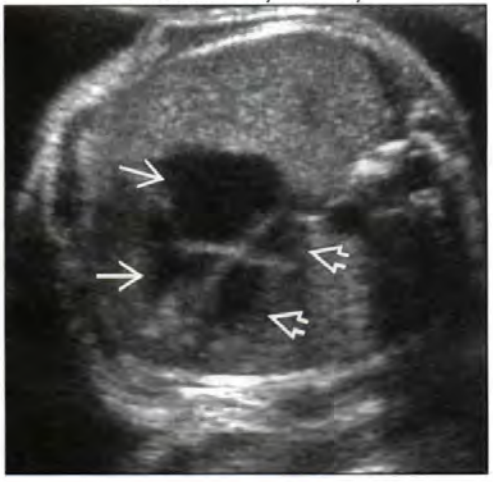


(Left) *Four chamber view shows the heart ➡ displaced to the right chest wall by a complex mass ➡ arising in the left hemithorax (S = spine). Calcifications were seen in other views.* ***(Right)*** *Axial ultrasound in a hydropic fetus with hypoplastic left heart syndrome shows a large anterior right ventricle (RV) and small left ventricle ➡ due to mitral atresia ➡. The axis (angle between lines connecting asterisks and Xs) is abnormal at almost 90 degrees.*

Chamber Asymmetry



Chamber Asymmetry



(Left) *Four chamber view shows an abnormal axis secondary to dramatic right atrial enlargement ➡ in a fetus with Ebstein anomaly. Note the inferior displacement of the septal tricuspid leaflet ➡, causing atrialization of the right ventricle, the functional part ➡ being very small.* ***(Right)*** *Four chamber view in a fetus with multiple anomalies including pulmonary stenosis shows that the right-sided cardiac chambers ➡ are larger than the left ➡. Final diagnosis was trisomy 18.*

ABNORMAL CARDIAC AXIS

Conotruncal Malformation



Conotruncal Malformation

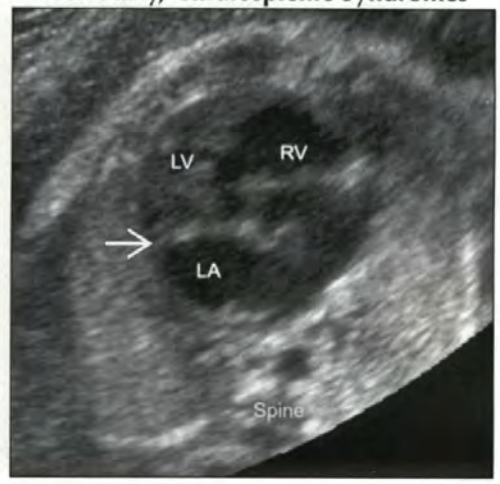


(Left) *Four chamber view ultrasound shows an abnormal axis and right atrial enlargement with normal placement of the tricuspid valve leaflets. Additional images demonstrated pulmonary atresia.* ***(Right)*** *RVOT ultrasound in a fetus with heterotaxy shows a double outlet right ventricle with parallel aorta (A) and pulmonary artery (P) exiting the right ventricle (RV). The cardiac axis was abnormal.*

Heterotaxy, Cardiosplenic Syndromes



Heterotaxy, Cardiosplenic Syndromes

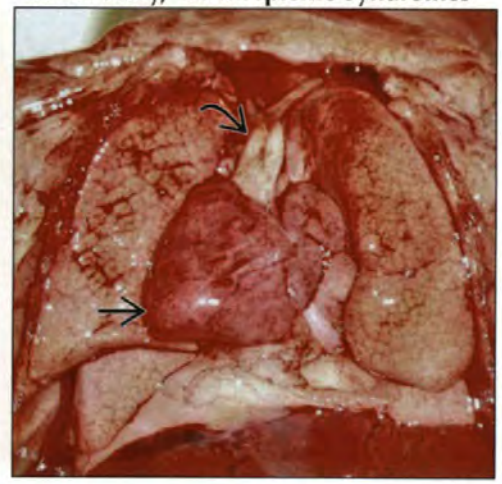


(Left) *Axial oblique ultrasound shows that the stomach and cardiac apex are on opposite sides of the abdomen.* ***(Right)*** *Four chamber view in the same case shows an abnormal axis with a small left ventricle (LV) secondary to mitral atresia. Left atrium (LA), right ventricle (RV).*

Heterotaxy, Cardiosplenic Syndromes



Heterotaxy, Cardiosplenic Syndromes



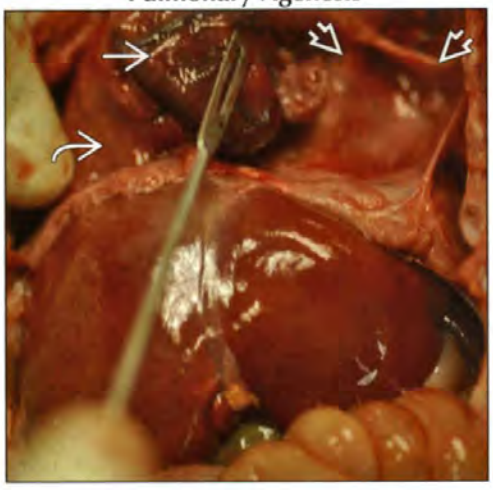
(Left) *Four chamber view at 16 weeks shows an abnormal axis, although the apex is pointed left. Final diagnosis was left-dominant, unbalanced atrioventricular septal defect and truncus arteriosus.* ***(Right)*** *Gross pathology shows dextrocardia and a right-sided aortic arch in a case of heterotaxy. There was also asplenia, total anomalous pulmonary venous return, and bowel malrotation.*

ABNORMAL CARDIAC AXIS

Pulmonary Agenesis



Pulmonary Agenesis

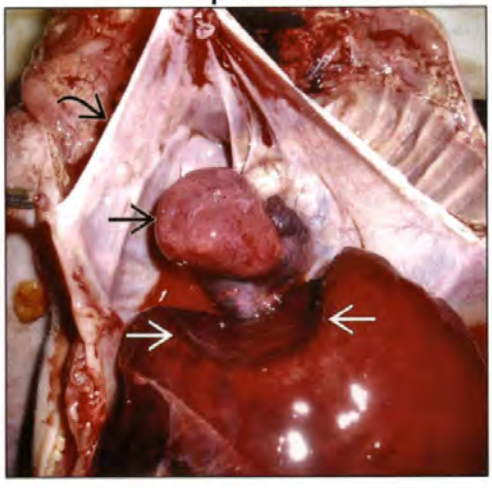


(Left) *Axial T2WI MR shows a normal right lung ➡ with the heart ➡ touching the left chest wall due to left pulmonary agenesis.* ***(Right)*** *Gross pathology in the same case shows the right lung ➡ and the "empty" left hemithorax ➡ with the heart ➡ retracted. The fetus had multiple other anomalies including a large encephalocele. The newborn died within minutes of delivery.*

Ectopia Cordis

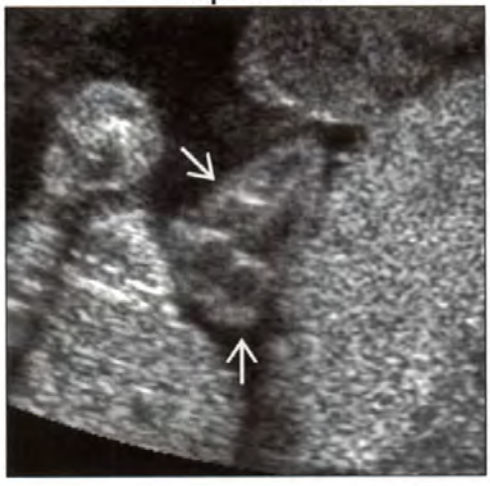


Ectopia Cordis



(Left) *Axial ultrasound through the fetal abdomen shows the heart ➡ within the abdomen, protruding through an abdominal wall defect ➡. The complexity of the vascular connections precluded successful repair.* ***(Right)*** *Gross pathology shows a different case of abdominal ectopia cordis. The heart ➡ is seen beneath the elevated and retracted diaphragm ➡. Note the mass effect ➡ on the dome of the liver.*

Ectopia Cordis



Ectopia Cordis



(Left) *Four chamber view in a fetus with amniotic band syndrome shows the extrathoracic location of the heart ➡, which is surrounded by amniotic fluid. Multiple other anomalies including a large facial cleft and exencephaly were also seen.* ***(Right)*** *Axial ultrasound shows a large omphalocele ➡ containing liver, small bowel ➡, and part of the stomach ➡. Further images showed the cardiac apex involved in the defect consistent with pentalogy of Cantrell.*

CHAMBER ASYMMETRY

DIFFERENTIAL DIAGNOSIS

Common

- Hypoplastic Left Heart Syndrome (HLHS)
- Tricuspid Atresia (TA)
- Pulmonary Valve Atresia with Intact Ventricular Septum
- Tricuspid Dysplasia
- Ebstein Anomaly

Less Common

- Atrioventricular Septal Defect (AVSD), Unbalanced
- Coarctation of the Aorta

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Are there one or two ventricles?
  - If only one, is it morphologically the right or left ventricle?
  - If two ventricles, do they both reach the apex of the heart?
- Are there one or two atrioventricular (AV) valves?
  - If only one AV valve, is it the anterior (tricuspid) or posterior (mitral) valve?
  - Are the AV valves normal in size?
  - Are the valves located in the same plane or off-set?
    - Normally tricuspid insertion is more apical than mitral
  - Is there AV valve regurgitation?
- Are there one or two great arteries?
  - Are they normal in size?
  - Does blood flow into both from the heart?
  - Is perfusion from the ductus?

Helpful Clues for Common Diagnoses

- **Hypoplastic Left Heart Syndrome (HLHS)**
  - Abnormal four chamber view with small, non-apex-forming left ventricle (LV)
    - May see brightly echogenic LV endocardium with endocardial fibroelastosis
    - LV function is poor
  - Inter-atrial septum bowed left to right as flow across foramen ovale is reversed
    - Little or no antegrade flow from LV so LA blood refluxes into RA
  - Aortic valve often atretic and/or very small
  - Ascending aorta very small
  - Transverse arch very small
  - Right ventricle is large
    - RV wraps around apex of LV
    - Function is typically very good, even hyperdynamic
- **Tricuspid Atresia (TA)**
  - Abnormal four chamber view
    - Small, non-apex-forming right ventricle
  - Left ventricle is normal to large in size with good function
  - Tricuspid valve appears "plate-like" with no movement
  - Ventricular septal defect (VSD) usually present to provide blood flow to great artery arising from RV
  - Size of great artery arising from RV depends on size of VSD
    - Larger VSD → bigger vessel
    - Vessel may be pulmonary artery or aorta
- **Pulmonary Valve Atresia with Intact Ventricular Septum**
  - Four chamber view is abnormal
    - RV very hypertrophied and small
  - RV pressure is usually greater than systemic
  - Tricuspid valve often hypoplastic
  - Look for abnormal coronary flow over RV
    - Indicates presence of coronary sinusoids
    - Low velocity flow in small vessels
  - Reversed flow in the ductus arteriosus
    - Pulmonary artery fills retrograde from aortic arch, not antegrade from right ventricle
  - Ductus arteriosus is more vertically oriented than usual
- **Tricuspid Dysplasia**
  - Valve leaflets are in normal position
  - Leaflets are thick, nodular, or irregular
  - Severe tricuspid regurgitation (TR) → right atrial enlargement
  - Often associated with pulmonary stenosis/atresia
  - LV normal in size with good function
- **Ebstein Anomaly**
  - Apical displacement of septal and mural tricuspid valve leaflets with attachments to ventricular septum
  - Anterior leaflet is often "sail-like"
  - "Atrialization" of the right ventricle
    - Significant right atrial enlargement
    - Functional RV is small
  - Variable degrees of TR

- Pulmonary artery is often small
  - Severe TR → lack of antegrade flow to RV

Helpful Clues for Less Common Diagnoses

- **Atrioventricular Septal Defect (AVSD), Unbalanced**
  - Missing "crux" of heart in four chamber view
    - Inlet ventricular septal defect
    - Primum atrial septal defect
  - Single AV valve is committed more to one ventricle than the other
    - Valve located in the same plane in a four chamber view is a tip-off
    - Normally tricuspid and mitral valves offset on interventricular septum
  - Ventricle lacking commitment will be hypoplastic
  - Additional cardiac malformations are common
  - Look for features of heterotaxy syndromes
    - Situs abnormalities (e.g., dextrocardia, right-sided stomach)
    - Midline liver with central portal vein bifurcation
    - Anomalous venous drainage especially azygous continuation of inferior vena cava
  - Look for signs of trisomy 21
    - Thick nuchal fold, absent nasal bone
    - Duodenal atresia, echogenic bowel
    - Short humerus, femur
    - Pyelectasis
    - Sandal gap toes, clinodactyly
- **Coarctation of the Aorta**
  - RV mildly enlarged compared to LV
  - Transverse arch hypoplasia is best clue
  - VSD with posterior deviation of the infundibular septum raises suspicion
  - Difficult prenatal diagnosis

Other Essential Information

- HLHS, TA, and unbalanced AVSD
  - All considered single ventricles and require 3 stage surgical palliation
  - Outcomes for HLHS have improved in the short-term but remain poor for long-term
  - Outcomes for TA are better than HLHS with survivors into their 40s and 50s
  - Outcomes for an unbalanced AVSD depend on which ventricle is dominant and associated anomalies
- Pulmonary atresia with intact ventricular septum can have a very poor prognosis
  - Presence of coronary sinusoids may prevent decompressing RV with catheter intervention
    - Increases the risk of sudden death
- Tricuspid valve dysplasia has a good prognosis
  - Oxygen and the natural lowering of the pulmonary vascular resistance often decreases the TR significantly
  - Surgery may not be necessary
- Coarctation of the aorta has an excellent long-term outcome
  - 85% of patients need no additional intervention in their lifetime

Hypoplastic Left Heart Syndrome (HLHS)



Four chamber view echocardiogram shows an enlarged right ventricle (RV) wrapping around the apex of the hypoplastic left ventricle. Note the echogenic endocardium seen in endocardial fibroelastosis.

Tricuspid Atresia (TA)



Four chamber view echocardiogram shows a "plate-like" tricuspid valve with a normal mitral valve and left ventricle (LV). The right ventricle is hypoplastic, and there is a VSD. Left atrium (LA).

CHAMBER ASYMMETRY

Tricuspid Atresia (TA)

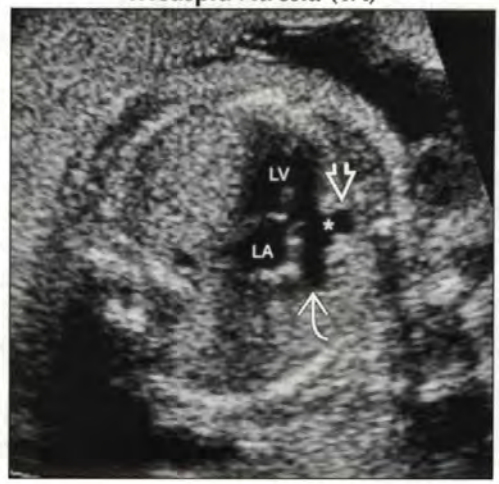


Tricuspid Atresia (TA)

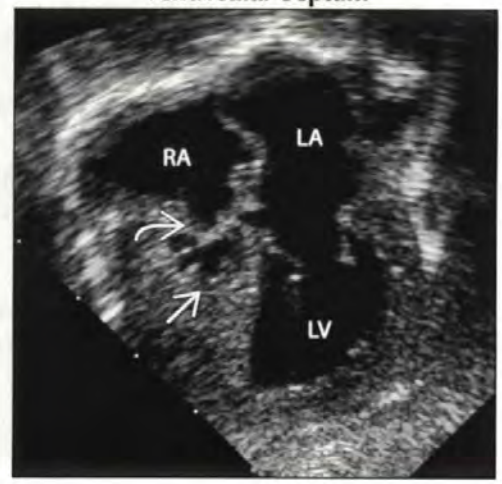


(Left) *Four chamber view shows a hypoplastic right ventricle with an atretic tricuspid valve. The left ventricle (LV) is mildly dilated. Left atrium (LA).* ***(Right)*** *Color Doppler echocardiogram in the same case shows flow from the right atrium, across the atrial septum to the left ventricle via the mitral valve. Blood cannot enter the right ventricle due to tricuspid atresia; therefore all right atrial blood crosses the foramen ovale to the left heart.*

Tricuspid Atresia (TA)



Pulmonary Valve Atresia with Intact Ventricular Septum



(Left) *LVOT view shows the aorta arising from the left ventricle (LV). A VSD (*) leads into the right ventricle, which is hypoplastic due to decreased inflow from tricuspid atresia.* ***(Right)*** *Four chamber view echocardiogram on day of life 1 shows a hypoplastic right ventricle with hypertrophied wall and hypoplastic tricuspid valve in a patient with pulmonary atresia. Left atrium (LA), right atrium (RA), left ventricle (LV).*

Pulmonary Valve Atresia with Intact Ventricular Septum



Tricuspid Dysplasia



(Left) *Color Doppler echocardiogram shows flow in a coronary sinusoid with retrograde perfusion from the right ventricle (RV) into a main coronary artery to the aorta (Ao).* ***(Right)*** *Four chamber view echocardiogram shows a very dilated right atrium (RA) due to a dysplastic tricuspid valve. Note the failure of coaptation of the leaflets in systole. Right ventricle (RV).*

CHAMBER ASYMMETRY

Tricuspid Dysplasia



Ebstein Anomaly

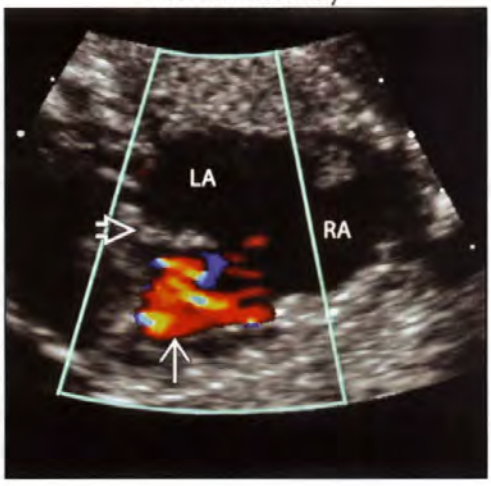


(Left) *Four chamber view color Doppler echocardiogram shows severe tricuspid regurgitation ➡ in a patient with tricuspid valve dysplasia on day of life 1. Right ventricle (RV).* ***(Right)*** *Radiograph shows the typical appearance of the chest X-ray in a child with Ebstein anomaly. This degree of cardiomegaly is called a "wall to wall" heart and is due to massive dilation of the right atrium.*

Ebstein Anomaly



Ebstein Anomaly

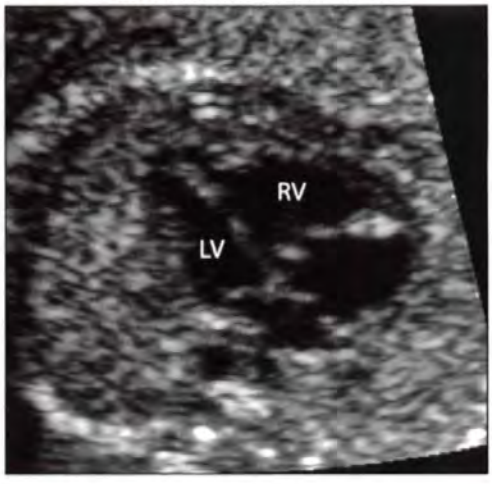


(Left) *Axial oblique ultrasound shows an Ebstein anomaly with downward displacement of the septal tricuspid leaflet ➡, resulting in "atrialization" of the right ventricle ➡ and a dilated right atrium (RA). Note the large atrial septal defect ➡. Left atrium (LA).* ***(Right)*** *Color Doppler echocardiogram in the same case shows tricuspid regurgitation ➡. This begins well below the level of the mitral valve ➡ due to the typical tricuspid valve position in Ebstein anomaly.*

Atrioventricular Septal Defect (AVSD), Unbalanced



Coarctation of the Aorta



(Left) *Four chamber view shows an unbalanced atrioventricular septal defect with the left ventricle ➡ larger than the right ➡. Note ventricular septal defect ➡ and lack of atrial septum ➡.* ***(Right)*** *Four chamber view echocardiogram shows the right ventricle (RV) to be much larger than the left (LV). Subsequent images revealed a small aorta and transverse arch hypoplasia. Coarctation of the aorta was confirmed at birth.*

CARDIOMEGALY

DIFFERENTIAL DIAGNOSIS

Common

- Cardiac
  - Ebstein Anomaly
  - Tricuspid Dysplasia
  - Dilated Cardiomyopathy
  - Unbalanced Atrioventricular Septal Defect
  - Tetralogy of Fallot with Absent Pulmonary Valve
- Non-Cardiac
  - Twin Related Heart Failure
    - Twin-Twin Transfusion Syndrome
    - Twin Reversed Arterial Perfusion
  - Vascular Shunting
    - Sacrococcygeal Teratoma
    - Chorioangioma
    - Vein of Galen Malformation

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Cardiomegaly defined as heart circumference > 50% of chest circumference or cardiac area > 33% of chest area
- Is it a singleton pregnancy or multiple gestation?
  - Twin-twin transfusion syndrome (TTTS) and twin reversed arterial perfusion (TRAP) sequence only occur in monochorionic twin pregnancies
    - TTTS: Two normal looking fetuses with asymmetric size, fluid distribution
    - TRAP: One normal, one very anomalous fetus
  - Twin demise in monochorionic pregnancy may result in ischemic cardiomyopathy in the survivor
    - Follow carefully after twin demise; may take 10-14 days to see full extent of damage
- Any other of the causes can occur in a singleton or in one of a dichorionic pair
- Is the fetus structurally normal?
  - Look for masses/arteriovenous malformation
- Is the intracardiac anatomy normal?
- Is the function normal?
- Are there one or two atrioventricular valves in the heart?
- Is there tricuspid regurgitation (TR)?
  - Are the leaflets normally located?
  - Are the leaflets thickened/dysplastic?

Helpful Clues for Common Diagnoses

- **Ebstein Anomaly**
  - Apical displacement of septal and posterior mural tricuspid valve leaflets with attachment to the ventricular septum
  - "Atrialization" of the right ventricle → right atrial enlargement
    - Anterior leaflet is often "sail-like"
    - TR may be severe
  - Pulmonary artery often small due to lack of antegrade flow in setting of severe TR
- **Tricuspid Dysplasia**
  - Thick, nodular, or irregular valve leaflets in normal position
  - Severe TR results in right atrial dilation
  - Often associated pulmonary stenosis/atresia
- **Dilated Cardiomyopathy**
  - No structural abnormality is present
  - Poor myocardial contractility
  - Myocardium often thin
  - Atrioventricular regurgitation
- **Unbalanced Atrioventricular Septal Defect**
  - Missing "crux" of heart in four chamber view
    - Inlet ventricular septal defect
    - Primum atrial septal defect
  - Single AV valve is committed more to one ventricle
  - Ventricle lacking commitment will be hypoplastic
  - Look for features of heterotaxy syndromes, especially anomalous venous drainage
- **Tetralogy of Fallot with Absent Pulmonary Valve**
  - Dilated aorta overriding a ventricular septal defect
  - Back and forth flow across pulmonary valve seen with color Doppler
  - Markedly enlarged main and branch pulmonary arteries
- **Twin-Twin Transfusion Syndrome**
  - Recipient twin is the one at risk for developing cardiomegaly
  - Monochorionic twins with asymmetric fluid distribution and growth
  - Donor shunts blood to recipient circulation → volume overload
    - Recipient: Polyhydramnios, larger size, ± abnormal Doppler, ± hydrops

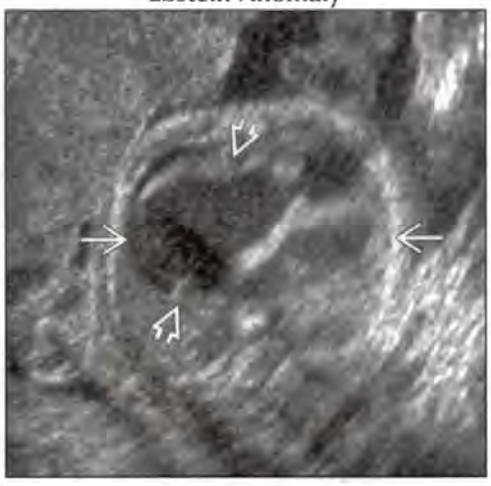
- Donor: Oligohydramnios, smaller size, ± absent bladder, ± abnormal Doppler
- Increased flow to the recipient twin causes high output failure
  - Biventricular hypertrophy
  - Tricuspid and mitral regurgitation
  - Abnormal ductus venosus flow

- **Twin Reversed Arterial Perfusion**
  - Pump twin is the one at risk for developing cardiomegaly
  - Monochorionic twins
    - One normal "pump twin"
    - One anomalous "acardiac" twin
  - Flow in "acardiac" umbilical artery is toward fetus
    - Often lacks cranium/upper extremities
    - May have rudimentary or absent heart
    - Grossly edematous
  - Normal twin develops cardiomegaly and failure from perfusing the abnormal twin, which can reach enormous size
- **Sacrococcygeal Teratoma**
  - Exophytic, mixed cystic/solid mass extending from sacrum
    - May contain calcifications
  - Polyhydramnios, placentomegaly
  - Solid tumors may have significant arteriovenous shunting
    - High output state → cardiomegaly → hydrops
    - Hydrops is very poor prognostic sign
- **Chorioangioma**
  - Well-defined hypoechoic placental mass, most often near cord insertion
  - Variable vascularity
    - Large, highly vascular ones more likely to cause cardiomegaly and hydrops
  - Fetal anemia may occur secondary to hemolysis
    - Monitor with middle cerebral artery peak systolic velocity
- **Vein of Galen Malformation**
  - Elongated midline cystic structure is dilated median prosencephalic vein
  - Doppler shows turbulent flow
  - May see enlarged carotid arteries in neck
  - High flow shunt → high output cardiac failure
  - Look for associated ischemic encephalomalacia and hydrocephalus

Other Essential Information

- Presence of hydrops confers poor prognosis in all cases
- Vein of Galen malformation is the most common extracardiac cause of high-output congestive heart failure in newborn period
- Twin-twin transfusion syndrome survival is improved with fetoscopic laser coagulation in severe cases
- Survival of pump twin in TRAP sequence is improved by radiofrequency ablation of anomalous twin's cord
- Fetal intervention considered for impending hydrops in sacrococcygeal teratoma

Ebstein Anomaly



Four chamber view shows massive cardiomegaly resulting in a "wall to wall" heart. This is mainly due to right atrial enlargement.

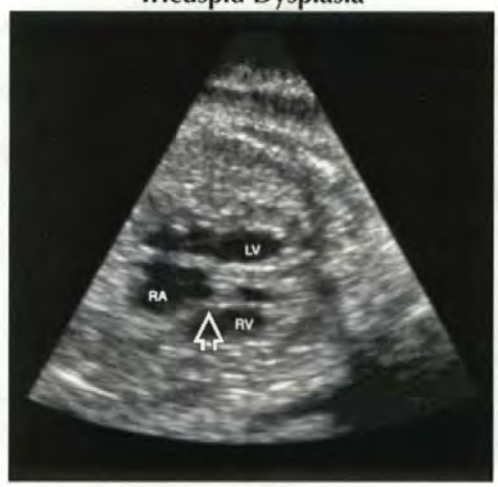
Ebstein Anomaly



Four chamber view echocardiogram shows significant downward displacement of the tricuspid septal leaflet. The coaptation point is very low in the ventricle, resulting in a small functional right ventricle.

CARDIOMEGALY

Tricuspid Dysplasia



(Left) *Four chamber view echocardiogram shows a thickened tricuspid valve that sits in the normal position. The right atrium (RA) is mildly enlarged. Right ventricle (RV), left ventricle (LV).* ***(Right)*** *Long axis echocardiogram shows a significantly dilated right ventricle (RV) with poor contractility in real time. The patient had positive genetic testing for Barth syndrome, an X-linked form of cardiomyopathy that causes a progressive dilated cardiomyopathy in infancy.*

Dilated Cardiomyopathy



Unbalanced Atrioventricular Septal Defect

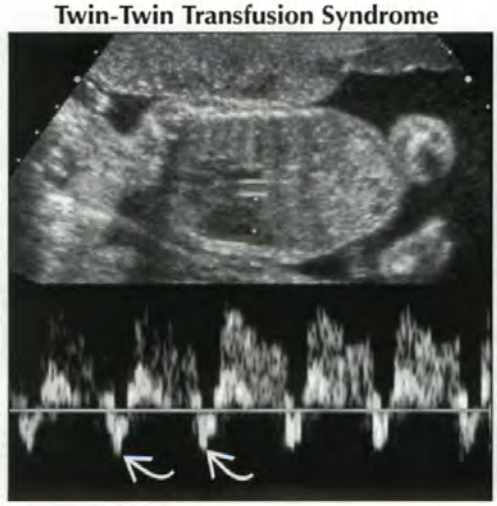


(Left) *Four chamber view shows a common atrioventricular valve opening preferentially to the right. The left ventricular (LV) is hypoplastic compared to the dilated right ventricle (RV). The interventricular septum is also visible.* ***(Right)*** *Echocardiogram shows marked dilation of the main and branch pulmonary arteries. A remnant of the pulmonary valve remains but does not prevent reversal of flow into the right ventricle (RV). Aorta (Ao).*

Tetralogy of Fallot with Absent Pulmonary Valve



Twin-Twin Transfusion Syndrome



(Left) *Pulsed Doppler ultrasound of the recipient in a case of TTTS. There is reversal of the ductus venosus A wave, indicating increased cardiac pressures and impending heart failure.* ***(Right)*** *Radiograph at birth shows massive cardiomegaly in the recipient twin in a case of twin-twin transfusion syndrome.*

Twin-Twin Transfusion Syndrome



Twin Reversed Arterial Perfusion

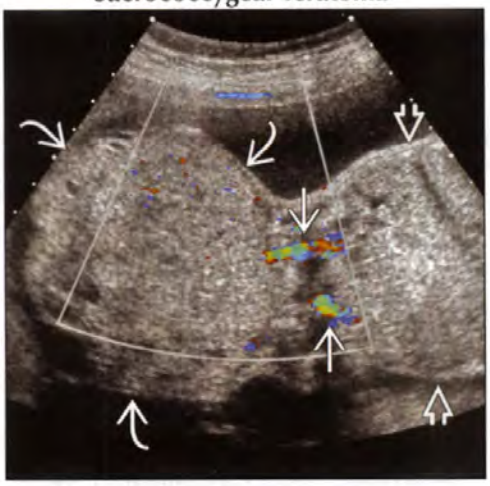


Twin Reversed Arterial Perfusion



(Left) *Four chamber view echocardiogram shows cardiomegaly in the pump twin of a pair with twin reversed arterial perfusion (TRAP) sequence. Following radiofrequency ablation of the TRAP twin's cord, the pump twin did well and was delivered at term with no apparent adverse consequences.* ***(Right)*** *Sagittal ultrasound shows the anomalous TRAP twin in the same case. Note the marked soft tissue edema, lack of cranial structures, and "empty" thoracic cavity.*

Sacrococcygeal Teratoma

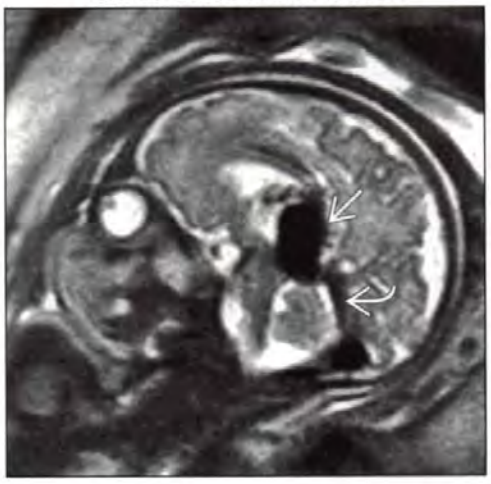


Sacrococcygeal Teratoma

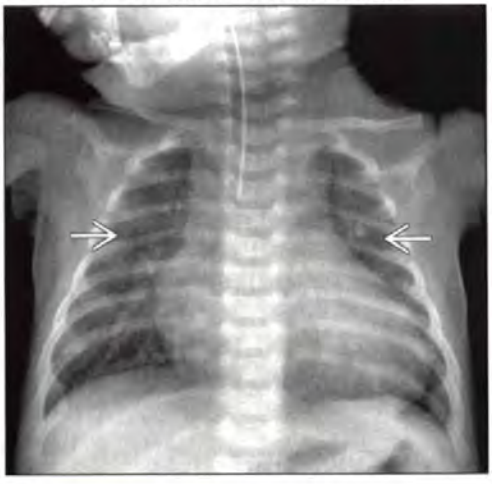


(Left) *Color Doppler ultrasound shows large vessels coursing from the abdomen into the solid sacrococcygeal teratoma, which is larger than the fetal body.* ***(Right)*** *Clinical photograph in the same case shows the large, bilobed mass, which is significantly larger than the infant's torso. The mass distorted the perineum but did not obstruct bowel or bladder. Perfusion of such a large hypervascular mass results in a high output state and may cause hydrops.*

Vein of Galen Malformation



Vein of Galen Malformation



(Left) *Sagittal T2WI MR shows a flow void in a dilated midline vascular structure, the typical appearance of a VGM. In this case, the malformation drains into the straight sinus.* ***(Right)*** *Radiograph performed shortly after birth demonstrates cardiomegaly and mild pulmonary edema. Although the patient's congestive heart failure was responding to medical therapy, progression of cerebral volume loss prompted transcatheter embolization of the fistula.*

SEPTAL DEFECT

DIFFERENTIAL DIAGNOSIS

Common

- Isolated Ventricular Septal Defect (VSD)
  - Muscular VSD
  - Perimembranous VSD
  - Inlet VSD
  - Outlet VSD
- VSD with Complex Congenital Heart Disease
- Atrioventricular Septal Defect
- Foramen Ovale Aneurysm

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Is the "crux" of the heart present?
  - If yes, atrioventricular septal defects (AVSD) are ruled out
  - If no, assess for balance of defect over ventricular chambers
    - Balanced AVSD: Both ventricles receive flow, symmetric in size
    - Unbalanced AVSD: AV valve committed to one ventricle, other ventricle becomes hypoplastic due to lack of blood flow
- Is the foramen ovale flap bulging into the left atrium normally?
- Are there additional holes in the atrial septum?
- Is there a hole in the ventricular septum and if so, where?
  - Muscular: In thick muscular part of septum, most often at the apex
  - Perimembranous: Located in LV outflow tract beneath the aortic valve, may extend into adjacent portions of muscular septum
  - Inlet: At plane of AV valves where atrial blood enters ventricles (i.e., at ventricular inlet), posterior and inferior to membranous septum
  - Outlet: In RV infundibulum beneath pulmonary valve (i.e., at ventricular outlet)
- Are there other cardiac lesions?
  - VSD is a very common component of complex congenital heart disease
    - Flow to a great vessel is often dependent on presence of VSD
  - Look carefully at outflow tracts
    - Normally cross as they exit heart
    - Normally symmetric in size
    - Aorta normally posterior to pulmonary artery (PA)
    - Aorta should arise from morphologic left ventricle (LV)
    - PA should arise from morphologic right ventricle (RV)
- Watch for potential "drop out" in septum due to the angle of insonation
  - From the apex, muscular to membranous transition looks like a VSD
  - Look in multiple different views and angles to confirm presence of a VSD
  - Color Doppler may be useful to confirm flow across suspected VSD

Helpful Clues for Common Diagnoses

- **Isolated Ventricular Septal Defect (VSD)**
  - **Muscular VSD**
    - Best seen on four chamber view
    - Small defects very difficult to see by 2-D
    - Larger defects easier to see with beam perpendicular to septum
    - In absence of outflow obstruction, no pressure gradient between RV and LV; therefore little flow and color Doppler often not helpful
  - **Perimembranous VSD**
    - Best viewed from the long axis or left ventricular outflow tract (LVOT) view with visualization of aortic valve
    - Common in association with other heart diseases, especially tetralogy of Fallot
    - Aneurysmal tissue from the tricuspid valve will partially or completely close this defect in many cases
  - **Inlet VSD**
    - Best viewed from the four chamber view with beam angled toward posterior diaphragm
    - Common in association with partial or complete AVSD
    - Gives impression that AV valves have nothing to attach to
  - **Outlet VSD**
    - Viewed best from the short axis or right ventricular outflow tract (RVOT) view
    - Defect is located so as to direct blood below pulmonary valve
    - Almost always found in association with other heart disease
- **VSD with Complex Congenital Heart Disease**
  - Tetralogy of Fallot

- Aorta overrides interventricular septum, straddling VSD
- With severe pulmonary stenosis or atresia, flow is directed toward aorta

- Truncus arteriosus
  - Single truncal valve straddles septum
  - Blood from both ventricles exits heart via truncus which gives rise to aorta and PA
- Double outlet right ventricle
  - Both great arteries arise over RV necessitating a VSD to provide blood flow to both body and lungs
- Interrupted aortic arch
  - VSD is necessary to carry oxygenated blood to the body via ductus arteriosus

- **Atrioventricular Septal Defect**
  - May be balanced or unbalanced over the ventricular septum
  - "Crux" of the heart is missing; only one AV valve present, not two
  - Primum atrial septal defect
    - Size is variable, but may appear to be a common atrium when large
    - If small, focus on valve being in the same plane to diagnose
  - Inlet ventricular septal defect
    - Size is variable, septum may actually appear to be absent in some cases
    - Focus again on valve being in same plane and presence of a primum atrial defect
  - AV valve regurgitation is very common
  - When AVSD is unbalanced, one ventricular chamber will be small
    - When severe, this may be confused with hypoplastic left heart or tricuspid atresia
- **Foramen Ovale Aneurysm**
  - Redundant tissue in foramen ovale flap → "balloon" appearance
    - Flap extends at least half way across left atrium
    - Flap excursion > 5 mm beyond plane of atrial septum
  - Flap demonstrates abnormal mobility
    - May make cyclical contact with left atrial wall and/or mitral valve
  - 5% of fetuses may have benign rhythm abnormalities

Other Essential Information

- Muscular and perimembranous VSDs in isolation have a high likelihood of closure
- Inlet and outlet VSDs tend to require surgical closure
- VSDs in association with other congenital heart disease always require closure if a two ventricle circulation is the goal
  - Patients who require palliation for a single ventricle will have their VSDs left alone and occasionally enlarged to assure adequate flow to the head and body
- Identification of a secundum atrial defect is very difficult, as location is the same as foramen ovale
- Isolated foramen ovale aneurysm is a benign entity

Muscular VSD



Four chamber view echocardiogram shows a small mid-septal muscular defect ➡. Ventricular sizes are normal, although the right atrium ➡ is increased in size. Left ventricle (LV), right ventricle (RV).

Muscular VSD



Four chamber view color Doppler echocardiogram shows the same small muscular ventricular septal defect ➡. The right atrium (RA) is enlarged. Right ventricle (RV).

SEPTAL DEFECT

Muscular VSD



Muscular VSD

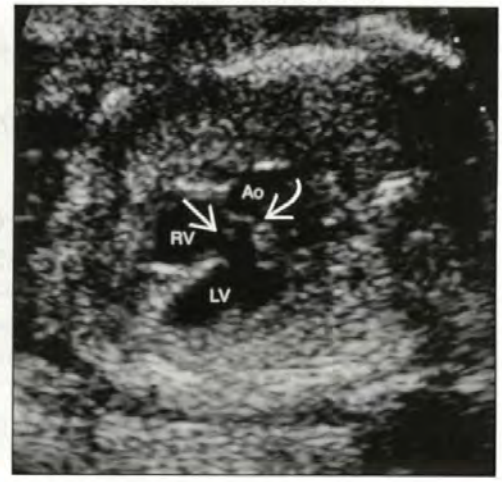


(Left) *LVOT echocardiogram shows an anterior muscular ventricular septal defect between the right and left (LV) ventricles. The ascending aorta and aortic valve are well seen in this LVOT projection. Left atrium (LA).* ***(Right)*** *LVOT color Doppler echocardiogram shows the same anterior muscular ventricular septal defect with flow into the right ventricle. Note laminar flow out the aortic valve. Left ventricle (LV).*

Muscular VSD



Perimembranous VSD

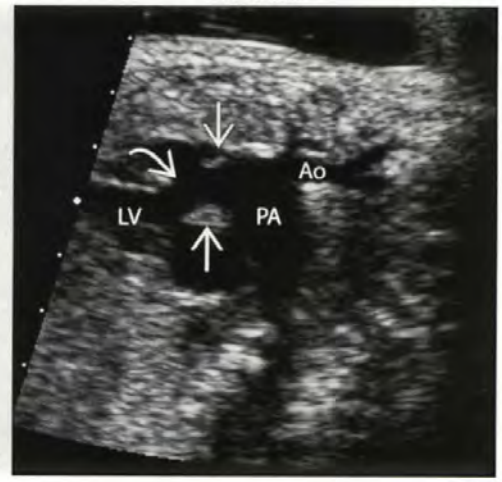


(Left) *Four chamber view color Doppler echocardiogram shows a muscular ventricular septal defect with flow between the right (RV) and left ventricles. Right atrium (RA).* ***(Right)*** *LVOT echocardiogram is angled to give the best view of a perimembranous ventricular septal defect beneath the aortic valve. Aorta (Ao), left ventricle (LV), right ventricle (RV).*

Inlet VSD



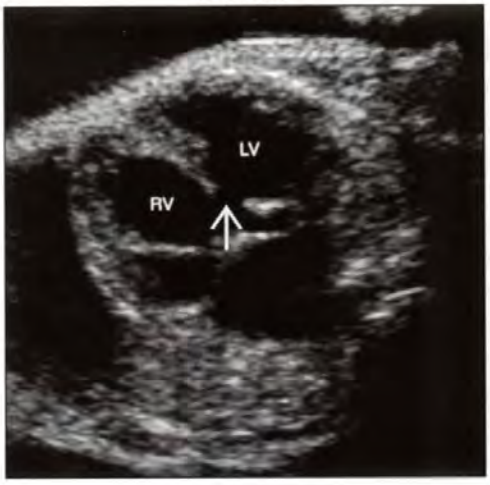
VSD with Complex Congenital Heart Disease



(Left) *Four chamber view echocardiogram with angulation of the beam toward the posterior diaphragm shows an inlet ventricular septal defect, which is identified beneath the septal leaflet of the tricuspid valve.* ***(Right)*** *Long axis echocardiogram (LVOT view) shows a large truncal valve overriding the ventricular septum with VSD. The truncus gives rise to an interrupted aortic arch (Ao) and a pulmonary artery (PA). Left ventricle (LV).*

SEPTAL DEFECT

VSD with Complex Congenital Heart Disease



VSD with Complex Congenital Heart Disease

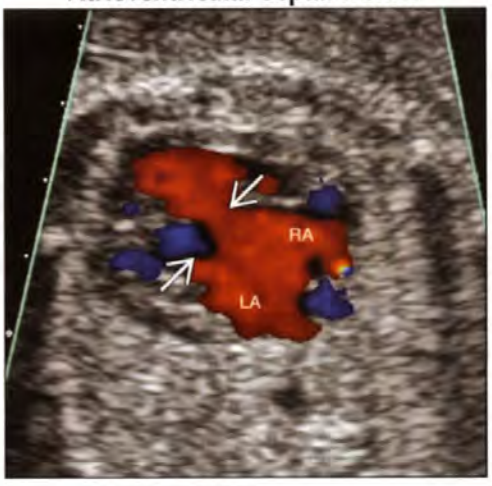


(Left) *Four chamber view echocardiogram shows a large, muscular ventricular septal defect ➡, and the right ventricle (RV) is dilated. Additional imaging showed this patient had tetralogy of Fallot with absent pulmonary valve.* ***(Right)*** *LVOT echocardiogram shows the aorta ➡ overriding the interventricular septum ➡. Additional imaging showed this patient had findings consistent with tetralogy of Fallot. Left ventricle (LV), right ventricle (RV).*

Atrioventricular Septal Defect

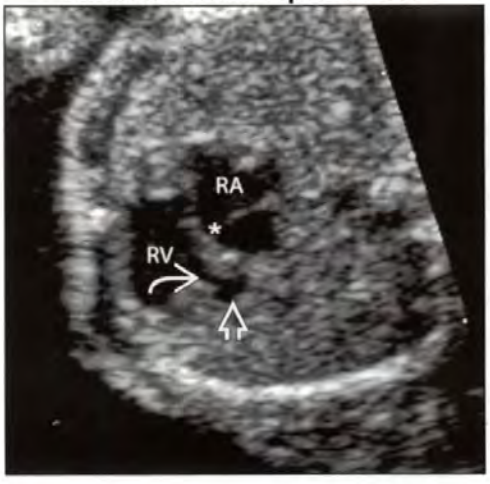


Atrioventricular Septal Defect



(Left) *Four chamber view echocardiogram shows a classic balanced atrioventricular septal defect with the "crux" of the heart ➡ missing in diastole. This indicates both a primum atrial defect and an inlet ventricular septal defect. Left ventricle (LV), right ventricle (RV), left atrium (LA), right atrium (RA).* ***(Right)*** *Four chamber view color Doppler echocardiogram shows the same image, with flow from each atrium (RA, LA) entering each ventricle without obstruction ➡.*

Atrioventricular Septal Defect



Foramen Ovale Aneurysm



(Left) *Four chamber view echocardiogram shows a right dominant atrioventricular septal defect. There is a primum atrial septal defect (*), an inlet ventricular septal defect ➡, and a hypoplastic left ventricle ➡. Right ventricle (RV), right atrium (RA).* ***(Right)*** *Four chamber view echocardiogram shows a foramen ovale aneurysm ➡ bowing well into the left atrium from the right atrium (RA).*

ABNORMAL OUTFLOW TRACTS

DIFFERENTIAL DIAGNOSIS

Common

- Hypoplastic Left Heart Syndrome (HLHS)
- Tricuspid Atresia (TA)
- Tetralogy of Fallot (TOF)
- Pulmonary Valve Stenosis, Atresia

Less Common

- Transposition of Great Arteries (TGA)
- Double Outlet Right Ventricle (DORV)
- Truncus Arteriosus

Rare but Important

- Interrupted Aortic Arch (IAA)

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Are there one or two ventricles?
  - If a ventricle is hypoplastic, the corresponding great vessel is typically small or atretic
- Is a ventricular septal defect (VSD) present?
  - Does the aorta override the interventricular septum?
- Do the great vessels exit the heart side by side?
  - Normal relationship is for the great vessels to cross as they exit the heart
- Are the aorta and pulmonary artery (PA) similar in size (i.e., normal), or is there a discrepancy?
- Are ductal and aortic arches present?
  - Ductal arch is often larger than aortic arch
  - Normal flow in the ductus arteriosus is right to left
    - Reversed in pulmonary atresia as PA fills retrograde from aorta
  - Retrograde flow in the aortic arch suggests left-sided obstruction
    - Aortic arch fills retrograde from PA via ductus

Helpful Clues for Common Diagnoses

- **Hypoplastic Left Heart Syndrome (HLHS)**
  - Abnormal four chamber view with small, non apex-forming left ventricle (LV)
    - May see brightly echogenic LV endocardium with endocardial fibroelastosis
    - Ventricular function is poor
    - Interatrial septum bowed left to right
  - Aortic valve often is atretic or very small
  - Ascending aorta and transverse arch are very small
  - Right ventricle (RV) is large and wraps around the LV apex
    - Function is typically very good
- **Tricuspid Atresia (TA)**
  - Abnormal four chamber view with small, non-apex-forming right ventricle
    - Tricuspid valve appears "plate-like" with no movement
    - VSD usually present → blood flow to great artery arising from RV
  - Size of great artery arising from RV (PA or aorta) depends on size of VSD
    - Larger VSD → bigger artery
  - Left ventricle is normal to large in size with good function
- **Tetralogy of Fallot (TOF)**
  - Dilated aortic root overriding a VSD
  - Pulmonary stenosis almost always present
    - Commonly due to anterior deviation of the infundibular septum
  - Both ventricles normal in size
    - RV may be hypertrophied if significant pulmonary stenosis
- **Pulmonary Valve Stenosis, Atresia**
  - Pulmonary valve stenosis typically → normal sized RV and small pulmonary valve
    - Turbulent, high velocity flow is noted across the valve
    - Often accompanied by tricuspid regurgitation
  - Pulmonary atresia with intact ventricular septum → hypoplastic right ventricle
    - Reverse flow in the ductus arteriosus
    - Ductus more vertically oriented than usual
    - Abnormal coronary flow suggests coronary sinusoids
    - Right ventricular pressure is greater than systemic
  - Pulmonary atresia with VSD
    - VSD is large
    - Aorta may override septum
    - Pulmonary arteries may be absent or very small
    - Collaterals off the descending aorta may be the only blood supply to the lungs

Helpful Clues for Less Common Diagnoses

- **Transposition of Great Arteries (TGA)**
  - Outflow tracts parallel as they exit heart
  - Aorta arises anteriorly from RV
  - Pulmonary artery arises posteriorly from LV
  - VSD common
  - Both ventricles are of normal size with good function
- **Double Outlet Right Ventricle (DORV)**
  - Prominent, dilated aortic root overriding a VSD
    - > 50% of aortic root arises from RV
  - Outflow tracts parallel as they exit the heart
    - Usually normally related with aorta posterior to PA
  - Ventricles are normal in size unless associated atrioventricular (AV) valve atresia
- **Truncus Arteriosus**
  - Single great artery (truncus) exits the heart
    - Branches into an aorta and pulmonary trunk
  - VSD almost always present
  - Truncal valve often abnormal with stenosis or insufficiency
    - Count valve leaflets of great artery, if > 3 ⇒ truncal valve
  - Both ventricles are normal in size with good function
  - Arch gives rise to one or more vessels which extend straight into neck
    - Normal "candy cane" arch view is not attainable
    - Blood supply is only to head → aorta is small in size
    - Ductus arteriosus supplies all blood to lower body
  - VSD usually present with posterior deviation of septum

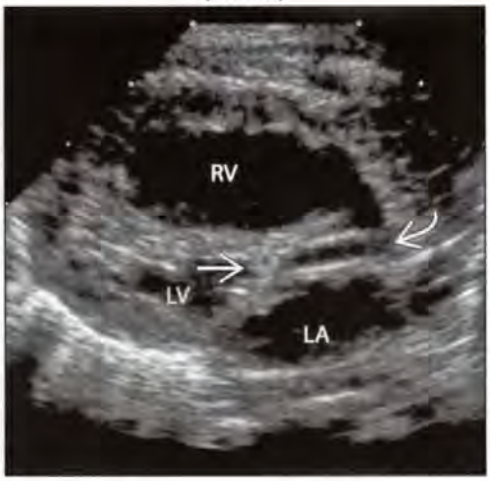
Other Essential Information

- Single ventricles (HLHS and truncus arteriosus) cannot be corrected, only palliated
  - Each require 3 surgeries, and life expectancy is shortened
  - Presence of AV valve regurgitation and an intact atrial septum increases surgical risk
- TGA, DORV, and TOF can be surgically corrected
  - Repair is within the first weeks to few months of life
  - Normal life expectancy in most cases
- IAA and pulmonary atresia are ductal dependent lesions
  - Newborn requires surgical or catheter intervention
  - IAA, once repaired, has a normal life expectancy without additional intervention

Helpful Clues for Rare Diagnoses

- **Interrupted Aortic Arch (IAA)**

Hypoplastic Left Heart Syndrome (HLHS)



Long axis echocardiogram day 1 of life shows a hypoplastic left ventricle (LV) with a thick, atretic aortic valve ➡ and small, hypoplastic ascending aorta ➡. Left atrium (LA), right ventricle (RV).

Tricuspid Atresia (TA)



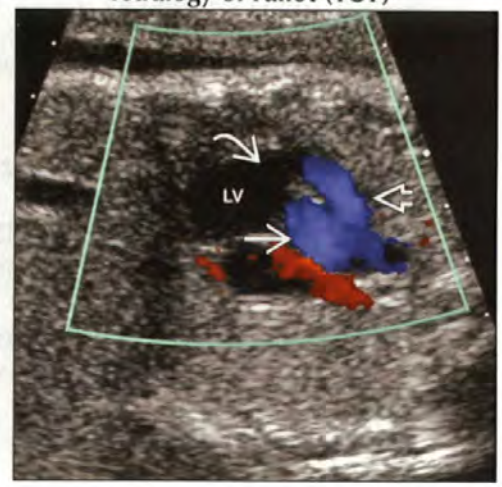
RVOT view day 1 of life shows a larger aorta (Ao) than RVOT. With tricuspid atresia, the only RVOT flow in utero was via a VSD. Note the thickened pulmonary valve ➡ and small branch pulmonary arteries (PAs).

ABNORMAL OUTFLOW TRACTS

Tetralogy of Fallot (TOF)



Tetralogy of Fallot (TOF)

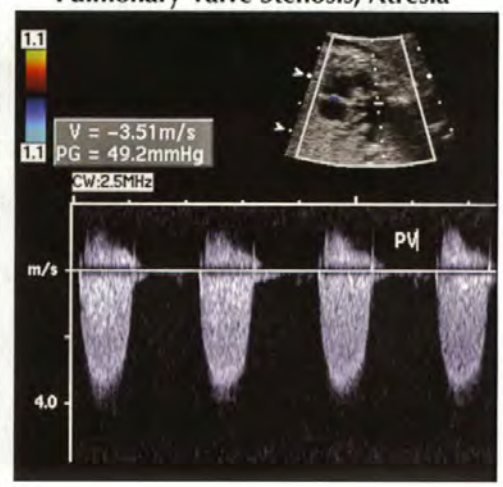


(Left) *Long axis echocardiogram shows anterior deviation of the infundibulum, causing a small PA despite the presence of a ventricular septal defect. The aorta comes off the left ventricle (LV) but there was override consistent with tetralogy of Fallot.* ***(Right)*** *Long axis color Doppler echocardiogram in the same case shows laminar flow out the aorta and pulmonary artery. A large VSD is easily seen. Left ventricle (LV).*

Pulmonary Valve Stenosis, Atresia



Pulmonary Valve Stenosis, Atresia



(Left) *RVOT echocardiogram shows a thick and dysplastic pulmonary valve with a dilated main pulmonary artery (MPA). Right ventricle (RV).* ***(Right)*** *RVOT pulsed Doppler echocardiogram shows a gradient across the pulmonary valve of 49 mmHg. This indicates at least moderate pulmonary stenosis.*

Pulmonary Valve Stenosis, Atresia



Pulmonary Valve Stenosis, Atresia



(Left) *RVOT echocardiogram shows the aorta arising from the anterior right ventricle and the pulmonary artery arising posterior to the aorta (Ao). The pulmonary valve appears atretic.* ***(Right)*** *RVOT color Doppler echocardiogram shows turbulent flow starting at the pulmonary valve and entering the main pulmonary artery, which is dilated.*

ABNORMAL OUTFLOW TRACTS

Transposition of Great Arteries (TGA)

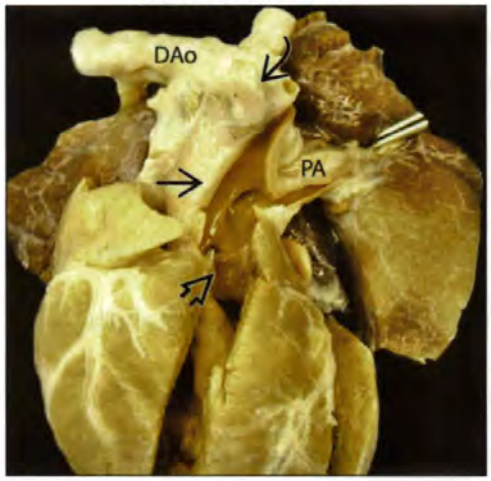


Double Outlet Right Ventricle (DORV)



(Left) *Transverse echocardiogram shows the aorta coming off the anterior right ventricle (RV) and the pulmonary artery ➡ arising from the posterior left ventricle (LV), consistent with transposition.* ***(Right)*** *Four chamber view echocardiogram shows both great arteries ➡, main pulmonary artery (MPA) and ascending aorta (AAo), coming off the right ventricle. There is also a large ventricular septal defect ➡. Left ventricle (LV), right ventricle (RV).*

Truncus Arteriosus



Truncus Arteriosus



(Left) *Gross pathology shows a large truncal valve ➡ and root giving rise to a pulmonary artery (PA) and aorta ➡ with head and neck vessels ➡. Descending aorta (DAo).* ***(Right)*** *Sagittal oblique echocardiogram shows a large truncal valve ➡ over a ventricular septal defect ➡. The truncus continues to the descending aorta (DAo).*

Truncus Arteriosus



Interrupted Aortic Arch (IAA)



(Left) *Echocardiogram of the outflow tracts shows the single truncus ➡, giving rise to a pulmonary artery ➡ and aorta ➡.* ***(Right)*** *Sagittal oblique echocardiogram shows the ascending aorta (AAo) heading straight to the head ➡. Note the distance between the ascending aorta and the ductus arteriosus ➡ and descending aorta (DAo). The subclavian artery ➡ arises from the descending aorta, making this a type B interruption of the aortic arch.*

ABNORMAL RATE OR RHYTHM

DIFFERENTIAL DIAGNOSIS

Common
- Premature Atrial Contraction
- Transient Bradycardia

Less Common
- Complete Heart Block
- Supraventricular Tachycardia
- Atrial Flutter

ESSENTIAL INFORMATION

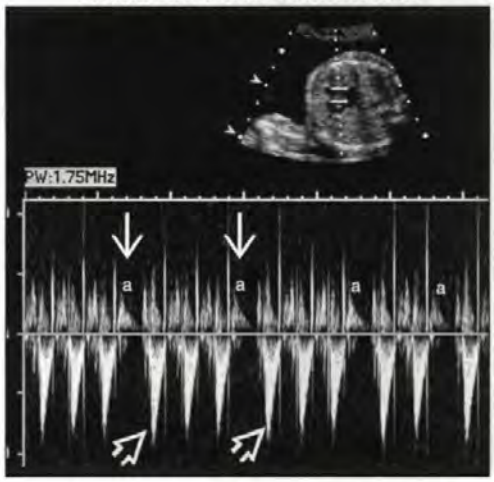
Key Differential Diagnosis Issues
- Normal heart rate: 100-180 beats per minute (bpm)
  - Irregular rhythm in this range most often benign premature atrial contractions (PACs)
  - Bradyarrhythmia is HR < 100 bpm
  - Tachyarrhythmia is HR > 180 bpm
    - Supraventricular tachycardia (SVT): HR 220-280 bpm
    - Atrial flutter: HR 300-600 bpm

Helpful Clues for Common Diagnoses
- **Premature Atrial Contraction**
  - Atrial contraction not conducted to ventricle,
    - Heart rate normal unless blocked PACs → ↓ ventricular rate
    - < 5% may develop SVT
- **Transient Bradycardia**
  - Anatomically normal heart, due to excessive transducer pressure

Helpful Clues for Less Common Diagnoses
- **Complete Heart Block**
  - Bradyarrhythmia with slow independent ventricular rate
  - Look for heterotaxy: Left atrial isomerism → no sinoatrial node
  - Look for ventricular inversion (i.e., corrected transposition of great arteries)
  - Check maternal history for connective tissue disease, Ro/La antibody status
    - Antibodies cross placenta → immune complex-mediated damage to conduction system
- **Supraventricular Tachycardia**
  - 1:1 AV relationship, intermittent or incessant, starts/stops in 1 beat
- **Atrial Flutter**
  - Atrial rate > ventricular rate, incessant with no variation in atrial rate
  - Variable degrees of AV block: Most common 2:1

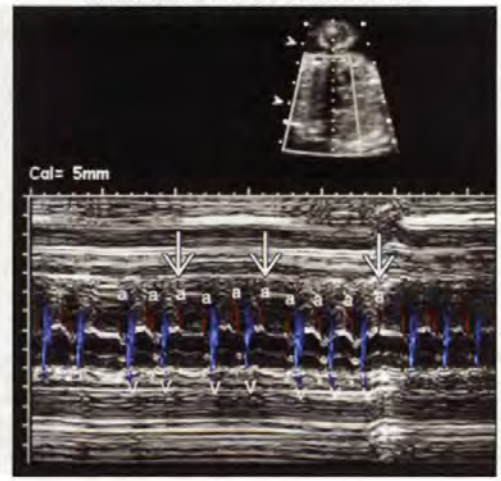
Other Essential Information
- PACs account for 85% of cases: Typically resolve without treatment
- Bradycardia is difficult to treat, often requires pacing after birth
  - Poor prognosis with congenital heart disease
  - Complete heart block associated with in-utero fetal demise
- Transplacental drug therapy highly successful for tachycardia
  - Mortality increased if hydrops develops

Premature Atrial Contraction



Pulsed Doppler echocardiogram shows normal sinus rhythm with occasional blocked premature atrial contractions ➡. The "a" is not followed by ventricular contraction ➡.

Premature Atrial Contraction

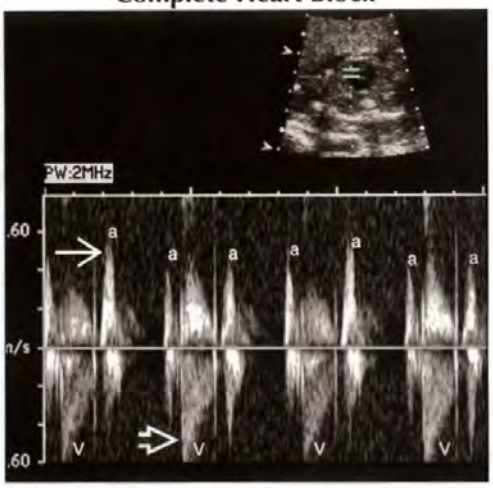


M-mode echocardiogram shows normal sinus rhythm with occasional blocked premature atrial contractions ➡. The "a" is not followed by ventricular contraction (v).

Complete Heart Block

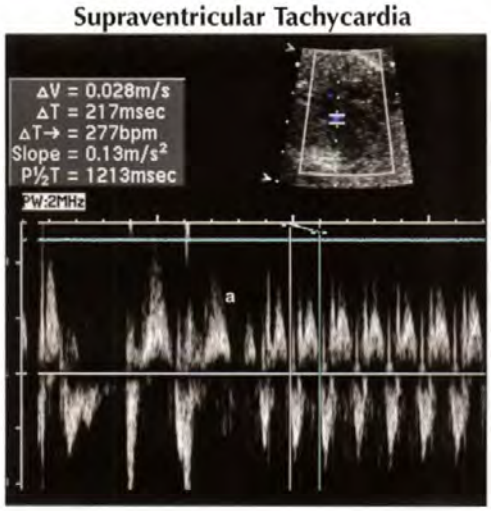


Complete Heart Block

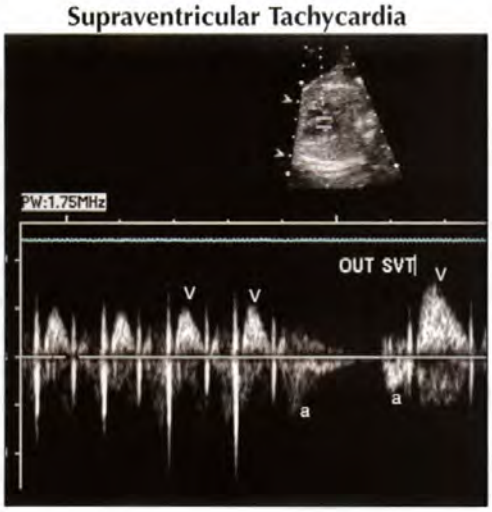


(Left) *Pulsed Doppler echocardiogram shows atrial contractions ➡ coming at regular intervals but not correlating to the ventricular contractions ➡. Some atrial contractions are not seen because they are occurring at the same time as the ventricular contractions.* ***(Right)*** *Pulsed Doppler echocardiogram shows mitral inflow ➡ and aortic outflow ➡. There are two atrial "a" beats for every ventricular "v" beat, demonstrating second degree heart block.*

Supraventricular Tachycardia

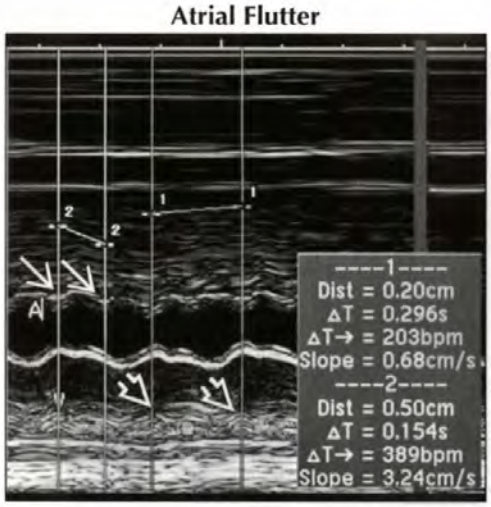


Supraventricular Tachycardia

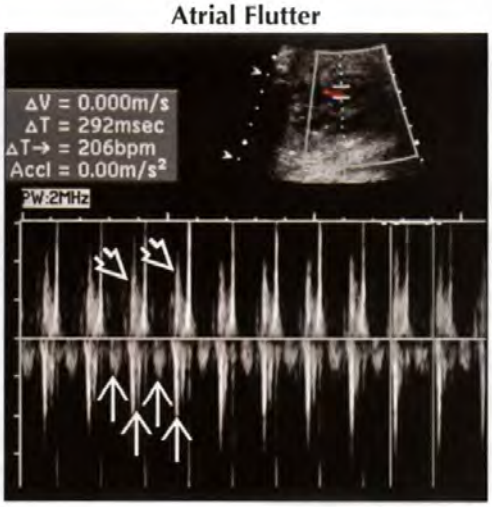


(Left) *Pulsed Doppler echocardiogram shows the onset of tachycardia at a rate of 277 bpm was caused by an atrial contraction "a".* ***(Right)*** *Pulsed Doppler echocardiogram shows termination of the tachycardia with an atrial contraction "a" and the start of normal sinus rhythm (an "a" followed by a "v").*

Atrial Flutter



Atrial Flutter



(Left) *M-mode echocardiogram shows an atrial rate ➡ of approximately 400 bpm (caliper set 2) with a ventricular rate ➡ of approximately 200 bpm (caliper set 1) indicating atrial flutter with 2:1 AV block.* ***(Right)*** *Pulsed Doppler echocardiogram shows rapid atrial contraction ➡ at 412 bpm with every other atrial beat being hidden. Ventricular rate ➡ is 206 bpm.*

CARDIAC MASS

DIFFERENTIAL DIAGNOSIS

Common
- Echogenic Cardiac Focus
- Moderator Band
- Papillary Muscle

Less Common
- Rhabdomyoma
- Hypertrophic Muscle

Rare but Important
- Teratoma
- Fibroma

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is the mass echogenic?
- Is it associated with the papillary muscle?
- Single or multiple masses?
- Where are they located?
  - Ventricle, septum, or on pericardium

Helpful Clues for Common Diagnoses
- **Echogenic Cardiac Focus**
  - Normal intracardiac anatomy
  - Focus is bright as bone
  - Small, associated with papillary muscle
  - Usually incidental finding but is a soft marker for trisomy 21
- **Moderator Band**
  - Distinguishing feature that identifies right ventricle
  - Normal muscle band from free wall to septum at 45° angle near apex
- **Papillary Muscle**
  - Normal feature of right and left ventricles
  - Left ventricular papillary muscles do not connect to septum: Defining feature of a morphologic left ventricle

Helpful Clues for Less Common Diagnoses
- **Rhabdomyoma**
  - Multiple, ventricular echogenic masses
  - Varying sizes
  - Typically resolve with time
  - Associated with tuberous sclerosis, look carefully at brain
- **Hypertrophic Muscle**
  - Often with asymmetric septal hypertrophy, not echogenic
  - Hypertrophy is uniform and concentric

Helpful Clues for Rare Diagnoses
- **Teratoma**
  - May be intra- or extrapericardial
  - Pericardial effusion common
- **Fibroma**
  - Typically a single tumor involving left ventricular free wall or septum
  - May be large causing outflow obstruction

Other Essential Information
- Echogenic cardiac focus is most commonly benign and not associated with cardiac pathology
  - If multiple may be associated with aneuploidy

Echogenic Cardiac Focus



Axial echocardiogram shows typical example of an ECF ➡ seen in a fetus with trisomy 21. This is usually an incidental finding, but if you see an ECF look for other findings associated with Down syndrome.

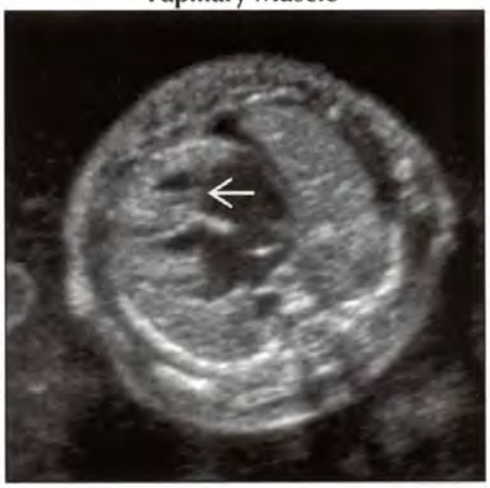
Moderator Band



LVOT echocardiogram shows the moderator band ➡ in the right ventricle (RV) as it attaches to the septum at nearly a 45 degree angle. Left ventricle (LV), aorta (Ao).

CARDIAC MASS

Papillary Muscle

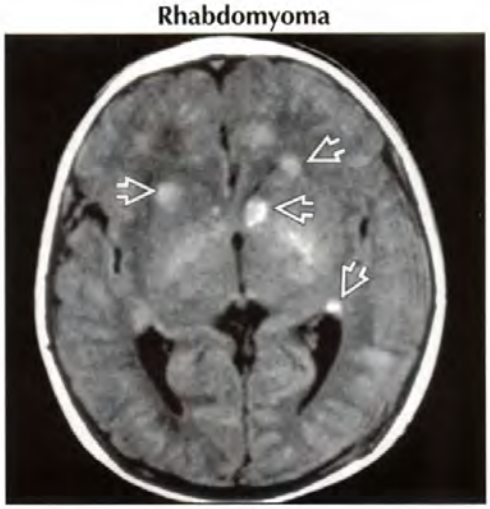


Rhabdomyoma



(Left) *Four chamber view of the heart shows a papillary muscle ➡ in the surviving twin of a monochorionic pair, complicated by twin demise. The heart dilated progressively and at autopsy ischemic brain and myocardial injury were confirmed.* ***(Right)*** *Four chamber view echocardiogram shows a massive rhabdomyoma ➡ involving the ventricular septum. Right atrium (RA), right ventricle (RV), left ventricle (LV).*

Rhabdomyoma

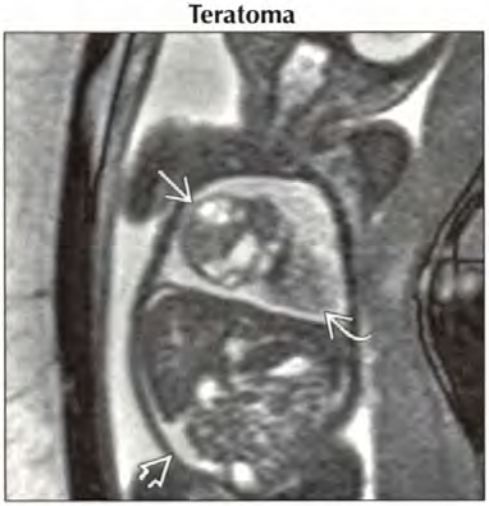


Hypertrophic Muscle

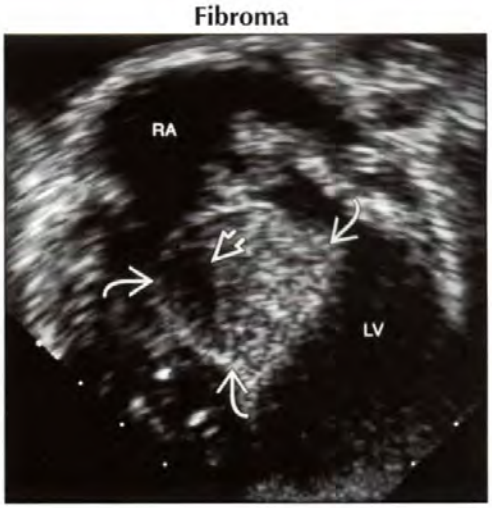


(Left) *Axial T1WI MR shows the typical appearance of subependymal nodules ➡ associated with tuberous sclerosis. Cardiac rhabdomyomas are the most common prenatal finding in tuberous sclerosis.* ***(Right)*** *Four chamber view echocardiogram shows a narrowed left ventricular outflow tract ➡ and the presence of a pericardial effusion ➡. The muscle is concentrically hypertrophied ➡. Left ventricle (LV), right ventricle (RV).*

Teratoma



Fibroma



(Left) *Coronal T2WI MR shows a large intrapericardial teratoma ➡ adjacent to the heart ➡. It is surrounded by a massive, high-signal pericardial effusion. No normal lungs are visualized. Ascites ➡ is also present.* ***(Right)*** *Four chamber view echocardiogram in a neonate shows a large cardiac fibroma ➡, which is virtually indistinguishable from the septum. There is an inhomogeneous area ➡ within the fibroma, which aids in the diagnosis.*

SECTION 6
Abdomen

Abdominal Wall

Abdominal Wall Defect 6-2

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ABDOMINAL WALL DEFECT

DIFFERENTIAL DIAGNOSIS

Common
- Gastroschisis
- Omphalocele
- Physiologic Gut Herniation (Mimic)

Less Common
- Amniotic Band Syndrome
- Body Stalk Anomaly

Rare but Important
- Bladder Exstrophy
- Cloacal Exstrophy
- Pentalogy of Cantrell
- OEIS Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Must document normal abdominal wall surrounding cord insertion in every case
- Umbilical cord insertion site key for making diagnosis
  - Gastroschisis: Defect to right of normal cord insertion
  - Omphalocele: Cord inserts on membrane surrounding defect
  - Bladder and cloacal exstrophy: Low defect below cord insertion
    - Cord insertion may be involved with cloacal extrophy
  - Pentalogy of Cantrell: High defect above and potentially involving the cord insertion
  - Amniotic band syndrome: No defined anatomic distribution

Helpful Clues for Common Diagnoses
- **Gastroschisis**
  - Bowel herniation through a right paramedian abdominal wall defect
  - Color Doppler shows umbilical cord insertion in normal location
  - No covering membrane
  - Small bowel always herniates through defect
    - Large bowel and stomach also reported
    - If liver or other solid organs are seen, gastroschisis unlikely (consider ruptured omphalocele, amniotics bands, body stalk, pentalogy of Cantrell)
  - Variable appearance of bowel
    - Both intra- and extra-abdominal loops may be dilated
    - Bowel wall may become thickened, echogenic, matted, and nodular from fibrinous peel on exposed bowel
- **Omphalocele**
  - Midline abdominal wall defect with herniation of abdominal contents into base of umbilical cord
  - Smooth mass protruding from central anterior abdominal wall with covering membrane
  - Umbilical cord inserts onto membrane
    - Usually centrally but may be eccentric
  - Liver and small bowel most common contents
  - Associated structural abnormalities are common
    - Omphaloceles containing small bowel have higher association of both chromosomal and structural malformations
    - Cardiac and gastrointestinal malformations are most common
    - Aneuploidy in 30-40%: Trisomy 18 most common
    - Also associated with syndromes including Beckwith-Wiedemann (omphalocele, organomegaly, macroglossia)
- **Physiologic Gut Herniation (Mimic)**
  - Be careful diagnosing an abdominal wall defect before 12 weeks
    - Bowel does not return to abdomen until 11.2 weeks
  - Normal bowel herniation should not extend more than 1 cm into cord
  - Never contains liver

Helpful Clues for Less Common Diagnoses
- **Amniotic Band Syndrome**
  - "Slash" defects that do not conform to developmental malformations
    - Asymmetric distribution of defects is hallmark of syndrome
  - Often involves multiple parts of the body
    - Defects may be isolated or multiple, but not in specific pattern
  - Craniofacial deformities are common and often severe
  - Bands in amniotic fluid appear as multiple thin membranes

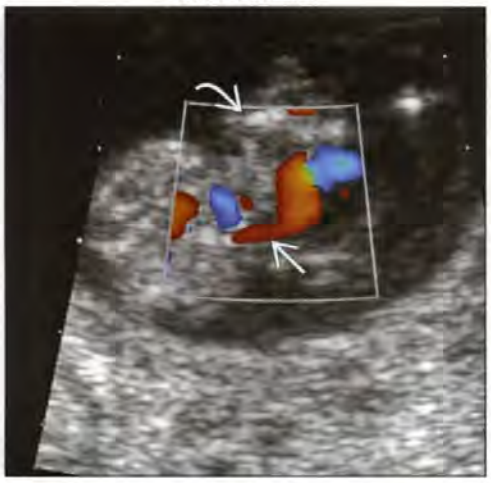
- Bands are often tightly adherent to fetus and may not be visible

- **Body Stalk Anomaly**
  - Lethal malformation characterized by attachment of visceral organs to the placenta
    - Short or absent umbilical cord
    - Vessels seen running from placental surface to fetal torso
    - No free floating cord identified
  - Gross distortion, with loss of anatomic landmarks
  - Scoliosis prominent feature
    - May have multiple acute angulation points

Helpful Clues for Rare Diagnoses

- **Bladder Exstrophy**
  - Failure of lower abdominal wall closure resulting in exposed bladder
  - Variable severity
    - Mild form associated with exstrophy of urethra and external sphincter
    - Severe form associated with wide diastasis of symphysis pubis and genital defects
  - Absence of bladder on prenatal ultrasound
  - Soft tissue mass on lower anterior abdominal wall, below cord insertion
    - Mass is posterior bladder wall
    - No extruded abdominal contents as in other abdominal wall defects
  - Beware of misdiagnosis in cases of a normal but empty bladder
    - Rescan after an interval of 10-15 minutes
- **Cloacal Exstrophy**
  - Spectrum of abnormalities resulting from abnormal development of cloacal membrane
  - Absence of normal bladder
  - Lower abdominal wall defect
    - Herniation of bowel between 2 halves of a split bladder
    - Appearance of prolapsed ileum described as looking like an elephant's trunk
    - Omphalocele forms upper part of defect
    - Males may have bifid scrotum and penis
  - Associated anomalies very common
    - Vertebral, myelomeningocele, urinary tract, gastrointestinal, clubfeet
- **Pentalogy of Cantrell**
  - Complex malformation with 5 components
    - Anterior diaphragmatic hernia
    - Midline abdominal wall defect
    - Cardiac anomalies
    - Defect of diaphragmatic pericardium
    - Lower sternal defect
  - Most likely diagnosis when there is a high omphalocele associated with ectopia cordis
- **OEIS Syndrome**
  - Full complex consists of 4 components
    - Omphalocele
    - Exstrophy of bladder
    - Imperforate anus
    - Spinal deformities
  - Some authors consider OEIS to be synonymous with cloacal exstrophy

Gastroschisis



Axial color Doppler ultrasound in a 12 week fetus shows a normal cord insertion ➡ on the abdominal wall, with adjacent loops of extruded bowel ➡.

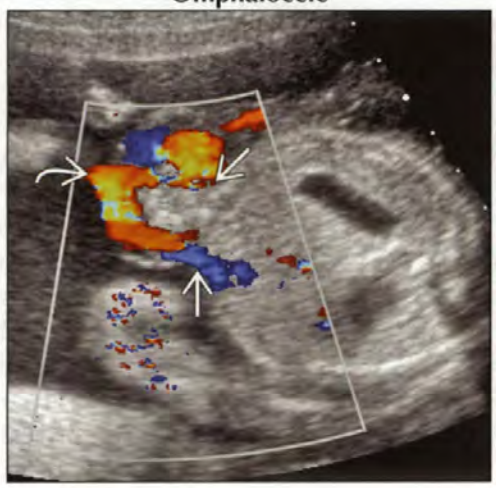
Gastroschisis



Axial ultrasound in a 2nd trimester fetus shows a large gastroschisis with free floating loops of non-dilated bowel ➡. The stomach ➡ is also partially herniated through the defect.

ABDOMINAL WALL DEFECT

Omphalocele



Physiologic Gut Herniation (Mimic)



(Left) *Axial color Doppler ultrasound of a 2nd trimester fetus shows a small, midline, bowel-containing omphalocele. Note how the cord inserts on the apex of the sac and then branches as it enters the fetus.* ***(Right)*** *Axial ultrasound in the 1st trimester has a similar appearance with bowel herniating into the base of the cord. This is a normal finding before 12 weeks and should not be confused with an omphalocele.*

Amniotic Band Syndrome



Amniotic Band Syndrome

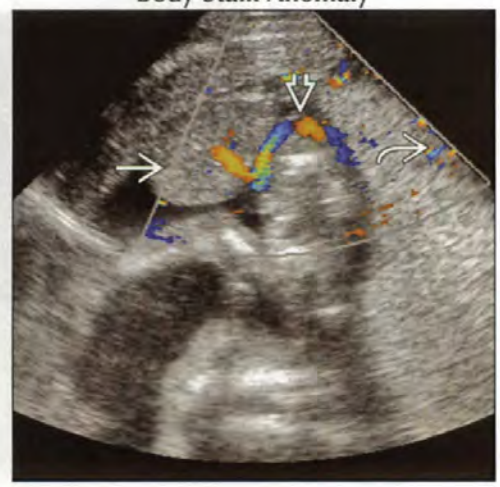


(Left) *Axial ultrasound shows a large abdominal defect, with the entire liver and a portion of the bowel extruded from the abdomen. A primary abdominal wall defect could be considered in the differential diagnosis, but craniofacial defects were also present, making amniotic bands more likely.* ***(Right)*** *Another image in the same case shows a band within the amniotic fluid. Bands are often tightly adherent to the fetus and difficult to see.*

Body Stalk Anomaly



Body Stalk Anomaly



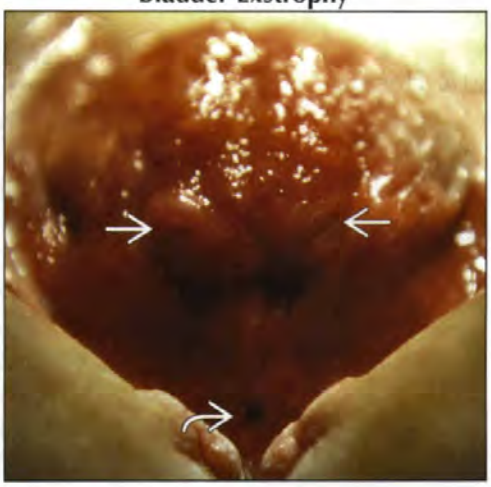
(Left) *Transabdominal ultrasound shows acute angulation of the fetal body. The extremities moved freely, but the spine and fetal location were fixed. Extruded small bowel could not be differentiated from the placenta.* ***(Right)*** *Transvaginal color Doppler in the same case shows the extruded fetal liver. There were no free loops of cord, and only a short segment could be seen going directly from the fetal body to the placenta.*

ABDOMINAL WALL DEFECT

Bladder Exstrophy

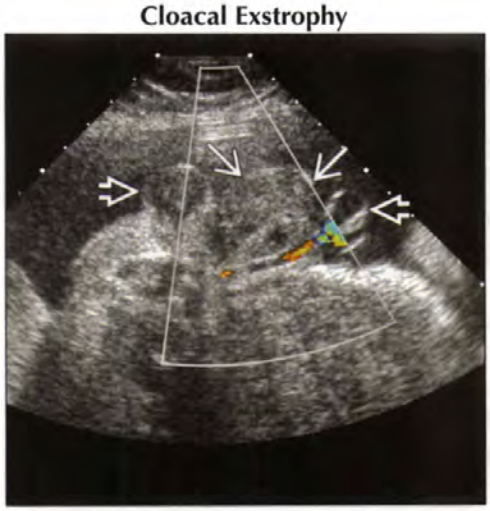


Bladder Exstrophy

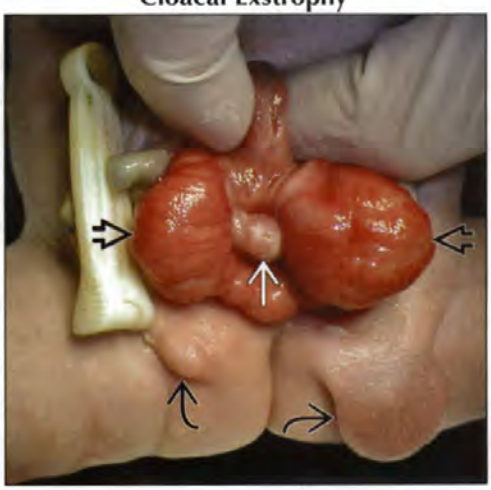


(Left) *Axial oblique ultrasound of the lower fetal abdomen shows an irregular, echogenic contour of the anterior abdominal wall. No normal bladder is seen.* ***(Right)*** *Clinical photograph after delivery shows a large, lower abdominal wall defect, with exposed mucosa of the posterior bladder wall. The ureteral orifices and urethral orifice are seen.*

Cloacal Exstrophy

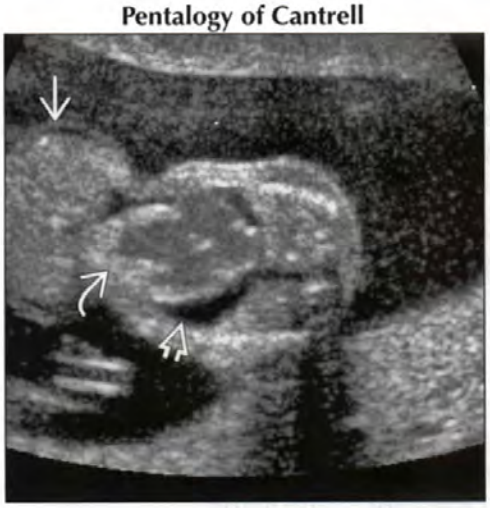


Cloacal Exstrophy

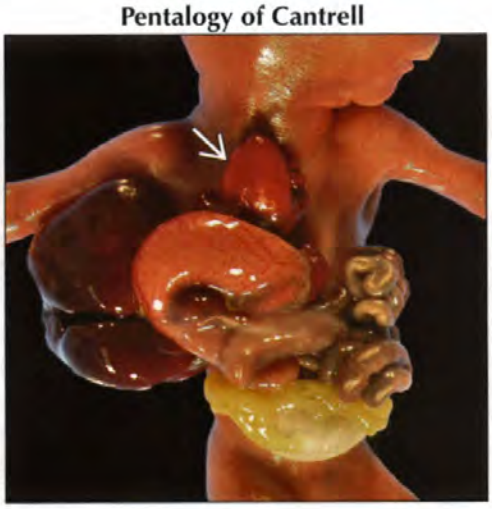


(Left) *Axial oblique image through the lower abdomen shows a soft tissue mass, which was inferior to the cord insertion site. The scrotum was bifid, and no bladder was seen.* ***(Right)*** *Clinical photograph in a different, but similar, case shows the extruded bowel being retracted, exposing the two halves of the split bladder and a small penis. There is a bifid scrotum. The right testis was undescended, accounting for the smaller size of that sac.*

Pentalogy of Cantrell



Pentalogy of Cantrell



(Left) *Axial ultrasound shows a herniated liver, with the cardiac apex protruding from the chest into the defect. There is also a pericardial effusion.* ***(Right)*** *Gross pathology from a similar case shows ectopia cordis, as well as abdominoschisis. Other findings included absence of the sternum, deficiency of the diaphragm and pericardium, and a ventricular septal defect. This constellation of findings is diagnostic of pentalogy of Cantrell.*

SMALL/ABSENT STOMACH

DIFFERENTIAL DIAGNOSIS

Common
- Normal Transient Finding
- Esophageal Atresia
- Congenital Diaphragmatic Hernia
- Oligohydramnios

Less Common
- Abnormal Swallowing
  - Arthrogryposis, Akinesia Sequence
  - Cleft Lip, Palate

Rare but Important
- Neck Mass
  - Goiter
  - Teratoma

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Often difficult to define when a stomach is "small"
  - No defined measurements
  - Stomach size varies between patients
  - Stomach size varies in same fetus over several hours
    - Related to swallowing and peristalsis
  - Requires experience
- More likely to be a true finding if polyhydramnios is present

Helpful Clues for Common Diagnoses
- **Normal Transient Finding**
  - 1% of normal fetuses have a small or absent stomach on initial scan, especially in 1st and 2nd trimester
  - Always get a follow-up exam if there is any question
- **Esophageal Atresia**
  - Atresia of esophagus often associated with tracheoesophageal fistula (TEF)
    - > 90% have a fistula
    - Proximal atresia with distal TEF most common type
  - Small or absent stomach
    - Complete absence suggests either no TEF or a very small, stenotic connection
    - Presence of fistula allows a small amount of fluid to enter stomach via the trachea
  - Look for esophageal "pouch" sign
    - Transient filling of proximal esophagus with swallowing
    - Best performed in a coronal plane
    - Use color Doppler to identify carotid and jugular vessels
  - IUGR seen in up to 40%
    - Ingested amniotic fluid important for growth in latter half of gestation
  - Polyhydramnios rarely develops before 20 weeks
    - Fetal swallowing not important part of amniotic fluid dynamics until that time
    - Polyhydramnios usually progressive after 20 weeks and may be severe in 3rd trimester
  - Part of VACTERL association
    - Vertebral anomalies
    - Anal atresia
    - Cardiac malformation
    - Tracheoesophageal fistula
    - Renal anomalies
    - Limb malformation (frequently radial ray)
  - Also associated with aneuploidy
    - Trisomy 18 > trisomy 21
- **Congenital Diaphragmatic Hernia**
  - Stomach in chest rather than normal location
  - May also have small bowel and liver in chest
  - Peristalsis within chest mass is pathognomonic
  - Abdominal circumference small
  - Polyhydramnios common
- **Oligohydramnios**
  - Stomach is normal but may not be visible because of lack of fluid to swallow
  - Stomach usually seen unless oligohydramnios is severe
    - Renal agenesis
    - Bilateral multicystic dysplastic kidneys
    - Severe autosomal recessive polycystic kidney disease
    - Posterior urethral valves

Helpful Clues for Less Common Diagnoses
- **Arthrogryposis, Akinesia Sequence**
  - Refers to a symptom complex caused by multiple different etiologies, resulting in lack of fetal movement
  - Also includes lack of facial movement
    - Open mouth posture
    - Recessed chin
    - Decreased fetal swallowing

- Unusual or persistent abnormal posturing of limbs
  - Persistent "pike" position of lower limbs with hyperextended knees
  - Cross-legged "tailor's position" of lower limbs, especially in a breech fetus
  - Clubfeet, may be very severe
  - Clenched hands that never open
- **Cleft Lip, Palate**
  - Significant clefts may cause impaired swallowing
  - 80% with cleft lip (CL) will also have cleft palate (CP)
  - Classification
    - Type 1: Unilateral CL without CP
    - Type 2: Unilateral CL with CP
    - Type 3: Bilateral CL/CP
    - Type 4: Midline CL/CP
  - Type 3-4 CL/CP often associated with aneuploidy/syndromes
    - Also more likely to have impaired swallowing

Helpful Clues for Rare Diagnoses

- **Goiter**
  - Homogeneous, smooth, anterior neck mass
    - Maintains thyroid contour
    - Similar appearance to a normal adult thyroid
  - May obstruct swallowing resulting in polyhydramnios and small stomach
  - If esophagus is obstructed, trachea is also likely compressed
    - May cause airway compromise at birth
  - May prevent normal fetal "chin tuck" → extended neck → obstructed labor
- **Teratoma**
  - Mixed echogenicity mass with cystic and solid components
  - Calcifications are virtually pathognomonic but not always present
  - Often very large, irregular shape
  - May exhibit rapid growth
  - Cervical teratoma
    - Located in the anterior neck
    - Frequently extends to involve surrounding structures
    - May extend into mediastinum
    - Head is often held in hyperextension
  - Epignathus
    - Nasopharyngeal teratoma
    - Fungating oral mass
    - May have intracranial extension

Other Essential Information

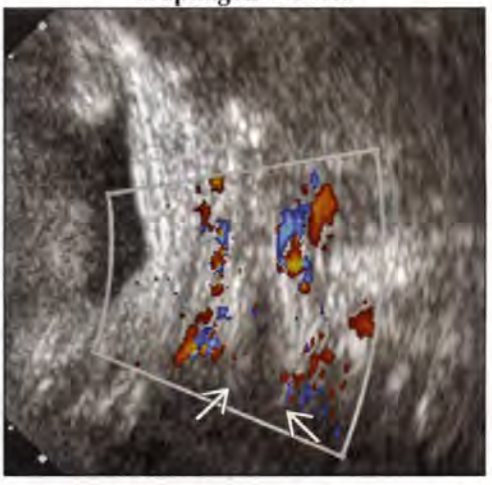
- Follow-up scans should be performed on all fetuses with a small stomach
  - Small stomach may be transient finding in normal fetus
  - Persistence on multiple exams more likely pathologic
  - Pathology likely if polyhydramnios develops
- Anything that impairs fetal swallowing may result in a small stomach and polyhydramnios

Esophageal Atresia



Axial ultrasound through the fetal abdomen shows a normal adrenal gland ⇨, but no stomach bubble. There is also polyhydramnios ⇨. A focused scan of the neck should be performed to evaluate the esophagus.

Esophageal Atresia



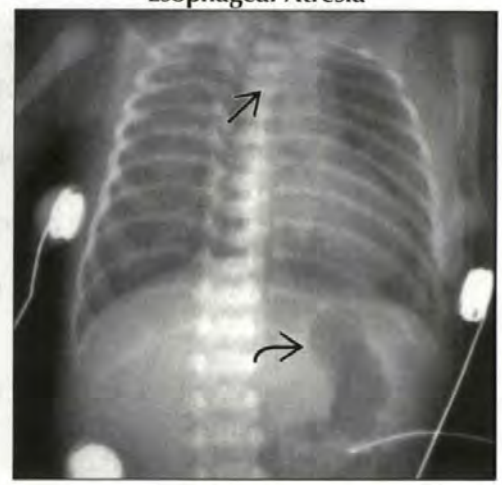
Coronal color Doppler ultrasound of the fetal neck in the same case shows the "pouch sign" ⇨ at the point of esophageal atresia. This blind-ending pouch showed expansion and contraction with fetal swallowing.

SMALL/ABSENT STOMACH

Esophageal Atresia



Esophageal Atresia



(Left) *Axial ultrasound of a 2nd trimester fetus shows that the stomach bubble is present, but smaller than expected. There is also polyhydramnios.* ***(Right)*** *Frontal radiograph shortly after delivery shows an orogastric tube curled within the esophagus and gas within the stomach, indicating there must be a distal tracheoesophageal fistula. The presence of a fistula allows some fluid to accumulate in the stomach prenatally.*

Congenital Diaphragmatic Hernia

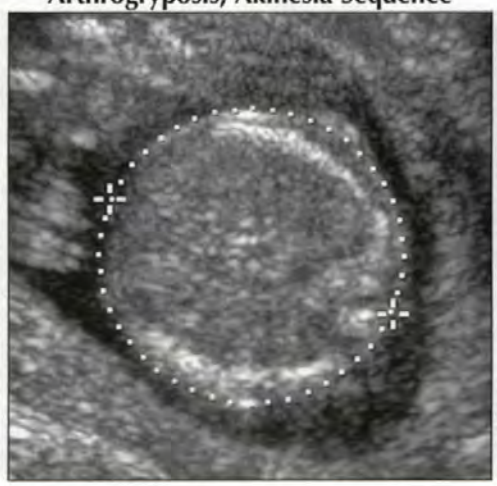


Congenital Diaphragmatic Hernia



(Left) *Axial ultrasound of the abdomen of a 36 week fetus shows no evidence of a stomach bubble. In addition, the abdominal circumference measures small, which is a common associated finding.* ***(Right)*** *Axial ultrasound of the chest in the same case shows stomach in the chest, with the heart displaced into the right hemithorax.*

Arthrogryposis, Akinesia Sequence

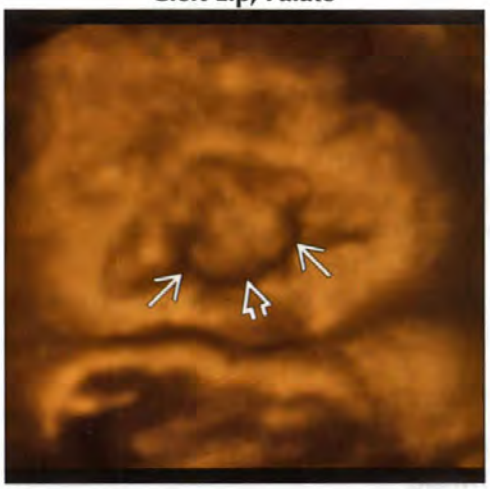


Arthrogryposis, Akinesia Sequence



(Left) *Axial transabdominal ultrasound at the level of the abdominal circumference shows an absent stomach bubble.* ***(Right)*** *Another image from the same case shows the hands, which were persistently held in an abnormal orientation to the wrist. The legs were held in extension, and the left foot was clubbed. Fetuses with arthrogryposis have impaired swallowing and therefore an absent or small stomach bubble. Amniocentesis showed trisomy 18.*

Cleft Lip, Palate



Cleft Lip, Palate

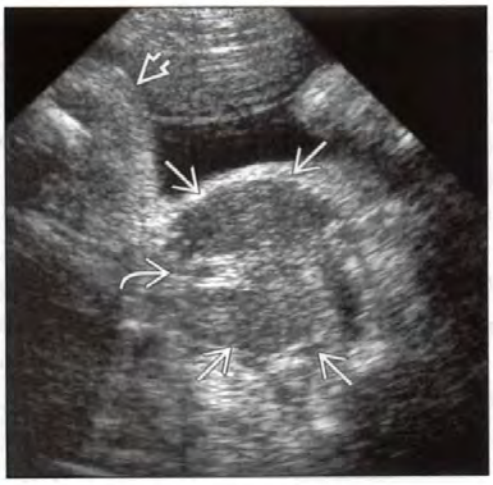


(Left) *Coronal 3D ultrasound shows typical facial features of bilateral cleft lip and palate, including the premaxillary protrusion.* ***(Right)*** *Clinical photograph of a stillborn with trisomy 13 shows a broad, abnormally-formed nose with a bilateral cleft lip. There is also micrognathia. These features impair fetal swallowing.*

Goiter



Goiter



(Left) *Sagittal T2WI MR shows a fetal goiter secondary to stimulation by maternal antibodies. The goiter has mass effect; it restricts fetal swallowing and causes distension of the oropharynx.* ***(Right)*** *Coronal ultrasound of the fetal neck shows both lobes of an enlarged thyroid. The chin is extended, and the trachea is persistently fluid-filled, increasing concern for potential airway obstruction at delivery.*

Teratoma



Teratoma



(Left) *Coronal ultrasound views through the fetal neck show a complex, anterior neck mass (calipers) that has both cystic and solid components. There was associated polyhydramnios and a small stomach from compression of the esophagus.* ***(Right)*** *Lateral radiograph after delivery shows a markedly narrowed airway.*

ECHOGENIC BOWEL

DIFFERENTIAL DIAGNOSIS

Common
- Idiopathic
- Trisomy 21
- Cystic Fibrosis
- Infection
  - Cytomegalovirus
  - Parvovirus
  - Toxoplasmosis
- Meconium Peritonitis, Pseudocyst

Less Common
- Ingested Blood
- Intrauterine Growth Restriction (IUGR)

Rare but Important
- Bowel Ischemia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Echogenic bowel (EB) grading
  - Grade 0: < Liver (normal)
  - Grade 1: > Liver, < bone (normal)
  - Grade 2: = Bone (potentially abnormal)
  - Grade 3: > Bone (potentially abnormal)
- Focal EB more likely pathologic than diffuse EB
- High frequency probe may cause false EB

Helpful Clues for Common Diagnoses
- **Idiopathic**
  - 0.6% incidence (unknown etiology)
- **Trisomy 21**
  - 6.7x ↑ maternal a priori risk when isolated
  - Look for other markers
- **Cystic Fibrosis**
  - ↑ In northern European Caucasians
  - ± Bowel obstruction (meconium ileus)
- **Infection**
  - Cytomegalovirus most common
    - Microcephaly (± calcifications)
  - Intrauterine growth restriction
  - Hydrops
- **Meconium Peritonitis, Pseudocyst**
  - Bowel perforation ⇒ peritonitis
  - Linear and punctate echoes/calcifications
    - Outline liver and bowel
  - Pseudocyst = walled-off fluid
  - Dilated bowel from associated atresia

Helpful Clues for Less Common Diagnoses
- **Ingested Blood**
  - Dependent layering in stomach
  - Often transient
  - Look for evidence of prior perigestational hemorrhage as source for blood
- **Intrauterine Growth Restriction (IUGR)**
  - 18% of 2nd trimester fetuses with EB develop IUGR
  - EB + ↑ Alpha fetoprotein = severe placental insufficiency

Helpful Clues for Rare Diagnoses
- **Bowel Ischemia**
  - From any fetal hypotensive event
  - Associated with twin-twin transfusion

Other Essential Information
- 6% adverse outcome when EB is isolated
- 50% adverse outcome when EB not isolated

Idiopathic



Coronal oblique ultrasound shows a 2nd trimester fetus with isolated grade 3 echogenic bowel. The EB is focal (calipers) and is as echogenic as the iliac crest ➡. Karyotype was normal.

Trisomy 21



Ultrasound shows focal areas of EB ➡ in a fetus with nuchal fold thickening ➡. The presence of two markers for trisomy 21 led to amniocentesis, which confirmed the diagnosis.

ECHOGENIC BOWEL

Cystic Fibrosis



Parvovirus

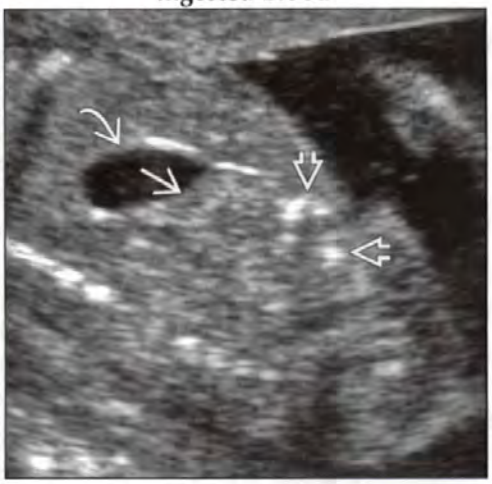


(Left) *Coronal ultrasound shows EB ➡ as bright as bone ➡. The parents were subsequently found to be cystic fibrosis carriers, and the fetus was diagnosed with cystic fibrosis at birth.* ***(Right)*** *Coronal oblique ultrasound shows EB ➡, ascites ➡, and body wall edema ➡ in a fetus subsequently proven to have parvovirus. The findings were transient.*

Meconium Peritonitis, Pseudocyst



Ingested Blood

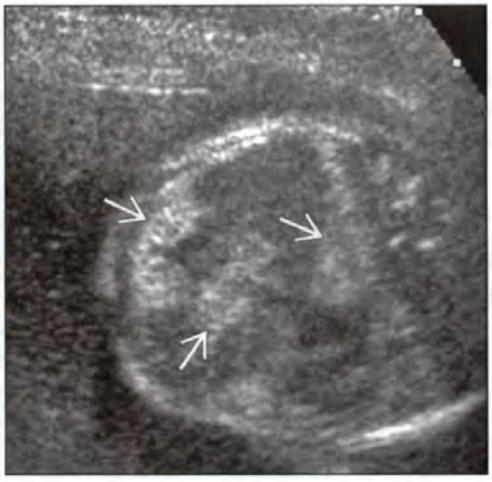


(Left) *Coronal ultrasound shows focal coarse linear echogenicities ➡ & a small hypoechoic fluid collection ➡ in the fetal abdomen. The findings represent peritoneal calcifications & a small pseudocyst.* ***(Right)*** *Sagittal oblique ultrasound shows EB ➡ & an echogenic fluid level ➡ in the fetal stomach ➡. The pregnancy was complicated by chronic abruption. The EB was caused from ingested intra-amniotic blood/proteinaceous material.*

Intrauterine Growth Restriction (IUGR)



Bowel Ischemia



(Left) *Sagittal oblique ultrasound shows EB ➡ in a pregnancy complicated by oligohydramnios and intrauterine growth restriction. The cause for the IUGR in this case was severe placental insufficiency.* ***(Right)*** *Axial oblique ultrasound shows diffusely echogenic bowel ➡ in a twin pregnancy complicated by twin-twin transfusion. The donor twin in this case suffered from bowel and brain ischemia.*

DILATED BOWEL

DIFFERENTIAL DIAGNOSIS

Common
- Normal Third Trimester Bowel
- Duodenal Atresia
- Jejunal, Ileal Atresia
- Meconium Ileus

Less Common
- Anal Atresia
- Volvulus

Rare but Important
- Cloacal Malformation

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Try to determine point of obstruction (proximal vs. distal)
- Assess peristalsis
  - Hyperperistalsis of obstructed bowel often seen in real-time evaluation
  - Infarcted bowel loses ability for peristalsis
- Polyhydramnios common with atresia, but timing & severity dependent on site of obstruction
  - Earlier presentation & more severe with proximal atresia
  - Usually not present before 24 weeks
    - Most present in 3rd trimester
  - Amniotic fluid is often echogenic from fetal regurgitation of intraluminal contents
- Intrauterine growth restriction (IUGR) common with atresia
  - Proximal atresias more likely to have IUGR
  - Ingested amniotic fluid important for fetal growth in latter half of gestation
- Always look for normal rectum/anus in setting of obstruction
  - Normal rectum has hypoechoic walls with a hyperechoic mucosa
  - Follow down to perineum in coronal or sagittal plane
  - "Target" appearance in axial plane

Helpful Clues for Common Diagnoses
- **Normal Third Trimester Bowel**
  - Colon, especially sigmoid, often prominent in 3rd trimester
  - Normal colon diameter ≤ 18 mm
- **Duodenal Atresia**
  - Fluid-filled stomach & duodenum create classic "double bubble" appearance in axial plane
  - Stomach & duodenum can be connected during real-time imaging
  - No fluid in distal bowel loops
  - 30% will have trisomy 21
  - May also have an associated esophageal atresia
    - If a tracheoesophageal fistula is not present, fluid may accumulate in distal esophagus, stomach, & duodenum, forming a "C loop"
    - Normal secretions accumulate in this closed loop
    - Accumulated secretions may cause marked dilatation, much greater than typically seen with just duodenal atresia
- **Jejunal, Ileal Atresia**
  - Roughly equal involvement between jejunum and ileum
  - "Triple bubble" for proximal jejunal atresia
  - "Sausage-shaped" bowel loops
  - Hyperperistalsis of obstructed segments often seen in real time
  - At risk for perforation and meconium peritonitis (≈ 6%)
- **Meconium Ileus**
  - Obstruction of distal ileum due to abnormally thick, tenacious meconium
  - Seen in fetuses with cystic fibrosis
  - Echogenic bowel in 2nd trimester
    - Increased echogenicity likely secondary to inspissated, mucus secretions in bowel lumen
  - Progresses to dilated small bowel in 3rd trimester
    - Appearance often indistinguishable from ileal atresia
  - May perforate & present with meconium peritonitis
  - Systic fibrosis is a autosomal recessive disorder
    - If diagnosis is suspected, test parents for carrier status
    - Can also do amniocentesis for direct detection of mutation in fetus
  - If not detected in utero, may present in newborn period with failure to pass meconium

- 10-20% of newborns with cystic fibrosis have meconium ileus

Helpful Clues for Less Common Diagnoses

- **Anal Atresia**
  - May go undetected prenatally
    - Dilatation does not typically occur until 3rd trimester
  - Difficult to distinguish large from small bowel
    - U- or V-shaped bowel in pelvis suggestive of anorectal atresia
  - Scan in both coronal and axial planes looking for normal rectum
    - May see rectum end above perineum
    - Normal "target" appearance will not be present
  - Often associated with urinary tract fistulae
    - Mixing of meconium with urine creates enteroliths (may see calcified meconium "marbles" moving within bowel)
  - Part of VACTERL association
    - Vertebral anomalies
    - Anal atresia
    - Cardiac anomalies
    - Tracheo-esophageal (TE) fistula
    - Renal anomalies
    - Limb malformations
- **Volvulus**
  - Single "kinked" loop is very suggestive but often difficult to determine
  - May see multiple, dilated loops from proximal obstruction
    - Often difficult to differentiate from bowel atresia
  - May have had a normal scan earlier in gestation
    - Volvulus is an abrupt event
  - Compromise of vascular supply leads to infarction & necrosis
    - Echogenic, intraluminal contents from sloughing of necrotic mucosa
    - Real-time evaluation important: Infarcted bowel loses ability for peristalsis

Helpful Clues for Rare Diagnoses

- **Cloacal Malformation**
  - Persistent cloaca
    - Failure of urorectal septum to reach perineum
    - Seen in female fetuses
    - Results in single perineal opening for urine, genital secretions, & meconium
    - Hydrometrocolpos may be seen secondary to communication of bladder with uterus and vagina
  - Variable in utero presentation
    - Cystic mass in pelvis
    - Dilated pelvic bowel loops; may see enteroliths from mixing of meconium and urine
    - Hydronephrosis
    - Lumbosacral anomalies
    - Abnormal genitalia with lack of normal labial/clitoral formation
    - Ascites reported in some cases

Normal Third Trimester Bowel



Coronal ultrasound in the 3rd trimester shows prominent, but normal, meconium-filled colon. Hepatic flexure, splenic flexure, and sigmoid colon are well seen.

Duodenal Atresia



Axial transabdominal ultrasound shows a dilated stomach, connecting with a dilated duodenum in a fetus with trisomy 21. Note the echogenic amniotic fluid, which is secondary to fetal regurgitation.

DILATED BOWEL

Jejunal, Ileal Atresia



Jejunal, Ileal Atresia



(Left) *Axial ultrasound shows the jejunum ➔, ending blindly in the left mid-abdomen. Polyhydramnios is present, and the amniotic fluid contains echogenic debris.* ***(Right)*** *Coronal oblique color Doppler ultrasound shows distended bowel filled with thick, echogenic fluid, which helps distinguish it from the bladder ➔ that is flanked by the umbilical arteries. Surgery confirmed a distal ileal atresia.*

Meconium Ileus



Meconium Ileus

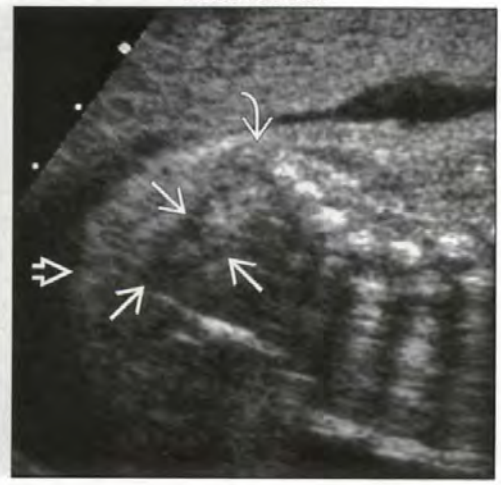


(Left) *Axial oblique ultrasound in a 2nd trimester fetus shows a focal area of echogenic bowel ➔.* ***(Right)*** *A follow-up scan in the 3rd trimester shows progression to dilated bowel loops ➔, consistent with meconium ileus. Genetic testing confirmed cystic fibrosis.*

Meconium Ileus



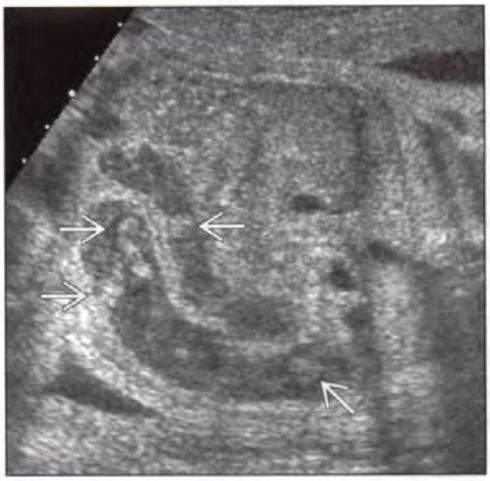
Anal Atresia



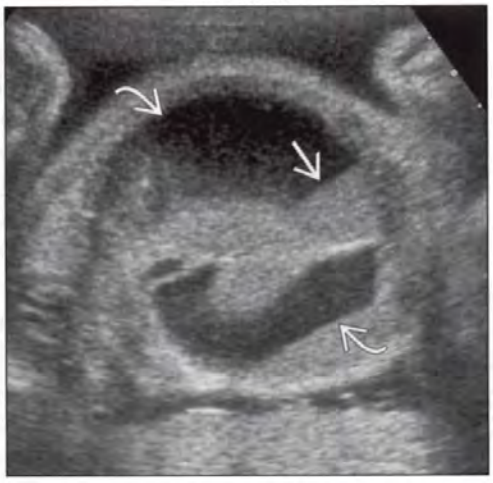
(Left) *Coronal ultrasound shows a slightly dilated loop of bowel with echogenic walls ➔ in this fetus with cystic fibrosis. Meconium ileus was confirmed at delivery.* ***(Right)*** *Sagittal ultrasound shows abrupt termination of the sacrum ➔, secondary to caudal regression. The hypoechoic rectal walls abruptly end in a V-shape ➔, well above where the anus should be ➔. VACTERL association was confirmed at birth.*

DILATED BOWEL

Anal Atresia

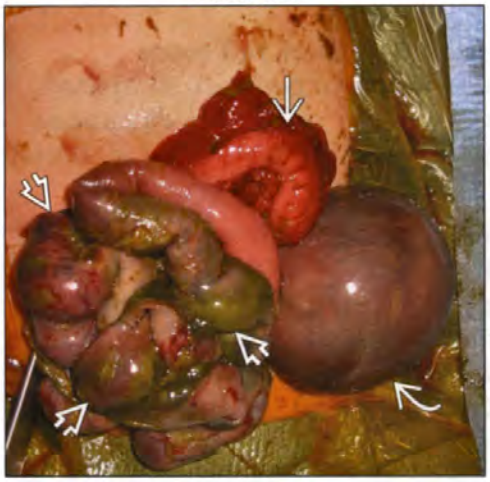


Volvulus



(Left) *Coronal ultrasound shows dilated bowel with echogenic, meconium "marbles" ➡, which moved on real-time exam. This appearance is suspicious for anal atresia with a vesicocolic fistula.* ***(Right)*** *Axial transabdominal ultrasound shows distended loops of bowel ➡. Note the fluid-debris level ➡ from sloughed, necrotic mucosa. There was lack of peristalsis on real-time exam. Postnatal surgery showed small bowel volvulus with extensive bowel infarction.*

Volvulus



Cloacal Malformation

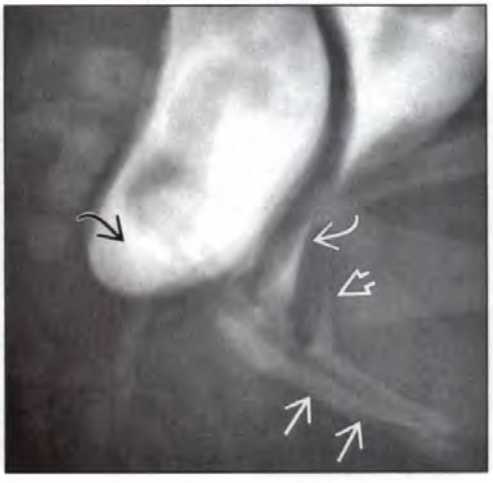


(Left) *Intra-operative photograph in a different case of mid-gut volvulus shows twisted and infarcted small bowel ➡, to the right of the colon ➡. Note the incidental large ovarian cyst ➡.* ***(Right)*** *Sagittal ultrasound shows an unusual appearing, dilated bowel loop ➡ in the pelvis of a fetus who was born with a cloacal malformation.*

Cloacal Malformation



Cloacal Malformation



(Left) *Clinical photograph of a newborn girl with a cloacal malformation shows abnormal genitalia, with a single perineal opening ➡, the cloaca, which is being cannulated.* ***(Right)*** *Lateral radiograph from a cloacagram, in a different newborn girl, shows a catheter introduced through the single perineal opening, which represents a persistent cloaca ➡. The urethra ➡, vagina ➡, and rectum ➡ all communicate with the cloaca.*

ASCITES

DIFFERENTIAL DIAGNOSIS

Common
- Pseudoascites (Mimic)
- Hydrops
- Bladder Rupture
- Bowel Perforation
- Infection

Less Common
- Tachyarrhythmia

ESSENTIAL INFORMATION

Helpful Clues for Common Diagnoses
- **Pseudoascites (Mimic)**
  - Abdominal wall musculature can mimic ascites
    - Surrounds the anterolateral liver margin but not seen posteriorly
  - More prominent with slightly oblique scan angle
  - True ascites is seen in other parts of abdomen, outlines organs
- **Hydrops**
  - Fluid in 2 body spaces
    - Skin edema, pleural effusion, ascites, pericardial effusion
  - Nonimmune or immune hydrops
  - Polyhydramnios and placentomegaly also commonly present
- **Bladder Rupture**
  - Initial ultrasound may show markedly enlarged bladder
    - Look for evidence of posterior urethral valves or urethral atresia
    - Less commonly due to prune belly or megacystis-microcolon
  - Bladder rupture results in urinary ascites
    - Thick-appearing bladder wall after decompression
  - Follow all fetuses with large bladder
    - Likely transient finding if otherwise normal urinary tract
- **Bowel Perforation**
  - Initial ultrasound may show dilated bowel
    - Underlying atresia(s), volvulus, intussusception, meconium ileus
  - Often perforation event is occult, with ascites being the only finding
  - Look for signs of meconium peritonitis to confirm diagnosis
    - Intraperitoneal calcifications
    - Meconium pseudocyst
- **Infection**
  - Ascites may be isolated, but more commonly, is part of generalized hydrops
  - Look for hepatic or cerebral calcifications
  - Correlate with clinical history for maternal signs of infection or exposures

Helpful Clues for Less Common Diagnoses
- **Tachyarrhythmia**
  - Sustained heart rate > 180-200 bpm
  - Ascites may be isolated and first sign of impending failure/hydrops
  - Always check heart rate in setting of isolated ascites

Pseudoascites (Mimic)



Axial oblique ultrasound shows a prominent hypoechoic linear area ➡, lateral to the liver, which represents abdominal wall musculature and not ascites. This was not seen in other scan planes.

Hydrops



Axial transabdominal ultrasound shows mild skin edema ➡ and ascites ➡. Note that the ascites is anechoic and extends anteriorly around the liver, distinguishing it from pseudoascites.

ASCITES

Hydrops

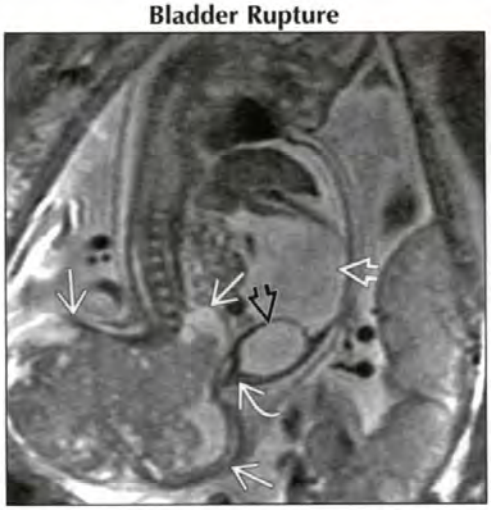


Hydrops



(Left) *Axial ultrasound shows massive ascites with a free floating membrane, likely representing the wall of the lesser sac. The fetus was delivered at 33.5 weeks for progressing hydrops and severe polyhydramnios. No underlying etiology was discovered.* ***(Right)*** *Sagittal transvaginal ultrasound in the first trimester shows skin edema and fluid in the abdomen. The couple elected to have chorionic villus sampling, which showed Turner syndrome.*

Bladder Rupture



Bowel Perforation

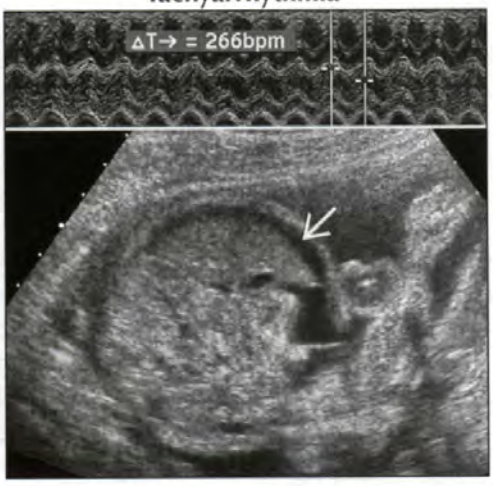


(Left) *Sagittal T2WI MR shows a fetus with a sacrococcygeal teratoma, extending into the pelvis and causing a bladder outlet obstruction. There is urinary ascites from bladder perforation. Note the relatively thick-walled, decompressed bladder.* ***(Right)*** *Sagittal ultrasound in a newborn shows peritoneal calcifications and ascites at the liver margin, supporting the prenatal suspicion of meconium peritonitis.*

Infection



Tachyarrhythmia



(Left) *Axial ultrasound of the abdomen shows ascites, as well as echogenic bowel. These findings were associated with fetal anemia in the clinical setting of maternal parvovirus infection.* ***(Right)*** *Axial ultrasound of the abdomen shows a small amount of ascites in a fetus with a sustained heart rate of 266 bpm. Ascites may be the first manifestation of impending failure and hydrops.*

ABDOMINAL CALCIFICATIONS

DIFFERENTIAL DIAGNOSIS

Common
- Meconium Peritonitis, Pseudocyst
- Gallstones
- Hepatic Calcifications

Less Common
- Intraluminal Calcified Meconium

Rare but Important
- Teratoma
- Fetus-in-Fetu

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Echogenic foci within fetal abdomen
  - May not have shadowing
  - Can be punctate or irregularly shaped
- Differential diagnosis for etiology based on location of calcifications
  - Liver
    - Intraparenchymal vs. liver capsule
    - Biliary
  - Bowel
    - Intraluminal vs. extraluminal
  - Peritoneum
  - Abdominal mass
- Look for any other associated findings for clues
  - Intracranial calcifications
    - Infection
  - Abnormal bowel
    - Meconium peritonitis
    - Anal dimple absent in anal atresia
  - Fluid collections
    - Ascites and pseudocysts with meconium peritonitis
  - Soft tissue mass
    - Teratoma
    - Fetus-in-fetu
    - Other fetal abdominal tumors do not typically calcify
  - Fetal anomalies

Helpful Clues for Common Diagnoses
- **Meconium Peritonitis, Pseudocyst**
  - Chemical peritonitis due to intrauterine bowel perforation
  - Intraperitoneal calcifications most specific finding
  - Ascites
  - Loculated fluid collection (pseudocyst) may also be present
    - Walled-off perforation
    - Contents have variable echogenicity
    - Walls often thick and may calcify
  - Dilated bowel present when secondary to obstruction
    - Not typical when secondary to ischemic bowel perforation
- **Gallstones**
  - Echogenic calcifications in the gallbladder
    - Shadowing may be present but not necessary to diagnose stones
    - "Comet tail" artifact sometimes present
    - Mobile
    - Not usually seen with biliary dilation
    - Calcifications not always dependent, and some may be within gallbladder wall
  - Most often seen 3rd trimester
    - Incidental finding during routine scan
  - If gallbladder is contracted, gallstones may mimic hepatic calcification
    - Focal linear area of echoes rather than diffusely spread through liver
    - Diagnosis more easily made when gallbladder filled
    - If calcifications seen in typical location of gallbladder, look again later during scan
    - When gallbladder fills, bile will outline stones
  - Usually spontaneously resolves within first year of life
    - Postnatal ultrasound can confirm presence of stones
    - Typically infants are asymptomatic
- **Hepatic Calcifications**
  - Intrahepatic, scattered, echogenic foci
    - Seen with infection
    - Association with chromosomal anomalies
  - Shadowing with larger calcifications
  - Occasionally seen incidentally, without known clinical consequences
    - Exclude other anatomic abnormalities
    - Exclude infectious etiology
    - If isolated, most neonates will be asymptomatic
  - Capsular calcifications more often related to meconium peritonitis

- Most commonly seen at liver dome or undersurface of liver
- Look for other calcifications in peritoneum

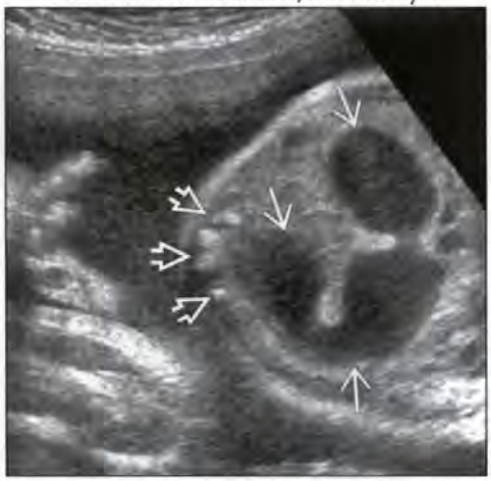
Helpful Clues for Less Common Diagnoses

- **Intraluminal Calcified Meconium**
  - Calcified, mobile material within bowel ("marbles")
  - Generally occur in setting of anal atresia with associated urinary tract fistula/anomalies
    - Urine mixes with meconium to cause calcifications
  - Additional findings of anal atresia
    - Absent anal dimple
    - Dilated bowel (usually only seen in 3rd trimester, if present)
    - May go undetected prenatally
  - Occasionally enteroliths form due to stasis of bowel contents
  - Look for other anomalies in VACTERL syndrome
    - Vertebral, anal atresia, cardiac, tracheo-esophageal fistula, renal, limb

Helpful Clues for Rare Diagnoses

- **Teratoma**
  - 3 germ cell layers present
    - Arise from growth of pluripotent stem cells without organization
    - Can have malignant potential
  - In spectrum with fetus-in-fetu
    - Fetus-in-fetu classically defined by presence of spine
    - Teratoma may still have limbs, digits, other organs
    - Can originate from retroperitoneum, as well as multiple other sites along midline
- **Fetus-in-Fetu**
  - Intra-abdominal fetiform mass
    - Classic definition includes spinal elements due to completion of initial steps of embryogenesis (vs. teratoma, which does not have spine formation)
    - Often complex mass with both cystic and solid components
    - Vascular supply typically from superior mesenteric artery
  - Majority reported are in upper retroperitoneum
    - Less commonly in other sites: Intracranial, pelvis, sacrum, liver
  - Theoretically results from inclusion of a monochorionic diamniotic twin within host twin
    - Grossly resembles acardiac twin in twin reversed arterial perfusion sequence (no cranial or cardiac development)
  - Can grow in size during gestation
  - Follow for evidence of hydrops
  - If not detected prenatally, may continue to grow slowly
    - Can present as asymptomatic abdominal mass on exam or symptoms from local mass effect
    - Rare reports of discovery in later adolescence and adulthood

Meconium Peritonitis, Pseudocyst



Axial ultrasound shows distended bowel ➡ in the mid-abdomen. Realtime peristalsis helps distinguish distended bowel from a pseudocyst. Peritoneal calcifications are present ➡.

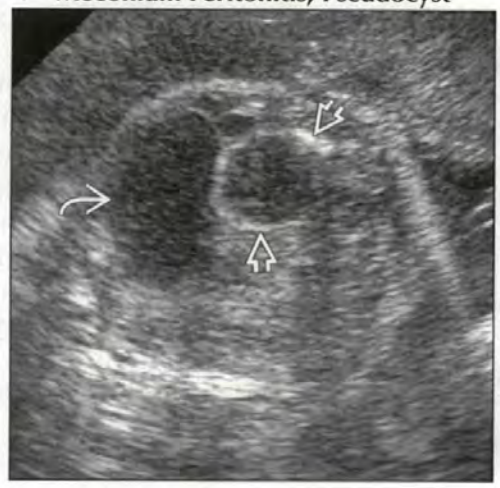
Meconium Peritonitis, Pseudocyst



Coronal ultrasound of the same fetus shows more peritoneal calcifications ➡ suggesting remote peritoneal spill of bowel contents with subsequent calcification. Jejunal atresia was confirmed after birth.

ABDOMINAL CALCIFICATIONS

Meconium Peritonitis, Pseudocyst



Meconium Peritonitis, Pseudocyst



(Left) *Axial ultrasound of the pelvis shows a calcified, thick-walled cyst and adjacent dilated bowel. This appearance is typical of pseudocyst formation following a perforated bowel obstruction.* ***(Right)*** *Radiograph of a newborn shows dense calcifications over the liver, which represent the sequelae of meconium peritonitis. There is nonobstructive bowel gas pattern and the infant began feeding without difficulty.*

Gallstones



Gallstones

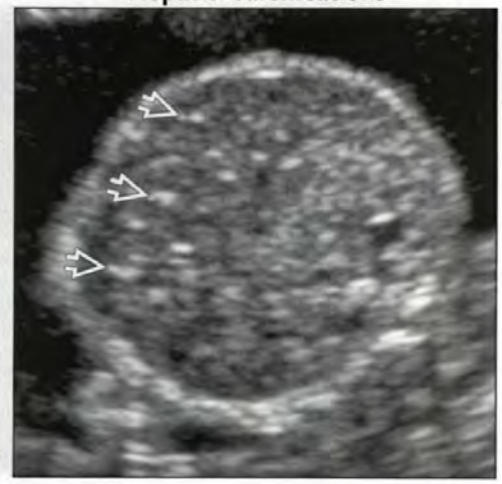


(Left) *Axial ultrasound shows innumerable gallstones in the fetal gallbladder in the third trimester, incidentally seen during a growth scan.* ***(Right)*** *Axial ultrasound of the same patient postnatally showed both mobile stones and non-dependent echogenic foci in the gallbladder wall with a comet tail artifact. The appearance is similar to that of an adult with adenomyomatosis, but when seen in a fetus or neonate, it generally resolves spontaneously.*

Gallstones



Hepatic Calcifications



(Left) *Axial ultrasound shows gallstones in a partially contracted gallbladder. If the gallbladder is not distended with bile, gallstones may be mistaken for hepatic parenchymal calcifications.* ***(Right)*** *Axial ultrasound shows multiple echogenic foci in the fetal liver. Polyhydramnios was also present. At 22 weeks the patient had premature rupture of membranes and delivered. The fetus expired shortly after birth. There was no proven etiology, but infection was suspected.*

ABDOMINAL CALCIFICATIONS

Hepatic Calcifications



Intraluminal Calcified Meconium

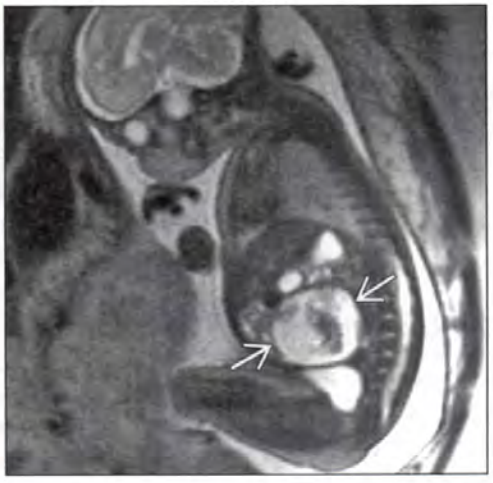


(Left) *Axial ultrasound shows incidental, coarse, hepatic calcifications ➡ with acoustic shadowing ➡ near the cord insertion. This was an isolated finding, & the neonate was normal at birth. Possible etiologies include early ischemic episode or occult bowel perforation.* ***(Right)*** *Axial ultrasound shows 2 views of dilated bowel ➡ with multiple internal "marbles" of calcified meconium ➡. A vesicocolic fistula and anal atresia were confirmed postnatally.*

Teratoma

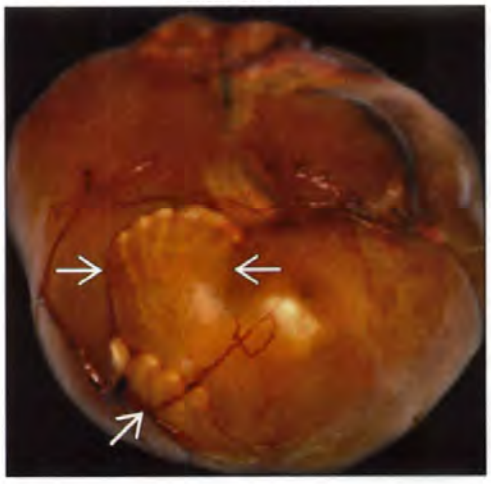


Teratoma

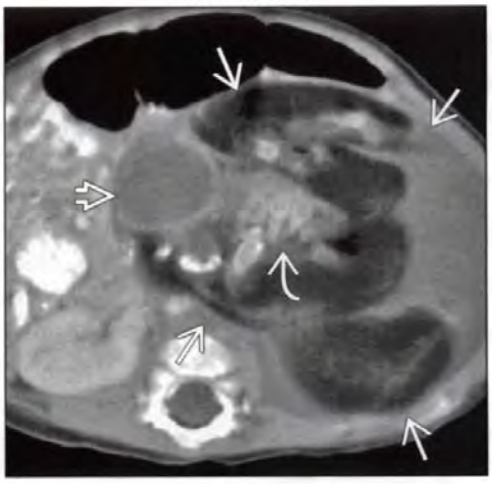


(Left) *Axial ultrasound shows a few calcifications ➡ in a mostly cystic fetal abdominal mass ➡. Bladder (BL).* ***(Right)*** *Sagittal T2WI MR of the same fetus shows a complex, cystic, and solid mass ➡ in the fetal abdomen. This mass had multiple osseous structures on excision, though no vertebrae. It is therefore classified as a teratoma rather than a fetus-in-fetu.*

Fetus-in-Fetu



Fetus-in-Fetu



(Left) *Gross pathology photograph shows an excised fetus-in-fetu after delivery. Note the two recognizable foot structures ➡.* ***(Right)*** *Axial CECT of a neonate shows a large intra-abdominal mass ➡ containing fat and fluid. Abnormal spine elements ➡ and a gallbladder ➡ are also present. Surgical excision confirmed a fetus-in-fetu.*

LARGE ABDOMINAL CIRCUMFERENCE

DIFFERENTIAL DIAGNOSIS

Common

- Fetal Overgrowth
  - Macrosomia
  - Diabetes
  - Beckwith-Wiedemann Syndrome
- Bowel Obstruction
  - Small Bowel Atresia
  - Volvulus
  - Meconium Pseudocyst
  - Meconium Ileus
- Urinary Tract Anomalies
  - Multicystic Dysplastic Kidneys
  - Autosomal Recessive Polycystic Kidney Disease (ARPKD)
  - Posterior Urethral Valves
  - Urethral Atresia
  - Prune Belly Syndrome
- Hydrops

Less Common

- Ascites, Massive
- Mesoblastic Nephroma
- Hepatomegaly, Splenomegaly

Rare but Important

- Liver Tumors
- Lymphangioma

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Is there generalized overgrowth, or is increased size limited to the abdomen?
- Is there increased fat deposition?
- Is there a history of maternal diabetes?
- Are there dilated loops of bowel or other masses in the abdomen?
- Are the kidneys enlarged or dilated?
- Is the bladder of normal size and shape?
- Is the amniotic fluid normal?
- Are there other structural anomalies?
- Is there associated hydrops or ascites?

Helpful Clues for Common Diagnoses

- **Fetal Overgrowth**
  - **Macrosomia**
    - Generalized increase in size of all parameters
    - Commonly seen in poorly controlled diabetes, but may be isolated
    - Associated polyhydramnios
    - 3rd trimester phenomenon
  - **Diabetes**
    - Overall increased size in fetus
    - Abdominal circumference increase often disproportionate
    - Greatest increase in growth velocity in 3rd trimester
    - Associated polyhydramnios
    - Growth acceleration associated with poor glycemic control, especially in gestational diabetes, but may also occur in pre-gestational diabetes
  - **Beckwith-Wiedemann Syndrome**
    - Generalized overgrowth
    - Visceromegaly with enlarged kidneys, liver, and spleen
    - Omphalocele common
    - Macroglossia can be seen prenatally
- **Bowel Obstruction**
  - **Small Bowel Atresia**
    - One or several loops of dilated bowel, depending on level of obstruction
    - May be seen as early as the late 2nd trimester
    - Hyperperistalsis seen in obstructed loops
  - **Volvulus**
    - Multiple loops of dilated bowel may fill abdomen
    - No peristalsis if bowel is infarcted
    - Evaluate presence/absence of anal dimple: Absence suggests associated anorectal malformation
  - **Meconium Pseudocyst**
    - Large, irregular fluid collection in the abdomen
    - Echogenic "rind"
    - Association with in-utero bowel perforation
    - May or may not be associated with fetal cystic fibrosis
  - **Meconium Ileus**
    - Focal area of echogenic bowel may precede appearance of dilated loops
    - Dilation may be severe
    - Associated ascites
    - Check for family history of cystic fibrosis
- **Urinary Tract Anomalies**
  - **Multicystic Dysplastic Kidneys**
    - Enlarged echogenic kidneys with multiple macrocysts of variable sizes
    - Bilateral or unilateral

- Bilateral with anhydramnios is lethal
- **Autosomal Recessive Polycystic Kidney Disease (ARPKD)**
  - Severe end of spectrum usually lethal, with markedly enlarged, echogenic kidneys
  - Small chest and anhydramnios with worst outcome
  - Gross cysts not seen on prenatal ultrasound
- **Posterior Urethral Valves**
  - Found only in male fetus
  - Dilated renal pelves may be severe
  - Bladder enlarged with "keyhole" shape due to dilated prostatic urethra
  - If severe, may be associated with complete bladder outlet obstruction early in gestation
- **Urethral Atresia**
  - Bladder distension massive with anhydramnios an early finding
  - Male and female fetuses
- **Prune Belly Syndrome**
  - Triad of abnormal abdominal muscle development, undescended testes, and marked bladder distension
  - Entire urinary tract dilated, with dramatic ureterectasis and hydronephrosis

- **Hydrops**
  - Ascites, pleural &/or pericardial effusions, and skin edema
  - Associated polyhydramnios or oligohydramnios
  - Cystic hygroma common association

Helpful Clues for Less Common Diagnoses

- **Ascites, Massive**
  - Isolated without other signs of hydrops
  - Poor prognosis for normal outcome
  - Associated bowel or renal abnormalities common
- **Mesoblastic Nephroma**
  - Large, homogeneous-appearing renal tumor
  - Association with polyhydramnios
  - Mass effect on surrounding vessels such as aorta
- **Hepatomegaly, Splenomegaly**
  - Association with edema from hydrops, heart failure, intrauterine infection
  - "Speckled" appearance concerning for infection

Helpful Clues for Rare Diagnoses

- **Liver Tumors**
  - Associated hepatomegaly
  - Cystic/vascular component more commonly associated with hemangioma or hemangioendothelioma
  - Solid tumor more likely to be hepatoblastoma
- **Lymphangioma**
  - Complex lesion with multiple cysts
  - Lesions may be superficial or within the chest or abdomen
  - Involvement of neck common

Macrosomia



Clinical photograph of a 12 lb 12 oz newborn. Note the protuberant abdomen and increased subcutaneous fat. The mother was not diabetic but did a have history of delivering large babies.

Diabetes



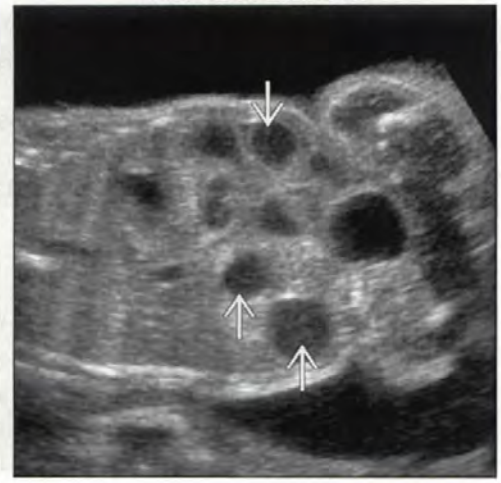
Coronal ultrasound shows a 33 week fetus with macrosomia related to poorly controlled maternal type II diabetes. The obviously enlarged abdomen ▶ is comparable in size to that of a 38 week fetus.

LARGE ABDOMINAL CIRCUMFERENCE

Beckwith-Wiedemann Syndrome



Small Bowel Atresia



(Left) *Axial ultrasound shows the enlarged kidneys ➔ seen in a 3rd trimester fetus with Beckwith-Wiedemann syndrome. The abdomen was large when compared to the other biometric parameters.* ***(Right)*** *Coronal ultrasound shows multiple loops of dilated bowel ➔ in a mid-trimester fetus with a bowel obstruction due to multiple atresias.*

Small Bowel Atresia



Small Bowel Atresia

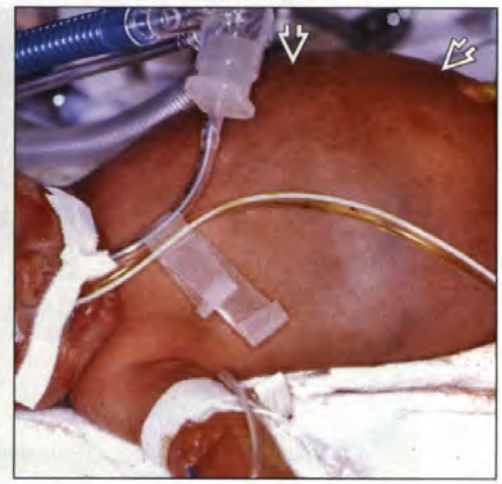


(Left) *Axial ultrasound shows the enlarged abdomen of a fetus in the third trimester with postnatally confirmed ileal atresia. Note the markedly dilated loops of bowel ➔.* ***(Right)*** *Clinical photograph shows the distended abdomen ➔ of the same infant at term. Ileal atresia was first diagnosed in the mid-trimester.*

Volvulus



Volvulus

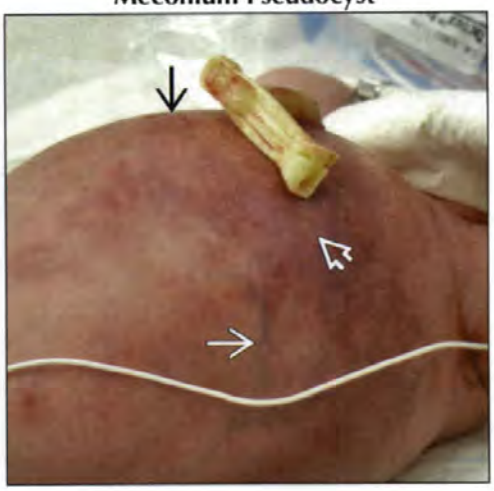


(Left) *Axial ultrasound shows multiple loops of dilated bowel ➔ in a mid-trimester fetus with a mid-gut volvulus due to a malrotation. Ascites ➔ is also seen between the bowel loops.* ***(Right)*** *Clinical photograph shows the massively distended abdomen ➔ in the same preterm infant. Mid-gut volvulus was confirmed at surgery.*

Meconium Pseudocyst



Meconium Pseudocyst



(Left) *Axial ultrasound shows a large, irregular meconium pseudocyst in the abdomen of a mid-trimester fetus. Note the echogenic rind ➔ around the pseudocyst.* ***(Right)*** *Clinical photograph shows the markedly distended abdomen ➔ in the same term infant. Note the ischemic skin changes ➔ and distended vessels ➔.*

Meconium Ileus



Multicystic Dysplastic Kidneys

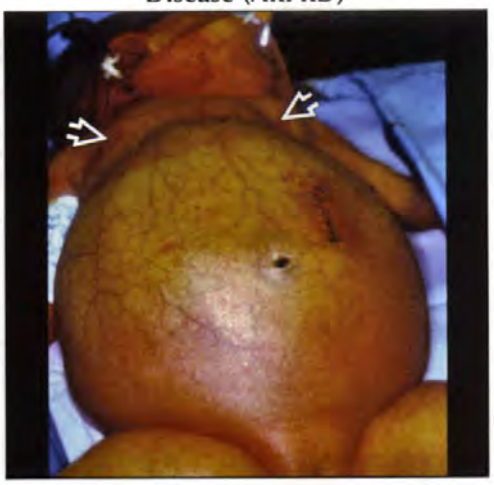


(Left) *Radiograph shows marked abdominal distention with multiple loops of dilated bowel ➔ in a newborn with failure to pass meconium. The in-utero appearance is often difficult to distinguish from ileal atresia. Always check for a family history of cystic fibrosis.* ***(Right)*** *Coronal ultrasound shows a 32 week fetus with bilateral multicystic dysplastic kidneys. The kidneys are very large ➔, with multiple cysts of varying sizes. The parenchyma ➔ is dysplastic, and there is anhydramnios.*

Autosomal Recessive Polycystic Kidney Disease (ARPKD)



Autosomal Recessive Polycystic Kidney Disease (ARPKD)



(Left) *Coronal ultrasound shows bilateral, markedly enlarged kidneys ➔ in a fetus with ARPKD. Note the increased medullary echogenicity ➔ and anhydramnios. The infant died shortly after birth due to pulmonary hypoplasia.* ***(Right)*** *Clinical photograph of a different infant with ARPKD shows massive enlargement of the abdomen. Note the very small chest ➔. There was severe oligohydramnios in utero, resulting in pulmonary hypoplasia.*

LARGE ABDOMINAL CIRCUMFERENCE

(Left) *Axial ultrasound shows a 3rd trimester male fetus with bilateral hydronephrosis due to posterior urethral valves. There is both renal pelvis and calyceal dilatation. Minimal residual renal parenchyma is visible and is echogenic, concerning for dysplasia.* ***(Right)*** *Coronal ultrasound of the same fetus shows one of the hydronephrotic kidneys and a distended bladder . The typical "keyhole" appearance represents the dilated prostatic urethra.*

Posterior Urethral Valves

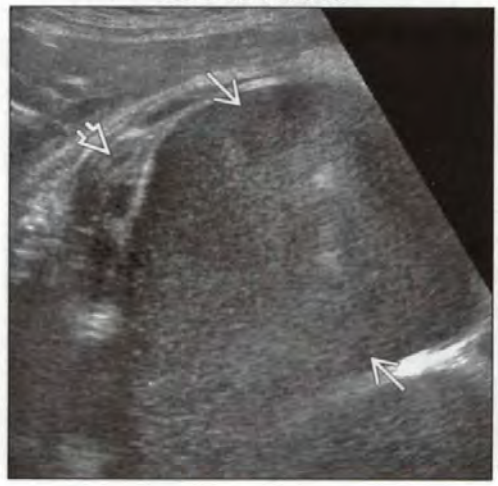


Posterior Urethral Valves

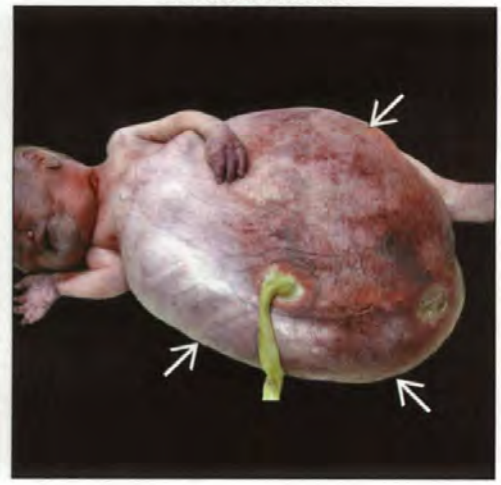


(Left) *Axial ultrasound shows a 3rd trimester fetus with urethral atresia. The bladder is massively distended, and there is associated anhydramnios. The kidneys are dysplastic due to long-standing obstruction.* ***(Right)*** *Clinical photograph shows the massively distended abdomen in the same preterm stillborn infant with urethral atresia. Vesicocentesis, with removal of 2.5 liters of fluid, was required in order to effect vaginal delivery.*

Urethral Atresia



Urethral Atresia



(Left) *Clinical photograph shows the lax abdominal musculature and undescended testes in this infant with prune belly syndrome. The distended bladder can also be appreciated .* ***(Right)*** *Coronal ultrasound shows the thorax and abdomen in a mid-trimester fetus with hydrops associated with trisomy 21. The classic features of hydrops including ascites , pleural effusions , and skin edema are noted.*

Prune Belly Syndrome



Hydrops



LARGE ABDOMINAL CIRCUMFERENCE

Ascites, Massive

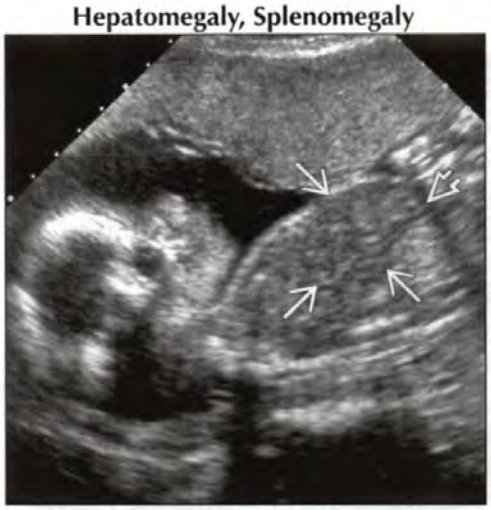


Mesoblastic Nephroma



(Left) *Coronal ultrasound shows an enlarged abdomen in an early 3rd trimester fetus with massive ascites. The etiology of the ascites was not clear, and no other evidence of hydrops was seen.* ***(Right)*** *Coronal ultrasound shows a fetus with a large, homogeneous-appearing mesoblastic nephroma. Note the mass effect on the aorta.*

Hepatomegaly, Splenomegaly



Hepatomegaly, Splenomegaly



(Left) *Sagittal ultrasound shows a 22 week fetus with hepatomegaly. The liver extends to the fetal pelvis. The liver parenchyma is somewhat "speckled" in appearance, concerning for intrauterine viral infection.* ***(Right)*** *Coronal ultrasound in the same fetus confirms hepatomegaly. The liver appearance is likely due to edema with developing intraparenchymal calcifications, both features of infection.*

Liver Tumors



Liver Tumors



(Left) *Coronal ultrasound shows a large mass that involves most of the liver. There are multiple vascular channels, which showed flow on color Doppler (not shown).* ***(Right)*** *Clinical photograph shows the obvious hepatomegaly, as demonstrated by the clinician's finger, in the same preterm infant with a massive liver hemangioendothelioma.*

HEPATOMEGALY

DIFFERENTIAL DIAGNOSIS

Common
- Hydrops
  - Immune Hydrops
  - Nonimmune Hydrops
- Infection

Less Common
- Beckwith-Wiedemann Syndrome
- Trisomy 21

Rare but Important
- Liver Tumors
  - Hemangioendothelioma
  - Mesenchymal Hamartoma
  - Hepatoblastoma
  - Leukemia
  - Neuroblastoma Metastases
- Glycogen Storage Disorder

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is the liver diffusely enlarged or is there a focal mass(es)?
  - Liver occupies a significant portion of the abdomen so an enlarged abdominal circumference may be first sign of diffuse enlargement
  - If focal mass, consider tumors (except leukemia)
    - Cystic mass more likely mesenchymal hamartoma
    - Marked vascularity more likely hemangioendothelioma
- Are there calcifications (in the liver and elsewhere) that would suggest infection?
- Is there organ overgrowth or other obvious anomalies?
- Look for signs of hydrops
  - Hepatomegaly may be one of first signs before florid hydrops develops
  - Skin/subcutaneous edema
  - Ascites
  - Pleural effusions
  - Pericardial effusion
  - Other findings
    - Placentomegaly (placental thickness > 40 mm)
    - Polyhydramnios

Helpful Clues for Common Diagnoses
- **Hydrops**
  - Vascular congestion may cause hepatomegaly
    - May occur before other more obvious signs
  - Important to recognize and institute treatment (if possible) before onset of hydrops, which often portends a poor outcome
  - **Immune Hydrops**
    - Maternal antibodies cross placenta and cause lysis of fetal red blood cells, leading to fetal anemia
    - Spleen often also involved (hepatosplenomegaly) but may be difficult to differentiate from liver
    - Hepatosplenomegaly not only from high cardiac output, but also may be secondary to extramedullary hematopoiesis in prolonged anemia
    - Anemia causes an elevated middle cerebral artery (MCA) peak systolic velocity (PSV)
  - **Nonimmune Hydrops**
    - Multiple causes, most of which have obvious other findings
    - Cardiac: Structural or arrhythmia
    - Fetal masses: Either vascular masses with arteriovenous shunting or masses which obstruct venous return
    - Placental chorioangioma
    - Aneuploidy
    - Twin-twin transfusion syndrome (TTTS)
- **Infection**
  - Cytomegalovirus (most common in utero infection), toxoplasmosis, parvovirus, varicella, HIV
  - Look for punctate, non-shadowing calcification within liver
    - May see calcifications elsewhere: Brain (periventricular, cortical), diaphragm, pericardium
  - Usually other findings also present
    - Ventriculomegaly, echogenic bowel, cardiomegaly, growth restriction, polyhydramnios, hydrops

Helpful Clues for Less Common Diagnoses
- **Beckwith-Wiedemann Syndrome**
  - Organomegaly, primarily hepatosplenomegaly and nephromegaly

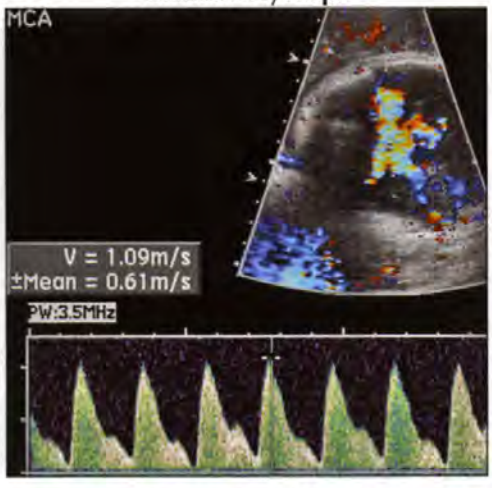
- Macroglossia
- Hemihypertrophy
- Omphalocele rarely

- **Trisomy 21**
  - Hepatomegaly may be from nonimmune hydrops or myeloproliferative disorder
    - Transient myeloproliferative disorder: Variable spectrum of severity
    - Congenital leukemia: Extensive organ involvement
  - Other markers of trisomy 21 more obvious

Helpful Clues for Rare Diagnoses

- **Hemangioendothelioma**
  - Variable appearance: Hypoechoic, hyperechoic, or mixed echogenicity
  - Increased flow on color Doppler
    - Flow void described on fetal MR
  - Hydrops common, resulting from two possible etiologies
    - Arteriovenous shunting
    - Kasabach-Merritt sequence: Hemolytic anemia, thrombocytopenia, and consumptive coagulopathy
- **Mesenchymal Hamartoma**
  - Benign, predominately cystic or mixed echogenicity liver tumor
  - Cysts vary in size from a few millimeters to several centimeters
    - Multiple small cysts may create "swiss cheese" appearance
  - Cysts may have internal septations
  - Avascular or hypovascular on color Doppler
- **Hepatoblastoma**
  - Malignant, solid, echogenic mass
  - Pseudocapsule around lesion creates well-defined borders
  - "Spoke-wheel" appearance described with alternating hypo- and hyperechoic areas
  - Color Doppler shows flow but not as vascular as hemangioendotheliomas
  - Calcifications occasionally seen
  - Can have spontaneous hemorrhage
- **Leukemia**
  - Hepatosplenomegaly most common finding
    - Result of either hydrops or extramedullary hematopoiesis
- **Neuroblastoma Metastases**
  - Most common primary fetal tumor to metastasize to liver
    - Other metastatic fetal tumors exceedingly rare
  - 25% of neuroblastoma cases have liver metastases
  - May be either infiltrative or focal
  - Look for suprarenal mass (primary tumor)
    - Solid neuroblastomas are more likely to metastasize than cystic ones
- **Glycogen Storage Disorder**
  - Gaucher disease (perinatal-lethal subtype)
    - Hepatosplenomegaly
    - Hydrops
    - Hypokinesia/arthrogryposis
    - Ichthyosis
    - Facial dysmorphism

Immune Hydrops



Pulsed Doppler ultrasound in a 34 week fetus with Rh incompatibility who had several prior transfusions. The MCA shows a markedly elevated peak systolic velocity of 109 cm/sec.

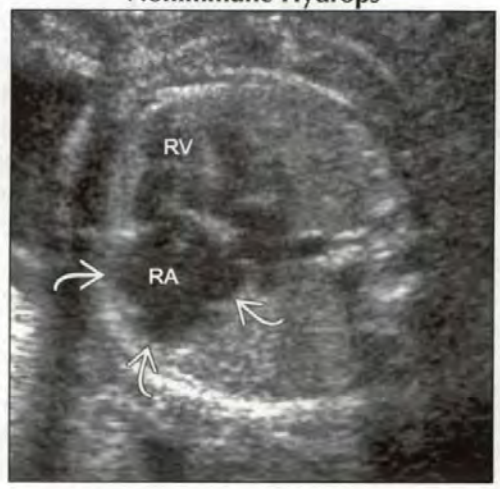
Immune Hydrops



Further imaging in the same fetus shows hepatomegaly ➡ a sign of early hydrops or extramedullary hematopoiesis. Given these findings & a nonreassuring cardiotocogram, the patient was delivered.

HEPATOMEGALY

Nonimmune Hydrops

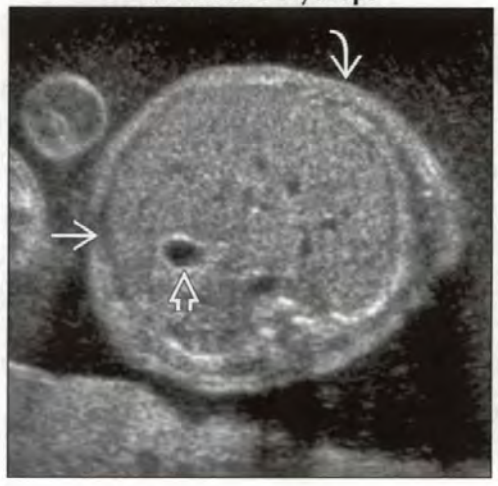


Nonimmune Hydrops

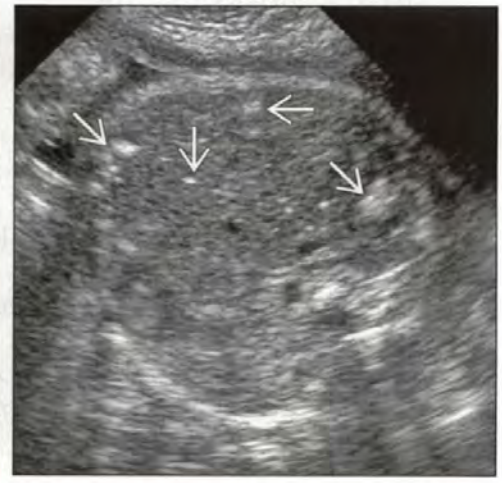


(Left) *Four chamber view shows an abnormal cardiac axis and cardiomegaly. The right atrium (RA) is significantly enlarged compared to the rest of the chambers, the result of pulmonary stenosis and tricuspid regurgitation. Right ventricle (RV).* ***(Right)*** *Coronal oblique ultrasound in the same case shows hepatomegaly. Tricuspid regurgitation led to the hepatic congestion and enlargement, an early sign of impending hydrops (enlarged right atrium).*

Nonimmune Hydrops



Infection



(Left) *Axial ultrasound shows hepatomegaly in the recipient twin in a TTTS case. The stomach is compressed by the markedly enlarged liver. There is also skin thickening and a thin rim of ascites.* ***(Right)*** *Axial oblique ultrasound shows hepatomegaly with multiple, non-shadowing, hyperechoic foci scattered throughout the liver. This finding is highly suggestive of infection.*

Beckwith-Wiedemann Syndrome

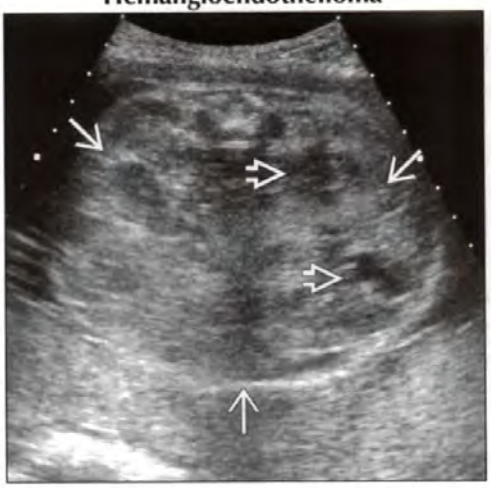


Beckwith-Wiedemann Syndrome

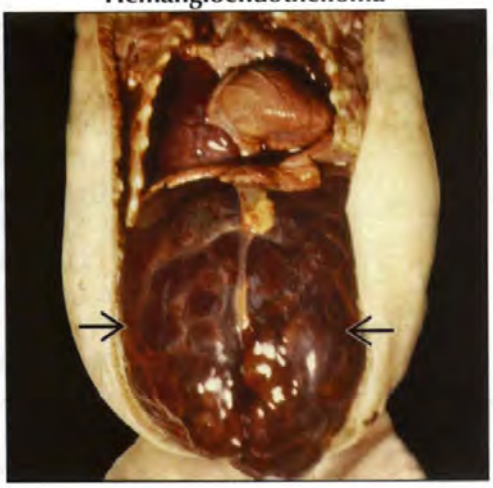


(Left) *Sagittal ultrasound shows macroglossia in a case of Beckwith-Wiedemann syndrome. The fetal tongue is enlarged and persistently protrudes from the mouth.* ***(Right)*** *Axial ultrasound in the same case shows the bowel displaced to the middle of the abdomen because both the spleen and liver are enlarged. In addition, there is a small amount of ascites (calipers).*

Hemangioendothelioma



Hemangioendothelioma



(Left) *Axial ultrasound shows a massive, heterogeneous liver mass. Note several cystic-appearing areas within this mass. Always use color Doppler to look for flow within these areas.* ***(Right)*** *Gross pathology at autopsy shows the liver is grossly enlarged and completely replaced by a lobular, vascular mass.*

Mesenchymal Hamartoma



Hepatoblastoma



(Left) *Axial T1WI MR with contrast shows a hypointense, multiseptated, cystic hepatic mass in a newborn. A right upper quadrant, avascular, cystic lesion was seen in utero. Cysts may vary from large, as in this case, to just a few millimeters giving the liver a "swiss cheese" appearance.* ***(Right)*** *Coronal ultrasound shows a large, solid, well-defined mass in the fetal liver. There was only moderate vascularity. These findings are typical of hepatoblastoma.*

Neuroblastoma Metastases



Neuroblastoma Metastases



(Left) *Axial ultrasound shows hepatomegaly with multiple subtle lesions throughout the liver. Other images showed a solid adrenal mass, which was the primary tumor. There was also hydrops (ascites), polyhydramnios, and placentomegaly.* ***(Right)*** *Gross pathology shows the liver studded with metastases. Liver metastases are rare in fetuses but, when they occur, they are most commonly from neuroblastoma.*

CYSTIC ABDOMINAL MASS

DIFFERENTIAL DIAGNOSIS

Common

- Urinary Tract
  - Multicystic Dysplastic Kidney (MCDK)
  - Ureteropelvic Junction Obstruction
  - Enlarged Bladder
  - Urinoma
- Gastrointestinal Tract
  - Bowel Atresia
  - Meconium Pseudocyst

Less Common

- Ovarian Cyst
- Lymphangioma
- Enteric Duplication Cyst

Rare but Important

- Choledochal Cyst
- Neuroblastoma
- Fetus-in-Fetu, Teratoma
- Urachal Anomalies
- Cloacal Malformation, Hydrocolpos

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Can the cystic mass be localized to a normal structure?
  - Most abdominal cystic masses are from the urinary tract
  - Gastrointestinal tract next most common
- Is it a simple cyst or a complex cystic mass?
  - Septations, internal echogenic debris
- What are the wall characteristics?
  - Thin-walled, thick-walled, calcified, "gut signature"
- Is it constant or does it change appearance during the exam, between exams?

Helpful Clues for Common Diagnoses

- **Multicystic Dysplastic Kidney (MCDK)**
  - Multiple cysts of varying sizes with no discernible renal parenchyma
  - Reniform shape is lost
  - Variable in utero course: May involute, remain stable, or grow
  - May be massive and cross midline
- **Ureteropelvic Junction Obstruction**
  - May present as large cyst if severe obstruction
  - Look for communication with dilated calyces
- **Enlarged Bladder**
  - Posterior urethral valves most common cause
    - Look for "keyhole" appearance created by the dilated posterior urethra
  - Prune belly syndrome and urethral atresia less common causes
  - Hydronephrosis also commonly seen
- **Urinoma**
  - Spontaneous rupture of the renal collecting system into retroperitoneum
  - Look for contained fluid collection adjacent to obstructed kidney
- **Bowel Atresia**
  - Can occur anywhere along gastrointestinal tract
  - Has a tubular, "sausage-shaped" appearance
  - Peristalsis within a cystic mass is pathognomonic
- **Meconium Pseudocyst**
  - Wall-off bowel perforation
  - Irregular, thick walls
  - Look for other signs of meconium peritonitis
    - Intraperitoneal calcifications
    - Dilated bowel
    - Ascites

Helpful Clues for Less Common Diagnoses

- **Ovarian Cyst**
  - Top consideration for a unilocular cyst in a 3rd trimester female fetus
  - "Daughter cyst" sign
    - Small cyst along wall of dominant cyst
    - Highly specific (up to 100%) sign for ovarian origin (82% sensitive)
  - May have occasional septations
  - If appearance becomes complex, with internal echoes, then there is concern for torsion
  - Occasionally found in upper abdomen
    - Supporting ligaments are lax, allowing for displacement
  - May occasionally be bilateral
- **Lymphangioma**
  - Thin-walled cystic mass
  - May be unilocular or multilocular, with one or multiple septations
  - Can be very complex, insinuating around organs and extending out of abdomen
  - Variable echogenicity of fluid, but usually anechoic

- **Enteric Duplication Cyst**
  - Solitary, thick-walled cyst
  - Look for "gut signature"
    - Layered appearance with echogenic mucosa, hypoechoic muscular layer, echogenic serosa
    - Often difficult to see in utero
  - Rarely bowel dilatation from obstruction

Helpful Clues for Rare Diagnoses

- **Choledochal Cyst**
  - Cystic dilatation of extrahepatic &/or intrahepatic bile ducts
  - Unilocular, simple, right upper quadrant cyst is most common presentation in fetus
    - Round in axial plane and fusiform in longitudinal plane
  - Following bile ducts into cyst confirms diagnosis
- **Neuroblastoma**
  - Arises from adrenal gland
  - Approximately 50% are cystic
  - Complex appearance with thick septations
  - Cystic neuroblastoma has an excellent prognosis
- **Fetus-in-Fetu, Teratoma**
  - Overlapping features between these two entities
    - Fetus-in-fetu more developed and must have spinal elements
  - Complex, with a large solid component encapsulated within a cyst
  - Calcifications, including well-formed bones, most specific finding
  - Majority reported in upper retroperitoneum
  - Fetus-in-fetu theoretically result of inclusion of a monochorionic diamniotic twin within a host twin
- **Urachal Anomalies**
  - Includes cysts and patent urachus
  - Communication with bladder confirms patent urachus
  - Bladder may appear elongated with a figure 8 or "waisted" configuration
  - May extend into base of umbilical cord
    - Associated with allantoic cord cysts
  - May resolve as gestation progresses
- **Cloacal Malformation, Hydrocolpos**
  - Persistent cloaca
    - Failure of urorectal septum to reach perineum
    - Seen in female fetuses
    - Results in single perineal opening for urine, genital secretions, and meconium
    - Bladder, vagina, and rectum may all communicate in utero
  - Variable presentation
    - Cystic mass in pelvis
    - Dilated pelvic bowel loops; may see enteroliths from mixing of meconium and urine
    - Hydronephrosis and lumbosacral anomalies may also be present
    - Abnormal genitalia with lack of normal labial/clitoral formation
    - Ascites reported in some cases

Multicystic Dysplastic Kidney (MCDK)



Coronal oblique ultrasound shows that the left kidney ➡ is enlarged and cystic, consistent with a MCDK. Most abdominal cystic masses in the fetus are related to the urinary tract.

Multicystic Dysplastic Kidney (MCDK)



Axial ultrasound shows a cystic mass filling the fetal abdomen. When large, a MCDK can have a confusing appearance. Only one normal kidney ➡ is seen, which is the key to the diagnosis.

CYSTIC ABDOMINAL MASS

Ureteropelvic Junction Obstruction



(Left) *Coronal ultrasound shows classic bilateral ureteropelvic junction obstruction in a 2nd trimester fetus.* ***(Right)*** *Axial ultrasound of the same fetus in the 3rd trimester shows dramatic progression, with massive left renal pelvis distention (calipers). When presenting late, this appearance may be confusing. Careful scanning may show dilated calyces.*

Ureteropelvic Junction Obstruction



Enlarged Bladder

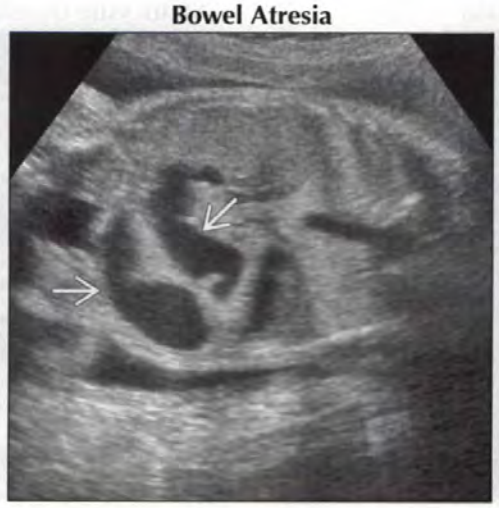


(Left) *Sagittal oblique ultrasound shows marked distention of the bladder in this male fetus with posterior urethral valves.* ***(Right)*** *Axial ultrasound shows a fetus with bilateral UPJ obstruction. There was a unilateral collecting system rupture, partially decompressing the left collecting system. Urine is collecting in the perirenal space. Note how the fluid is surrounding and compressing the kidney, classic features of a urinoma.*

Urinoma

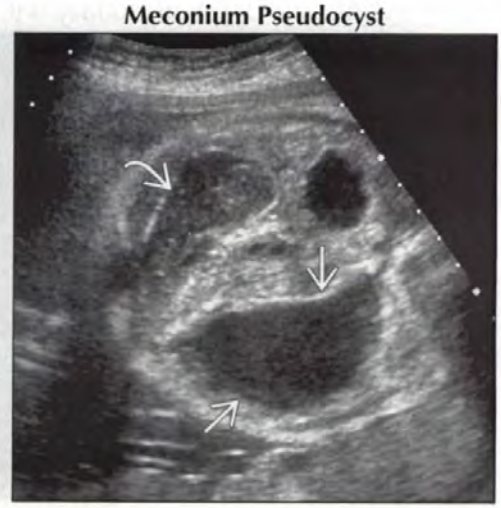


Bowel Atresia



(Left) *Coronal ultrasound shows a tubular, cystic mass in a fetus with jejunal atresia. Seeing peristalsis confirms a cystic mass is bowel.* ***(Right)*** *Axial ultrasound shows a dilated loop of bowel, filled with echogenic material. There has been an in utero perforation with formation of a large, irregular, thick-walled meconium pseudocyst.*

Meconium Pseudocyst



CYSTIC ABDOMINAL MASS

Meconium Pseudocyst



Ovarian Cyst

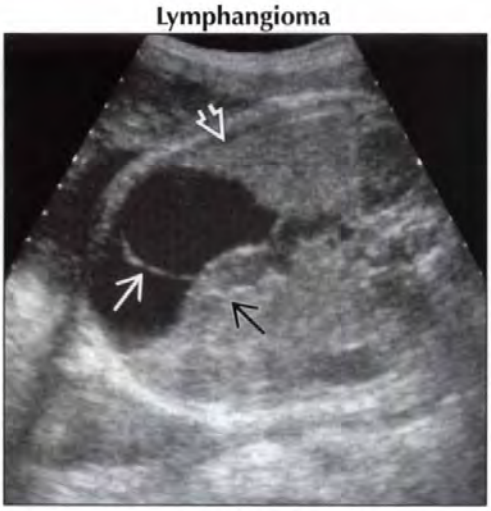


(Left) *Axial ultrasound shows a large, irregular cystic mass. Note the thick, echogenic walls. Surgery confirmed an ileal atresia with perforation, complicated by meconium peritonitis and pseudocyst formation. Pseudocysts will often have an irregular, angular contour.* ***(Right)*** *Coronal ultrasound in a female fetus shows a large cyst above the bladder. Note the small "daughter cyst" along the wall. This sign is highly specific for an ovarian cyst.*

Ovarian Cyst



Lymphangioma



(Left) *Coronal ultrasound of a 3rd trimester female fetus shows bilateral, simple pelvic cysts. These were confirmed to be ovarian cysts. Most ovarian cysts regress by 6 months of age.* ***(Right)*** *Axial oblique ultrasound shows a septation within a thin-walled abdominal cyst. The cyst is insinuated between the liver and colon. The appearance of mesenteric lymphangiomas is quite variable, ranging from a unilocular cyst to a large complex mass.*

Enteric Duplication Cyst



Enteric Duplication Cyst



(Left) *Axial ultrasound shows a thick-walled cyst within the peritoneal cavity. Careful evaluation of the cyst wall should be done looking for a layered appearance ("gut signature"), but this is often difficult in the fetus.* ***(Right)*** *Photograph of the resected ileum shows the thick-walled cyst. Note how the intestinal lumen is narrowed. Duplication cysts can occasionally present with bowel obstruction.*

CYSTIC ABDOMINAL MASS

Choledochal Cyst



Choledochal Cyst



(Left) *Axial ultrasound shows a choledochal cyst located adjacent to the gallbladder. A small bile duct is seen entering the cyst.* ***(Right)*** *Coronal oblique ultrasound in a different patient shows the bile ducts exiting the liver and entering the dilated common bile duct. Fusiform dilatation is the most common fetal presentation of a choledochal cyst. Showing bile ducts entering a right upper quadrant cyst confirms the diagnosis.*

Choledochal Cyst



Neuroblastoma

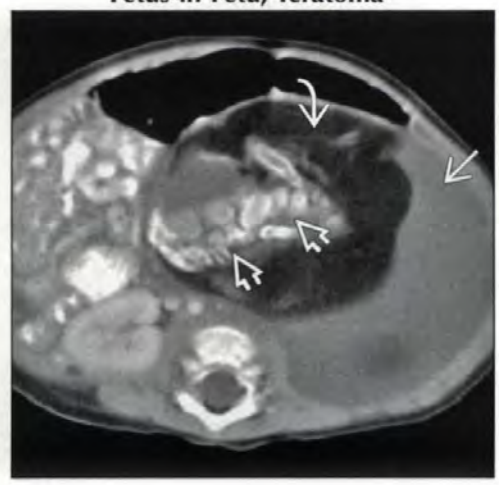


(Left) *Intra-operative photograph shows a large choledochal cyst adjacent to the gallbladder.* ***(Right)*** *Coronal ultrasound of a cystic neuroblastoma shows a complex cystic mass with thick septations, above the kidney. The majority of fetal neuroblastomas arise from the adrenal gland, so location is the key to making the proper diagnosis.*

Fetus-in-Fetu, Teratoma



Fetus-in-Fetu, Teratoma



(Left) *Axial ultrasound shows a cystic mass with a large, central, solid component. Calcifications were also present. Resection after delivery showed a teratoma.* ***(Right)*** *CECT in a newborn shows a complex mass within the left upper quadrant containing fluid, fat, and a well-differentiated spine. This meets the criteria for a fetus-in-fetu, which may result from an aberration in twinning.*

Urachal Anomalies

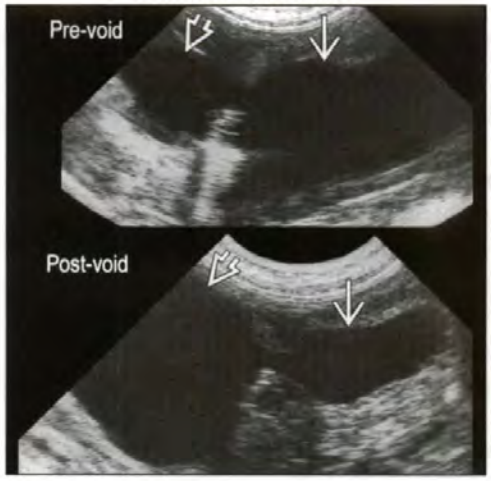


Urachal Anomalies

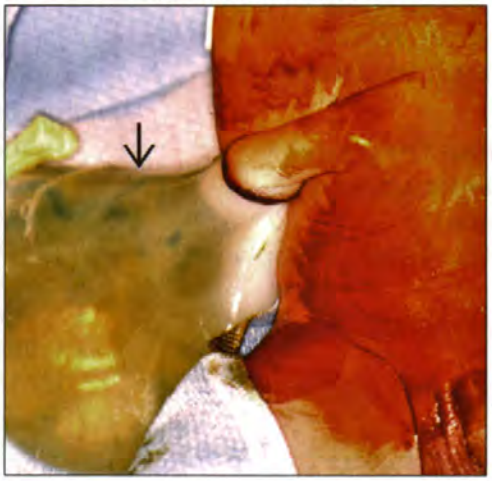


(Left) *Coronal oblique ultrasound shows an unusual configuration of the bladder ⇨: It narrows and then balloons out ⇨ as it extends up toward the umbilicus. This appearance is typical for a patent urachus.* ***(Right)*** *Voiding cystourethrogram in a different patient shows the bladder ⇨ in direct continuity superiorly with the patent urachus ⇨.*

Urachal Anomalies

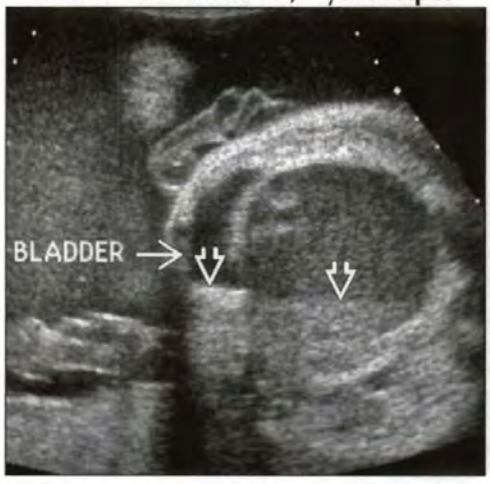


Urachal Anomalies

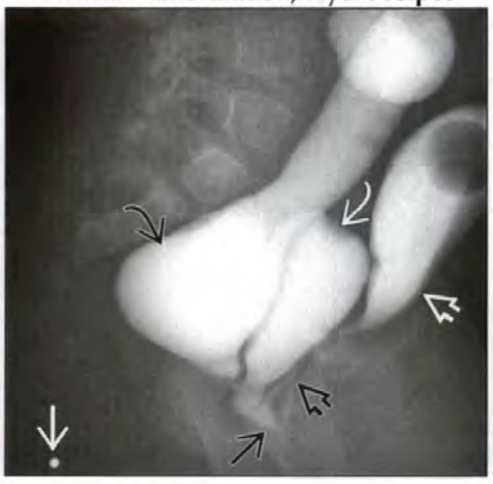


(Left) *Sagittal ultrasound of a newborn with a patent urachus shows the bladder ⇨ communicating with urachus ⇨. Note how, on the post-void image (bottom), the urachus expands as the bladder decompresses. This explains the changing appearance that can be seen in utero.* ***(Right)*** *Intra-operative photograph shows an allantoic cyst ⇨ within the base of the umbilical cord. The cyst is filled with urine and communicates with the bladder via a patent urachus.*

Cloacal Malformation, Hydrocolpos



Cloacal Malformation, Hydrocolpos



(Left) *Axial ultrasound shows the bladder ⇨ being compressed by a large cystic mass, both of which have a fluid-debris level ⇨. A persistent cloaca allows communication between the rectum, vagina, and bladder.* ***(Right)*** *Lateral pre-operative contrast study shows filling of the urethra ⇨, bladder ⇨, vagina ⇨, and rectum ⇨, all communicating with the cloaca ⇨, which opens onto the perineum as a single orifice. The BB ⇨ marks the expected location of the anal sphincter.*

SOLID ABDOMINAL MASS

DIFFERENTIAL DIAGNOSIS

Common
- Bronchopulmonary Sequestration
- Mesoblastic Nephroma

Less Common
- Neuroblastoma

Rare but Important
- Fetus-in-Fetu, Teratoma
- Liver Tumors

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Key to diagnosis is careful anatomic survey to determine origin
  - Kidney, adrenal, liver, not within a defined organ
- Color Doppler assessment important
  - Patterns of perfusion are different
  - Solid masses often have increased vascularity that can result in arteriovenous shunting & hydrops

Helpful Clues for Common Diagnoses
- **Bronchopulmonary Sequestration**
  - 10-15% subdiaphragmatic
  - Majority left-sided, with stomach displaced anteriorly
  - Typically an echogenic, solid mass, but may see cystic areas
  - Look for dominant feeding vessel from aorta
  - Usually presents in 2nd trimester
- **Mesoblastic Nephroma**
  - Large, solid renal mass
  - Look for mass being fed by renal artery
  - Polyhydramnios in ≈ 70%, often severe

Helpful Clues for Less Common Diagnoses
- **Neuroblastoma**
  - May be solid or cystic
  - 60% on right
  - Separate from kidney & adrenal
  - No dominant feeding vessel
  - Usually not seen until 3rd trimester

Helpful Clues for Rare Diagnoses
- **Fetus-in-Fetu, Teratoma**
  - Overlapping features between these two entities
    - Fetus-in-fetu more developed & must have spinal elements
  - Usually not completely solid
    - May have large, solid component encapsulated within a cyst
  - Calcifications, including well-formed bones, most specific finding
  - Superior mesenteric artery is predominant vascular supply
- **Liver Tumors**
  - Generally large, causing hepatomegaly & increased abdominal circumference
  - Hemangioendothelioma most common
    - Large vessels within mass
    - Follow carefully for development of hydrops
  - Hepatoblastoma, leukemic infiltration, & metastases may also appear as solid masses

Bronchopulmonary Sequestration



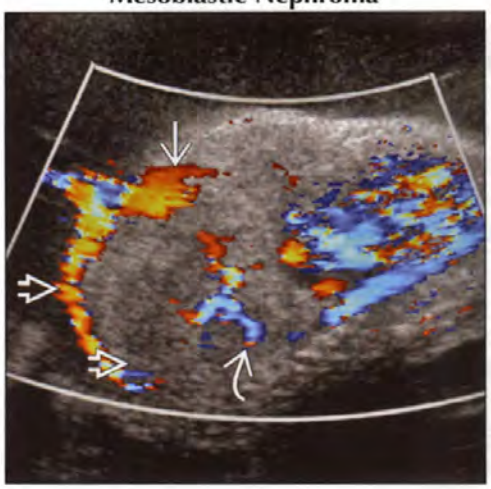
Axial ultrasound in a 2nd trimester fetus shows a small, solid, echogenic mass (calipers) posterior to the stomach.

Bronchopulmonary Sequestration



Coronal power Doppler ultrasound shows two large vessels perfusing a suprarenal mass (kidney). Identifying the vascular supply is important for making the correct diagnosis.

Mesoblastic Nephroma



Mesoblastic Nephroma

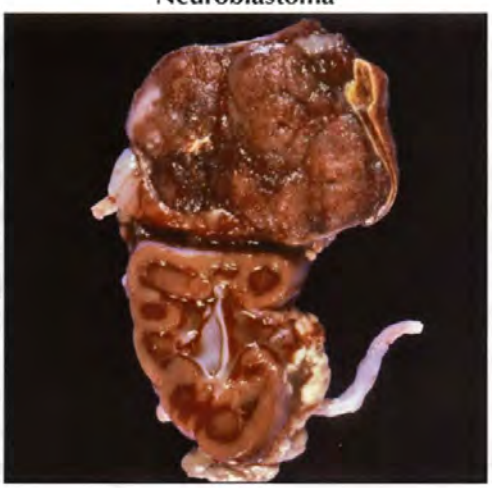


(Left) *Sagittal color Doppler ultrasound shows the umbilical vein ➡ and artery ➡ splayed around a solid abdominal mass. The mass was being supplied by the renal artery ➡. There was also polyhydramnios, which is often a prominent feature.* ***(Right)*** *Coronal ultrasound in a different case shows a large, solid abdominal mass ➡, which extends down to the bladder ➡. No kidney could be identified on that side. Postnatal excision confirmed a mesoblastic nephroma.*

Neuroblastoma

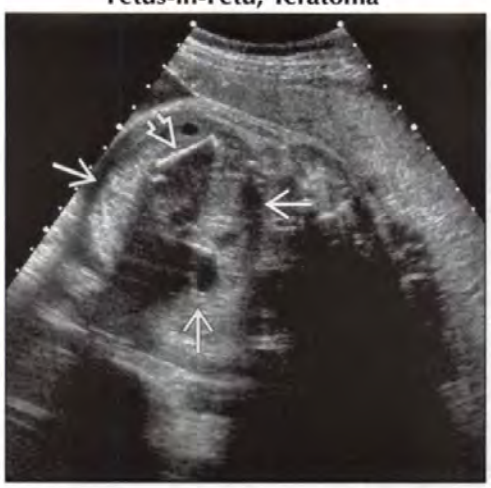


Neuroblastoma



(Left) *Coronal ultrasound shows a solid, echogenic mass (calipers) superior to the right kidney ➡. Note the mass effect that it exerts on the upper pole of the kidney. No dominant perfusing vessel was identified.* ***(Right)*** *Gross pathology in the same case shows the adrenal mass compressing the upper pole of the kidney. Histology confirmed a neuroblastoma, which was confined to the adrenal gland.*

Fetus-in-Fetu, Teratoma



Liver Tumors



(Left) *Axial oblique ultrasound shows a large, complex mass ➡ within the fetal abdomen, which contains a well-formed bone ➡.* ***(Right)*** *Axial T2WI MR shows a large, solid mass ➡ involving the left lobe of the liver. It is higher in signal than the normal liver ➡. This is a large hemangioendothelioma. The infant is being treated conservatively and is doing well.*

MILD PELVIECTASIS

DIFFERENTIAL DIAGNOSIS

Common
- Normal, Idiopathic
- Trisomy 21 (T21)
- Ureteropelvic Junction Obstruction, Early

Less Common
- Duplicated Collecting System

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Routine renal views
  - Axial at level of renal pelvis
    - Measure anterior-posterior renal pelvis diameter (RPD)
  - Longitudinal (sagittal or coronal)
    - Best for renal morphology
    - Look for calyx distention
    - Look for ureter distention
  - Use color Doppler to identify renal arteries
    - Helpful for small or absent kidneys
- Definition of mild pelviectasis (MP)
  - RPD on axial view
    - > 4 mm at 14-22 wks
    - > 5 mm at 22-32 wks
    - > 7 mm after 32 wks
  - RPD should be < 1/3 renal diameter
  - No calyx or ureter distention

Helpful Clues for Common Diagnoses
- **Normal, Idiopathic**
  - 3% of normal fetuses have MP
  - Often resolves in utero
  - Look for other anomalies
- **Trisomy 21 (T21)**
  - 1.6x ↑ risk for T21 when finding present
    - Compare with maternal a priori risk
  - Look for other T21 markers
    - Increased nuchal fold
    - Echogenic bowel
    - Short femur/humerus
    - Echogenic intracardiac focus
- **Ureteropelvic Junction Obstruction, Early**
  - MP can be first sign of ureteropelvic junction (UPJ) obstruction
    - Unilateral or asymmetric MP
  - Findings
    - Progressively ↑ RPD
    - Calyceal dilatation
    - Dilatation ends abruptly at UPJ

Helpful Clues for Less Common Diagnoses
- **Duplicated Collecting System**
  - Separate upper pole and lower pole collecting systems
  - Variable amount of duplication
    - Partial: 2 renal pelves fuse proximally
    - Complete: 2 separate full ureters
  - Weigert-Meyer rule
    - Upper pole with ectopic ureter or ureterocele
    - Ectopic ureter inserts inferior to trigone
    - Lower pole with reflux

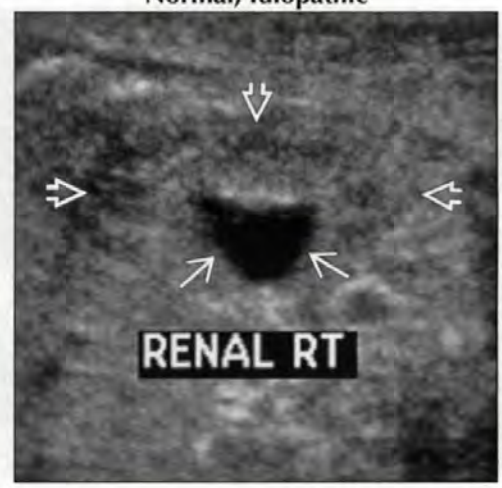
Other Essential Information
- Isolated MP, maternal age, and aneuploidy
  - < 36 yo; 1:303 risk for aneuploidy (0.33%)
  - > 36 yo; 1:45 risk for aneuploidy (2.2%)

Normal, Idiopathic



Axial ultrasound of the renal pelvis diameters shows mild pelviectasis in this 18 week fetus. The finding was isolated and resolved on follow-up.

Normal, Idiopathic



Coronal ultrasound shows renal pelvis distention ➡. Otherwise normal kidney morphology ➡ is seen well on this coronal view. Note the lack of calyx and ureter distention.

Trisomy 21 (T21)

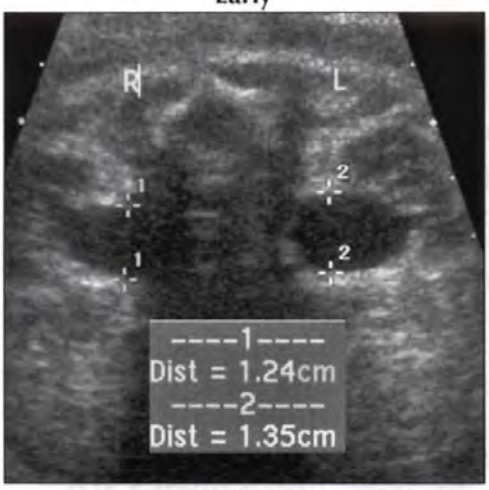


Trisomy 21 (T21)

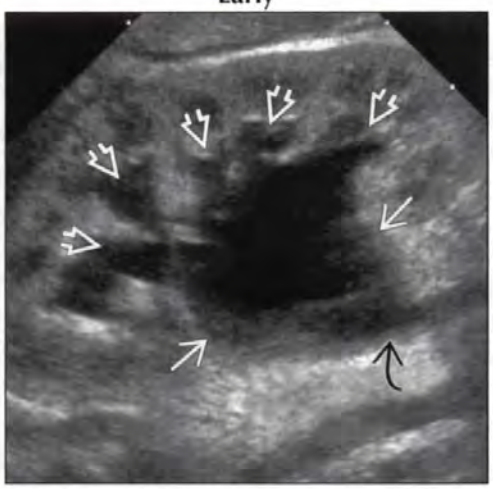


(Left) *Axial ultrasound shows bilateral mild renal pelvis distention. **(Right)** Axial ultrasound through the fetal heart, in the same case, shows a large atrioventricular septal defect. The fetus also had clinodactyly and sandal gap foot deformity. The mild pelviectasis, hand, and foot findings are considered minor markers for T21, while the atrioventricular septal is a major cardiac defect, also associated with T21.*

Ureteropelvic Junction Obstruction, Early



Ureteropelvic Junction Obstruction, Early

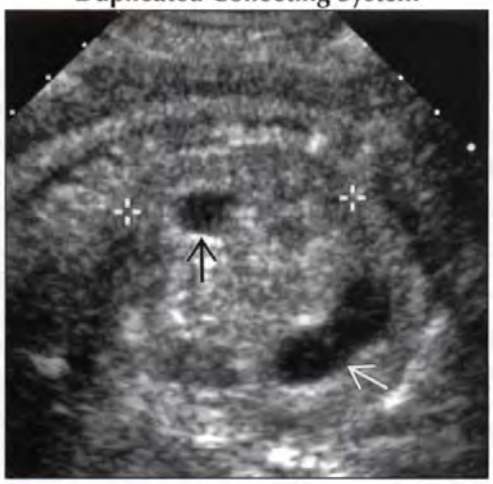


(Left) *Axial ultrasound of a 3rd trimester fetus shows bilateral renal pelvis distention without significant calyceal distention. Postnatal ultrasound was recommended. **(Right)** Coronal ultrasound of the same fetus as a neonate shows progressive hydronephrosis. The renal pelvis is markedly distended, and there is an abrupt narrowing at the UPJ. Moderate calyceal distention is also present. UPJ obstruction often initially presents as mild pelviectasis.*

Duplicated Collecting System



Duplicated Collecting System



(Left) *Coronal ultrasound shows a distended upper pole renal pelvis. Notice the large amount of lower renal tissue that is not drained by the upper pole collecting system. **(Right)** Sagittal oblique ultrasound in another fetus shows a distended lower moiety of the left kidney. The stomach is also seen. While upper pole distention is more common, isolated lower pole MP can be seen with duplicated kidneys. The lower pole system is more prone to reflux.*

FETAL HYDRONEPHROSIS

DIFFERENTIAL DIAGNOSIS

Common
- Ureteropelvic Junction Obstruction
- Posterior Urethral Valves (PUV)

Less Common
- Duplicated Collecting System with Obstruction
- Ureterovesicle Junction Obstruction
- Ureterocele
- Vesicoureteral Reflux

Rare but Important
- Prune Belly Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Renal pelvis diameter (RPD) distention
  - Obtain midrenal axial view
    - Measure anterior-posterior RPD
  - Abnormal RPD measurements
    - > 3 mm in first trimester
    - > 4 mm at 14-22 wks
    - > 5 mm at 22-32 wks
    - > 7 mm after 32 wks
    - RPD > 1/3 renal diameter
  - Use coronal and sagittal views
    - Rule out calyceal/ureteral distention
- Evaluate collecting system methodically to determine point of obstruction
  - Pelviectasis
    - ↑ Renal pelvis only (normal calyces)
  - Hydronephrosis
    - ↑ Renal pelvis + caliectasis
  - Hydroureter
    - Hydronephrosis + dilated ureter
  - Persistent bladder distention
  - Urethra distention
    - Best seen on sagittal view

Helpful Clues for Common Diagnoses
- **Ureteropelvic Junction Obstruction**
  - Hydronephrosis without ureter distention
    - Partial or complete obstruction
  - Ultrasound findings
    - ↑ Renal pelvis ends abruptly at ureteropelvic junction (UPJ)
    - "Bullet-shaped" renal pelvis
    - Distended calyces
  - Associated anomalies
    - 10% bilateral UPJ
    - 25% contralateral renal anomaly
    - 10% non-genitourinary anomaly
  - Etiology
    - Accessory crossing vessel in 1/3
    - Abnormal muscle layer at UPJ
    - Abnormal neural innervation at UPJ
- **Posterior Urethral Valves (PUV)**
  - Obstructive posterior urethral membrane
    - Acts as valves
    - Partial or complete obstruction
    - Seen in male fetuses only
  - "Keyhole" bladder is hallmark finding
    - Distended bladder + distended posterior urethra
    - Bladder wall often thick
    - Bladder may be massive
  - Variable hydronephrosis/hydroureter
  - Variable oligohydramnios
    - Partial vs. complete obstruction
    - Renal functioning ability
    - Pulmonary hypoplasia if severe/early
  - Associated anomalies in 43%
    - VACTERL association
    - Cardiac malformations

Helpful Clues for Less Common Diagnoses
- **Duplicated Collecting System with Obstruction**
  - Duplicated renal parenchyma
    - Separate upper and lower pole moieties
  - Variable collecting system duplication
    - Complete = 2 separate ureters
  - Upper pole drained by ectopic ureter
    - Ectopic ureterocele at distal end
    - Upper pole ureter inserts inferior and medial to lower pole ureter (Weigert-Meyer rule)
    - Upper moiety hydronephrosis common
  - Lower pole ureter refluxes
    - Milder lower moiety hydronephrosis
- **Ureterovesicle Junction Obstruction**
  - Primary megaureter
  - Congenital stenosis at ureterovesicle junction (UVJ)
    - Hypoplasia/atrophy muscle fibers
    - Paucity of ganglion cells
  - Prenatal ultrasound clue
    - Hydroureter without ureterocele or duplication
  - Differential diagnosis
    - Vesicoureteral reflux
    - Non-visualized ureterocele

- **Ureterocele**
  - Cystic dilatation of distal submucosal ureter at bladder insertion
    - Partial or complete obstruction
    - Bilateral in 10%
  - Prenatal ultrasound findings
    - Anechoic, thin-walled cyst in bladder
    - Distended ureter "balloons" into bladder
    - Associated hydroureter + hydronephrosis
  - Orthotopic vs. ectopic ureterocele
    - Orthotopic associated with single collecting system
    - Ectopic associated with renal duplication
    - Ectopic 3x more common
- **Vesicoureteral Reflux**
  - Retrograde flow of urine
    - Bladder ⇒ ureter or kidney
  - Ultrasound findings
    - Variable/intermittent hydronephrosis
    - ↑ Dilatation immediately after voiding
  - Definitive diagnosis made after delivery
    - Voiding cystourethrography (VCUG)
    - Nuclear cystography
  - Grading system
    - I: Reflux into ureter only
    - II: Reflux reaches pelvis (normal calyces)
    - III: Mild caliceal blunting
    - IV: Progressive caliceal dilatation
    - V: Dilated tortuous collecting system with severe caliceal dilatation
  - 80% outgrow reflux
  - Surgical treatment for persistent reflux
    - Ureteral reimplantation
    - Endoscopic periureteral injection

Helpful Clues for Rare Diagnoses

- **Prune Belly Syndrome**
  - Three defining features
    - Dramatic collecting system dilatation
    - Deficient abdominal musculature
    - Cryptorchidism
  - Prenatal ultrasound findings
    - Large, thin-walled bladder
    - Bilateral hydroureter and hydronephrosis
    - Ureter often markedly tortuous
    - Diffuse urethral dilatation (no "keyhole")
    - No obvious point of obstruction
    - ± Oligohydramnios
  - Very difficult to differentiate from PUV

Other Essential Information

- Pitfalls
  - Renal pyramids may mimic caliectasis
  - Distended calyces may mimic renal cysts
    - Look for connection with renal pelvis
  - Ureteroceles may prolapse in and out of bladder
  - Large ureteroceles may fill entire bladder and can be missed
    - Especially true after fetal voiding when bladder collapses around ureterocele
- Look for secondary renal cystic dysplasia
  - Hydronephrosis + renal cysts
  - Suggests significant renal damage
  - Can mimic multicystic renal dysplasia
- Obstruction can decompress spontaneously
  - Urinary ascites
  - Urinoma

Ureteropelvic Junction Obstruction



Sagittal ultrasound shows renal pelvis ➡ and calyceal ➡ dilatation in a 2nd trimester fetus. The renal pelvis is "bullet-shaped" as the distention ends abruptly at the UPJ ➡.

Ureteropelvic Junction Obstruction



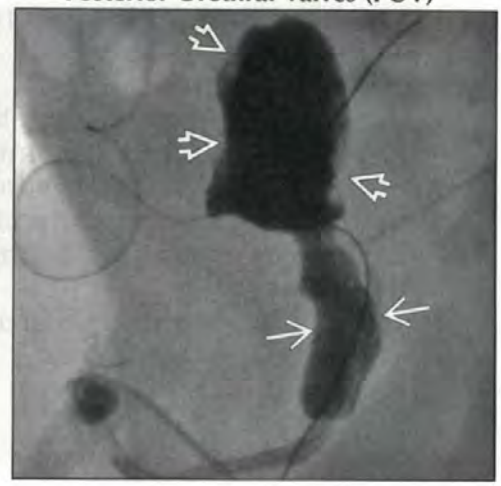
Coronal CECT of a child with a UPJ obstruction shows a crossing vessel ⇨ at the level of the UPJ, causing the massive dilatation of the left renal pelvis ➡. Crossing vessels can be arteries or veins.

FETAL HYDRONEPHROSIS

Posterior Urethral Valves (PUV)



Posterior Urethral Valves (PUV)

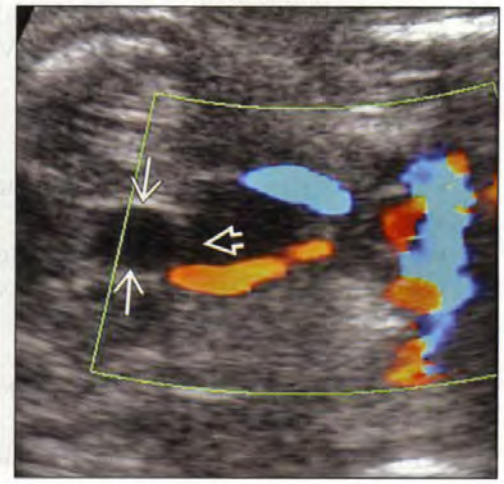


(Left) *Coronal ultrasound shows a markedly distended bladder, as well as bilateral hydroureters and a distended urethra. Focal dilatation of the distal posterior urethra suggests the bladder outlet obstruction is from posterior urethral valves.* ***(Right)*** *Sagittal oblique radiograph during a VCUG in another case shows a distended posterior urethra secondary to posterior urethral valves. Although subtle, bladder wall thickening is also seen.*

Duplicated Collecting System with Obstruction



Duplicated Collecting System with Obstruction



(Left) *Coronal oblique ultrasound shows an obstructed upper pole collecting system. The ureter associated with the upper pole is markedly dilated and tortuous.* ***(Right)*** *Coronal color Doppler ultrasound of the UVJ in the same fetus shows a ureterocele. The distal part of the distended ureter balloons into the bladder. The ectopic ureterocele associated with the upper pole moiety is the cause of the obstruction.*

Ureterovesicle Junction Obstruction



Ureterovesicle Junction Obstruction



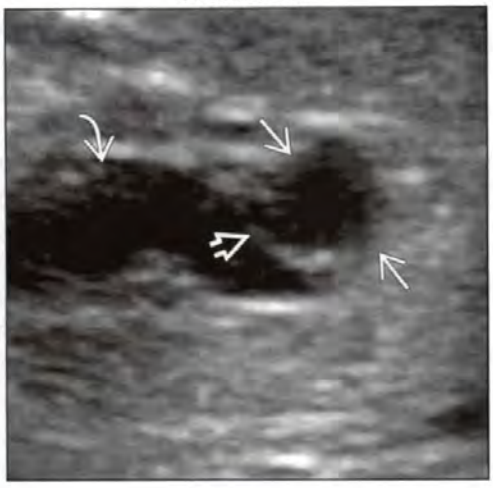
(Left) *Coronal ultrasound shows a single renal collecting system and a distended ureter extending to the bladder.* ***(Right)*** *Coronal ultrasound of the same case, focusing on the UVJ, shows focal narrowing of the distal ureter. There is no ureterocele or renal duplication in this case. Reflux is also within the differential, but was ruled out on postnatal evaluation.*

FETAL HYDRONEPHROSIS

Ureterocele



Ureterocele

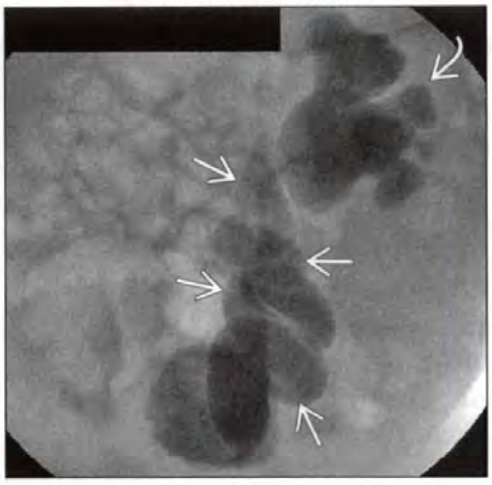


(Left) *Coronal ultrasound shows a non-duplicated, hydronephrotic kidney with ureteral dilatation and a focal "cystic" distention at the UVJ.* ***(Right)*** *Axial ultrasound focused on the partially filled bladder shows the distended distal ureter "ballooning" into the bladder. This is an example of an orthotopic ureterocele, not associated with duplication.*

Vesicoureteral Reflux

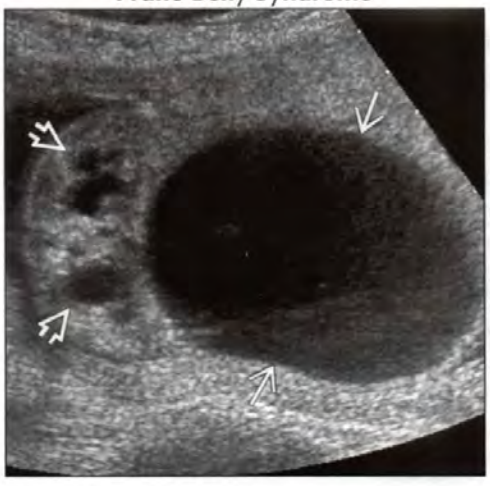


Vesicoureteral Reflux



(Left) *Coronal ultrasound of a neonate with prenatal diagnosis of hydronephrosis confirms calyceal and renal pelvis distention. Immediately after the ultrasound, a VCUG was performed.* ***(Right)*** *Coronal radiograph of the VCUG, obtained immediately after voiding, shows significant reflux. The ureter is distended and tortuous. The renal pelvis and calyces are markedly dilated as well.*

Prune Belly Syndrome



Prune Belly Syndrome



(Left) *Axial ultrasound shows a large bladder and hydronephrosis. Although similar to PUV, the postnatal diagnosis was prune belly syndrome.* ***(Right)*** *Axial color Doppler ultrasound of the bladder and penis in another case shows a vesico-amniotic shunt catheter placed for what was thought to be obstruction from PUV. However, color Doppler at the tip of the penis shows that the baby is urinating easily. Final diagnosis was prune belly syndrome.*

RENAL ENLARGEMENT

DIFFERENTIAL DIAGNOSIS

Common
- Multicystic Dysplastic Kidney (MCDK)
- Autosomal Recessive Polycystic Kidney Disease (ARPKD)

Less Common
- Duplicated Collecting System
- Crossed Fused Ectopia

Rare but Important
- Meckel-Gruber Syndrome
- Beckwith-Wiedemann Syndrome
- Mesoblastic Nephroma
- Compensatory Hypertrophy from Unilateral Renal Agenesis

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- If subjectively large, measure renal length and compare to nomograms
- Ratio of renal circumference to abdominal circumference ranges from 0.27-0.30
  - Ratio remains stable throughout pregnancy
- Are one or both kidneys enlarged?
  - Bilateral disease may be asymmetric
- Are there other anomalies?

Helpful Clues for Common Diagnoses
- **Multicystic Dysplastic Kidney (MCDK)**
  - Multiple, variable-sized cysts in renal fossa
  - Renal length > 95th percentile in 90% of cases
    - May be massive and cross midline
  - Reniform shape is lost
  - Normal renal parenchyma not discernible
  - Unilateral in 80%
    - Contralateral renal anomaly in 40%: Vesicoureteric reflux (most common), ureteropelvic junction obstruction, renal agenesis or hypoplasia
    - Usually normal amniotic fluid unless there is a significant contralateral abnormality
    - Variable in utero course with some involuting and others increasing in size
  - Bilateral in 20%
    - Anhydramnios
    - Lethal anomaly secondary to pulmonary hypoplasia
- **Autosomal Recessive Polycystic Kidney Disease (ARPKD)**
  - Renal enlargement may not occur until mid 2nd trimester but is often progressive later in pregnancy
  - Normal hypoechoic cortex may be present
    - Look for thin hypoechoic rim around echogenic medulla
  - Kidneys are diffusely hyperechoic in severe disease
  - Asymmetric involvement occurs
  - Small macroscopic cysts may be seen
  - Oligohydramnios variable depending on severity of renal involvement
    - Early onset oligohydramnios → poor prognosis
  - Autosomal recessive, with 25% recurrence risk

Helpful Clues for Less Common Diagnoses
- **Duplicated Collecting System**
  - Upper and lower pole moieties separated by band of renal parenchyma
    - Two separate ureters drain upper and lower poles
  - Asymmetric renal size
    - Affected kidney larger than contralateral side (unless both are duplicated)
    - Unilateral renal enlargement may be only clue that duplication is present
  - Upper pole drained by ectopic ureter
    - Ectopic ureter inserts inferior and medial to normotopic ureter (Weigert-Meyer rule)
    - Look for ureterocele in bladder
    - Prone to obstruction
  - Lower pole drained by normotopic ureter
    - Prone to reflux
- **Crossed Fused Ectopia**
  - One empty renal fossa
  - Ectopic kidney crossed to opposite side
    - Creates a large, bilobed kidney
    - May appear as a pelvic mass
  - Left crosses to right more often
    - 95% fused, 5% unfused

Helpful Clues for Rare Diagnoses
- **Meckel-Gruber Syndrome**
  - Triad of classic findings (2 findings required for diagnosis)
    - Renal cystic dysplasia most consistent finding, present in 95-100%
    - Encephalocele in 60-80%

- Postaxial polydactyly in 55-75%
- Renal involvement has variable appearance but often severe
  - Grossly enlarged, echogenic kidneys most common
  - May have macroscopic cysts
  - 10-20x normal size, with enlarged abdominal circumference
  - Bladder may be small or absent
  - Oligohydramnios common and often severe
- Autosomal recessive, with 25% recurrence risk

- **Beckwith-Wiedemann Syndrome**
  - Fetal macrosomia with organomegaly
  - Kidneys frequently involved
    - Renal size enlarged but normal echogenicity and morphology
  - Other prenatal findings
    - Macroglossia
    - Omphalocele
    - Hepatosplenomegaly
    - Hemihypertrophy
  - Definitive diagnosis may not be made until after delivery
- **Mesoblastic Nephroma**
  - Benign mesenchymal renal tumor
  - Solid renal mass on ultrasound
    - Iso- to slightly hyperechoic compared to normal renal parenchyma
    - May rarely have cystic areas
  - May be very large and increase abdominal circumference
  - Polyhydramnios in ≈ 70%, often severe
  - Vascular on Doppler imaging
  - Hydrops may occur with significant arteriovenous shunting or from obstruction of venous return
- **Compensatory Hypertrophy from Unilateral Renal Agenesis**
  - One empty renal fossa
  - Normal kidney compensates for absent kidney
    - Size > 95th percentile
    - Seen in 44% of cases, as early as 22 weeks
  - Fetal adrenal gland is a potential pitfall in diagnosis
    - Fetal adrenal is large and easily mistaken for a kidney, especially in 1st and 2nd trimester
    - In renal agenesis, the adrenal has a flattened, discoid, "lying down" appearance

Alternative Differential Approaches

- Bilateral renal enlargement
  - Autosomal recessive polycystic kidney disease
  - Meckel-Gruber syndrome
  - Beckwith-Wiedemann syndrome
  - Multicystic dysplastic kidneys (in 20%)
- Unilateral renal enlargement
  - Multicystic dysplastic kidney
  - Duplicated collecting system
  - Mesoblastic nephroma
  - Compensatory hypertrophy from unilateral renal agenesis

Multicystic Dysplastic Kidney (MCDK)



Axial ultrasound shows a large, complex mass ➡ with multiple cysts of varying sizes. It has lost its reniform shape and is extending across the midline. The spine ➡ is noted.

Multicystic Dysplastic Kidney (MCDK)



Gross pathology from a different case shows a MCDK ➡. Note the multiple cysts of varying size, with no discernible renal parenchyma. MCDK can become quite large, enlarging the abdominal circumference.

RENAL ENLARGEMENT

Multicystic Dysplastic Kidney (MCDK)



Autosomal Recessive Polycystic Kidney Disease (ARPKD)

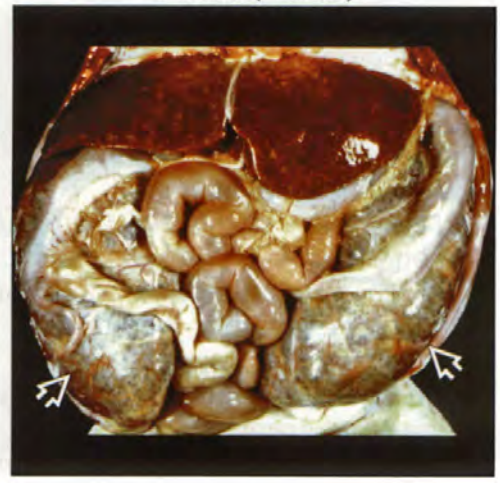


(Left) *Coronal ultrasound shows bilateral, large kidneys ➡ with multiple macroscopic cysts. There is also anhydramnios. When bilateral, MCDK is a lethal malformation.* ***(Right)*** *Coronal ultrasound shows bilaterally enlarged, echogenic kidneys ➡ distending the abdomen. Note how small the chest ➡ appears in comparison. There are scattered, small macroscopic cysts, but the appearance is significantly different from that of bilateral MCDKs.*

Autosomal Recessive Polycystic Kidney Disease (ARPKD)

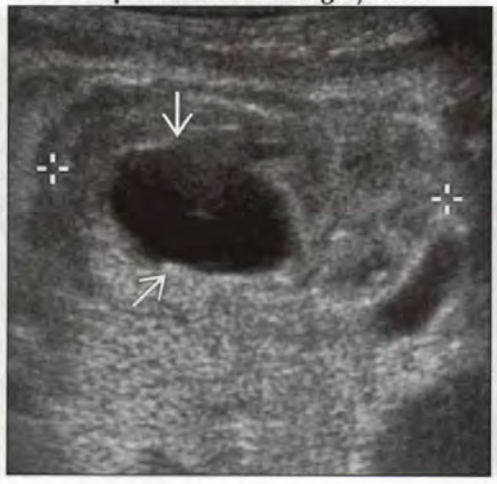


Autosomal Recessive Polycystic Kidney Disease (ARPKD)



(Left) *Axial ultrasound shows bilateral, enlarged, echogenic kidneys ➡. In this case, they are asymmetric in size, which is a variable presentation of ARPKD. Renal enlargement progresses throughout pregnancy.* ***(Right)*** *Gross pathology from an autopsy shows grossly enlarged kidneys ➡ in a case of ARPKD. This fetus had severe oligohydramnios in utero and died of pulmonary hypoplasia.*

Duplicated Collecting System

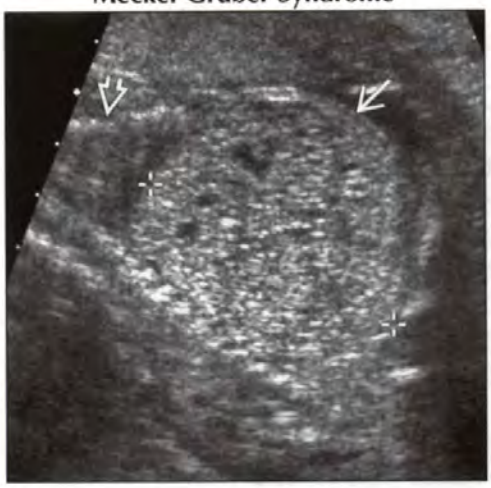


Crossed Fused Ectopia



(Left) *Sagittal ultrasound shows a moderately enlarged kidney (calipers) with hydronephrosis of the upper pole ➡. This is typical of a duplicated collecting system with upper pole obstruction. The bladder should be evaluated for an ectopic ureterocele.* ***(Right)*** *Axial oblique ultrasound shows crossed fused renal ectopia. The left and right kidneys are fused and form a bilobed single kidney ➡. The ectopic crossed kidney may appear as a midline mass.*

Meckel-Gruber Syndrome



Beckwith-Wiedemann Syndrome



(Left) *Sagittal ultrasound shows dramatic renal enlargement (calipers), distending the abdomen ➡ (compare to chest ➡). The appearance is similar to ARPKD, but polydactyly & an encephalocele were also present, which are typical features of Meckel-Gruber syndrome.* ***(Right)*** *Axial ultrasound shows bilateral but slightly asymmetric renal enlargement ➡ in this fetus with Beckwith-Wiedemann syndrome. Note that renal echogenicity & contour are preserved.*

Mesoblastic Nephroma

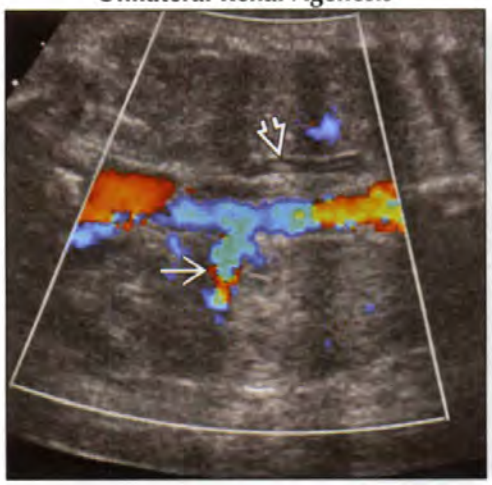


Mesoblastic Nephroma



(Left) *Axial ultrasound shows a large, unilateral, predominately solid mass ➡.* ***(Right)*** *Coronal color Doppler ultrasound of the same mass ➡ shows displacement of the aorta ➡. The mass is supplied by the renal artery ➡, confirming the mass is renal in origin. Polyhydramnios was also present, a common associated finding with mesoblastic nephroma.*

Compensatory Hypertrophy from Unilateral Renal Agenesis



Compensatory Hypertrophy from Unilateral Renal Agenesis



(Left) *Coronal color Doppler ultrasound shows an absent right kidney with a "lying down" adrenal gland ➡ in the right renal fossa. There is a left renal artery ➡, but none is seen on the right.* ***(Right)*** *Coronal ultrasound focused on the solitary left kidney shows a large, but morphologically normal, kidney (calipers). Average renal length at 35 weeks gestation is 3.3 cm. This is compensatory hypertrophy in the setting of unilateral renal agenesis.*

ECHOGENIC KIDNEYS

DIFFERENTIAL DIAGNOSIS

Common

- Autosomal Recessive Polycystic Kidney Disease (ARPKD)
- Obstructive Cystic Dysplasia

Less Common

- Trisomy 13
- Meckel-Gruber Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Are there other anomalies?
  - Trisomy 13 and Meckel-Gruber syndrome have anomalies in addition to the kidneys

Helpful Clues for Common Diagnoses

- **Autosomal Recessive Polycystic Kidney Disease (ARPKD)**
  - Echogenic, large kidneys
  - Normal hypoechoic cortex may be present
    - Look for thin, hypoechoic rim around echogenic medulla
  - Diffusely hyperechoic in severe disease
  - May see small, scattered macroscopic cysts
  - Oligohydramnios variable depending on severity of renal involvement
    - Early onset oligohydramnios → poor prognosis
- **Obstructive Cystic Dysplasia**
  - Chronic obstruction disrupts normal nephron tubular induction
  - Cortical echogenicity often increased from microscopic cysts
  - Macroscopic cortical cysts usually present, but occasionally, kidneys will be appear small and echogenic

Helpful Clues for Less Common Diagnoses

- **Trisomy 13**
  - Cystic dysplasia seen in 50%
    - Kidneys usually echogenic and enlarged; cysts may be visible
  - Multiple major anomalies in > 90%
    - Brain/face: Holoprosencephaly, cyclopia, proboscis, hypotelorism, midline or bilateral cleft lip
    - Body: Postaxial polydactyly, cardiac defects, intrauterine growth restriction
- **Meckel-Gruber Syndrome**
  - Triad of findings (2 findings required for diagnosis)
    - Renal cystic dysplasia most consistent finding, present in 95-100%
    - Encephalocele in 60-80%
    - Postaxial polydactyly in 55-75%
  - Renal involvement has variable appearance, but often severe
    - Grossly enlarged, echogenic kidneys most common
    - May have macroscopic cysts
    - Oligohydramnios common and often severe
    - Appearance may be identical ARPKD so important to look for associated findings

Autosomal Recessive Polycystic Kidney Disease (ARPKD)



Coronal ultrasound shows bilateral, enlarged echogenic kidneys in a second trimester fetus with ARPKD.

Autosomal Recessive Polycystic Kidney Disease (ARPKD)



Axial ultrasound in a 36 week fetus with ARPKD shows renal enlargement, with increased renal medullary echogenicity, but preservation of some normal cortex.

Obstructive Cystic Dysplasia



Obstructive Cystic Dysplasia

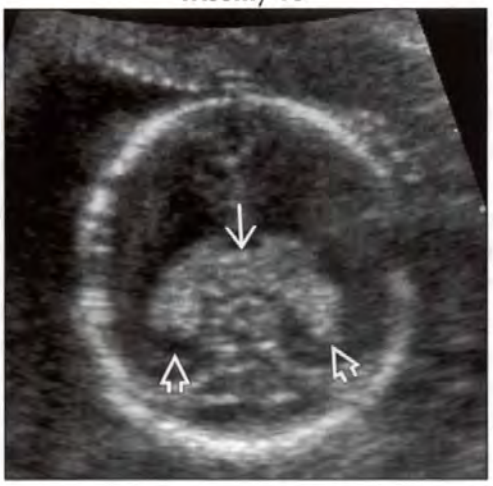


(Left) *Axial ultrasound shows small, echogenic kidneys* ➡. ***(Right)*** *Sagittal ultrasound of the pelvis, in the same case, shows an enlarged bladder* ➡, *a dilated posterior urethra* ➡, *and male genitalia* ➡. *These are typical findings of posterior urethral valves, with resulting renal dysplasia from long-standing obstruction.*

Trisomy 13



Trisomy 13

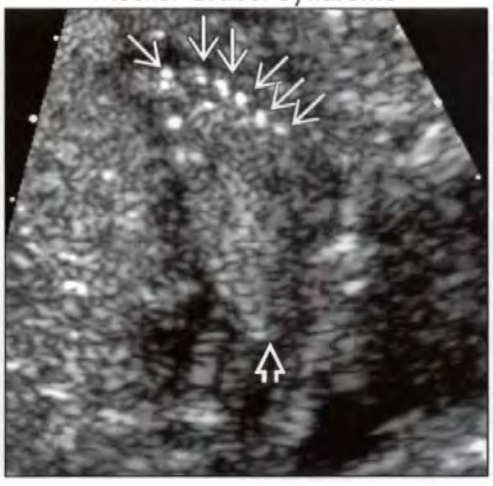


(Left) *Sagittal ultrasound of a second trimester fetus with trisomy 13 shows a mildly enlarged, echogenic kidney* ➡. ***(Right)*** *Axial ultrasound of the brain, in the same fetus, shows semilobar holoprosencephaly, with differentiation of occipital lobes* ➡ *and ventricular communication across the midline* ➡ *anteriorly. Other findings included polydactyly and hypotelorism. Cystic dysplasia is seen in approximately half of all trisomy 13 cases.*

Meckel-Gruber Syndrome



Meckel-Gruber Syndrome



(Left) *Axial ultrasound shows markedly enlarged, echogenic kidneys* ➡. *There is also severe oligohydramnios.* ***(Right)*** *Coronal ultrasound through the foot, in the same case, shows 6 toes* ➡. *The heel* ➡ *is also noted. Additionally, an encephalocele was present. Cystic dysplasia is the most consistent finding in Meckel-Gruber syndrome.*

CYSTIC KIDNEY

DIFFERENTIAL DIAGNOSIS

Common

- Multicystic Dysplastic Kidney (MCDK)
- Hydronephrosis (Mimic)
- Obstructive Cystic Dysplasia
- Duplicated Collecting System with Obstruction (Mimic)

Rare but Important

- Simple Cyst
- Meckel-Gruber Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Do the "cysts" connect?
  - Real-time evaluation is essential for differentiating an obstructed system from true renal cystic disease

Helpful Clues for Common Diagnoses

- **Multicystic Dysplastic Kidney (MCDK)**
  - Multiple, variable-sized cysts in renal fossa
  - Reniform shape is lost
  - Often large, distorting normal abdominal anatomy
  - Variable in-utero course; some involute and others increase in size
- **Hydronephrosis (Mimic)**
  - Distended calyces may appear "cyst-like"
  - Must show that calyces connect with renal pelvis (longitudinal views best)
  - Causes: Ureteropelvic junction (UPJ) or bladder outlet obstruction
- **Obstructive Cystic Dysplasia**
  - Cystic parenchymal change from chronic obstruction
    - Hydronephrosis → cortical cysts
    - Reflects nephron damage and decreased renal function
  - Cysts are often cortical
    - Form in subcapsular nephrogenic zone
  - Kidneys become echogenic
  - Reniform shape generally retained
  - Rarely can appear identical to MCDK
- **Duplicated Collecting System with Obstruction (Mimic)**
  - Upper and lower pole moieties separated by band of renal parenchyma
  - Upper pole prone to obstruction and can appear as cystic mass
  - Look for ureterocele in bladder

Helpful Clues for Rare Diagnoses

- **Simple Cyst**
  - Isolated, unilocular renal cyst
  - Vast majority resolve during pregnancy
    - 4% progress to MCDK
- **Meckel-Gruber Syndrome**
  - Triad of classic findings (2 findings required for diagnosis)
  - Renal cystic dysplasia most consistent finding, present in 95-100%
    - Grossly enlarged, echogenic kidneys most common, but may present with bilateral, large cysts
  - Encephalocele in 60-80%
  - Post-axial polydactyly in 55-75%

Multicystic Dysplastic Kidney (MCDK)



Coronal ultrasound shows a MCDK on the right and a normal kidney on the left. A central cyst could be confused for a renal pelvis, but none of the cysts connected during real-time evaluation.

Hydronephrosis (Mimic)



Coronal ultrasound shows dilated calyces surrounding the renal pelves in a fetus with bilateral UPJ obstructions. Real-time evaluation is essential to show calyces connecting with the pelvis.

CYSTIC KIDNEY

Obstructive Cystic Dysplasia

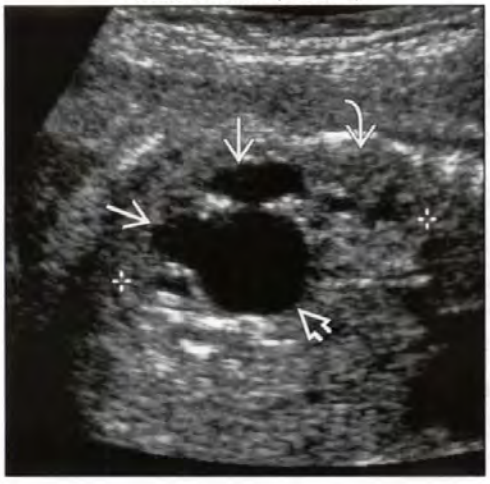


Obstructive Cystic Dysplasia



(Left) *Axial ultrasound shows a UPJ obstruction, resulting in cystic dysplasia. Note the small cortical cysts, indicating nephron damage from the obstruction.* ***(Right)*** *Sagittal ultrasound after delivery shows progression of dysplasia with a generalized increase in echogenicity, loss of normal corticomedullary differentiation, and progression of parenchymal cysts. Note the abrupt, blunted appearance at the UPJ, a classic appearance of UPJ obstruction.*

Duplicated Collecting System with Obstruction (Mimic)



Simple Cyst



(Left) *Coronal ultrasound of the kidney (calipers) shows dilated upper pole calyces connecting with the renal pelvis. The lower pole of the kidney is normal. Always consider a duplicated system when only a portion of the kidney is involved.* ***(Right)*** *Axial ultrasound shows a unilateral, small cyst within what are otherwise normal kidneys. This is generally an isolated, benign finding, but should be followed up to rule out progression.*

Meckel-Gruber Syndrome



Meckel-Gruber Syndrome



(Left) *Axial ultrasound of the kidneys in a case of Meckel-Gruber syndrome shows bilateral enlargement, with the renal parenchyma replaced by multiple, macroscopic cysts. This appearance is less common than enlarged, echogenic kidneys.* ***(Right)*** *Radiograph in the same case shows typical features of Meckel-Gruber syndrome, including an encephalocele, post-axial polydactyly, and a markedly protuberant abdomen from the enlarged, cystic kidneys.*

ABSENT KIDNEY

DIFFERENTIAL DIAGNOSIS

Common

- Renal Agenesis
  - Bilateral Renal Agenesis
  - Unilateral Renal Agenesis

Less Common

- Mimics for Renal Agenesis
  - Pelvic Kidney
  - Crossed Fused Ectopia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Is the kidney truly absent?
  - Always search for ectopic location
- Is the fluid normal?
  - Bilateral renal agenesis will have anhydramnios
  - Remainder have normal fluid
- Fetal adrenal is large and easily mistaken for a kidney, especially in 1st and 2nd trimester
  - Normal adrenal has an "ice cream sandwich" appearance
    - Hypoechoic cortex surrounding a hyperechoic medulla
  - In renal agenesis adrenal has a flattened, discoid, "lying down" appearance
    - Adrenal gland does not fold into "Y" or "tricorn hat" configuration if no kidney

Helpful Clues for Common Diagnoses

- **Bilateral Renal Agenesis**
  - No demonstrable renal tissue
  - No urine in fetal bladder
  - Anhydramnios
  - "Lying down" adrenals in renal fossa, although this may be difficult to see in setting of no fluid
  - Look for renal arteries, but be aware of pitfalls
    - Lumbar arteries can easily be mistaken for renal arteries
  - MR very helpful for confirmation of diagnosis
- **Unilateral Renal Agenesis**
  - One kidney seen, which may show compensatory hypertrophy
  - May see "lying down" adrenal ipsilateral to absent kidney
  - Bladder seen to fill and empty
  - Normal amniotic fluid volume

Helpful Clues for Less Common Diagnoses

- **Pelvic Kidney**
  - Empty renal fossa
  - Kidney in fetal pelvis, superior to bladder
    - May be difficult to see as echogenicity similar to bowel
  - Contralateral kidney is normal-sized
- **Crossed Fused Ectopia**
  - Ectopic kidney located in opposite flank creating a large bilobed kidney
    - 95% fused, 5% unfused
  - Left crosses to right most often

Other Essential Information

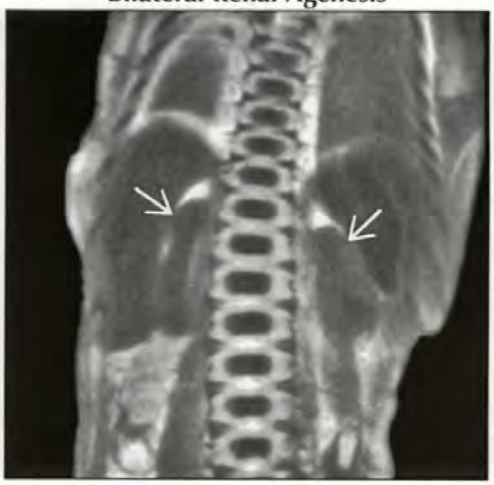
- Uterine anomalies associated with renal anomalies, especially renal agenesis

Bilateral Renal Agenesis



Coronal color Doppler ultrasound of a fetus with anhydramnios, which makes visualization of fetal anatomy difficult. Flow is seen in the aorta ⇨ and iliac arteries ⇨, but no renal arteries were identified.

Bilateral Renal Agenesis



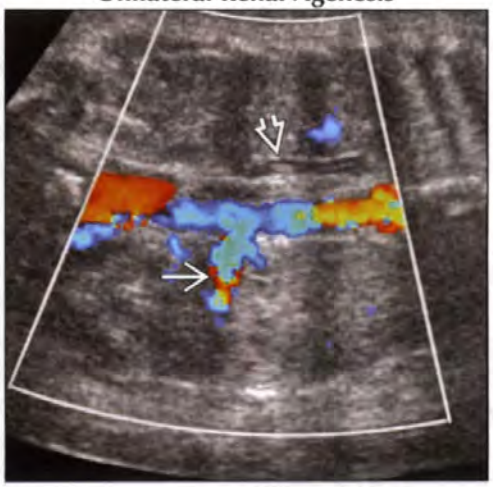
Coronal postmortem T2WI of a fetus with bilateral renal agenesis shows elongated, "lying down" adrenal glands ⇨ in the renal fossa. In the absence of kidneys, the adrenals no longer have the normal "Y" shape.

ABSENT KIDNEY

Unilateral Renal Agenesis



Unilateral Renal Agenesis



(Left) *Axial ultrasound shows the left kidney, which is easily identified by the presence of a small amount of fluid in the renal pelvis. The right renal fossa is filled with liver. The kidney was large for gestational age, typical of compensatory hypertrophy.* ***(Right)*** *Coronal color Doppler ultrasound in the same case shows only the left renal artery arising from the abdominal aorta. There is a "lying down" adrenal gland in the right renal fossa.*

Unilateral Renal Agenesis



Unilateral Renal Agenesis



(Left) *Axial ultrasound shows a single left kidney, with liver in the right renal fossa. This is a female fetus, so ultrasound of the uterus was recommended after delivery.* ***(Right)*** *Postnatal ultrasound of the pelvis in the same patient shows a septate uterus. There are two endometrial cavities separated by a thick septum. The external uterine contour is normal, distinguishing it from either a didelphys or bicornuate uterus.*

Pelvic Kidney



Crossed Fused Ectopia



(Left) *Sagittal oblique ultrasound shows a pelvic kidney, adjacent to the bladder. This was smaller than the orthotopic kidney and difficult to differentiate from surrounding bowel.* ***(Right)*** *Postnatal ultrasound of the right flank, in a different patient, shows a crossed fused ectopic kidney. The ectopic left kidney is fused to the lower pole of the orthotopic right kidney.*

LARGE BLADDER

DIFFERENTIAL DIAGNOSIS

Common
- Normal
- Posterior Urethral Valves (PUV)

Less Common
- Prune Belly Syndrome
- First Trimester Megacystis

Rare but Important
- Urethral Atresia
- Megacystis Microcolon

ESSENTIAL INFORMATION

Helpful Clues for Common Diagnoses
- **Normal**
  - Transient finding with otherwise normal urinary tract & amniotic fluid volume
  - Fetus will usually void during exam
    - Follow-up if bladder fails to decompress
- **Posterior Urethral Valves (PUV)**
  - Urethral membrane acts as valve, resulting in bladder outlet obstruction
  - Occurs exclusively in males
  - "Keyhole" sign: Distended bladder "funnels" into dilated posterior urethra
  - Bladder often thick-walled, with degree of distention depending on severity of obstruction
  - Hydronephrosis common with potential development of renal dysplasia
  - Typically oligohydramnios, or even anhydramnios, in severe obstruction

Helpful Clues for Less Common Diagnoses
- **Prune Belly Syndrome**
  - Triad of dramatic collecting system dilatation, deficiency of abdominal musculature & cryptorchidism
  - Often difficult to differentiate from PUV
  - Look carefully at urethra
    - Entire urethra may be dilated
    - Does not terminate at posterior urethra
    - May see spontaneous voiding
- **First Trimester Megacystis**
  - Bladder length > 7 mm at 10-14 weeks
  - 25% reported to have aneuploidy (trisomy 13, trisomy 18 most common)
  - Of those that are chromosomally normal, 90% regress while 10% progress to obstructive uropathy

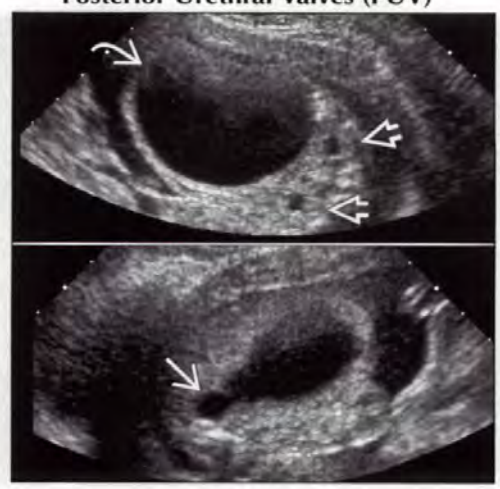
Helpful Clues for Rare Diagnoses
- **Urethral Atresia**
  - Complete obstruction, therefore massive bladder dilatation and anhydramnios
  - Occur in either males or females, but oligohydramnios often precludes ability to determine sex
  - Often indistinguishable from severe PUV
- **Megacystis Microcolon**
  - Dilated bladder with normal to increased amniotic fluid
    - Differentiates it from other causes of large bladder
  - Intestinal hypoperistalsis may result in dilated small bowel
  - More common in females (M:F, 1:4)

Posterior Urethral Valves (PUV)



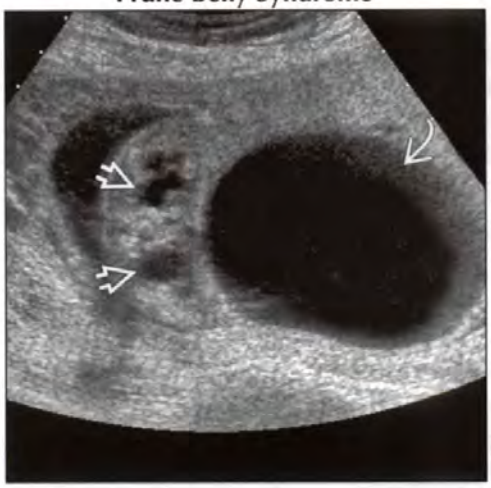
Coronal oblique ultrasound shows a dilated bladder "funneling" into a dilated posterior urethra ➡ ("keyhole" sign). There is also ureteral dilatation ➡ and hydronephrosis ➡.

Posterior Urethral Valves (PUV)



A composite image shows marked bladder distention ➡ with small, echogenic kidneys ➡. The diagnosis is made by angling the transducer towards the bladder base, showing the dilated posterior urethra ➡.

Prune Belly Syndrome



Prune Belly Syndrome

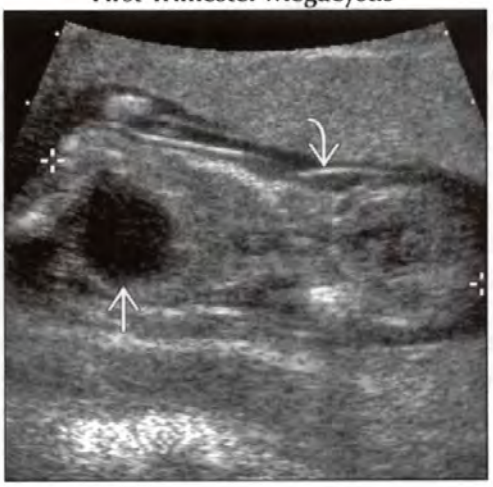


(Left) *Axial oblique ultrasound shows marked bladder distention and bilateral hydronephrosis.* ***(Right)*** *Ultrasound focused on the penis, in the same case, shows dilatation of the entire urethra. There was no "keyhole" configuration as seen with PUV. No testes were seen in the scrotum. These features help to distinguish prune belly syndrome from PUV.*

Prune Belly Syndrome



First Trimester Megacystis

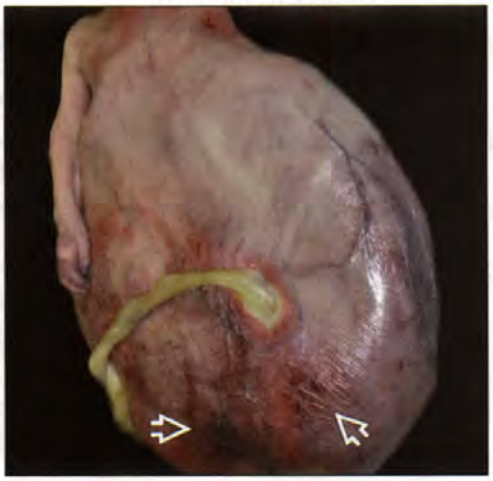


(Left) *Autopsy in a different case shows a virtually translucent abdominal wall secondary to lack of the abdominal musculature. There is a grossly distended bladder, hydronephrosis, & undescended testes, all typical features of prune belly syndrome.* ***(Right)*** *Sagittal ultrasound shows fetal bladder distension & increased nuchal translucency in a fetus with trisomy 18. Approximately 25% of 1st trimester fetuses with megacystis have aneuploidy.*

Urethral Atresia



Urethral Atresia



(Left) *Ultrasound in a 27 week fetus shows massive bladder distention (compare to the femur, which is being measured). There was anhydramnios making visualization of any other fetal structures difficult.* ***(Right)*** *Gross pathology from the same case shows dramatic abdominal distention. The abdominal wall has been stretched so thin, areas were becoming ischemic. Autopsy confirmed urethral atresia.*

ABSENT/SMALL BLADDER

DIFFERENTIAL DIAGNOSIS

Common
- Normal
- Bilateral Renal Anomalies
  - Bilateral Multicystic Dysplastic Kidneys
  - Bilateral Ureteropelvic Junction Obstructions
  - Bilateral Renal Agenesis
- Autosomal Recessive Polycystic Kidney Disease

Less Common
- Severe IUGR
- Twin-Twin Transfusion Syndrome

Rare but Important
- Bladder Rupture
- Bladder Exstrophy
- Cloacal Exstrophy

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Reasons for absent/small bladder
  - Urine is not be produced
    - Primary renal anomaly
    - Poor fetal perfusion leading to poor fetal urine production: Twin-twin transfusion syndrome (TTTS), severe intrauterine growth restriction (IUGR)
    - Bladder not truly absent, just not seen because of lack of distention
  - Bladder is "leaking"
    - Cloacal exstrophy
    - Bladder exstrophy
    - Bladder rupture
- Amniotic fluid assessment is important part of identifying cause of absent/small bladder
  - Oligohydramnios/anhydramnios in singleton
    - Renal agenesis
    - Bilateral multicystic dysplastic kidneys (MCDK)
    - Bilateral uteropelvic junction (UPJ) obstructions
    - Autosomal recessive polycystic kidney disease (ARPKD)
    - Intrauterine growth restriction (IUGR)
  - Twin with oligohydramnios
    - TTTS or any of the above fetal anomalies
  - Normal or increased fluid suggests abdominal wall defect as cause of absent bladder
    - Bladder exstrophy
    - Cloacal exstrophy
- Document bladder on every exam
  - Normal bladder will be flanked on either side by the umbilical arteries
    - Seen best in axial plane

Helpful Clues for Common Diagnoses
- **Normal**
  - Bladder filling/voiding is dynamic & changes during course of examination
    - Recheck at end of exam if bladder is empty at beginning
  - If amniotic fluid & kidneys are otherwise normal, then a pathologic process is unlikely
- **Bilateral Multicystic Dysplastic Kidneys**
  - Renal tissue replaced by cysts of varying sizes
  - Affected kidneys are non-functional
  - Unilateral MCDK has normal bladder & amniotic fluid
  - Bilateral MCDK has no visualized bladder or amniotic fluid
    - Bilateral in 20%
  - Contralateral renal anomaly (non-MCDK) in 40%
    - If severe (e.g., agenesis), may also result in no urine production
- **Bilateral Ureteropelvic Junction Obstructions**
  - Calyceal & pelvis distention which ends abruptly at UPJ
  - ↑ Risk of renal impairment if prenatal AP diameter > 10 mm
  - 10% bilateral
    - Can progress causing complete obstruction with "absent" bladder & anhydramnios
- **Bilateral Renal Agenesis**
  - No demonstrable renal tissue
  - Flattened, discoid adrenals in renal fossa
  - No urine in fetal bladder
  - Anhydramnios
- **Autosomal Recessive Polycystic Kidney Disease**
  - Bilateral, large, echogenic kidneys
    - Normal hypoechoic cortex may be seen

- Look for thin hypoechoic rim around echogenic medulla
- Degree of bladder filling & amniotic fluid is variable depending on severity of disease

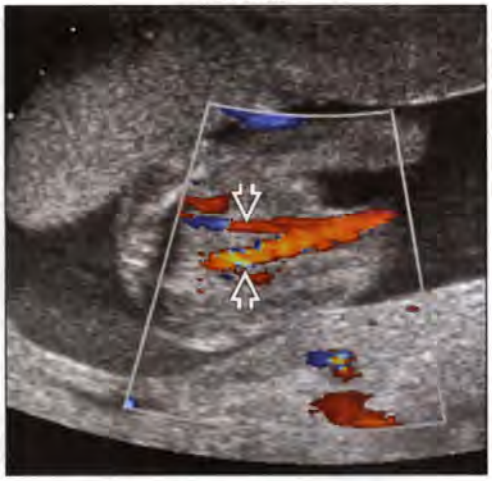
Helpful Clues for Less Common Diagnoses

- **Severe IUGR**
  - Placental insufficiency most common cause
    - Usually late onset, asymmetric IUGR
  - High resistance placental perfusion
    - Blood returning to fetus shunted to cerebral and coronary circulations
    - ↓ Renal perfusion → ↓ urine production → "absent" bladder + oligohydramnios
- **Twin-Twin Transfusion Syndrome**
  - Monochorionic twins with artery-to-vein anastomoses in placenta
  - Donor twin partly perfuses recipient twin
  - Fetuses often discordant in size
    - Donor small, recipient large
  - Donor twin: Oligemic → ↓ urine production → "absent" bladder + oligohydramnios
    - Amniotic membrane tightly adherent, giving a "shrink-wrapped" appearance
    - May appear suspended from uterine wall
  - Recipient twin: Plethoric, polyhydramnios, at risk for hydrops
  - Presence of urine in bladder important in staging TTTS
    - Stage 1: Donor bladder visible, normal Doppler
    - Stage 2: Donor bladder empty, normal Doppler
    - Stage 3: Donor bladder empty, abnormal Doppler
    - Stage 4: Hydrops in recipient
    - Stage 5: Demise of one or both

Helpful Clues for Rare Diagnoses

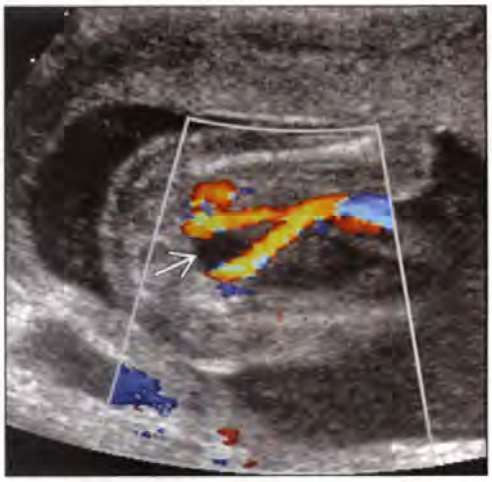
- **Bladder Rupture**
  - Initial ultrasound may show markedly enlarged bladder
  - Rupture results in urinary ascites
  - Thick-appearing bladder wall after decompression
- **Bladder Exstrophy**
  - Failure of abdominal wall closure resulting in exposed posterior bladder wall
  - Absence of bladder on prenatal ultrasound most consistent finding
  - Soft tissue mass/irregularity along abdominal wall, below cord insertion
    - No extruded bowel as in cloacal exstrophy
- **Cloacal Exstrophy**
  - Spectrum of abnormalities resulting from abnormal development of cloacal membrane
  - Absence of normal bladder
  - Lower abdominal wall defect
    - Herniation of bowel between 2 halves of split bladder
    - Omphalocele forms upper part of defect
    - Abnormal genitalia: Males may have bifid scrotum and penis

Normal



Axial color Doppler ultrasound shows bilateral umbilical arteries ⇨. This is the anatomic landmark for the fetal bladder, but none was seen. Normal amniotic fluid and kidneys make an anomaly unlikely.

Normal



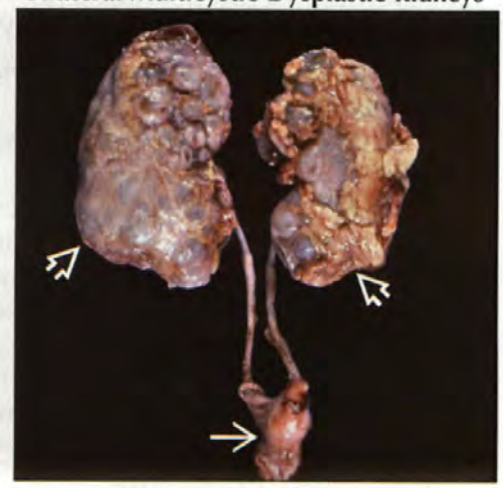
Axial color Doppler ultrasound 7 minutes later shows the bladder ⇨ starting to fill, confirming that this fetus is indeed normal. The bladder often changes appearance during the examination.

ABSENT/SMALL BLADDER

Bilateral Multicystic Dysplastic Kidneys



Bilateral Multicystic Dysplastic Kidneys

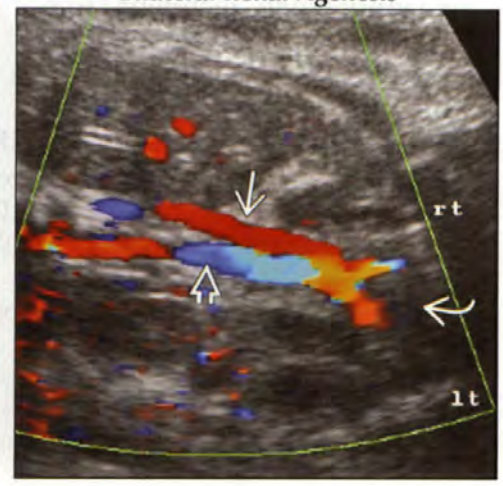


(Left) *Coronal ultrasound shows bilateral, enlarged, cystic kidneys with no fluid-filled bladder in the expected location. MCDK are nonfunctioning, so when bilateral, there is no urine production. Anhydramnios results in pulmonary hypoplasia, which is lethal.* ***(Right)*** *Gross pathology in a different case shows bilateral, large, cystic kidneys. The bladder is atretic. It is present, but not seen, in cases of anuria because of lack of distention.*

Bilateral Ureteropelvic Junction Obstructions



Bilateral Renal Agenesis



(Left) *Axial ultrasound shows bilateral, severe UPJ obstructions resulting in oligohydramnios. UPJ obstructions have a variable in utero course with those > 10 mm more likely to be significant.* ***(Right)*** *Coronal color Doppler ultrasound of the aorta and inferior vena cava shows absent renal arteries and veins. There is anhydramnios and an "absent" bladder.*

Autosomal Recessive Polycystic Kidney Disease



Severe IUGR



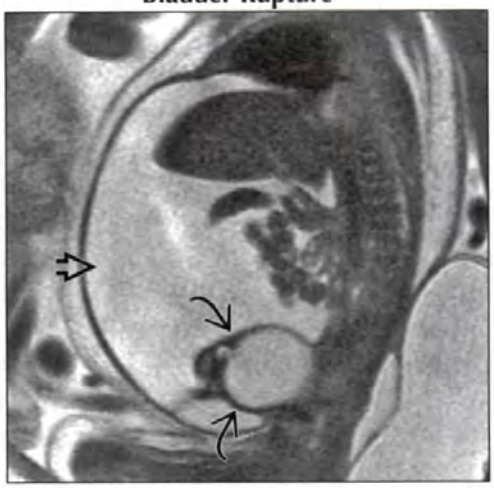
(Left) *Coronal ultrasound shows bilateral, large, echogenic kidneys in a fetus with ARPKD. The renal function is compromised, resulting in an absent bladder and anhydramnios.* ***(Right)*** *Sagittal ultrasound shows a case of severe, early onset, asymmetric IUGR. The spine and pelvis are visible despite anhydramnios; no urine was seen in the bladder. The placenta had massive fibrin deposition, causing severe placental insufficiency.*

ABSENT/SMALL BLADDER

Twin-Twin Transfusion Syndrome



Bladder Rupture



(Left) *Sagittal US of a donor twin, in a case of TTTS, shows oligohydramnios, with the donor suspended from the uterus by a tightly "shrink-wrapped" membrane. There is no urine within the bladder, making this at least stage 2 TTTS.* ***(Right)*** *Sagittal T2WI MR shows abdominal distention from urinary ascites. The decompressed bladder has a thick wall, the sequela of having been massively stretched. Multiple other anomalies were present.*

Bladder Exstrophy

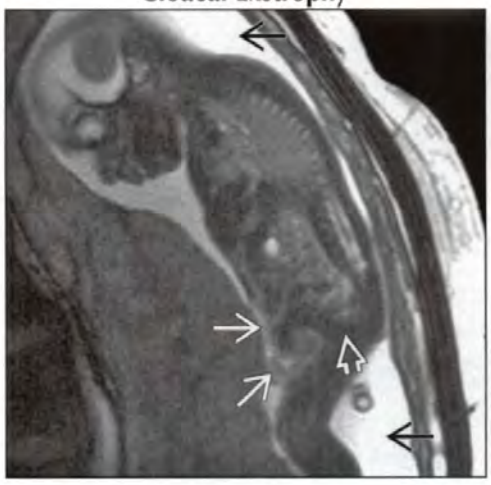


Cloacal Exstrophy

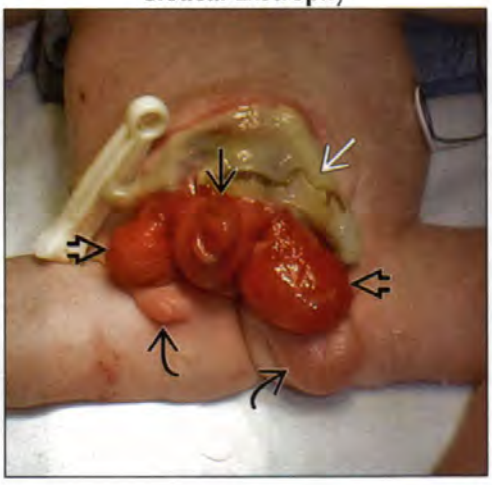


(Left) *Sagittal ultrasound of the lower fetal abdomen shows a normal cord insertion. No bladder was seen, and the anterior abdominal wall has irregular, echogenic contour, which represents the posterior wall of the exposed bladder. The amniotic fluid was normal.* ***(Right)*** *Sagittal ultrasound shows an absent bladder and an abdominal wall defect with extruded bowel inferior to the cord insertion, typical findings of cloacal exstrophy.*

Cloacal Exstrophy



Cloacal Exstrophy



(Left) *Sagittal T2WI MR of a fetus with cloacal exstrophy shows a low abdominal wall defect with non-visualization of the bladder. The amniotic fluid is normal.* ***(Right)*** *Clinical photograph in the same case shows the ruptured omphalocele, with bowel herniating between the two halves of the exposed bladder. There is also a split scrotum.*

ADRENAL MASS

DIFFERENTIAL DIAGNOSIS

Common
- Bronchopulmonary Sequestration (Mimic)
- Neuroblastoma

Rare but Important
- Adrenal Hemorrhage

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Search carefully for an adrenal gland
  - Neuroblastoma and hemorrhage involve the adrenal gland, while a sequestration is separate from it
- Normal adrenal gland has an "ice cream sandwich" appearance
  - Hypoechoic cortex, hyperechoic medulla

Helpful Clues for Common Diagnoses
- **Bronchopulmonary Sequestration (Mimic)**
  - 10-15% subdiaphragmatic
  - Majority left-sided
    - Stomach is displaced anteriorly
  - Typically an echogenic, solid mass
    - May see small cystic areas
  - Separate from adrenal gland
  - Color Doppler important for diagnosis
    - Look for dominant feeding vessel from aorta
  - Usually presents in 2nd trimester
- **Neuroblastoma**
  - May be either cystic or solid
    - Approximately equal occurrence
  - 60% are right-sided
  - No identifiable adrenal gland on side of mass
  - Displaces kidney inferiorly
  - Color Doppler may show diffuse vascularity but no dominant feeding vessel
  - Usually presents in 3rd trimester
  - Solid masses are more likely to metastasize
    - Liver most common site for metastases in utero
    - Hydrops may develop with large masses or metastatic disease
  - Cystic masses are usually complex, with thick septations

Helpful Clues for Rare Diagnoses
- **Adrenal Hemorrhage**
  - Reported in utero but uncommon
  - Can be solid or cystic
    - Appearance variable and changes with evolution of blood products
  - Will involute over time
  - No color flow within mass
  - MR can confirm blood products

Other Essential Information
- Neuroblastoma has variable, but generally favorable, in utero course
  - > 90% overall survival
    - Tumors remain stable or even spontaneously resolve
    - Cystic masses have best prognosis and may represent involuting tumor
  - Minority of cases progress to hydrops and even death

Bronchopulmonary Sequestration (Mimic)



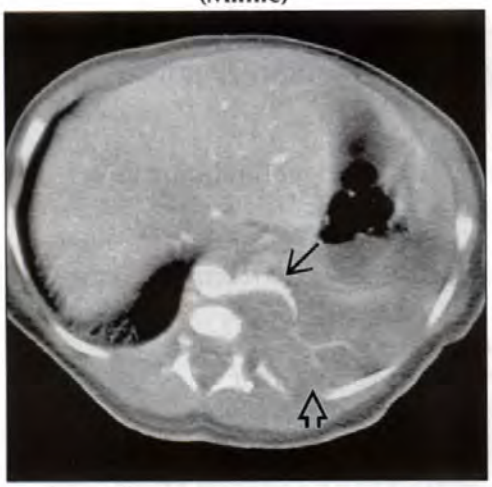
Coronal ultrasound shows a predominately solid suprarenal mass (calipers), separate from the kidney ➡. Use Doppler to look for a dominant feeding vessel.

Bronchopulmonary Sequestration (Mimic)

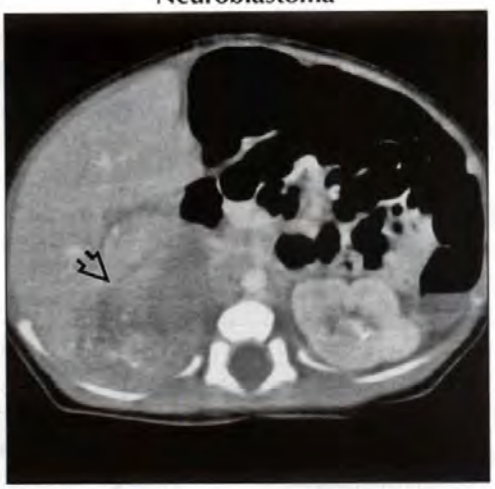


Sagittal ultrasound after delivery shows the same mass ➡, clearly separate from the adrenal gland ➡. Resection confirmed a sequestration.

Bronchopulmonary Sequestration (Mimic)



Neuroblastoma

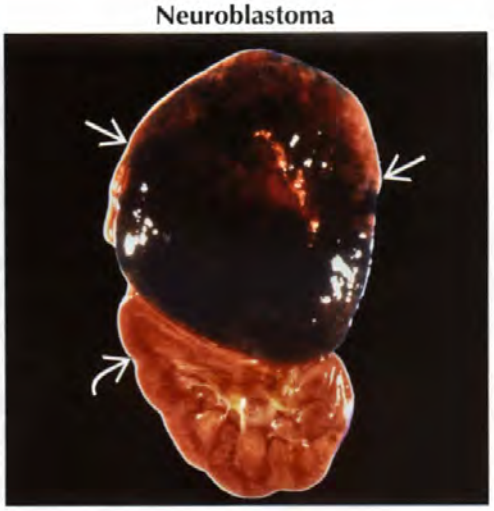


(Left) *Axial CECT in a newborn with a left-sided suprarenal mass shows a very prominent feeding vessel arising from the aorta. This is an important finding in a sequestration.* ***(Right)*** *Axial CECT in a newborn with a right-sided suprarenal mass shows heterogeneous enhancement and no feeding vessel. Biopsy showed a neuroblastoma.*

Neuroblastoma



Neuroblastoma

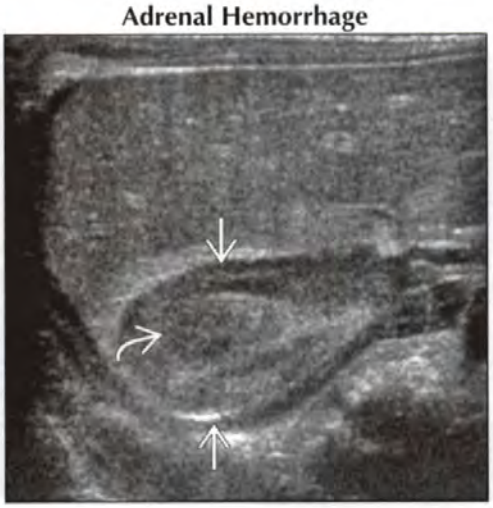


(Left) *Sagittal ultrasound shows a large, solid suprarenal mass that displaces the kidney inferiorly. In addition, there were multiple liver lesions, consistent with metastatic neuroblastoma.* ***(Right)*** *Gross pathology from the autopsy in the same case shows the large, solid adrenal tumor, which is compressing the upper pole of the kidney. This correlates with the ultrasound appearance.*

Adrenal Hemorrhage



Adrenal Hemorrhage



(Left) *Coronal ultrasound shows a cyst by the upper pole of the kidney. It is difficult to tell, with certainty, whether the cyst is adrenal or renal in origin, but no normal adrenal gland could be seen on that side. Blood products were identified on a fetal MR.* ***(Right)*** *Axial ultrasound after delivery shows a mildly enlarged adrenal gland, with a residual hypoechoic area. This completely resolved on follow-up scans, which is typical for adrenal hemorrhage.*

AMBIGUOUS GENITALIA

DIFFERENTIAL DIAGNOSIS

Common
- Clitoromegaly
- Microphallus

Less Common
- Hypospadias

Rare but Important
- Cloacal Exstrophy/Malformation

ESSENTIAL INFORMATION

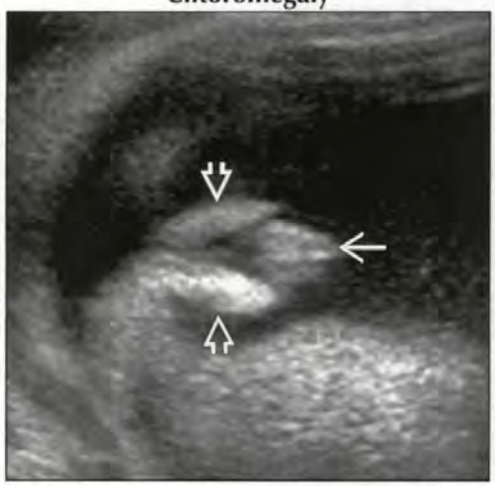
Key Differential Diagnosis Issues
- Accurate prenatal diagnosis difficult
  - Do not assign gender prenatally
  - Genetic amniocentesis helpful
  - Look carefully for other anomalies
- Aneuploidy/syndrome associations
  - Trisomy 13
  - Triploidy
  - Deletion/translocation abnormalities
  - Syndromes
    - Smith-Lemli-Opitz
    - Prader-Willi
- Abnormal hormone influence
  - Female pseudohermaphrodite (46,XX)
    - Excess androgenesis
    - Congenital adrenal hyperplasia
  - Male pseudohermaphrodite (46,XY)
    - Abnormal response to testosterone
    - Deficient testosterone production
  - True hermaphrodite
    - Extremely rare
    - Genetic female with Y chromatin
    - Mosaic (46,XX/46,XY)
  - Mixed or pure gonadal dysgenesis
    - Variable karyotypes (mosaics common)

Helpful Clues for Common Diagnoses
- **Clitoromegaly**
  - Idiopathic when mild
  - Enlarged clitoris between labial folds
    - Can mimic penis
  - Clitoris points inferiorly on sagittal views
    - May help differentiate from small penis, which points cranially
- **Microphallus**
  - Often with cryptorchidism
    - Empty scrotum mimics labia
  - Can look identical to clitoromegaly

Helpful Clues for Less Common Diagnoses
- **Hypospadias**
  - Urethra opens on ventral side of penis
  - Common associated genital anomalies
    - Small penis
    - Chordee (curved penis)
    - Cryptorchidism

Helpful Clues for Rare Diagnoses
- **Cloacal Extrophy/Malformation**
  - Infraumbilical abdominal wall defect
  - Absent bladder
    - Exposed bladder in 2 halves
  - Splayed symphysis pubis
  - Markedly abnormal genitalia
    - Bifid penis, split scrotum, cryptorchidism
    - Split labia, duplicated Müllerian structures, abnormal vagina

Clitoromegaly



Axial ultrasound shows an enlarged clitoris ➡ between normal labial folds ➡. Amniocentesis results were 46,XX and at delivery, the finding was thought to be mild and no treatment was warranted.

Microphallus

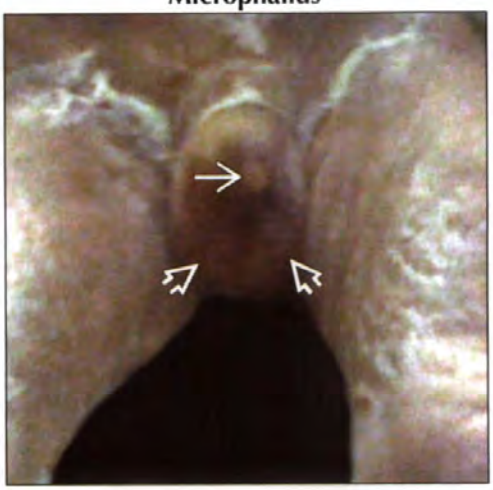


Axial ultrasound shows a small penis ➡ and empty scrotal sacs ➡, which mimic labial folds. The fetus also had a cardiac defect and hydrocephalus. Karyotype was 46,XY.

Microphallus

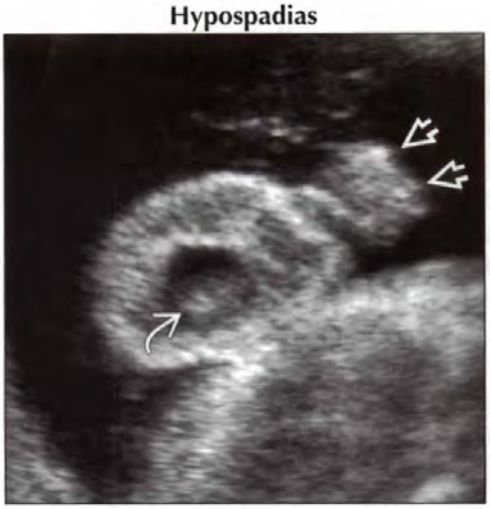


Microphallus

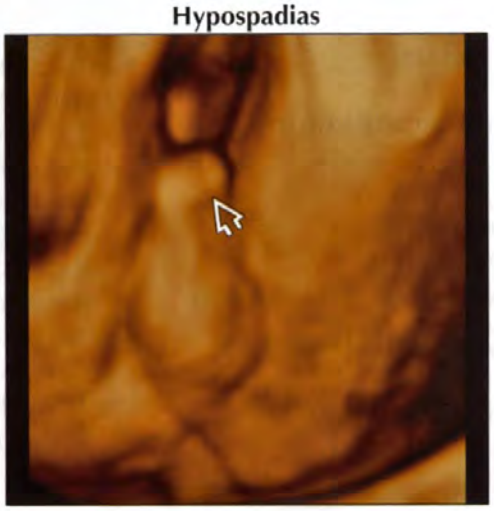


(Left) *Axial ultrasound shows ambiguous genitalia in a male fetus with trisomy 13. It was difficult to tell whether this was a micropenis or a clitoris. The fetus also had other severe anomalies.* ***(Right)*** *Gross pathology image of the same fetus shows the micropenis and the empty scrotal sac. Cryptorchism is often seen in association with microphallus.*

Hypospadias



Hypospadias



(Left) *Axial ultrasound of a 32 week fetus with ambiguous genitalia at 20 weeks shows a single descended testis surrounded by fluid. In addition, the tip of the penis is blunted, rather than normally tapered. The blunted appearance suggests hypospadias.* ***(Right)*** *3D ultrasound confirms the blunted appearance of the penile tip. Also mild ventral curvature of the tip of the penis is appreciated. The findings were confirmed after delivery.*

Cloacal Exstrophy/Malformation



Cloacal Exstrophy/Malformation



(Left) *Axial ultrasound of the genitalia in a fetus with cloacal extrophy and female karyotype shows abnormal labial folds.* ***(Right)*** *Clinical photograph after delivery shows the inferior abdominal wall defect and splayed labia majora. An atretic vagina was present. The split, externalized bladder mucosa and the midline omphalocele are hallmarks of cloacal extrophy.*

SCROTAL MASS

DIFFERENTIAL DIAGNOSIS

Common
- Hydrocele

Less Common
- Testicular Torsion
- Inguinal Hernia

ESSENTIAL INFORMATION

Helpful Clues for Common Diagnoses
- **Hydrocele**
  - Simple hydrocele
    - Anechoic fluid
    - Fluid forms "half moon" crescent around testis
    - Large hydrocele may completely surround testis
    - May be isolated or part of generalized hydrops
    - 2/3 unilateral, 1/3 bilateral
    - Testes normal
    - Usually transient finding with most resolved by birth
  - Complex hydrocele
    - Fluid with linear/focal echoes
    - Suggests a secondary process: Hemorrhage, testicular infarction/torsion

Helpful Clues for Less Common Diagnoses
- **Testicular Torsion**
  - Testis may be either large (acute) or small (chronic)
  - Variable echogenicity
    - Diffusely hypoechoic from edema
    - Heterogeneous from infarction
  - Scrotal edema
  - Complex hydrocele from hemorrhage or inflammatory reaction
  - "Double ring hemorrhage" variant: Hemorrhage trapped in two spaces
    - Between visceral and parietal tunica vaginalis
    - Between tunica vaginalis and scrotum
  - Doppler rarely helpful, unless obvious flow in normal testis
- **Inguinal Hernia**
  - Bowel herniates through inguinal canal
  - Cystic/echogenic mass in scrotum
  - Look for peristalsis
  - Hydrocele common
  - Look for a normal testis adjacent to mass

Other Essential Information
- Normal testicular descent at 25-32 wks
- Processus vaginalis forms from extension of peritoneal cavity and aids in descent of testis
  - Normally obliterates and becomes tunica vaginalis
  - Hydrocele forms if persistent patent processus vaginalis or fluid not resorbed
  - Patent processus vaginalis also risk factor for inguinal hernia
- Always consider torsion in setting of complex hydrocele
  - Testis is rarely saved when torsion diagnosed in utero

Hydrocele



Axial ultrasound shows bilateral, asymmetric, simple hydroceles. The testes ➡ are displaced posteriorly. These resolved spontaneously and the scrotum was normal on the newborn exam.

Hydrocele

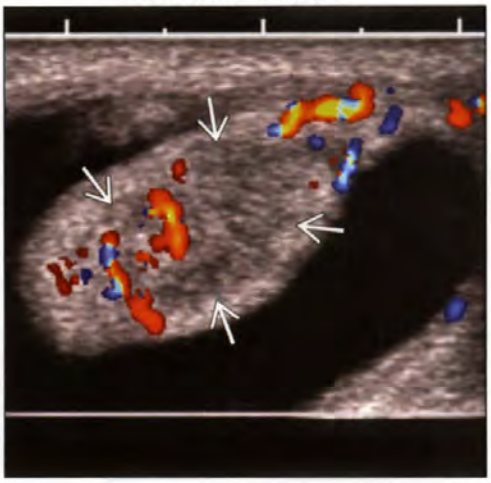


Sagittal ultrasound of the scrotum shows a hydrocele ➡ resulting from ascites extending through a patent processus vaginalis ➡ (penis ➡).

Testicular Torsion



Testicular Torsion



(Left) *Coronal ultrasound of the fetal scrotum shows a unilateral right hydrocele with internal echoes raising concern for a hemorrhagic or inflammatory process.* ***(Right)*** *Sagittal color Doppler ultrasound after delivery shows an enlarged right testis with hypoechoic avascular areas, consistent with infarction from intermittent torsion. The left testis was normal.*

Testicular Torsion

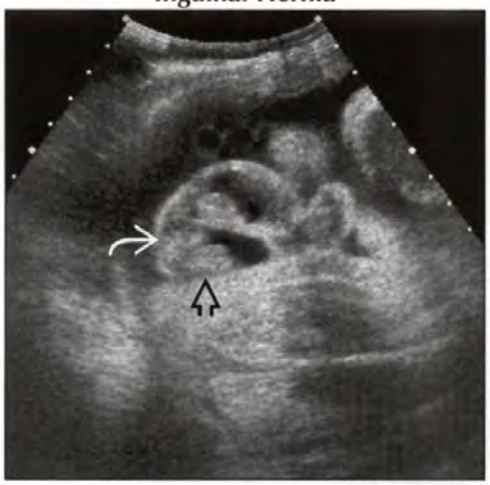


Testicular Torsion

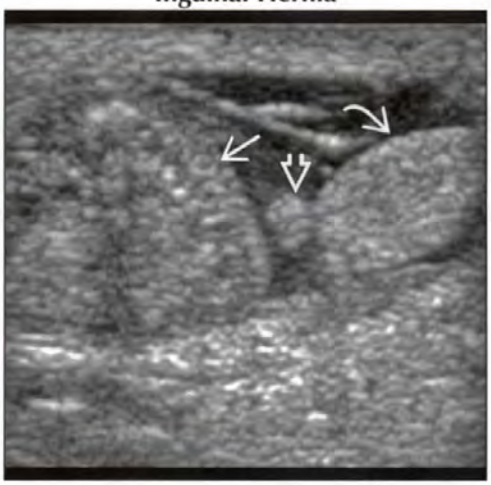


(Left) *Axial power Doppler ultrasound in a newborn with a dusky, swollen scrotum on physical exam shows no flow within a very abnormal, heterogeneous testis.* ***(Right)*** *Gross pathology following orchiectomy shows hemorrhagic infarction, which resulted from in utero torsion.*

Inguinal Hernia



Inguinal Hernia



(Left) *Axial ultrasound of the fetal scrotum shows simple, bilateral hydroceles. The presence of scrotal fluid aids in the detection of a soft tissue mass adjacent to the right testis.* ***(Right)*** *Sagittal ultrasound performed after delivery shows a bowel loop adjacent to the normal testis and epididymis. Peristalsis was seen during the scan confirming bowel herniation.*

SECTION 7
Musculoskeletal

Abnormal Fetal Posture/Movement	7-2
Mildly Short Femur/Humerus	7-8
Severe Limb Shortening	7-12
Angulated Bones	7-18
Abnormal Ossification	7-24
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Abnormal Foot	7-30
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ABNORMAL FETAL POSTURE/MOVEMENT

DIFFERENTIAL DIAGNOSIS

Common
- Spina Bifida
- Trisomy 18
- Arthrogryposis, Akinesia Sequence
- Fetal Constraint

Less Common
- Body Stalk Anomaly
- Caudal Regression Sequence
- Fetal Hypoxia/Severe Hypotonia
- Amniotic Bands
- Fetal Neck Masses
- Joint Dislocation
- Vertebral Segmentation Abnormalities

Rare but Important
- Iniencephaly
- Multiple Pterygium Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Postural abnormalities
  - Is the abnormality fixed or does the position normalize with fetal movement?
  - Is the abnormal body posture associated with an obvious anomaly?
- Fetal movement abnormalities
  - Is the movement abnormality progressive over time or an acute change?
  - Is there evidence of arthrogryposis?
  - Is there associated oligohydramnios, polyhydramnios, osteopenia or edema/hydrops?
  - Is the fetus normally grown?
  - Can normal movement be elicited by acoustic stimulation?
    - Normal sleep cycle vs. pathologic lack of movement

Helpful Clues for Common Diagnoses
- **Spina Bifida**
  - Lack of movement of the lower extremities associated with clubfeet ± ventriculomegaly
  - Abnormal calvarium shape ("lemon" sign) with small abnormal posterior fossa ("banana" sign)
- **Trisomy 18**
  - Arthrogryposis of multiple joints may be seen
  - Severe symmetrical growth restriction (IUGR) and multiple anomalies common
- **Arthrogryposis, Akinesia Sequence**
  - Fixed contractures of multiple joints, often severe
  - Joint abnormalities due to fetal akinesia
    - Bilateral and symmetrical
  - Upper and lower extremities may be equally affected or discordant in severity
  - Associated polyhydramnios due to decreased swallowing
  - Skin edema, osteopenia, frank hydrops often late findings, especially in lethal cases
- **Fetal Constraint**
  - Multiple gestation
    - Postural abnormalities due to crowding
    - True deformations may result (clubfeet, torticollis, plagiocephaly)
  - Fibroids
    - Large submucosal or multiple smaller fibroids
  - Uterine anomaly
    - Associated malposition common
  - Severe oligohydramnios
    - Premature rupture of membranes
    - Twin-twin transfusion donor twin
    - Severe IUGR, genitourinary anomalies

Helpful Clues for Less Common Diagnoses
- **Body Stalk Anomaly**
  - Severe postural abnormalities
  - Rotary scoliosis with unusual angulation of extremities from the body axis
  - Lack of free floating umbilical cord
  - Large schisis defects of abdomen &/or thorax seen, often with adherence of defect to the placenta
- **Caudal Regression Sequence**
  - Absent sacrum with hypoplastic lower extremities
  - Legs held in a fixed, "Buddha" or "crossed-legged tailor's" posture
  - More common in poorly controlled diabetes
- **Fetal Hypoxia/Severe Hypotonia**
  - Decreased or absent fetal breathing, tone and movement
  - Increased risk of fetal/neonatal birth asphyxia, neurologic injury, seizures
  - Chronic hypoxia from severe uteroplacental insufficiency

- IUGR, oligohydramnios
- Abnormal Dopplers with absent or reversed end diastolic flow, pulsatile ductus venosus

- Causes of acute hypoxia
  - Abruptio placenta
  - Severe maternal hypoxemia (trauma, cardiopulmonary arrest, asphyxia)
- Severe hypotonia: May be acute or chronic
  - Hyperextended or hyperflexed neck
  - Usually due to underlying neurologic abnormality

- **Amniotic Bands**
  - Wide spectrum of disruptions, often associated with postural abnormalities
    - Fetus may appear tethered
  - Membrane strands may be visible in amniotic cavity
- **Fetal Neck Masses**
  - Postural abnormality of neck may be progressive
  - Decreased fetal swallowing → development of polyhydramnios → increased risk of airway obstruction
  - Goiter
    - Neck hyperextension with large goiter
    - Sagittal view to evaluate position of head and neck; mode of delivery or airway at birth unlikely to be affected if normal neck flexion observed
  - Cystic hygroma (lymphangioma)
    - Large and asymmetrical masses lead to significant postural abnormality of head and neck
- **Joint Dislocation**
  - More common in hips, knees
- **Vertebral Segmentation Abnormalities**
  - Hemivertebrae, missing vertebral segments, abnormal ribs associated with scoliosis

Helpful Clues for Rare Diagnoses

- **Iniencephaly**
  - Persistent "stargazer" posture of head, neck due to fixed cervical hyperextension, cervical neural tube defect
  - Other malformations common
- **Multiple Pterygium Syndrome**
  - Fixed joint contractures associated with abnormal posture
  - Pterygia may not be visualized on ultrasound
  - Cystic hygroma and hydrops in lethal type

Other Essential Information

- Hydrops and polyhydramnios with arthrogryposis predict high risk for lethality
- If decreased or absent fetal movement, search for evidence of acute or chronic condition
  - If acute, fetal hypoxia is likely and delivery may be life saving
- If abnormal posture, evaluate for evidence of associated fetal or uterine abnormality to determine underlying cause

Spina Bifida



Axial ultrasound shows a sac ⇦ overlying a large lumbosacral myelomeningocele ⇨ in a mid-trimester fetus.

Spina Bifida



Clinical photograph shows term newborn with lumbosacral neural tube defect. Note the bilateral clubfeet ⇦ and atrophic legs ⇨ held in an abnormal posture.

ABNORMAL FETAL POSTURE/MOVEMENT

Trisomy 18



Trisomy 18



(Left) *Transabdominal ultrasound shows a typical case of arthrogryposis associated with trisomy 18. The hand was persistently held in an abnormal orientation to the wrist. The legs were held in extension, and the left foot was clubbed.* ***(Right)*** *Clinical photograph shows a stillborn term infant with trisomy 18. Arthrogryposis with multiple joint contractures can be seen. A large omphalocele is also apparent.*

Arthrogryposis, Akinesia Sequence

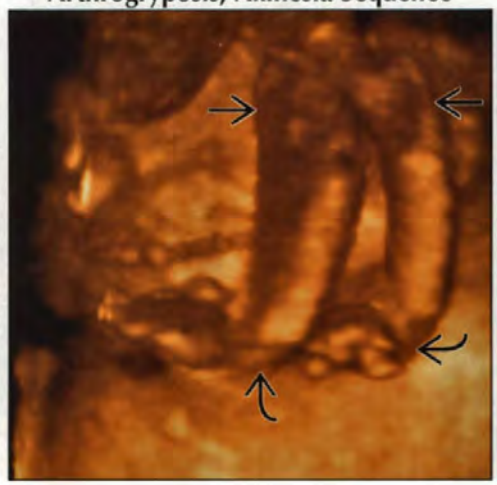


Arthrogryposis, Akinesia Sequence

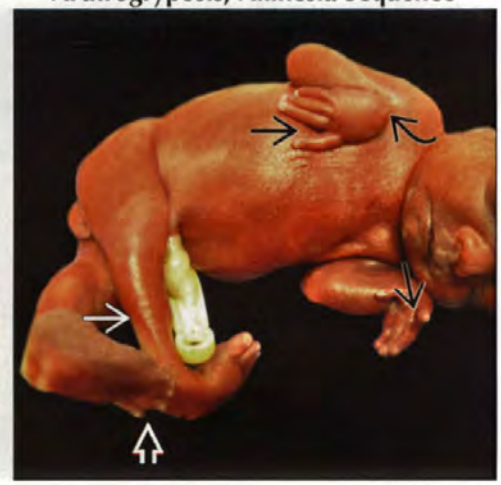


(Left) *Ultrasound shows a fixed, extended knee and clubfoot in a mid-trimester fetus. Both upper and lower extremities were affected.* ***(Right)*** *Clinical photograph of another infant who had amyoplasia shows flexed wrists, hyperextended elbows, and severely atrophic hands. The lower extremities were normal.*

Arthrogryposis, Akinesia Sequence



Arthrogryposis, Akinesia Sequence



(Left) *Sagittal 3D ultrasound shows hyperextended knees in another midtrimester fetus with severe, progressive fetal akinesia sequence. Bilateral clubfeet are also seen. Polyhydramnios was present.* ***(Right)*** *Clinical photograph shows the same stillborn fetus at 23 weeks. Note the atrophic legs and clubfeet. The wrists are flexed, and the fingers extended with a gap between the 2nd and 3rd fingers.*

ABNORMAL FETAL POSTURE/MOVEMENT

Fetal Constraint

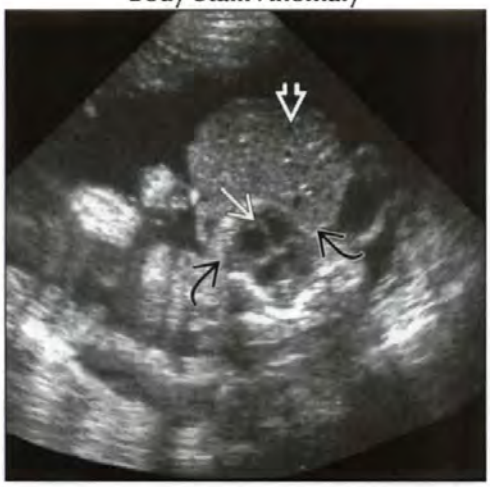


Fetal Constraint



(Left) *Coronal ultrasound shows oligohydramnios in a donor fetus with twin-twin transfusion. Normal fetal movement is prevented by the lack of fluid and tight membrane* ➡. ***(Right)*** *Clinical photograph of a newborn with renal agnesis shows typical Potter facies including wrinkled skin, flattened facial features, low-set ears* ➡, *and a small chin* ➡. *Clubbed feet and muscle atrophy* ➡ *are also present, all the result of anhydramnios causing restricted fetal movement.*

Body Stalk Anomaly



Body Stalk Anomaly



(Left) *Axial ultrasound shows extensive thoracoabdominal defect* ➡ *with ectopia cordis* ➡ *and extrusion of the liver* ➡ *in a fetus with a body stalk anomaly.* ***(Right)*** *Clinical photograph shows a preterm stillborn fetus with a body wall complex. Severe rotary scoliosis is apparent* ➡, *as is a large sacral lipomeningocele* ➡. *The substantial abdominal wall defect* ➡ *is adherent to the placenta* ➡. *The limbs are normal. There is no free cord visible.*

Caudal Regression Sequence



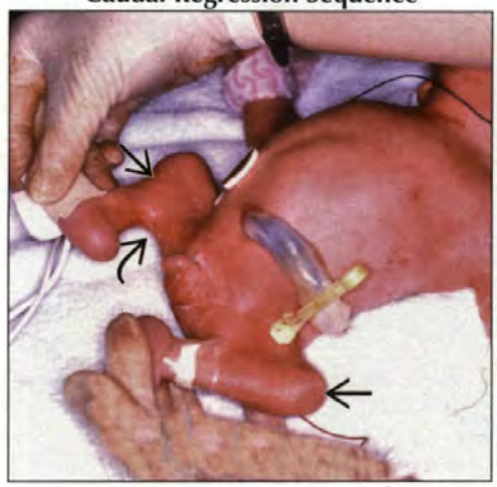
Caudal Regression Sequence



(Left) *Coronal ultrasound shows a mid-trimester fetus with sacral agenesis and caudal dysplasia. The sacral spine is absent below S1* ➡. ***(Right)*** *Ultrasound of the lower extremities showed the legs* ➡ *in a persistently flexed and crossed position. This has been called the "cross-legged tailor's" posture or "Buddha" pose. This fetus had triploidy.*

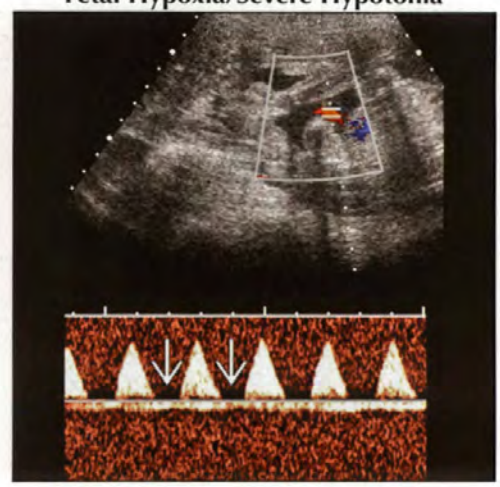
ABNORMAL FETAL POSTURE/MOVEMENT

Caudal Regression Sequence

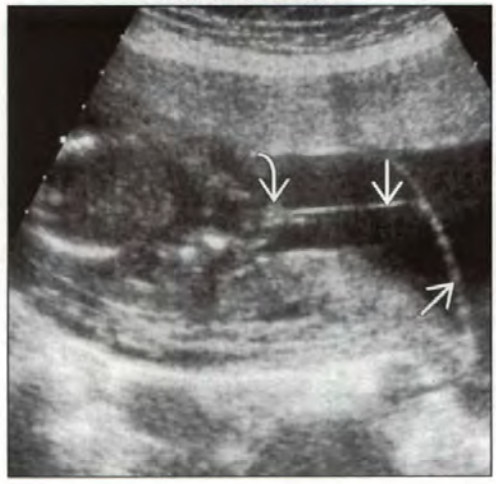


(Left) *Clinical photograph shows a preterm infant of a diabetic mother with caudal dysplasia. The lower extremities are hypoplastic, and the legs are held in a fixed "cross-legged tailor's" position. Note the popliteal pterygia from lack of movement in utero.* ***(Right)*** *Pulsed Doppler ultrasound shows absent end diastolic flow in a severely growth restricted fetus. Abnormal cord Doppler in IUGR is concerning for fetal hypoxia.*

Fetal Hypoxia/Severe Hypotonia



Amniotic Bands



(Left) *Ultrasound shows multiple strands of membrane in a mid-trimester case of amniotic bands. The strands are attached to fetal parts and limit fetal movement.* ***(Right)*** *Coronal T2WI MR shows a large multicystic mass in the anterolateral neck of this early third trimester fetus. The mass, of lymphatic origin, invaded the mouth of the fetus.*

Fetal Neck Masses



Fetal Neck Masses



(Left) *Clinical photograph shows residual redundant nuchal skin in a newborn diagnosed with a large goiter in utero. The fetus was successfully treated in utero with resolution of neck hyperextension.* ***(Right)*** *Clinical photograph shows a large lymphangioma in a newborn. Significant lateral flexion of the neck is seen.*

Fetal Neck Masses



ABNORMAL FETAL POSTURE/MOVEMENT

Joint Dislocation



Joint Dislocation



(Left) *Sagittal ultrasound shows a hyperextended knee in a mid-trimester fetus. The foot and leg were otherwise normal. Movement was seen involving the joint, but the leg never straightened normally.* ***(Right)*** *Lateral clinical photograph shows the lower extremities of the same infant at birth. The knee dislocation was treated with splinting and surgery after birth.*

Vertebral Segmentation Abnormalities



Iniencephaly

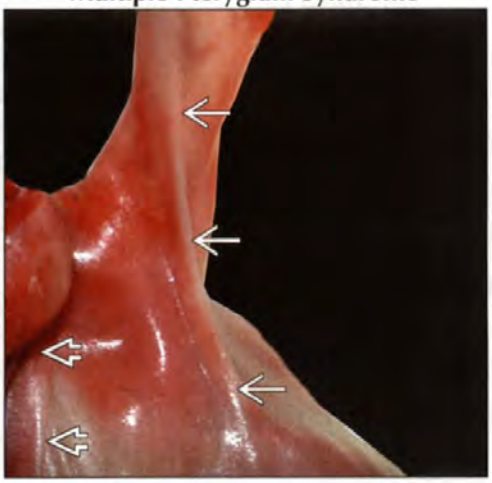


(Left) *Radiograph shows complex vertebral segmentation defects in a newborn with congenital scoliosis. A hemivertebrae, bifid rib, and absent rib are noted.* ***(Right)*** *Sagittal ultrasound shows a 1st trimester fetus with iniencephaly. The head is directly contiguous with the body, with no apparent neck, giving the appearance of the head being too big for the body. Facial bones are seen but no obvious cranium or posterior spinal elements (rachischisis).*

Iniencephaly



Multiple Pterygium Syndrome



(Left) *Clinical photograph shows a stillborn mid-trimester fetus with iniencephaly. Note the classic "stargazer" posture of the head, the short neck, and the cervical neural tube defect. The head appears disproportionately large for the fetal body.* ***(Right)*** *Clinical photograph shows axillary and anterior nuchal pterygia in a stillborn infant with multiple pterygium syndrome.*

MILDLY SHORT FEMUR/HUMERUS

DIFFERENTIAL DIAGNOSIS

Common

- Idiopathic
- Chromosome Abnormality
  - Trisomy 21
  - Turner Syndrome (XO)
- Early Onset IUGR

Less Common

- Heterozygous Achondroplasia
- Osteogenesis Imperfecta
- Proximal Focal Femoral Dysplasia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Is the short femur or humerus present bilaterally versus isolated asymmetric?
  - Measure humerus and femur on both sides to compare
  - Consider measuring all extremity bones if humerus/femur is dramatically short
    - More likely a skeletal dysplasia
  - If isolated to one long bone, could be focal defect of that limb
    - Check morphology of affected extremity to exclude in utero amputation/amniotic bands
- Assess morphology of osseous structures
  - Are the long bones straight versus bent?
    - Bent bones suggests in utero fractures and/or abnormal bone density
  - Skull shape
    - Abnormal can aid in assessment of skeletal dysplasia
  - Bone density
    - Can be difficult to assess in utero until later in gestation
    - Poor ossification of skull can be useful clue for abnormal bone density
- Many syndromes are associated with mildly short long bones
  - Ranges from chromosomal abnormalities to multisystem disorders

Helpful Clues for Common Diagnoses

- **Idiopathic**
  - Often constitutional if symmetric and parents are short
  - May be seen in fetuses of Asian or Hispanic descent
    - Shorter average femur and humerus than white or African American pregnancies
  - Biologic inherent variation has most impact in the third trimester
- **Trisomy 21**
  - Minor marker for trisomy 21 (T21)
  - Short humerus length (HL) more sensitive than short femur length (FL)
  - FL and HL compared to BPD
    - Expected FL = -9.3 + 0.90 (BPD)
    - Expected HL = -7.9 + 0.84 (BPD)
    - Abnormal ⇒ measured:expected FL ≤ 0.91 or HL ≤ 0.90
  - Look for other signs of trisomy 21
    - ≥ 1 minor markers seen in 50-70% of T21 fetuses
    - Major anomalies in 25-30% (cardiac, duodenal atresia, etc.)
  - Correlate with serum screen and first trimester ultrasound
    - Serum screen risk for T21 reported for maternal serum quadruple test or integrated screening
    - Nuchal translucency in first trimester always should be < 3 mm
- **Turner Syndrome (XO)**
  - Mild rhizomelia present at time of second trimester screening
  - Nuchal cystic hygroma is the predominant obvious finding
    - Usually large fluid collection in lateral and posterior neck
    - Can have multiple thin septations
  - Can present with non-immune hydrops
  - Associated with cardiovascular anomalies (60%)
    - Coarctation of aorta
    - Hypoplastic left heart
  - May have early symmetric growth restriction
- **Early Onset IUGR**
  - All biometric parameters should be equally affected
  - Symmetric IUGR has strong association with aneuploidy
    - Search carefully for anatomic abnormalities to correlate
    - Unlike asymmetric IUGR, more often fetal etiology rather than maternal/placental
  - Oligohydramnios may be present

- Especially in cases of trisomy 13, 18

Helpful Clues for Less Common Diagnoses

- **Heterozygous Achondroplasia**
  - Long bone shortening noted in late second or third trimester
    - Early scan may be normal with progressively discrepant bone growth
    - Humerus affected more severely than femur
  - Normal ossification and morphology
    - No fractures or bowing
  - Look for other signs to make diagnosis
    - Macrocephaly, frontal bossing
    - Thoracolumbar kyphosis
    - Trident hands
  - Autosomal dominant
    - Check parental chromosomes to assess for recurrence risk
    - 80% are new mutations
    - Homozygous achondroplasia is lethal with early severe findings
- **Osteogenesis Imperfecta**
  - Bent bones due to in utero fractures
  - Decreased mineralization
    - Skull can be manually deformed from transducer pressure
  - Small chest circumference with "beading"
    - Due to multiple rib fractures
  - Measure all long bones in suspected fetus
    - Severe shortening in OI type II
    - Other types less affected
  - Genetic counseling indicated for recurrence risk
    - Most mutations autosomal dominant
- **Proximal Focal Femoral Dysplasia**
  - Consider if focal isolated defect of proximal femur(s)
    - Most commonly unilateral (80%)
    - Affected femur may have acute varus angulation due to discontinuity of proximal femur
    - Femoral head can be absent in severe cases
  - Hemipelvis usually affected
    - Ranging from shallow acetabulum → hemipelvis hypoplasia
  - Occasionally seen with fibular hypoplasia/aplasia
  - Associated with diabetic embryopathy in the setting of femoral-facial syndrome
    - Check for other anomalies seen with maternal diabetes
    - Caudal regression, cardiac anomalies, brain anomalies, etc.
  - Depending on severity of defect and postnatal clinical follow-up, can have excellent prognosis with correction

Other Essential Information

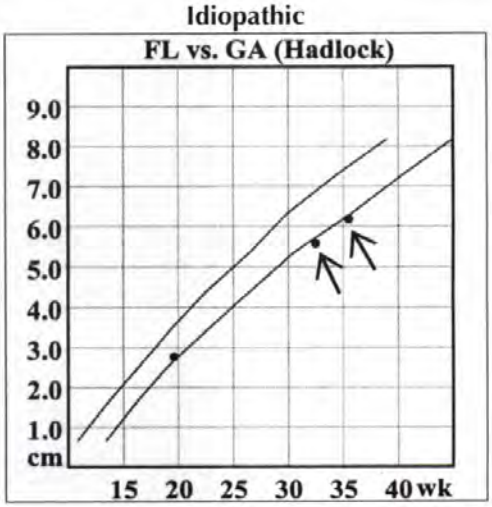
- If femur is not exactly in scan plane during measurement, can easily simulate mild shortening
- Measure upside femur for most accurate length
- If one femur is short, measure the contralateral side to assess whether finding is bilateral

Idiopathic



Ultrasound shows a relatively short femur in an otherwise normal fetus of Asian descent. At this 32.5 week scan, the femur measures about 29 weeks according to standard growth curves.

Idiopathic

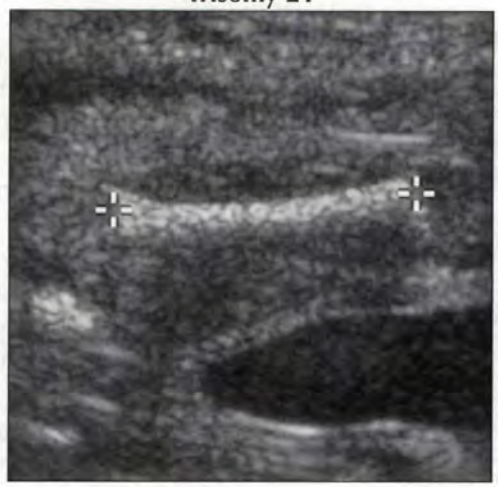


Growth chart plotting femur length (FL) against gestational age (GA) in the same fetus shows the mildly short femur. Note the difference is more pronounced in third trimester ⇨, which is usually the case.

MILDLY SHORT FEMUR/HUMERUS

(Left) *Axial ultrasound shows a morphologically normal femur (calipers), measuring 19.5 weeks though the fetus was 24 weeks gestation. Always remember to measure the humerus if trisomy 21 is suspected, as it is more affected than femur length.* ***(Right)*** *Coronal ultrasound of the same fetus shows clinodactyly ➔. Additional findings of trisomy 21 also were present. Amniocentesis confirmed the diagnosis.*

Trisomy 21



Trisomy 21

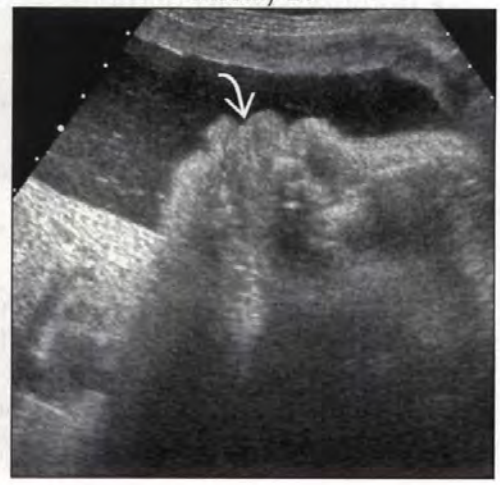


(Left) *Axial ultrasound shows a straight, unremarkable femur measuring 26 weeks in a 35 week gestation.* ***(Right)*** *Sagittal ultrasound of the same fetus shows a protuberant tongue ➔, which was seen in realtime and could not retract fully into the oral cavity. This fetus had trisomy 21.*

Trisomy 21



Trisomy 21

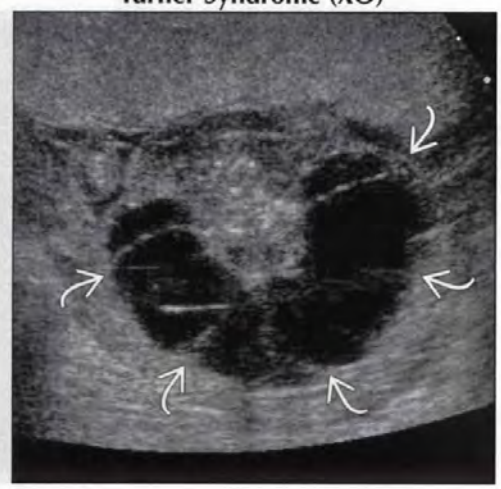


(Left) *Sagittal ultrasound of the femur shows a normal femur (calipers), though short for gestational age, measuring 15 weeks at 17 weeks gestation. Note the soft tissue edema of the thigh ➔.* ***(Right)*** *Axial ultrasound of the same female fetus at the level of the cervical spine shows the associated large cystic hygroma ➔, characteristic for Turner syndrome.*

Turner Syndrome (XO)



Turner Syndrome (XO)



MILDLY SHORT FEMUR/HUMERUS

Early Onset IUGR



Heterozygous Achondroplasia

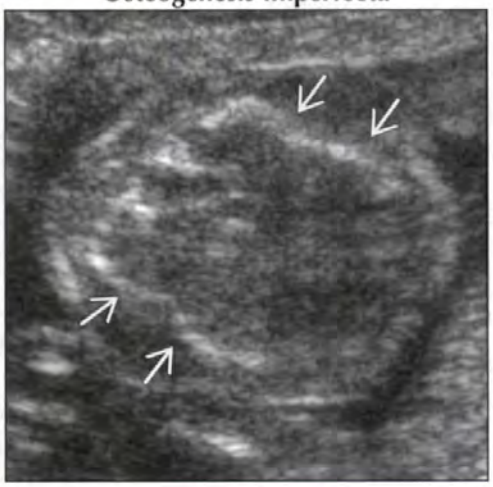


(Left) *Ultrasound of the distal femur in a fetus with IUGR shows ossification of the distal femoral epiphysis, indicating the fetus is at least 32 weeks gestation. The femur measured 33 weeks and the humerus 32 weeks in a 35.6 week fetus.* ***(Right)*** *Clinical photograph shows typical facial findings of achondroplasia with a flat midface, depressed nasal bridge, and upturned nasal tip. Note the trident hand and short humerus. In utero the femur measured 24 weeks at 31 weeks gestation.*

Osteogenesis Imperfecta



Osteogenesis Imperfecta



(Left) *Axial ultrasound shows an irregular contour to the femur (calipers), consistent with multiple healed in utero fractures. The femur can vary from mildly to severely shortened depending on the type of OI.* ***(Right)*** *Axial ultrasound of the same fetus shows the irregular "beaded" appearance of the fetal ribs, again due to multiple fractures. There is also poor mineralization of the osseous structures.*

Proximal Focal Femoral Dysplasia



Proximal Focal Femoral Dysplasia



(Left) *Axial ultrasound shows gross asymmetry of the left femur compared to the right at the time of screening for fetal anomalies.* ***(Right)*** *Anteroposterior radiograph of the same child after delivery shows a hypoplastic left proximal femur with slight varus angulation, a shallow acetabulum, and laterally dislocated femoral head (not ossified). Note the femur length discrepancy as well.*

SEVERE LIMB SHORTENING

DIFFERENTIAL DIAGNOSIS

Common
- Thanatophoric Dysplasia
- Osteogenesis Imperfecta
- Diabetic Embryopathy

Less Common
- Achondrogenesis
- Limb Reduction Defect

Rare but Important
- Hypophosphatasia
- Homozygous Achondroplasia
- Amelia, Micromelia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is the shortening limited to a single bone, single limb, or generalized?
- Are the long bones mildly shortened (1-3 standard deviations below the mean) or severely shortened?
- Is the ossification normal?
- Is the calvarium normally shaped?
- Are there fractures?
- Is there angulation or curvature of the long bones?
- Are there other structural anomalies?
- Is there a cystic hygroma or hydrops?

Helpful Clues for Common Diagnoses
- **Thanatophoric Dysplasia**
  - Most common prenatally diagnosed lethal skeletal dysplasia
  - Polyhydramnios, often severe and progressive
  - Type I
    - Normally shaped calvarium
    - Micromelia
    - "Telephone receiver" femur
    - Normal ossification, without fractures
    - Platyspondyly
    - Small chest with short ribs
    - Trident hand
    - Other malformations rare
  - Type II
    - Cloverleaf-shaped calvarium (Kleeblattschädel)
    - Femora slightly longer and straighter than type I
    - Other features similar to type I
- **Osteogenesis Imperfecta**
  - Multiple subtypes with most severe manifestations in perinatal lethal type II
  - Micromelia
  - Generalized decrease in ossification of all bones
  - Multiple fractures in utero
  - Ribs with "beaded" appearance due to fractures
  - Bones with irregular angulation due to fractures
  - Superb visualization of brain by ultrasound due to underossified calvarium
- **Diabetic Embryopathy**
  - Uncontrolled diabetes is most common human teratogen
  - Femoral hypoplasia common manifestation of embryopathy
    - Shortening may be severe
  - Usually bilateral but may be asymmetrical
  - Preaxial polydactyly
  - Other structural anomalies involving central nervous system, heart, skeletal common
  - Caudal regression
    - More common in uncontrolled diabetes
    - Sacral agenesis with shortened legs in fixed, crossed leg "tailor's posture"

Helpful Clues for Less Common Diagnoses
- **Achondrogenesis**
  - Severe micromelia
  - Vertebral ossification absent
  - Small chest with short flared ribs, severe pulmonary hypoplasia
  - Disproportionately large head with normal or deficient ossification
  - Hydrops and cystic hygroma common in early gestation
  - Type IA most severely affected with unossified spine and skull, multiple rib fractures
  - Type IB without rib fractures
  - Type II with normal skull ossification but unossified spine
- **Limb Reduction Defect**
  - May affect any segment of any limb
  - Usually single limb affected
  - Varying degrees of limb shortening
  - Longitudinal, intercalary and terminal transverse defects possible

- Greatest shortening with transverse defects
- May be seen with varicella exposure
- Amniotic band related amputation may have similar appearance

Helpful Clues for Rare Diagnoses

- **Hypophosphatasia**
  - 3 main subtypes: Perinatal, infantile, adult onset
  - Perinatal onset associated with most severe limb shortening
  - Micromelia and hypomineralization, often severe, often lethal
  - Long bones thin and bowed
  - Calvarium may be severely underossified
- **Homozygous Achondroplasia**
  - Autosomal dominant
  - Both parents must have achondroplasia
    - 1/4 offspring with homozygous achondroplasia (lethal)
    - 1/4 with average stature (unaffected)
    - 1/2 with achondroplasia (heterozygous)
  - Limb shortening severe, onset in the mid-trimester
  - Chest is very small with associated pulmonary hypoplasia
- **Amelia, Micromelia**
  - Amelia: Absence of 1 or more limbs
    - Tetra-amelia rare
    - Associated anomalies very common (cardiac, orofacial clefts, genitourinary)
    - High risk pulmonary hypoplasia, perinatal death
  - Micromelia: Shortening of both proximal and distal segments of limb
    - Association with severe, generalized syndromes
    - High risk perinatal lethality
  - Phocomelia: Shortening of limb with hand or foot arising near trunk

Other Essential Information

- Most important approach in prenatal evaluation of short limbs is to determine lethal vs. non-lethal condition
- Severity of limb shortening and chest size most informative
- Severe limb shortening associated with evidence of a generalized chondrodystrophy usually lethal
- Small chest correlated with risk of pulmonary hypoplasia
- Severe polyhydramnios common in lethal conditions
- Pattern of involvement is critical in formulating differential diagnosis
  - Are limb anomalies symmetric or asymmetric?
  - Are upper or lower limbs more severely affected?
  - Are the hands and feet present or absent; normal or abnormal?
- Careful evaluation for other non-skeletal anomalies
- Search for evidence of amniotic bands

Thanatophoric Dysplasia



Clinical photograph shows a stillborn infant with thanatophoric dysplasia (TD). Micromelia and a "trident" hand are seen. The chest is bell-shaped.

Thanatophoric Dysplasia



Coronal ultrasound shows a characteristic "trident" hand of a fetus with thanatophoric dysplasia.

SEVERE LIMB SHORTENING

(Left) *Ultrasound shows the very short radius and ulna ➡ in the forearm of a mid-trimester fetus with TD.* ***(Right)*** *Axial ultrasound shows the normally ossified calvarium of a mid-trimester fetus with TD. The parietal prominences ➡ seen are characteristic of the "cloverleaf-shaped" skull seen in type II TD.*

Thanatophoric Dysplasia



Thanatophoric Dysplasia

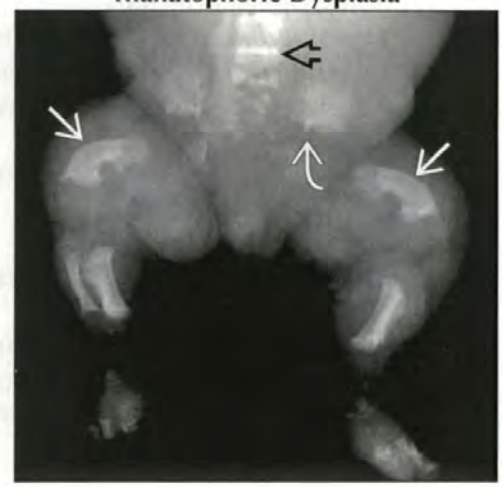


(Left) *Clinical photograph shows the body of a stillborn fetus with type II TD. The very short limbs ➡ are characterized by straighter long bones than is typical of type I TD. The narrow chest ➡ is also seen.* ***(Right)*** *Anteroposterior radiograph shows the typical "telephone receiver" femora ➡, platyspondyly ➡ and spiculations ➡ of the iliac wings in a stillborn infant with type I TD.*

Thanatophoric Dysplasia

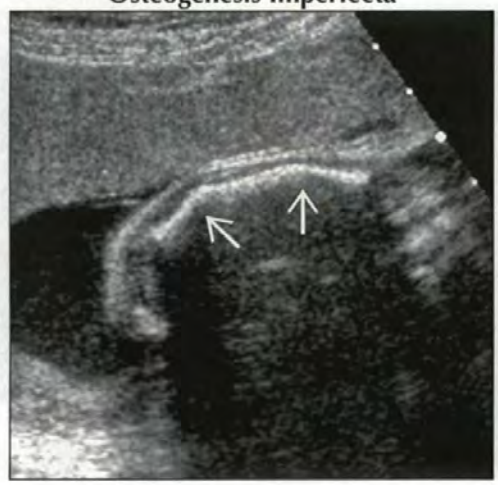


Thanatophoric Dysplasia



(Left) *Ultrasound shows the angulated femur ➡ of a late gestation fetus with type IV osteogenesis imperfecta. The bone is quite thin.* ***(Right)*** *Sagittal radiograph shows the severely underossified calvarium ➡ of a stillborn infant with perinatal lethal or type II osteogenesis imperfecta (OI). The short angulated humerus ➡ and typical "beaded" appearance of the ribs ➡ are due to multiple fractures.*

Osteogenesis Imperfecta

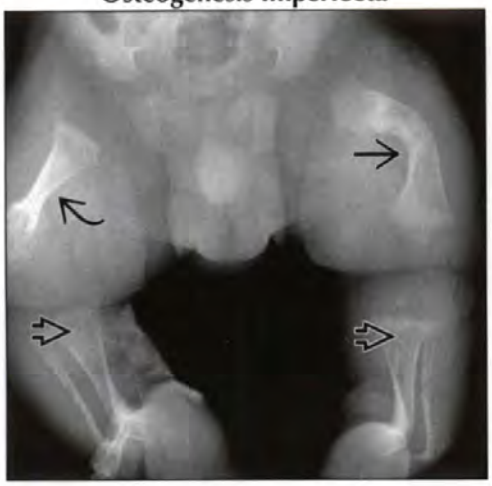


Osteogenesis Imperfecta



SEVERE LIMB SHORTENING

Osteogenesis Imperfecta



Osteogenesis Imperfecta



(Left) *Anteroposterior radiograph shows the pelvis and legs of a liveborn infant with type IV osteogenesis imperfecta. Note the angulated left femur and the straight right femur. Generalized osteopenia is also seen. **(Right)** Axial ultrasound shows the unusual appearance of the hypomineralized calvarium of a fetus with a non-lethal form of OI. The flattening of the lateral skull is due to normal pressure from the ultrasound transducer.*

Diabetic Embryopathy

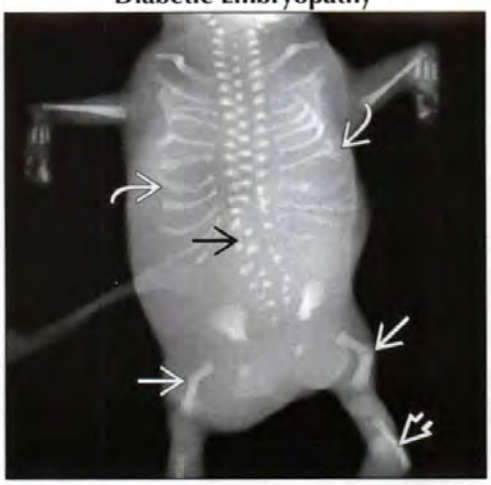


Diabetic Embryopathy



(Left) *Clinical photograph shows bilateral femoral hypoplasia in the infant of a poorly controlled diabetic mother. Preaxial polydactyly is also seen. **(Right)** Axial oblique ultrasound shows a large orofacial cleft in a fetus with holoprosencephaly related to poorly controlled diabetes. The tongue is seen protruding from the cleft.*

Diabetic Embryopathy



Diabetic Embryopathy



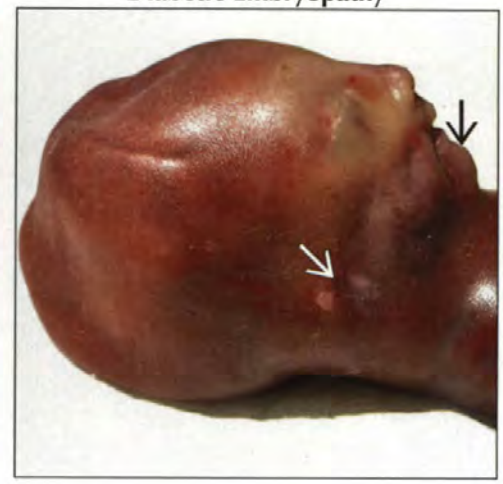
(Left) *Anteroposterior radiograph shows multiple severe skeletal anomalies in an infant with diabetic embryopathy. Multiple segmentation abnormalities of the spine, rib anomalies and dysplastic femurs are seen. Tibial aplasia is noted in the distal lower extremities. **(Right)** Sagittal radiograph shows sacral agenesis in an infant of a poorly controlled diabetic. The infant also had an imperforate anus and bilateral short femora.*

SEVERE LIMB SHORTENING

Diabetic Embryopathy



Diabetic Embryopathy

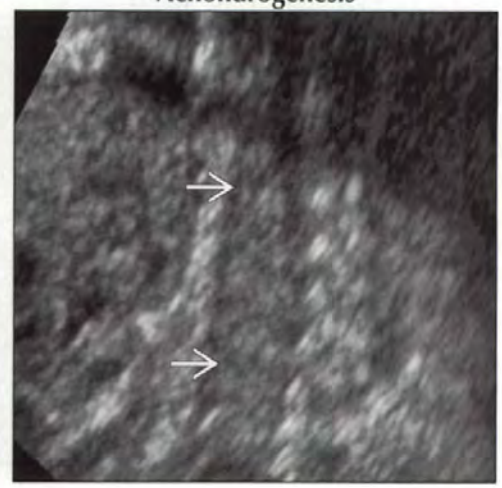


(Left) *Clinical photograph shows a severely dysplastic lower extremity in a stillborn mid-trimester fetus with diabetic embryopathy. Preaxial polydactyly is also seen, as well as syndactyly of toes 5-6.* ***(Right)*** *Lateral clinical photograph shows the same fetus with bilateral microtia and micrognathia.*

Achondrogenesis



Achondrogenesis

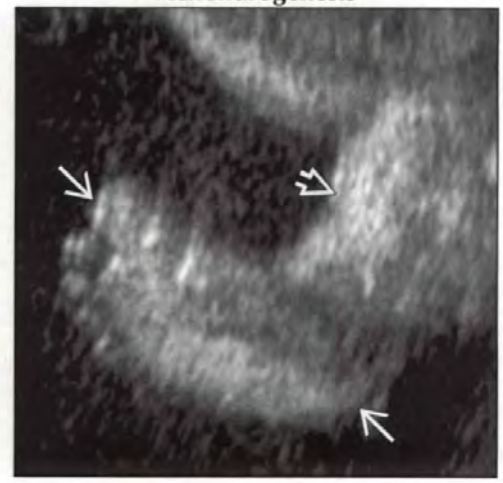


(Left) *Sagittal ultrasound shows severe hypomineralization in the spine of a fetus with achondrogenesis. The small chest and protuberant abdomen are also seen.* ***(Right)*** *Coronal ultrasound shows the underossified spine in the same fetus with achondrogenesis.*

Achondrogenesis



Achondrogenesis



(Left) *Clinical photograph shows the typical appearance of a stillborn infant with achondrogenesis. Note the severe micromelia, hypoplastic midface, disproportionately large head, and small chest.* ***(Right)*** *Ultrasound shows severe lower extremity micromelia in a fetus with achondrogenesis. The foot is as long as the entire leg.*

SEVERE LIMB SHORTENING

Limb Reduction Defect



Limb Reduction Defect

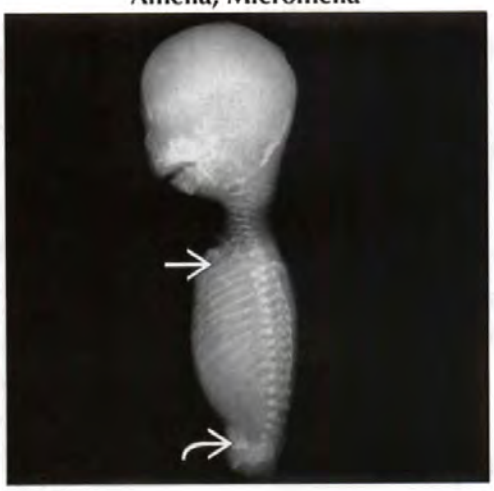


(Left) *Anteroposterior radiograph shows a severely shortened radius and ulna and missing hand in an infant with a terminal transverse limb defect. This fetus had fetal varicella syndrome.* ***(Right)*** *Clinical photograph shows a term infant with a terminal transverse limb defect attributed to amniotic bands. Rudimentary digits are seen.*

Hypophosphatasia



Amelia, Micromelia

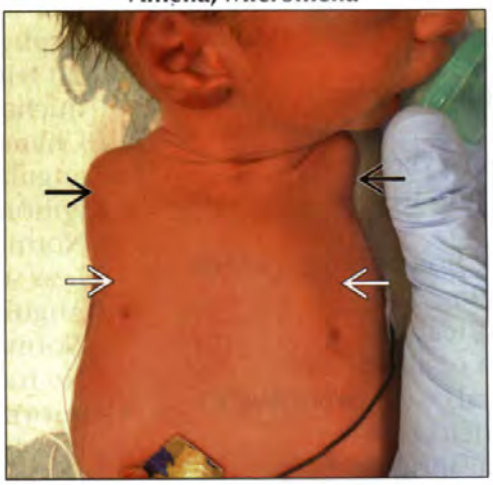


(Left) *Axial ultrasound shows a severely hypomineralized calvarium in a fetus with hypophosphatasia.* ***(Right)*** *Sagittal oblique radiograph shows a stillborn infant with tetra-amelia. The shoulder girdle and pelvis are severely dysplastic. All four extremities are absent.*

Amelia, Micromelia



Amelia, Micromelia



(Left) *Anteroposterior radiograph shows the thorax of a term infant with upper extremity amelia. Lower extremity phocomelia with severe limb shortening was present.* ***(Right)*** *Clinical photograph shows the same infant with upper extremity amelia. Both arms are missing. The chest musculature is hypoplastic and asymmetric.*

ANGULATED BONES

DIFFERENTIAL DIAGNOSIS

Common
- Thanatophoric Dysplasia
- Osteogenesis Imperfecta
- Diabetic Embryopathy

Less Common
- Campomelic Dysplasia
- Kyphomelic Dysplasia
- Abnormal Joint Angulation

Rare but Important
- Hypophosphatasia
- Fetal Trauma

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Are there fractures?
- Is the ossification normal?
- Is the angulation mid-shaft or at a joint?
- Is the distal limb normal?
- Is one limb affected or all?
- Are both segments of the limb affected?
- Are the abnormalities limited to the long bones or are other skeletal elements affected?
- Are there other structural anomalies?

Helpful Clues for Common Diagnoses
- **Thanatophoric Dysplasia**
  - Micromelia
  - Normal ossification
  - No fractures
  - Short ribs with bell-shaped thorax
  - Platyspondyly
  - Lumbar kyphosis common
  - "Telephone receiver" femur in type I
  - Normal calvarium in type I
  - Femora less curved in type II
  - Cloverleaf-shaped skull (Kleeblattschädel) in type II
  - Polyhydramnios often severe and progressive in the second trimester
  - Other anomalies rare
  - Lethal within first few hours-to-days of life
- **Osteogenesis Imperfecta**
  - Fractures a prominent feature
  - Decreased ossification of all bones
  - Type II (perinatal lethal) with extensive in utero fractures, limb deformities
  - "Beaded" ribs due to healing fractures
  - Deformable skull with pressure from ultrasound transducer
  - Non-lethal types associated with less severe limb shortening, fewer in utero fractures
  - Progressive deformation, shortening may occur in type III/IV
  - Type III/IV may present with isolated bent femur in utero
  - Size of chest correlated with risk of lethal outcome
- **Diabetic Embryopathy**
  - Uncontrolled diabetes most prevalent human teratogen
  - Abnormal femur common
    - Usually bilateral femur abnormality, but often discordant
    - Short, angulated or curved femur
  - Associated tibia-fibula abnormality
  - Preaxial polydactyly
  - Other structural defects common in uncontrolled diabetes
    - Cardiac
    - Central nervous system: Anencephaly, holoprosencephaly, spina bifida
    - Anorectal malformation

Helpful Clues for Less Common Diagnoses
- **Campomelic Dysplasia**
  - Severe angulation of femora, tibiae, fibulae
    - Anterolateral bowing especially common
  - Scapula absent or hypoplastic
  - XY sex reversal (male to female) or ambiguity
    - Genotypic males appear phenotypically as females
  - Normal ossification
  - No fractures
  - Bell-shaped chest
  - Kyphoscoliosis
  - 1st trimester cystic hygroma or increased nuchal translucency
  - Characteristic skin dimpling over area of angulation
- **Kyphomelic Dysplasia**
  - Normal chest size
  - Less severe long bone shortening
  - Angulation or curvature of long bones
  - Normal ossification
  - No fractures
- **Abnormal Joint Angulation**
  - Fixed vs. moveable joint

- Normal distal extremity associated with dislocated joint
  - Knees, hips most commonly affected
  - Movement at the joint often observed in utero despite dislocation
  - May be unilateral or bilateral
  - May be associated with fetal malpresentation
  - Prolonged dislocation may result in dysplastic joint
- Abnormal distal extremity often associated with abnormal joint or proximal bone
  - Joint usually without spontaneous movement
  - Wrist most commonly affected, but ankle also possible
  - Angle of deviation predicts which bone is hypo- or aplastic; angulation is toward the hypoplastic element
  - Radial deviation associated with hypoplasia or aplasia of radius and thumb
  - Ulnar deviation less common; associated with ulnar hypoplasia
  - Tibial or fibular hypoplasia or aplasia associated with fixed angulation of ankle
  - Associated oligodactyly common

Helpful Clues for Rare Diagnoses

- **Hypophosphatasia**
  - Multiple sub-types including perinatal lethal, infantile and late onset (adult)
  - In general the later the onset, the less severe the clinical course
  - Undermineralization of calvarium results in brain being seen "too well" on ultrasound
  - Perinatal lethal type with prominent midtrimester ultrasound findings of severe undermineralization and micromelia of all long bones and calvarium
  - In general long bones thin and bowed with absent posterior shadowing
  - Spurs often seen along mid-shaft of long bones
- **Fetal Trauma**
  - Isolated fractures due to fetal trauma rare in absence of severe maternal trauma

Other Essential Information

- Distinguish between angulated bones and angulated joints when evaluating the fetus
- Curvature of multiple bones predicts generalized osteochondrodystrophy
  - Severity of associated limb length shortening and chest size will predict lethal vs. non-lethal skeletal dysplasia

Alternative Differential Approaches

- Presence of fractures of major importance
  - Consider osseous fragility syndromes including osteogenesis imperfecta and hypophosphatasia
  - Severity and number of in utero fractures may help distinguish lethal vs. non-lethal disorder
  - Rib fractures without long bone fractures seen in type IA achondrogenesis

Thanatophoric Dysplasia



Coronal ultrasound shows a short, curved, "telephone receiver" femur seen in type I thanatophoric dysplasia (TD).

Thanatophoric Dysplasia



Clinical photograph shows the same infant after birth. Note the normally shaped, but disproportionately large calvarium, micromelia, small chest, and trident hands.

ANGULATED BONES

Thanatophoric Dysplasia



(Left) Anteroposterior radiograph shows the short curved femur ⇨ typical of type I TD. Note also the spicules on the inferior iliac wing ⇨ and the platyspondyly ⇨ involving the lumbar spine. ***(Right)*** *Sagittal ultrasound shows lumbar lordosis ⇨ in a fetus with thanatophoric dysplasia. The platyspondyly ⇨ is also prominent.*

Thanatophoric Dysplasia



Osteogenesis Imperfecta



(Left) Sagittal radiograph shows "beaded" appearance of the ribs ⇨ due to multiple healing fractures, typical of perinatal lethal osteogenesis imperfecta (OI). Irregular curvature of the humerus ⇨ and femur ⇨ is also due to multiple fractures. ***(Right)*** *Ultrasound shows a curved femur ⇨ with callus formation ⇨ due to a healed fracture in a fetus with type IV OI.*

Osteogenesis Imperfecta



Osteogenesis Imperfecta



(Left) Clinical photograph shows typical appearance of the lower extremities in perinatal lethal OI type II. Pseudoarthroses ⇨ are due to multiple fractures in utero. ***(Right)*** *Sagittal ultrasound shows bowing of the lower leg ⇨ in a fetus with type IV osteogenesis imperfecta.*

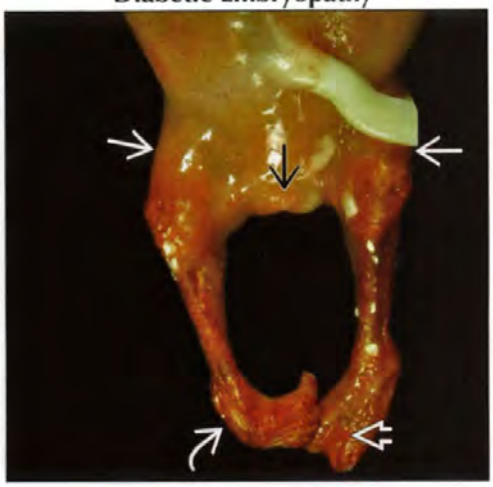
Osteogenesis Imperfecta



Diabetic Embryopathy



Diabetic Embryopathy



(Left) *Anteroposterior radiograph shows a midtrimester fetal loss of a poorly controlled diabetic. Note bilateral hypoplastic femora, angulation of one femur, and sacral agenesis. **(Right)** Clinical photograph shows the same fetus. In addition to the femoral hypoplasia, extremities appear quite atrophic. There is a clubfoot and complex syndactyly of the toes. The genitalia are ambiguous and an imperforate anus was present.*

Diabetic Embryopathy

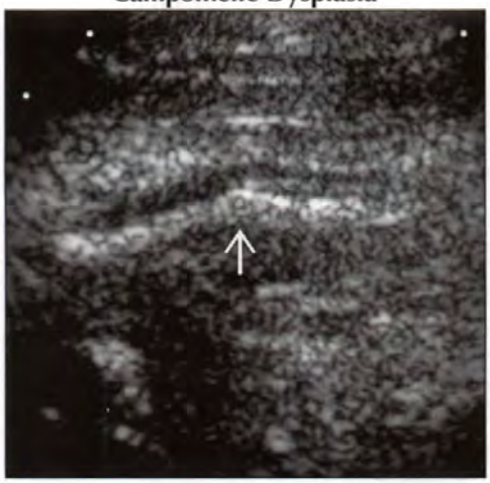


Diabetic Embryopathy



(Left) *Clinical photograph shows severe lower extremity anomalies in the infant of a poorly controlled diabetic. Femoral hypoplasia with absent tibia and fibula, abnormal angulation of the "ankle" and preaxial polydactyly are seen in this infant. **(Right)** Coronal T2WI MR shows the monoventricle of alobar holoprosencephaly in a fetus of a mother with poorly controlled diabetes.*

Campomelic Dysplasia



Campomelic Dysplasia



(Left) *Coronal ultrasound shows mid shaft angulation of the femur in a midtrimester fetus with campomelic dysplasia. The bone is normally ossified and without evidence of fractures. Prominent angulation of the tibia and fibula was also noted. **(Right)** Coronal oblique radiograph shows typical hypoplastic scapulae in a newborn infant with campomelic dysplasia. The chest is mildly bell-shaped and ossification is normal.*

ANGULATED BONES

(Left) *Coronal ultrasound shows an angulated femur in a fetus diagnosed after birth with kyphomelic dysplasia. Ossification is normal, and no fractures were seen in the mildly shortened long bones.* ***(Right)*** *Coronal ultrasound shows a normally ossified scapula in the same fetus ruling out campomelic dysplasia.*

Kyphomelic Dysplasia



Kyphomelic Dysplasia



(Left) *Ultrasound shows a fetus with absent ulna, hypoplastic radius, fixed ulnar deviation of the wrist, and abnormal hand. The defect was bilateral and limited to the upper extremities.* ***(Right)*** *Clinical photograph shows the same infant with symphalangism at birth. Note the ulnar deviation of the wrists and the oligodactyly of the hands. The infant was otherwise normal.*

Abnormal Joint Angulation

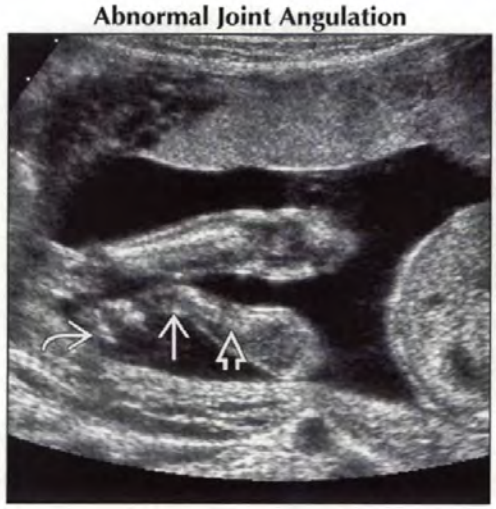


Abnormal Joint Angulation

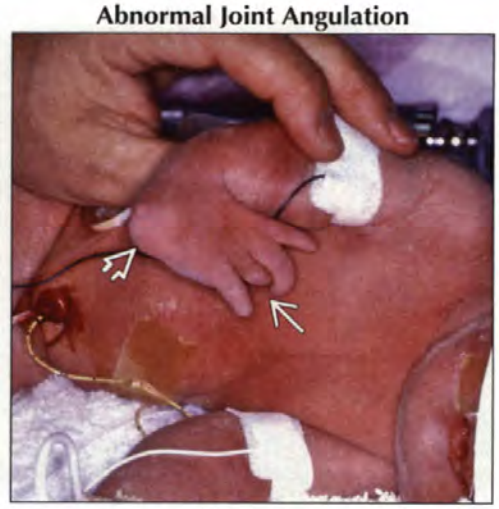


(Left) *Coronal ultrasound shows unilateral fibular hemimelia with a fixed laterally deviated ankle joint. The tibia is short and dysplastic, while the fibula appears to be absent. Oligodactyly of the foot is seen.* ***(Right)*** *Clinical photograph shows an infant with a radial ray defect with a radially deviated wrist due to an absent radius. The ulna was also hypoplastic and the thumb was absent. Other digits also appear dysplastic.*

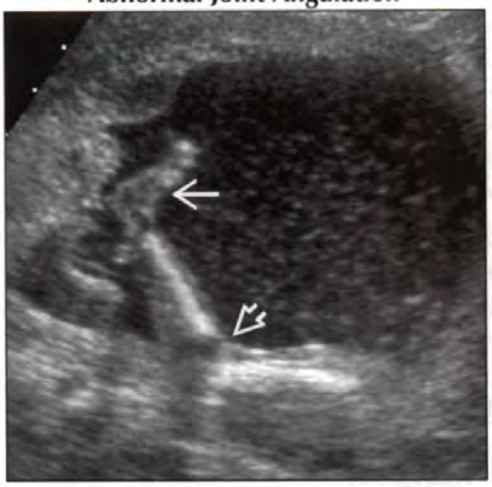
Abnormal Joint Angulation



Abnormal Joint Angulation



Abnormal Joint Angulation

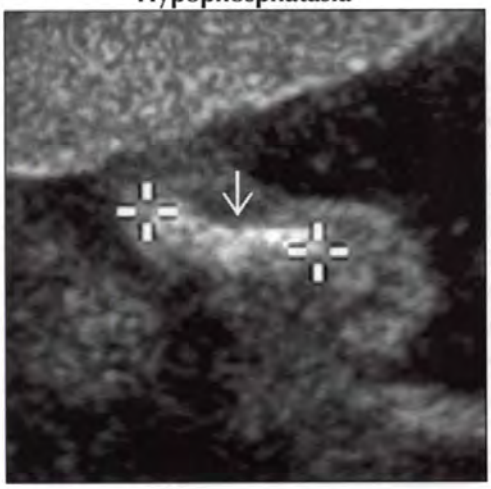


Abnormal Joint Angulation

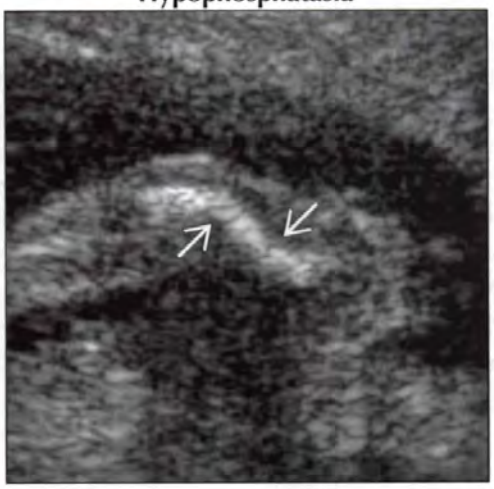


(Left) *Sagittal ultrasound shows a congenitally dislocated knee ➡ with hyperextension of the lower leg ➡. First noted at the patient's 18 week ultrasound, the fetal leg, including the joint, moved but never was seen in a normal position.* ***(Right)*** *Clinical photograph shows the same fetus at birth with a congenitally dislocated knee ➡. Splinting therapy was unsuccessful, and the infant required surgical correction.*

Hypophosphatasia



Hypophosphatasia



(Left) *Ultrasound shows a curved femur ➡ in a mid-trimester fetus with the infantile form of hypophosphatasia. At this gestation, the ossification appears nearly normal.* ***(Right)*** *Coronal ultrasound shows complex curvature ➡ of the humerus in the late 2nd trimester in the same fetus with hypophosphatasia. The abnormal angulation is likely due to healed fractures.*

Fetal Trauma



Fetal Trauma



(Left) *Clinical photograph shows severe maternal abdominal bruising ➡ from a motor vehicle accident.* ***(Right)*** *Clinical photograph shows complete placental abruption ➡ with death of the fetus, due to direct maternal abdominal trauma in a motor vehicle accident. Fetal fractures are uncommon in the absence of significant maternal trauma.*

ABNORMAL OSSIFICATION

DIFFERENTIAL DIAGNOSIS

Common
- Arthrogryposis, Akinesia Sequence
- Osteogenesis Imperfecta

Less Common
- Achondrogenesis
- Hypochondrogenesis

Rare but Important
- Hypophosphatasia
- Atelosteogenesis

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Are there fractures?
  - Presence or absence of fractures most important clue for differential
- Are the fractures generalized or limited to a portion of the skeleton (e.g., ribs)?
- Do the long bones appear short?
- If short long bones, is there micromelia?
- Is there angulation or curvature of the long bones without fractures?
- Is the underossification generalized or limited to part of the skeleton?
- Is the calvarium involved?
- Does the fetus move normally or is there evidence of arthrogryposis/akinesia?

Helpful Clues for Common Diagnoses
- **Arthrogryposis, Akinesia Sequence**
  - Lack of movement → decreased mineralization
  - Fractures rare in utero; may occur at the time of birth
- **Osteogenesis Imperfecta**
  - Perinatal lethal form with multiple fractures in utero
  - "Beaded" appearance of ribs due to multiple fractures
  - Deformable calvarium due to underossification

Helpful Clues for Less Common Diagnoses
- **Achondrogenesis**
  - Large calvarium, micromelia
  - Severely decreased ossification of spine, calvarium in IA, IB
  - Rib fractures in type IA; none in IB
  - Normal cranial ossification, absence of rib fractures in type II
- **Hypochondrogenesis**
  - Part of a spectrum of achondrogenesis type II, but less severe findings
  - Fractures rare

Helpful Clues for Rare Diagnoses
- **Hypophosphatasia**
  - Generalized lack of ossification
  - Calvarium poorly ossified
  - Small thorax, short limbs
  - Fractures rare, but may involve ribs which may have a "rachitic rosary" appearance
- **Atelosteogenesis**
  - Severely underossified humerus, fibula, femur; bowed tibia
  - Narrow thorax, proximal limb shortening

Arthrogryposis, Akinesia Sequence



Ultrasound shows the clenched hand ➡ of a mid-trimester fetus with fetal akinesia sequence. Note the lack of ossification of the bones of the forearm ➡.

Arthrogryposis, Akinesia Sequence



Ultrasound shows hyperextended elbows ➡ and flexed wrists ➡ of another mid-trimester fetus with akinesia sequence. The bones appear thin ➡, but without fractures.

Osteogenesis Imperfecta



Osteogenesis Imperfecta



(Left) *Axial ultrasound shows the chest of a third trimester fetus with type II osteogenesis imperfecta. The chest is small with obvious displaced rib fractures* ➡. ***(Right)*** *Axial ultrasound shows the underossified calvarium* ➡ *that appears flattened by the pressure of the ultrasound transducer. Note also the fracture of the humerus* ➡ *and thinning of the bones* ➡ *of the distal upper extremity.*

Achondrogenesis



Hypochondrogenesis



(Left) *Sagittal ultrasound shows a mid-trimester fetus with achondrogenesis. Note the severely underossified spine* ➡. ***(Right)*** *Sagittal ultrasound shows the small chest* ➡ *of a third trimester fetus with hypochondrogenesis. Underossification of the spine is also seen* ➡. *Platyspondyly is apparent* ➡.

Hypochondrogenesis



Hypophosphatasia



(Left) *Coronal ultrasound shows short, straight ribs* ➡ *in a fetus with hypochondrogenesis. No fractures are seen.* ***(Right)*** *Ultrasound shows the forearm of a third trimester fetus with hypophosphatasia. The significant underossification of the bones of the hand* ➡ *and forearm* ➡ *can be seen.*

RADIAL RAY MALFORMATION

DIFFERENTIAL DIAGNOSIS

Common
- Isolated
- VACTERL Association
- Trisomy 18
- Diabetic Embryopathy
- Syndromal

Less Common
- Holt Oram Syndrome
- Thrombocytopenia-Absent Radius Syndrome
- Fanconi Anemia
- Fetal Valproate Syndrome

Rare but Important
- Cornelia de Lange Syndrome
- Roberts Syndrome
- Aase Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is the anomaly unilateral or bilateral?
- Is there radial or ulnar deviation of the wrist?
  - Direction of deviation will predict the particular bony abnormality of the forearm
- Are there other skeletal anomalies present?
- Are the thumbs present or absent?
- Does the fetus move normally, or is there evidence of arthrogryposis of other joints?
- Are there other structural anomalies?
- Is there a relevant family history?
- Is the fetus normally grown?

Helpful Clues for Common Diagnoses
- **Isolated**
  - Unilateral or bilateral abnormality
  - Variable thumb defects including absence, triphalangeal, hypoplastic
- **VACTERL Association**
  - Nonrandom association of defects, usually sporadic
    - Vertebral anomalies
    - Anorectal malformation
    - Cardiac defects
    - Tracheoesophageal fistula with esophageal atresia
    - Radial ray anomaly with or without thumb abnormality
    - Renal anomaly
  - Associated hydrocephalus (aqueductal stenosis) also reported and may be familial
- **Trisomy 18**
  - Severe prenatal onset growth restriction
  - Multiple anomalies common including cardiac, gastrointestinal, central nervous system, skeletal
  - Bilateral radial ray defect, often discordant in severity
  - Thumbs may be absent or abnormal with radial hypoplasia/aplasia
- **Diabetic Embryopathy**
  - Risk highest in poorly controlled diabetic
  - Multiple anomalies common including skeletal, cardiac, central nervous system, renal
  - Preaxial polydactyly
- **Syndromal**
  - Radial ray defects found in many syndromes
  - May be seen with other structural anomalies without a unifying diagnosis

Helpful Clues for Less Common Diagnoses
- **Holt Oram Syndrome**
  - Bilateral upper extremity defects with cardiac anomaly
  - Lower extremities uninvolved
  - Wide spectrum of upper extremity malformation
    - Radial aplasia, hypoplasia, phocomelia (10%)
    - Thumb: Absent, hypoplastic, triphalangeal, bifid
    - Asymmetry of defects, with those on the left more severe
    - Other anomalies of upper limbs common including narrow shoulders, clinodactyly, syndactyly, defects of ulna, humerus, clavicle, sternum
  - Range of cardiac defects; ventricular septal defect and atrial septal defect (secundum) most common
    - Conduction defects
- **Thrombocytopenia-Absent Radius Syndrome**
  - Radial aplasia in 100%
  - Variable degrees of hypoplasia, absence of ulnae, humeri
  - Thumbs always present
  - Abnormal shoulders

- Variable anomalies of the lower extremities in 50%
- 1/4-1/3 with cardiac anomalies, predominantly atrial septal defect or tetralogy of Fallot
- Hematologic abnormalities may be severe in early infancy
- Autosomal recessive
- Prenatal or postnatal diagnosis confirmed by inducible chromosomal breakage

- **Fanconi Anemia**
  - Prenatal onset growth restriction, including microcephaly
  - Radial ray defect in half of cases
  - Varying degrees of thumb abnormality including hypoplasia, aplasia, triphalangeal, duplication
  - Renal anomalies
  - Hematologic abnormalities manifest in childhood
  - Autosomal recessive
- **Fetal Valproate Syndrome**
  - Limb anomalies in 45-65%, including radial ray defects
  - Neural tube defect in 1-2%
  - Prenatal growth restriction

Helpful Clues for Rare Diagnoses

- **Cornelia de Lange Syndrome**
  - Severe prenatal onset growth deficiency
  - Variable degrees of upper limb deficiency including oligodactyly, phocomelia, micromelia
  - Radial hypoplasia/aplasia with abnormal thumbs
  - Long philtrum with micrognathia seen on prenatal ultrasound
  - Other structural anomalies including cardiac anomalies and diaphragmatic hernia
- **Roberts Syndrome**
  - Varying degrees of limb deficiency varying from tetra-amelia to tetraphocomelia to less severe limb reduction defects
  - Radial aplasia/hypoplasia in majority
  - Severe prenatal onset growth restriction with microcephaly
  - Cleft lip with or without cleft palate
  - Premature centromere separation in most cases confirms diagnosis
  - Autosomal recessive
- **Aase Syndrome**
  - Radial hypoplasia, triphalangeal thumbs
  - Diamond-Blackfan anemia (pure red cell aplasia)

Other Essential Information

- Association of radial ray anomalies and hematologic disorders
  - Awareness at delivery important
- Association of radial ray anomalies and cardiac malformations
  - Fetal echocardiography indicated
- 86% of patients with hypoplastic thumbs have other anomalies
- Careful search for other anomalies essential for syndromal diagnoses

Isolated



Ultrasound shows an isolated radial ray defect in a mid-trimester fetus. The angulation of the wrist ➡ is seen as well as a single bone in the forearm ➡. This fetus was otherwise normal.

VACTERL Association



Coronal ultrasound shows multiple segmentation defects ➡ of the lower spine in a fetus with VACTERL syndrome. Bilateral radial ray defects were also present.

RADIAL RAY MALFORMATION

Trisomy 18



Diabetic Embryopathy

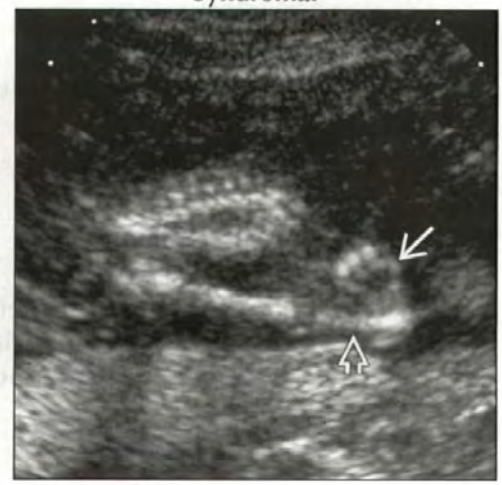


(Left) *Clinical photograph at autopsy shows the typical appearance of a radial ray defect in a fetus with trisomy 18. The forearm is shortened, and there is acute radial angulation at the wrist. The thumb is missing, and there is 2-3 syndactyly.* ***(Right)*** *Radiograph shows a stillborn fetus with severe diabetic embryopathy. Complete radial/ulnar aplasia is noted, as well as oligodactyly. The humerus is also shown.*

Diabetic Embryopathy



Syndromal



(Left) *Clinical photograph shows a radial ray defect in a term infant with diabetic embryopathy. Radial deviation is due to radial hypoplasia. The infant also had microtia and imperforate anus.* ***(Right)*** *Ultrasound shows a radial ray defect in an acardiac fetus (twin reversed arterial perfusion sequence). The single bone in the distal arm and abnormal hand are noted.*

Holt Oram Syndrome



Holt Oram Syndrome

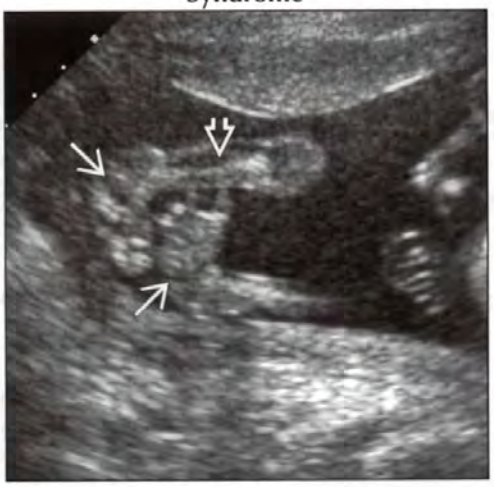


(Left) *Clinical photograph shows the hand of a woman with Holt Oram syndrome. Absence of thumb and thenar hypoplasia are noted. Abnormal palmar creases are seen. Clinodactyly is also present.* ***(Right)*** *Clinical photograph shows the hands of the infant of the same woman, also diagnosed with Holt Oram syndrome. A proximally implanted triphalangeal thumb is noted. Thenar hypoplasia is also present, as well as mild radial hypoplasia.*

Thrombocytopenia-Absent Radius Syndrome

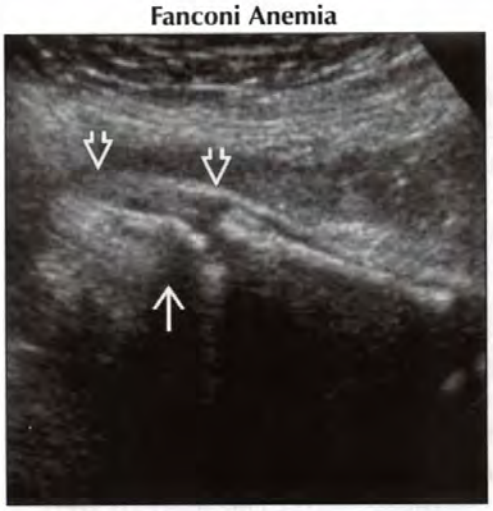


Thrombocytopenia-Absent Radius Syndrome



(Left) *Ultrasound shows the radial ray defect in a fetus with thrombocytopenia-absent radius syndrome (TAR). Radial hypoplasia is noted. Important in the diagnosis of TAR is the presence of thumbs.* ***(Right)*** *Ultrasound shows the bilateral abnormal lower extremities of a fetus with thrombocytopenia-absent radius syndrome. Clubfeet are noted as well as a single bone in the lower extremities.*

Fanconi Anemia



Fetal Valproate Syndrome



(Left) *Ultrasound shows the arm of a fetus diagnosed postnatally with Fanconi anemia. The hypoplastic forearm and severe radial angulation of the wrist and hand are seen.* ***(Right)*** *Ultrasound shows a 2nd trimester fetus with severe valproate embryopathy. Bilateral radial ray defects with radial aplasia, fixed radial deviation of the wrist, and absent thumbs are noted. Sacral spina bifida, microcephaly, and dysmorphic facies were also present.*

Cornelia de Lange Syndrome



Cornelia de Lange Syndrome



(Left) *Ultrasound shows a severely abnormal upper limb in a mid-trimester fetus with Cornelia de Lange syndrome. Radial and ulnar hypoplasia and monodactyly are noted.* ***(Right)*** *Clinical photograph shows a close-up of the limb reduction defect with monodactyly. Note the formed fingernail.*

ABNORMAL FOOT

DIFFERENTIAL DIAGNOSIS

Common
- Clubfoot
- Rockerbottom Foot
- Sandal Gap Foot
- Edema of Feet and Extremities

Less Common
- Amniotic Band Syndrome
- Ectrodactyly
- Abnormal Toes
- Terminal Transverse Limb Defect

Rare but Important
- Sirenomelia
- Amelia, Micromelia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is the anomaly an isolated finding?
- Are there multiple anomalies?
- Bilateral vs. unilateral
- Are hands similarly affected?
- Is the finding associated with abnormal or lack of fetal movement?
  - Lack of movement within the amniotic cavity?
  - Does the fetus appear "stuck" to the placenta?

Helpful Clues for Common Diagnoses
- **Clubfoot**
  - Plane of imaging important
    - Coronal view: Ankles should be straight
    - Lateral view: Bones of foot and tibia/fibula should not be seen in same plane
    - "Footprint" view: Clubfoot associated with abnormal shape/shortening
  - Bilateral vs. unilateral
  - Limitations to diagnosis
    - Gestational age: Clubfoot difficult to diagnose past 2nd trimester
    - Oligohydramnios
    - Crowding in multiple gestation
    - Footling breech position limits visualization of feet
- **Rockerbottom Foot**
  - Prominent heel associated with rounded bottom of foot
  - Best seen in lateral view
  - May be associated with dorsiflexed great toe, short hallux
  - Look carefully for other anomalies associated with trisomy 18
    - Persistently clenched hands or overlapping digits
- **Sandal Gap Foot**
  - Gap between great toe and 2nd toe
  - May be normal finding with fetal "flare" of toes while moving feet
  - Look for persistence of gap even when fetus at rest
  - Most often seen in normal fetuses but is a minor marker in trisomy 21
- **Edema of Feet and Extremities**
  - Pattern of edema important for establishing differential diagnosis
  - Edema of extremities vs. body edema
  - Unilateral edema concerning for obstructive phenomenon such as amniotic bands or tumor

Helpful Clues for Less Common Diagnoses
- **Amniotic Band Syndrome**
  - Amputations of digits or feet/hands
  - Usually asymmetrical
  - Associated disruptions of face, calvarium, body wall
  - Constriction rings of extremities
  - Adherence to placenta in severe cases
  - Extremity edema distal to constriction band is common
- **Ectrodactyly**
  - Split hand-split foot malformation
    - Fusion of digits with deep median cleft ("lobster claw")
  - May be unilateral or bilateral
  - May involve upper, lower, or both
  - Variable appearance between extremities
- **Abnormal Toes**
  - Polydactyly
    - Often difficult to diagnose, especially in early gestation
    - Foot usually appears wider than normal
    - Look at hands for extra fingers
    - Post-axial more common than pre-axial except in diabetic embryopathy
  - Syndactyly
    - Usually not diagnosed prenatally
    - May be associated with abnormal shape of foot
  - Oligodactyly

- Foot appears narrow
- Often associated with abnormality of tibia or fibula
- Abnormal position of foot common

- **Terminal Transverse Limb Defect**
  - Varying degree of limb deficiency
  - May be seen with congenital varicella infection

Helpful Clues for Rare Diagnoses

- **Sirenomelia**
  - Legs may appear "stuck" together
  - Single femur or fewer than 4 lower extremity long bones valuable clues
  - Foot/feet may be absent, single, or a complex fusion of structures
  - Usually associated with bilateral renal agenesis
  - May be difficult to visualize due to oligohydramnios
- **Amelia, Micromelia**
  - Extremities may be absent (amelia) vs. severely shortened (micromelia)
  - May be limited to upper or lower extremities
  - Digits may be present
  - Symmetrical micromelia associated with severe skeletal dysplasias

Other Essential Information

- If multiple anomalies, consider aneuploidy syndromes
  - Trisomy 18
    - Rockerbottom feet classic finding
    - Hands with overlapping digits
    - Multiple other anomalies including cardiac, orofacial, extremity
    - Severe growth restriction the rule by late 2nd trimester
  - Trisomy 21
    - Extremity findings are often subtle "soft marker" findings
    - Extremity anomalies: "Sandal gap" foot & clinodactyly, short humeri & femora
    - Pattern of edema in hydrops rarely involves hands and feet
    - Increased nuchal thickening in 2nd trimester
    - Flat mid-face with hypoplastic nasal bone common
    - May see tongue thrusting behavior in 3rd trimester
  - Turner syndrome
    - Pattern of edema characteristic
    - Edema of the dorsum of hands & feet
    - Anasarca common
    - Often with large cystic hygroma
    - Female gender
- Fetus with multiple anomalies: Transfer of care to a tertiary care center

Alternative Differential Approaches

- Clubfoot with abnormal calvarium shape: Neural tube defect
- Severe foot and extremity edema with cystic hygroma: Turner syndrome
- Rockerbottom feet with other anomalies and growth restriction: Trisomy 18

Clubfoot



Coronal ultrasound shows a clubfoot in a mid-trimester fetus. The bones of the distal lower extremity are seen in the same plane as those of the foot.

Clubfoot



Clinical photograph shows a severe clubfoot, which was bilateral in this infant.

ABNORMAL FOOT

Clubfoot

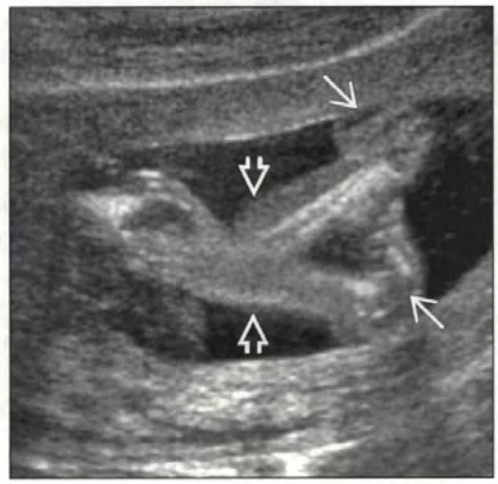


Clubfoot



(Left) *Coronal 3D ultrasound shows a posterior view of the leg. Note the inward deviation the heel and foot. The fetus had abnormal in utero movement and tested positive for myotonic dystrophy.* ***(Right)*** *Clinical photograph shows a unilateral clubfoot and shortened extremity in an infant with multiple unilateral anomalies.*

Clubfoot



Rockerbottom Foot



(Left) *Coronal ultrasound shows severe bilateral clubfeet in a mid-trimester fetus with fetal akinesia sequence. The persistent "scissored" posture of the legs is commonly seen in fetuses with hypertonia or neurologic abnormality.* ***(Right)*** *Clinical photograph shows bilateral rockerbottom feet in a premature stillborn neonate with trisomy 13. Note the prominent calcaneus and dorsiflexed great toes.*

Rockerbottom Foot



Rockerbottom Foot



(Left) *Coronal ultrasound of the lower leg shows a prominent heel with a rounded appearance to the bottom of the foot, characteristic of rockerbottom foot.* ***(Right)*** *Clinical photograph shows a rockerbottom foot in a full-term newborn with trisomy 18. Note the wrinkled skin due to the lack of subcutaneous tissue in this growth restricted infant. The great toe appears short because of a short 1st metatarsal.*

ABNORMAL FOOT

Sandal Gap Foot



Sandal Gap Foot



(Left) *Ultrasound shows a typical appearance in the mid-trimester of a sandal gap foot in a fetus with trisomy 21. Note the gap between the 1st and 2nd toes.* ***(Right)*** *Clinical photograph shows bilateral sandal gap feet in a stillborn mid-trimester fetus with trisomy 21. The gap between the great and second toes can be seen.*

Edema of Feet and Extremities



Edema of Feet and Extremities

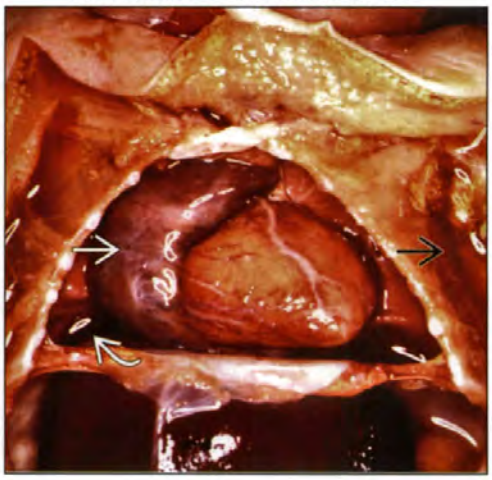


(Left) *Lateral ultrasound shows the lower extremities in a fetus with severe hydrops due to monosomy X (Turner syndrome). The dorsal pedal edema and edema of the lower leg are characteristic.* ***(Right)*** *Clinical photograph shows a mid-trimester fetal demise with Turner syndrome. Hydrops is apparent with a huge cystic hygroma and a protuberant abdomen due to ascites. Edema of the dorsum of the hands and feet is typical in Turner syndrome.*

Edema of Feet and Extremities



Edema of Feet and Extremities



(Left) *Clinical photograph shows severe extremity and dorsal pedal edema (all soft tissues were equally affected) in a term newborn with hydrops due to supraventricular tachycardia. Although classically described with Turner syndrome, dorsal pedal edema can be seen with generalized, severe, diffuse edema.* ***(Right)*** *Autopsy photograph of the same infant shows cardiomegaly, subcutaneous edema, and pleural fluid.*

ABNORMAL FOOT

Amniotic Band Syndrome

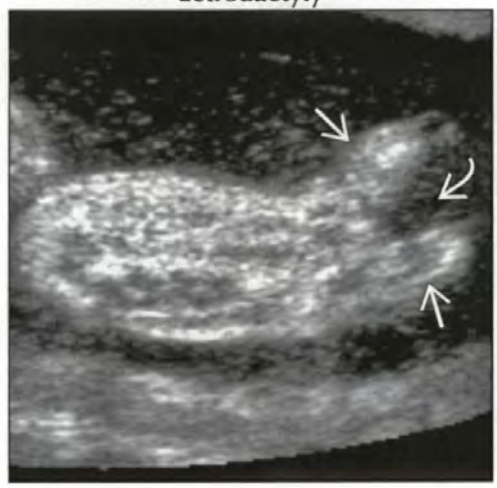


Amniotic Band Syndrome



(Left) *Ultrasound of the right lower extremity shows a clubfoot ➡ with a cleft created by missing amputated toes ➡. Multiple other extremity and body deformities were seen, including acrania, abdominal wall defect, and facial cleft. Bands are often tightly adherent to the fetus and not visible by sonography.* ***(Right)*** *Gross pathology shows the extremity deformities (➡ corresponds to the finding on ultrasound), as well as a large abdominal wall defect.*

Ectrodactyly



Abnormal Toes

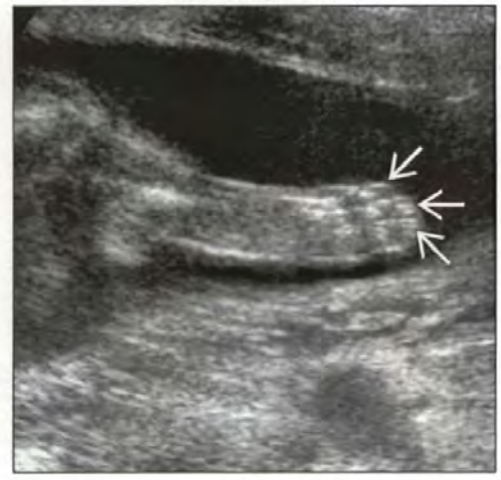


(Left) *Ultrasound of the foot in a mid-trimester fetus shows a median defect ➡ with syndactyly of the remaining digits ➡ resulting in a "lobster claw" appearance. This condition may be either unilateral or bilateral and may also involve the hands.* ***(Right)*** *Radiograph shows complex polysyndactyly in an infant with Pfeiffer syndrome. Note the partial duplication and fusion of the great toe ➡ and 1st metatarsal ➡.*

Abnormal Toes



Abnormal Toes



(Left) *Clinical photograph shows bilateral clubfeet in this newborn with bilateral postaxial polydactyly ➡. Note that the feet appear abnormally wide.* ***(Right)*** *Coronal ultrasound shows oligodactyly ➡ in a foot of a fetus with tibial hemimelia. The foot appeared narrow in comparison with the normal foot.*

ABNORMAL FOOT

Abnormal Toes



Abnormal Toes



(Left) *Ultrasound shows significant brachydactyly ➡ in this mid-trimester fetus with a nonlethal skeletal dysplasia.* ***(Right)*** *Clinical photograph shows 2-3 toe syndactyly in this newborn's foot. 2-3 syndactyly is the most common type of syndactyly and is autosomal dominant in some families. The degree of fusion is highly variable, but it most often involves only soft tissues.*

Terminal Transverse Limb Defect



Sirenomelia

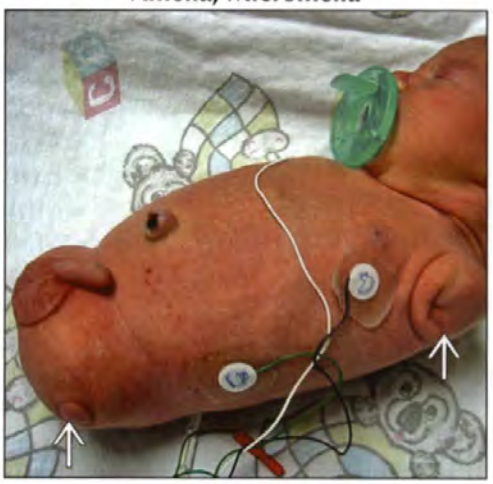


(Left) *Clinical photograph shows a terminal transverse limb defect ⇨ in this term newborn. The anterior portion of the foot, including the metatarsals and all the toes, was absent.* ***(Right)*** *Clinical photograph shows complex abnormalities of the feet ➡ in this stillborn fetus with bilateral renal agenesis and sirenomelia.*

Sirenomelia



Amelia, Micromelia



(Left) *Sagittal ultrasound shows the very abnormal lower extremity in this mid-trimester fetus with sirenomelia. There is a single femur ➡ and a single lower leg bone ➡ without a foot.* ***(Right)*** *Clinical photograph shows a newborn with tetra-amelia. Small mounds of tissue ➡ are noted without any definable extremities.*

ABNORMAL DIGITS

DIFFERENTIAL DIAGNOSIS

Common

- Polydactyly
  - Trisomy 13
  - Meckel-Gruber Syndrome
  - Familial
  - Diabetic Embryopathy
- Syndactyly
  - Familial
  - Amniotic Band Syndrome
  - Triploidy
  - Apert Syndrome
  - Carpenter Syndrome
- Abnormal Hands
  - Clinodactyly
  - Oligodactyly
  - Camptodactyly
  - Brachydactyly
  - Trident Hand
  - Arachnodactyly
  - Broad Thumbs
  - Radial Ray Malformation
  - Clenched Fingers
    - Trisomy 18
    - Arthrogryposis, Akinesia Sequence
  - "Hitchhiker Thumb"
    - Diastrophic Dysplasia
- Abnormal Feet
  - Sandal Gap Foot
  - Broad Toes
  - Brachydactyly
  - Overlapping Toes

Less Common

- Ectrodactyly

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Does the abnormality affect upper or lower extremities, or both?
- Is the abnormality unilateral or bilateral?
- Is there an associated hand, foot, or limb anomaly or does the change affect the digits only?
- Are there other structural anomalies of the fetus?
- Are there changes suggestive of a skeletal dysplasia?
- Is the fetus normally grown or is there associated severe growth restriction?
- Is there evidence of amniotic bands?
- Does the fetal move normally?

Helpful Clues for Common Diagnoses

- **Polydactyly**
  - **Trisomy 13**
    - Often bilateral, affecting hands and feet, but may also be limited to 1 or 2 limbs
    - Most commonly postaxial
    - Associated growth restriction
    - Other structural anomalies common: Orofacial cleft, holoprosencephaly, cardiac, omphalocele, genitourinary
  - **Meckel-Gruber Syndrome**
    - Classic triad of posterior encephalocele, dysplastic kidneys and polydactyly
    - Most commonly postaxial
    - May affect any or all extremities
  - **Familial**
    - Most commonly postaxial
    - Increased prevalence in African Americans
    - Hands more commonly affected than feet, but may affect all extremities
  - **Diabetic Embryopathy**
    - Preaxial polydactyly
    - Multiple anomalies: Cardiac, neural tube defects, femoral hypoplasia
- **Syndactyly**
  - **Familial**
    - Most commonly 2-3 toe syndactyly
    - Autosomal dominant
    - Usually isolated
  - **Amniotic Band Syndrome**
    - Pseudosyndactyly
    - Often associated with band entwining digits
    - Extremities discordant for defects
  - **Triploidy**
    - Syndactyly of digits 3 and 4 of hands most common
    - Association with partial molar gestation
    - Early onset, severe growth restriction
    - Multiple structural anomalies
    - Maternal severe preeclampsia in the mid-trimester
    - Maternal bilateral, enlarged cystic ovaries (theca lutein cysts)
  - **Apert Syndrome**
    - "Mitten" syndactyly of hands and feet
    - Severe soft tissue and bony syndactyly
    - Coronal craniosynostosis

- **Carpenter Syndrome**
  - Complex syndactyly, associated with broad, deviated toes and thumbs
  - Abnormal calvarial shape due to complex craniosynostosis

- **Abnormal Hands**
  - **Clinodactyly**
    - 5th finger most common, but can affect any digit
    - Commonly seen in Down syndrome
  - **Oligodactyly**
    - Hand or foot may appear "too narrow"
    - May be seen in cases of amniotic bands
    - May be an isolated defect vs. part of complex limb reduction defect
  - **Camptodactyly**
    - Inability to fully extend fingers
    - Usually involves multiple fingers
  - **Brachydactyly**
    - Shortened digits
    - Often bilateral; may affect both hands and feet
    - Common in skeletal dysplasias
  - **Trident Hand**
    - Fingers appear to be of a similar length
    - Achondroplasia
    - Thanatophoric dysplasia
  - **Arachnodactyly**
    - Long "spider-like" fingers associated with Marfan syndrome and homocystinuria
    - Not usually appreciated on prenatal ultrasound
  - **Broad Thumbs**
    - ± Abnormal deviation of digit
  - **Radial Ray Malformation**
    - Associated with hypoplasia or aplasia of one or both thumbs
    - Radial deviation of wrists
    - Isolated anomaly vs. part of a syndrome
  - **Clenched Fingers**
    - Trisomy 18
    - Arthrogryposis/akinesia sequence
  - **"Hitchhiker Thumb"**
    - Adducted thumb (also great toe)
    - Seen in diastrophic dysplasia
    - Generalized shortening of all long bones

- **Abnormal Feet**
  - **Sandal Gap Foot**
    - Gap between the 1st and 2nd toes
    - "Soft marker" for Down syndrome
  - **Broad Toes**
    - Associated with deviation of toes
  - **Brachydactyly**
    - May be associated with brachydactyly of fingers, shortened long bones
  - **Overlapping Toes**
    - May appear "crowded" or "jumbled" on ultrasound

Helpful Clues for Less Common Diagnoses

- **Ectrodactyly**
  - Split hand-split foot malformation ("lobster claw")
  - Characterized by deficiency/hypoplasia of digits, deep median cleft and fusion of remaining digits
  - Defects often asymmetrical, discordant

Polydactyly



Clinical photograph shows bilateral postaxial polydactyly ➔ in an infant with a normal karyotype. This was an isolated finding.

Trisomy 13



Clinical photograph shows a term growth restricted infant with trisomy 13. Several anomalies are noted including postaxial polydactyly ➔, a small omphalocele ➔, and bilateral orofacial clefts ➔.

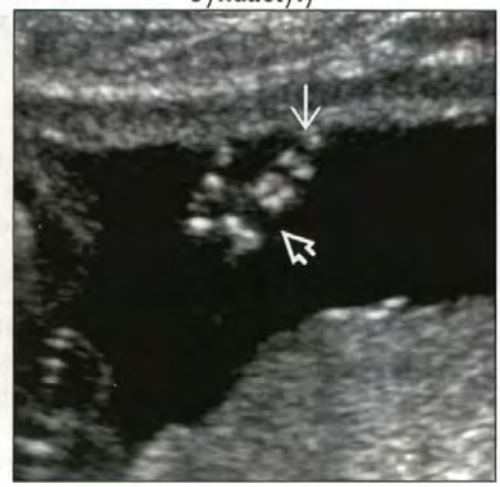
ABNORMAL DIGITS

Diabetic Embryopathy



(Left) *Clinical photograph shows both preaxial polydactyly and syndactyly in an infant with bilateral femoral hypoplasia due to diabetic embryopathy. Nail hypoplasia is also noted on the syndactylous digits.* ***(Right)*** *Ultrasound shows a complex hand malformation in a mid-trimester fetus. There is soft tissue syndactyly of the 2nd and 3rd digits. Ulnar deficiency resulted in oligodactyly with missing digits 4 and 5.*

Syndactyly



Triploidy



(Left) *Coronal ultrasound shows overlapping toes in a fetus with syndactyly associated with triploidy. Abnormal leg posture due to sacral agenesis and ambiguous genitalia in this male fetus are also evident.* ***(Right)*** *Radiograph shows the hand of an infant with multiple anomalies. Clinodactyly of the 5th and 2nd fingers can be seen. Both abnormalities are associated with hypoplasia of the middle phalanges. Also note the short broad thumb.*

Clinodactyly



Oligodactyly



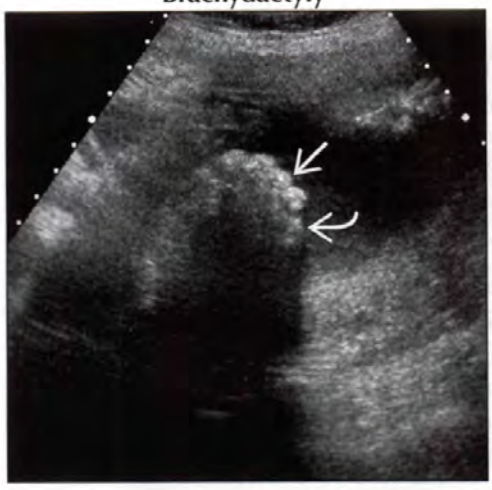
(Left) *Ultrasound shows a very abnormal hand in a fetus with multiple anomalies. Only 4 digits were present with absence of the thumb. Camptodactyly of the remaining digits was noted. The ulnar deviation of the wrist is unusual with a radial deficiency.* ***(Right)*** *Clinical photograph shows the same infant after birth with absent thumb and camptodactyly of the remaining digits.*

Camptodactyly



ABNORMAL DIGITS

Brachydactyly



Trident Hand



(Left) *Ultrasound shows very short toes ➡ in a 3rd trimester fetus with a nonlethal skeletal dysplasia. There is a proximally placed, short great toe ➡.* ***(Right)*** *Clinical photograph shows significant brachydactyly and a "trident hand" ➡ in a stillborn fetus with thanatophoric dysplasia. Note the tiny chest ➪ associated with severe pulmonary hypoplasia.*

Radial Ray Malformation

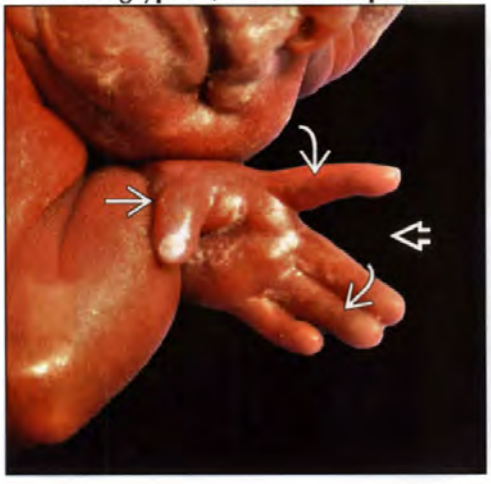


Trisomy 18



(Left) *Clinical photograph shows a radial ray abnormality in an infant with multiple anomalies. The thumb ➪ is hypoplastic and deviated. Camptodactyly ➪ of the other digits is also apparent.* ***(Right)*** *3D ultrasound shows the typical abnormal hand posture of a 3rd trimester fetus with trisomy 18. There is a clenched fist with an overriding 2nd finger ➡.*

Arthrogryposis, Akinesia Sequence



Sandal Gap Foot



(Left) *Photograph of a stillborn with fetal akinesia sequence shows an adducted, hypoplastic thumb ➡. Fingers are extended, with a wide gap between 2nd & 3rd fingers ➪. The absence of flexion creases ➡ proves lack of fetal movement in utero.* ***(Right)*** *US shows a sandal gap foot in a mid-trimester fetus at risk for trisomy 21 based on an abnormal maternal serum screen. Note wide gap between 1st & 2nd toes ➡. This was persistent despite fetal movement.*

POLYDACTYLY

DIFFERENTIAL DIAGNOSIS

Common
- Familial
- Trisomy 13
- Diabetic Embryopathy

Less Common
- Meckel-Gruber Syndrome
- Smith-Lemli-Opitz Syndrome
- Carpenter Syndrome
- Asphyxiating Thoracic Dysplasia

Rare but Important
- Short Rib-Polydactyly
- Ellis-van Creveld
- Majewski Syndrome
- Mohr Syndrome (Oral-Facial-Digital Syndrome)
- Bardet-Biedel Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Clinical history important
  - Maternal history of diabetes, advanced maternal age (AMA)
  - Family history of polydactyly
- Are there other associated limb anomalies?
- Are there other structural anomalies?
- Is there evidence of a skeletal dysplasia?
- Is there growth restriction (IUGR)?
- Where is the extra digit (preaxial vs. postaxial)?

Helpful Clues for Common Diagnoses
- **Familial**
  - Most commonly postaxial
  - Increased prevalence in African Americans
- **Trisomy 13**
  - Multiple anomalies: Orofacial clefts, holoprosencephaly, cardiac, IUGR, AMA
- **Diabetic Embryopathy**
  - Multiple anomalies: Neural tube defects, holoprosencephaly, anorectal, cardiac

Helpful Clues for Less Common Diagnoses
- **Meckel-Gruber Syndrome**
  - Classic triad of polydactyly, posterior encephalocele, dysplastic kidneys
- **Smith-Lemli-Opitz Syndrome**
  - Severe early onset IUGR, cardiac defects, holoprosencephaly, very low estriol
- **Carpenter Syndrome**
  - Craniosynostosis, complex digital anomalies, cardiac anomalies
- **Asphyxiating Thoracic Dysplasia**
  - Short horizontal ribs, cystic kidneys, postaxial polydactyly, short extremities

Helpful Clues for Rare Diagnoses
- **Short Rib-Polydactyly**
  - Pre- and postaxial polydactyly, short horizontal ribs, micromelia, hydrops
- **Ellis-van Creveld**
  - Postaxial polydactyly, cardiac anomalies, short extremities, small thorax

Familial



Clinical photograph shows a newborn infant, one of monozygotic twins, born with preaxial polydactyly. The infant was otherwise normal and had a normal karyotype.

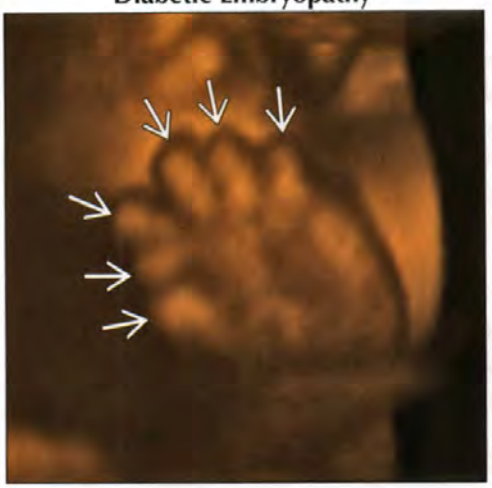
Trisomy 13



Clinical photograph shows an unusual insertional polydactyly in a stillborn fetus with trisomy 13. Note the extra hypoplastic appearing digit. Polydactyly in trisomy 13 is more commonly postaxial.

POLYDACTYLY

Diabetic Embryopathy



Diabetic Embryopathy



(Left) *Coronal 3D ultrasound in this fetus of a poorly controlled diabetic clearly shows polydactyly. There was also a short tibia and fibula with severe angulation at the ankle.* ***(Right)*** *Clinical photograph in the same case confirms polydactyly and clubbing of both feet. Polydactyly is one of the many anomalies seen in diabetic embryopathy.*

Smith-Lemli-Opitz Syndrome



Smith-Lemli-Opitz Syndrome



(Left) *Ultrasound shows the hand of a mid-trimester fetus with Smith-Lemli-Opitz syndrome. The "jumbled" appearance of multiple digits is commonly seen in polydactyly in the fetus. Six digits can be seen and correlated with the postnatal appearance of the hand.* ***(Right)*** *Clinical photograph shows the hand of the same infant at birth. Postaxial polydactyly is seen as well as camptodactyly of multiple digits.*

Carpenter Syndrome



Carpenter Syndrome



(Left) *Ultrasound shows the hands of a mid-trimester fetus with Carpenter syndrome. The hands appear clenched. Preaxial polydactyly can be suspected on one of the hands, which appears "too wide".* ***(Right)*** *Clinical photograph shows the hand of the same Carpenter syndrome infant at birth with preaxial polydactyly. Extensive soft tissue syndactyly was also apparent and likely correlated with the prenatal appearance of persistently clenched hands.*

SYNDACTYLY

DIFFERENTIAL DIAGNOSIS

Common
- Syndromic Syndactyly
  - Apert Syndrome
  - Carpenter Syndrome
  - Triploidy
  - Pfeiffer Syndrome
  - Smith-Lemli-Opitz Syndrome
- Non-Syndromic Syndactyly
  - Amniotic Band Syndrome
  - Familial

Less Common
- Diabetic Embryopathy

Rare but Important
- Greig Cephalopolysyndactyly
- Ectrodactyly

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Are there associated limb anomalies?
- Are there other structural anomalies?
- Abnormal calvarium/craniosynostosis?
- Evidence of amniotic bands?
- Is there growth restriction (IUGR)?
- Must look carefully
  - Syndactyly is frequently missed by prenatal ultrasound

Helpful Clues for Common Diagnoses
- **Syndromic Syndactyly**
  - **Apert Syndrome**
    - "Mitten" syndactyly of hands and feet
    - Acrocephaly
  - **Carpenter Syndrome**
    - Craniosynostosis
    - Brachydactyly with clinodactyly, camptodactyly and syndactyly
  - **Triploidy**
    - Classic 3-4 syndactyly of the hands
  - **Pfeiffer Syndrome**
    - Craniosynostosis/cloverleaf skull
    - Complex partial syndactyly of hands, feet
  - **Smith-Lemli-Opitz Syndrome**
    - 2-3 toe syndactyly
- **Non-Syndromic Syndactyly**
  - **Amniotic Band Syndrome**
    - Distal digits "stuck" together by bands
    - Actual band may be difficult to see
    - May cause amputations & "slash" defects
  - **Familial**
    - 2-3 toe syndactyly

Helpful Clues for Less Common Diagnoses
- **Diabetic Embryopathy**
  - Preaxial polydactyly & multiple anomalies

Helpful Clues for Rare Diagnoses
- **Greig Cephalopolysyndactyly**
  - Postaxial polydactyly of hands with 3-4 syndactyly
  - Preaxial polydactyly of feet with 1-3 syndactyly
- **Ectrodactyly**
  - Split hand-split foot malformation
  - Subtle syndactyly present in otherwise normal carriers

Apert Syndrome



Clinical photograph shows a stillborn fetus with Apert syndrome. The characteristic "mitten" syndactyly ⇨ is seen, as well as the acrocephaly ⇨, which is due to coronal synostosis.

Carpenter Syndrome



Ultrasound shows the broad foot of a mid-trimester fetus with Carpenter syndrome. Extensive syndactyly of all the toes ➡ is noted.

SYNDACTYLY

Triploidy



Triploidy

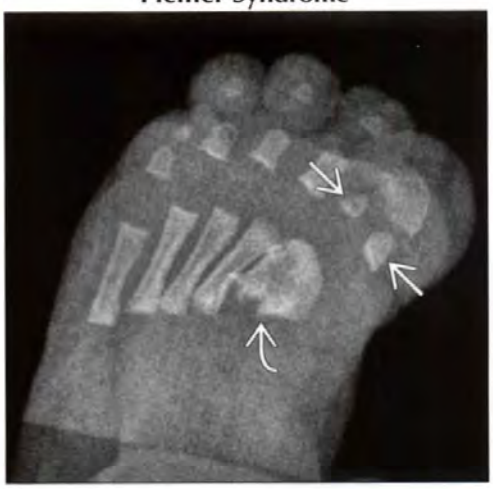


(Left) *Ultrasound shows the hand of a mid-trimester fetus with triploidy. Syndactyly of digits 3 and 4 is noted. Digit 5 is closely approximated to 4, suggestive of further syndactyly.* ***(Right)*** *Clinical photograph shows the hand of a stillborn mid-trimester fetus with triploidy. The classic 3-4 syndactyly is seen. In addition, partial syndactyly between digits 4 and 5 can be seen. Clinodactyly of digit 2 is also apparent.*

Pfeiffer Syndrome



Pfeiffer Syndrome



(Left) *Clinical photograph shows extensive soft tissue syndactyly between all the toes in a newborn with Pfeiffer syndrome. Note also the broad great toe, which is actually duplicated.* ***(Right)*** *Radiograph shows the foot of the same infant with Pfeiffer syndrome. The soft tissue syndactyly seen clinically is difficult to appreciate. Note the duplicated metatarsal and proximal phalanx of the great toe.*

Smith-Lemli-Opitz Syndrome



Smith-Lemli-Opitz Syndrome



(Left) *Ultrasound shows the foot of a fetus with Smith-Lemli-Opitz syndrome. Syndactyly of the toes is difficult to diagnose on ultrasound, but the "crowded" appearance of the toes correlated with syndactyly at birth.* ***(Right)*** *Clinical photograph shows the feet of a young adult with Smith-Lemli-Opitz syndrome. Characteristic 2-3 toe syndactyly can be seen.*

SECTION 8
Multiple Gestations

Membranes in Multiple Gestations	8-2
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Asymmetric Fluid Distribution	8-10
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Conjoined Twins	8-20

MEMBRANES IN MULTIPLE GESTATIONS

DIFFERENTIAL DIAGNOSIS

Common
- Dichorionic Diamniotic Twins
- Monochorionic Diamniotic Twins
- Monochorionic Monoamniotic Twins

Less Common
- Bleed Between Membranes
- Synechiae
- Chorioamniotic Separation
- Amniotic Band Syndrome

Rare but Important
- Uterine Duplication

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Twin types
  - 70% dizygotic: Two separate fertilized ova
  - 30% monozygotic: Single zygote splits at various times post conception
    - Zygote splits at < 3 days post conception → dichorionic twins (30%)
    - Split occurs 4-8 days post conception → monochorionic diamniotic twins (60-65%)
    - Split occurs 8-12 days post conception → monoamniotic twins (5-10%)
    - Split occurs > 13 days post conception → conjoined twins (< 1%)
- Dichorionic twins have best prognosis with high likelihood of two liveborn infants
- Important to recognize monochorionic twins due to specific complications
  - Twin twin transfusion syndrome (TTTS)
  - Twin reversed arterial perfusion (TRAP) sequence
- Important to recognize monoamniotic twins as high risk of cord accidents, as well as other complications of monochorionic twinning

Helpful Clues for Common Diagnoses
- **Dichorionic Diamniotic Twins**
  - First trimester: Two complete thick echogenic chorionic rings
  - Thick membrane
    - 2 layers chorion + 2 layers amnion = 4 layers = thick membrane
    - In second and third trimester use high frequency transducer to count layers if chorionicity otherwise unclear
  - "Twin peak" or "lambda" sign
    - Echogenic chorionic tissue extends between layers of amnion
    - Triangle shape with base on placental surface
    - Apex of triangle fades into inter-twin membrane
  - Two placentas
    - Can be difficult to determine if placentas implanted side by side or fused
  - Different genders = dizygotic = dichorionic by definition
- **Monochorionic Diamniotic Twins**
  - First trimester: One thick echogenic outer ring containing two thin rings of amnion
    - Count yolk sacs if amnion not yet visible
    - Number of yolk sacs = number of amnions
  - Thin membrane
    - 2 layers of amnion + no chorion = thin membrane
  - "T" sign
    - Thin membrane abuts placental surface at 90° without echogenic triangle of chorionic tissue
  - Single placenta
    - Beware pitfall of fused placentas in dichorionic twins
    - Do not confuse succenturiate lobe with second complete placenta
  - Monochorionic twins must be same gender
- **Monochorionic Monoamniotic Twins**
  - First trimester: One thick echogenic ring containing two embryos inside a single amniotic sac with single yolk sac
  - No inter-twin membrane
    - Beware pitfall of TTTS with anhydramnios of one twin
    - "Stuck" twin stays adherent to uterine wall
    - Membrane difficult to see: Look carefully between extremities or where head is adjacent to uterine wall
  - Monoamniotic twins free to move around each other
    - Cord entanglement only occurs in absence of membrane

- Single placenta
  - Cord insertion sites often closely approximated

Helpful Clues for Less Common Diagnoses

- **Bleed Between Membranes**
  - Apparent thick membrane but echogenicity < chorion
  - Echogenicity and thickness changes quite rapidly as clot breaks down
- **Synechiae**
  - Ask patient about prior surgeries
  - In first trimester, can cause distorted sac shape
  - Synechiae cross uterine cavity, may look like thick membrane
    - In singleton pregnancy, look for fetus straddling synechia or placenta on one side/fetus on the other
    - In twin pregnancy look at relationship to placenta: Membranes reflect off placenta, synechial attachment to myometrium can be anywhere
- **Chorioamniotic Separation**
  - Visualization of chorion and amnion as separate layers after 14-16 weeks
  - May be associated with aneuploidy when seen early in second trimester
  - More usually complication of intervention
    - Amniocentesis
    - Amnioreduction in TTTS or for polyhydramnios in one twin
    - Hysteroscopic laser ablation of abnormal vascular connections in TTTS or TRAP
  - Chorioamniotic separation in singleton pregnancy
    - One fetus with "membrane" tending to parallel margins of cavity rather than divide it
    - Do not confuse with failed twin gestation
- **Amniotic Band Syndrome**
  - Very variable presentation: Constriction bands on extremities to major amputation/destructive defects
  - Look for fine filaments in amniotic fluid
  - Fetus often "tethered" by bands; may need to vary maternal position to float fetus away from uterine wall

Helpful Clues for Rare Diagnoses

- **Uterine Duplication**
  - In first trimester, use 3D to create coronal image for fundal contour
  - Beware pregnancy in rudimentary horn → risk rupture with catastrophic hemorrhage

Other Essential Information

- Twins account for 1.1% of births in USA but 10% of perinatal morbidity and mortality
- Prognosis in multiple gestation relates to chorionicity
- Best imaging tool for membrane assessment/determination of chorionicity is first trimester transvaginal ultrasound
- "Twin peak" sign is reliable indicator of dichorionicity but is not absolute
- "T" sign most often seen in monochorionic twins but does not exclude dichorionicity

Dichorionic Diamniotic Twins



Transvaginal ultrasound shows two thick, echogenic rings ➡ typical of dichorionic twinning. By the second trimester the membrane looked thin, but chorionicity had been firmly established in the first trimester.

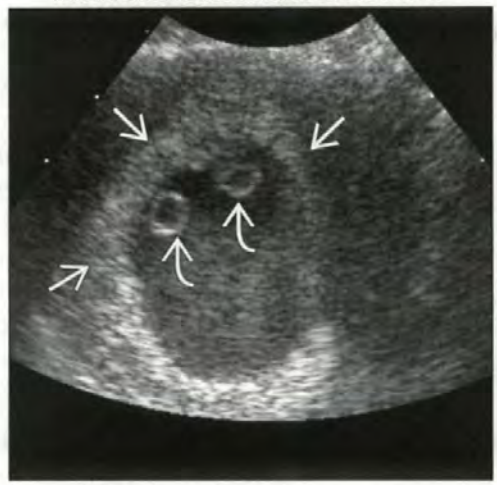
Dichorionic Diamniotic Twins



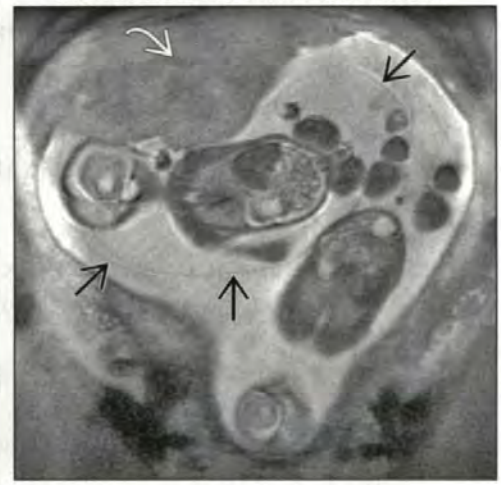
Ultrasound shows a thick membrane ➡ and "twin peak" ➪ sign. However in this case, the different genders indicate that the twins are dizygotic and therefore dichorionic by definition.

MEMBRANES IN MULTIPLE GESTATIONS

Monochorionic Diamniotic Twins

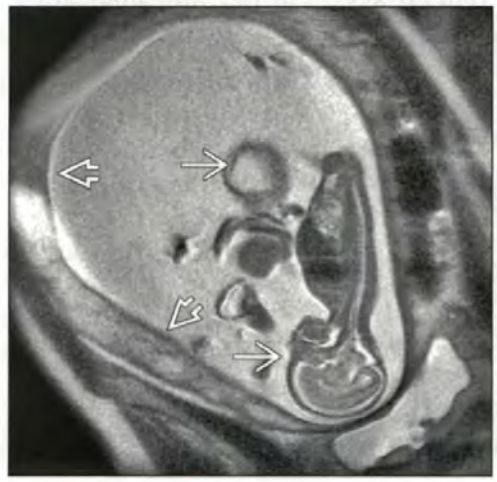


Monochorionic Diamniotic Twins



(Left) *Transabdominal ultrasound shows two yolk sacs within a single chorionic sac. The amnion is not yet visible but two yolk sacs correlates strongly with diamnionicity. Monochorionic diamniotic twinning was confirmed at follow-up.* ***(Right)*** *T2WI MR following successful shunt obliteration for twin-twin transfusion syndrome shows two fetuses, a single placental mass, and a thin inter-twin membrane with symmetric amniotic fluid distribution.*

Monochorionic Monoamniotic Twins



Monochorionic Monoamniotic Twins



(Left) *T2WI MR shows two fetuses with a single placenta and no inter-twin membrane. Note fetal proximity despite polyhydramnios. Real time observation excluded conjoined twins.* ***(Right)*** *Gross pathology shows a cord knot in a pair of monoamniotic twins with demise of one twin at 27 weeks. The dark cord was thrombosed and belonged to the dead twin. The living twin was premature but did well.*

Synechiae

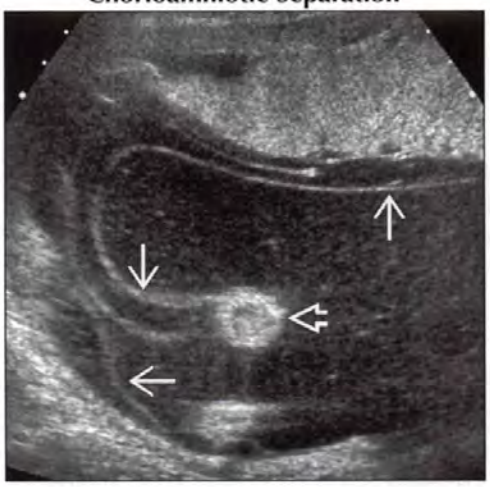


Synechiae

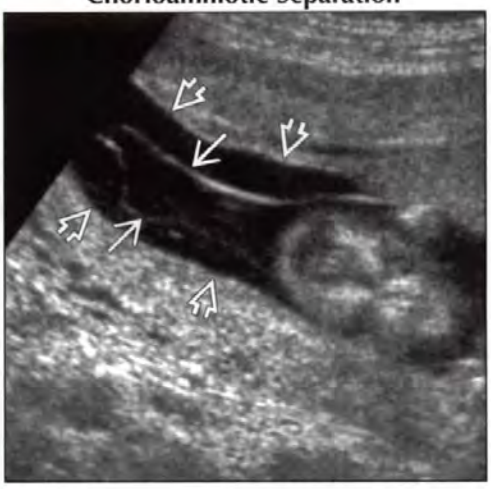


(Left) *Axial ultrasound shows an apparent inter-twin membrane and twin peak sign. In fact this is a singleton in a patient with synechiae. Fetal parts are seen on both sides of the synechia. The fetus was freely mobile, distinguishing this from amniotic bands.* ***(Right)*** *Axial ultrasound early in the second trimester shows the edge of the placenta apparently lifted off the uterine wall. This appearance is secondary to placental implantation upon a uterine synechia.*

Chorioamniotic Separation

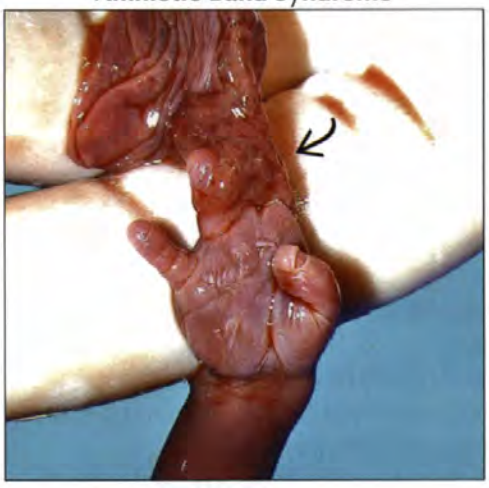


Chorioamniotic Separation



(Left) *Transabdominal ultrasound shows the thin amnion separated from the chorion and draped over the fetal extremities. This patient had been treated with serial amnioreduction.* ***(Right)*** *Sagittal ultrasound at 16 weeks shows delayed fusion of the amnion with the chorion. Note how the membrane parallels the uterine wall. Multiple associated anomalies were due to trisomy 18.*

Amniotic Band Syndrome

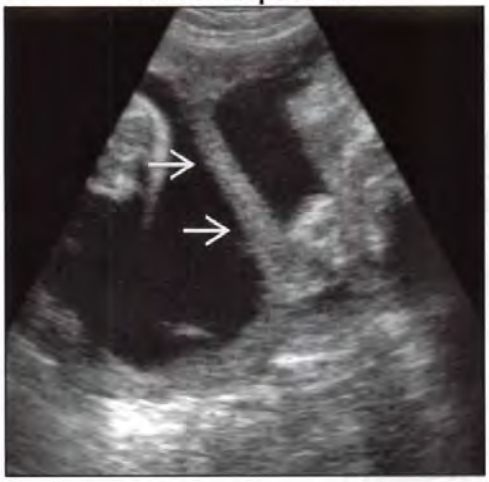


Amniotic Band Syndrome

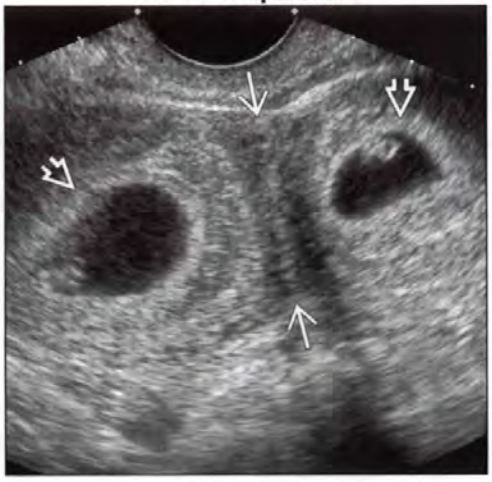


(Left) *Gross pathology shows a band trapping fetal fingers. In this case, a band constricted the cord causing fetal demise. Inter-twin membranes do not attach to the fetus. Even with a "stuck" twin the membranes are closely applied to, but not adherent to, the fetus.* ***(Right)*** *Transabdominal ultrasound shows a band within the amniotic fluid. Bands extend from the uterine wall to the fetal surface often "anchoring" the fetus in a fixed position.*

Uterine Duplication



Uterine Duplication



(Left) *Ultrasound shows an apparent very thick inter-twin "membrane". In fact, this is a uterine septum. It is essential to follow all linear echoes in the amniotic fluid to differentiate membranes from synechia from septa.* ***(Right)*** *Axial transvaginal ultrasound shows a dichorionic twin pregnancy. The separating "membrane" is much thicker and more hypoechoic than expected and is actually myometrium. This is a twin pregnancy in a bicornuate uterus.*

DISCORDANT TWIN GROWTH

DIFFERENTIAL DIAGNOSIS

Common
- Idiopathic and Normal
- Intrauterine Growth Restriction of One Twin
- Twin-Twin Transfusion Syndrome
- Unequal Placental Sharing

Less Common
- Anomaly of One Twin

Rare but Important
- Twin Reversed Arterial Perfusion

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Must differentiate small but normal twin from abnormally grown twin
  - By convention term "discordance" is used when one twin meets criteria for intrauterine growth restriction (IUGR)
    - Estimated fetal weight (EFW) < 10th percentile
  - Size difference in twins not clinically significant if both show appropriate interval growth and normal fluid
- Determination of chorionicity essential for management
  - Disparate size not uncommon in dichorionic gestation
    - Exclude anomaly/aneuploidy
    - Check placental cord insertion sites
    - Track growth/deepest fluid pockets at 3-4 week intervals
    - Doppler studies if either twin meets criteria for IUGR
  - Certain conditions only occur in monochorionic (MC) gestations
    - Twin-twin transfusion syndrome (TTTS)
    - Twin reversed arterial perfusion sequence (TRAP)
    - Unequal placental sharing
  - In MC twins, demise of one twin has profound consequences for the survivor
    - Demise of one MC twin may → "twin embolization syndrome" (better thought of in terms of exsanguination or hypotension rather than embolization)
    - Results in ischemic brain/cardiac injury in survivor
    - Preterm delivery confers risks of prematurity to both but may be a better option than continuing the pregnancy with risk of demise of one twin
  - In dichorionic twins with one at-risk fetus, goal is to maximize outcome for healthy fetus

Helpful Clues for Common Diagnoses
- **Idiopathic and Normal**
  - Appropriate interval growth even if at low end of normal range
  - Normal amniotic fluid volume, normal Doppler studies, no anomalies
- **Intrauterine Growth Restriction of One Twin**
  - Less than expected interval growth
  - Associated with unequal placental sharing
    - Look for velamentous or marginal cord insertion
  - Look at placental location
    - Implantation on septum or over large fibroids
  - Look for evidence of abruption
  - Early onset concerning for anomaly, syndrome or aneuploidy
    - Consider amniocentesis
  - Late onset more suggestive of placental insufficiency
    - Use Doppler to evaluate fetoplacental circulation
- **Twin-Twin Transfusion Syndrome**
  - Monochorionic twins with artery-to-vein anastomoses in the placenta
  - Asymmetric size with true discordance in many cases
  - Recipient: Larger twin with polyhydramnios
  - Donor: Smaller twin with oligohydramnios
  - Staging of TTTS
    - Stage 1: Donor bladder visible, Doppler normal
    - Stage 2: Donor bladder empty, Doppler normal
    - Stage 3: Donor bladder empty, Doppler abnormal
    - Stage 4: Hydrops in recipient
    - Stage 5: Demise of one or both twins
- **Unequal Placental Sharing**
  - Velamentous cord insertion
    - Cord inserts onto membranes away from placental margin

- Strong association with TTTS; monitor carefully for fluid volume/growth
- Look for associated vasa previa; if present mandates delivery by cesarean section before onset of labor

- Marginal cord insertion
  - Cord inserts at edge of placenta rather than onto placental disc
- Eccentric cord insertion on placenta
  - One twin has less than half of placenta

Helpful Clues for Less Common Diagnoses

- **Anomaly of One Twin**
  - Anomalies more common in multiples than in singletons
    - Severe structural malformations seen in 2.6% of monochorionic twins in one series
  - Careful anatomic survey mandatory
  - Multiple anomalies increase suspicion for aneuploidy or syndrome
  - Amniocentesis may be offered
  - Fetal reduction may be offered

Helpful Clues for Rare Diagnoses

- **Twin Reversed Arterial Perfusion**
  - TRAP twin often looks very abnormal
    - Absent cranial structures/upper extremities strongly suggests this diagnosis
    - Heart maybe "absent" or rudimentary
    - Extensive subcutaneous edema often with cystic spaces
  - Pathognomonic feature of TRAP sequence is umbilical artery flow away from placenta toward abnormal fetus
  - Unlike most other anomalous twins, TRAP fetus is often very large due to edema

Other Essential Information

- Risk of aneuploidy in one twin is greater than a priori risk for singleton
- Birth defects rate in twins higher than for singletons
- Birth defects rate in twins conceived with assisted reproduction higher than for those conceived naturally
- MC diamniotic twins with unequal placental sharing have 9.8x likelihood of birth weight discordance compared to twins with equal placental sharing
- Fetal intervention possible in some circumstances
  - TTTS: Laser of anomalous vascular connections now preferred over serial amnioreduction
  - TRAP: Radiofrequency ablation of TRAP twin cord obliterates shunt → protects pump twin from hydrops

Alternative Differential Approaches

- Small twin with no anomaly, normal fluid
  - Idiopathic and normal
- Small twin with oligohydramnios/small bladder
  - Renal anomaly
  - IUGR
  - TTTS (only seen in monochorionic twins)

Idiopathic and Normal



Clinical photograph shows twins who were delivered at 35 weeks when the smaller fetus' estimated fetal weight dipped below the 10th percentile.

Idiopathic and Normal



Photograph of the same twins at age 3 shows continued size discrepancy but otherwise rambunctious, good health in both boys.

DISCORDANT TWIN GROWTH

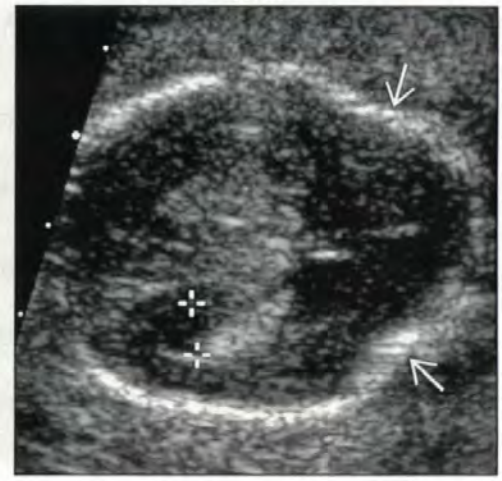
Intrauterine Growth Restriction of One Twin

(Left) *Table shows a typical example of severe asymmetric IUGR of one fetus in dichorionic twins. Twin B is markedly small with the abdominal circumference and femur length lagging behind the head measurements.* ***(Right)*** *Axial ultrasound of the head of twin B shows mild ventriculomegaly (calipers) and exaggerated frontal bone concavity ➡. Hypertelorism was shown on other images.*

Fetus A MA = 24w0d ±10d
→Fetus B← MA = 18w0d ±8d

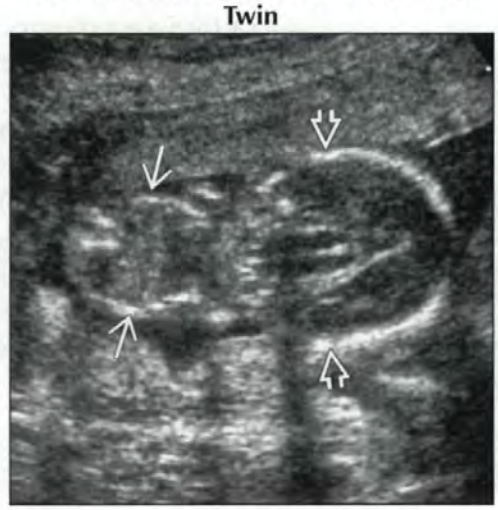
BPD	19w5d±12d	4.55cm
HC	19w4d±10d	16.97cm
AC	15w0d±12d	8.67cm
FL	17w6d±10d	2.59cm

Intrauterine Growth Restriction of One Twin

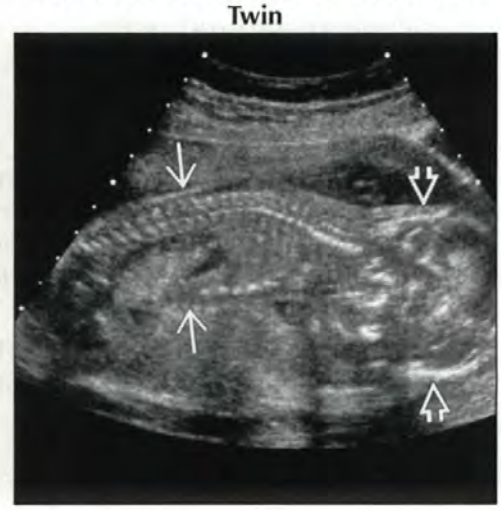


Intrauterine Growth Restriction of One Twin

(Left) *Coronal ultrasound in the same case as previous image, shows that the torso ➡ of twin B appears very small compared to the head ➡. Surrounding fluid volume is low.* ***(Right)*** *Sagittal ultrasound of twin A shows a normal head ➡ to abdomen ➡ ratio and normal surrounding fluid volume. Amniocentesis revealed normal chromosomes for twin A and triploidy for twin B.*



Intrauterine Growth Restriction of One Twin



Twin-Twin Transfusion Syndrome

(Left) *Chart shows unusually marked discordance between the twins in this case of twin-twin transfusion syndrome.* ***(Right)*** *Ultrasound shows the "stuck" smaller donor twin with no urine in the bladder ➡ and the membrane ➡ tightly draped around the extremities. Cord Doppler showed absent end-diastolic flow. This is stage 3 TTTS.*

Fetus A MA = 24w1d±13d
Fetus B MA = 19w3d±10d

→Fetus A←		→Fetus B←	
	MA ±SD		MA ±SD
BPD	25w0d±15d	BPD	21w1d±12d
HC	23w6d±10d	HC	20w1d±10d
AC	24w1d±15d	AC	19w5d±14d
FL	24w5d±15d	FL	18w0d±13d

Twin-Twin Transfusion Syndrome



DISCORDANT TWIN GROWTH

Unequal Placental Sharing

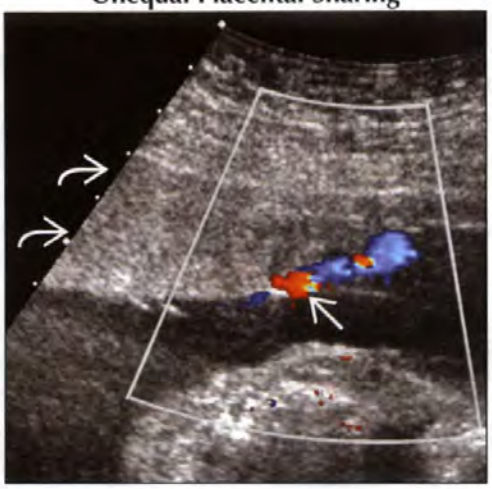
Fetus A LMP% <10%

BPD 24w2d±15d
HC 23w0d±10d
AC 20w6d±14d
FL 20w4d±13d

Fetus B LMP% 35%

BPD 24w5d±15d
HC 24w3d±14d
AC 23w6d±14d
FL 24w6d±15d

Unequal Placental Sharing

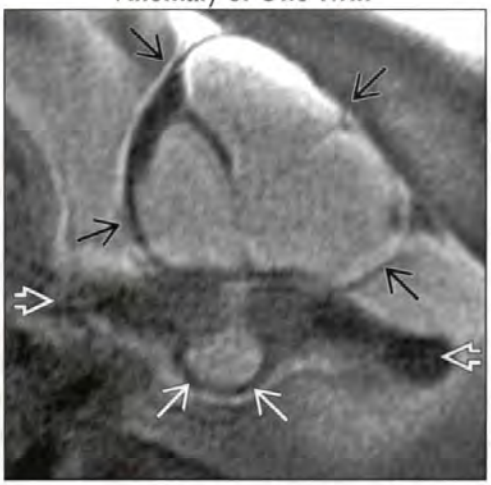


(Left) *Growth data from a twin gestation shows fetus A with an EFW < 10th percentile. No structural anomalies were seen and there was no evidence of twin-twin transfusion syndrome.* ***(Right)*** *Color Doppler ultrasound shows a marginal cord insertion with the cord for twin A ➡ inserting at the edge of the placental disc ➡. Twin B's cord was centrally placed. Discordant growth was from unequal sharing of the placenta, not artery-to-vein anastomoses as in TTTS.*

Unequal Placental Sharing



Anomaly of One Twin

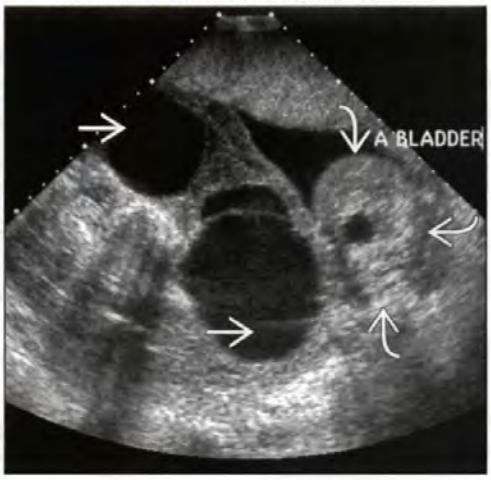


(Left) *Transabdominal ultrasound in the same case, shows reduction of fluid volume around twin A ➡ compared to that around the normally grown twin B ➡.* ***(Right)*** *Axial T2WI MR through the torso of a monoamniotic twin shows a large omphalocele ➡ and a meningocele ➡ on either side of the lower extremities ➡. Other images showed a large neural tube defect and ventriculomegaly. The bladder was never identified. Thus, this fetus has OEIS complex.*

Anomaly of One Twin



Twin Reversed Arterial Perfusion



(Left) *T2WI MR in the same case as previous image, shows the normal twin's cranium ➡ and the abnormal twin's torso ➡. There was polyhydramnios as well as IUGR of the abnormal twin. Chromosomes were normal.* ***(Right)*** *Axial ultrasound shows large cystic areas ➡ within the edematous skin of the TRAP twin, which became so large that it compressed the normal twin ➡ against the uterine wall.*

ASYMMETRIC FLUID DISTRIBUTION

DIFFERENTIAL DIAGNOSIS

Common

- Twin-Twin Transfusion Syndrome
- Discordant Twin Growth
- Unequal Placental Sharing
- Oligohydramnios of One Twin
- Polyhydramnios of One Twin

Less Common

- Premature Rupture of Membranes
- Myomatous Uterus

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Asymmetric fluid distribution may be caused by primary abnormality in one twin or complication of placental sharing
- Determination of chorionicity vital to management
  - Anomaly in one dichorionic twin
    - Management focused on maximizing outcome for normal fetus
    - Early intervention for anomalous twin confers additional risks of prematurity on normal twin
  - Monochorionic twins
    - Demise of one twin has potentially disastrous consequences for survivor
    - Intervention may be appropriate despite risks of prematurity
  - Twin-twin transfusion syndrome (TTTS) has high mortality for both twins if untreated
  - Twin reversed arterial perfusion (TRAP) sequence
    - Management is focused on maximizing outcome for pump twin

Helpful Clues for Common Diagnoses

- **Twin-Twin Transfusion Syndrome**
  - Monochorionic placentation
  - Fetuses often discordant in size: Donor small/recipient large
  - Donor twin: Oligemic → ↓ urine production → "absent" bladder + oligohydramnios
  - Recipient twin: Plethoric, polyhydramnios, at risk for hydrops
- **Discordant Twin Growth**
  - Smaller twin may be structurally abnormal ± aneuploid
  - Look for signs of growth restriction
    - Decreased abdominal circumference, estimated fetal weight < 10th percentile, oligohydramnios
  - Differentiate from normal but asymmetric twins
    - Serial evaluation shows appropriate interval growth with normal fluid volume around both
- **Unequal Placental Sharing**
  - Best time to look at placental cord insertion site is late first trimester
  - Look for eccentric cord placement
    - Twin with smaller area of placenta at risk for growth restriction/oligohydramnios
  - Velamentous cord: Cord inserts onto membranes away from placental disc
  - Marginal cord: Cord inserts at edge of placental disc
- **Oligohydramnios of One Twin**
  - Demise
  - Bilateral renal anomaly
    - Renal agenesis: Absent kidneys, absent renal arteries, "lying down" adrenals
    - Multicystic dysplastic kidneys: Variable sized cysts, no normal renal parenchyma
    - Severe UPJ obstruction: Dilated pelves/calyces without hydroureter
    - Obstructive cystic dysplasia: Multiple cysts within echogenic parenchyma ± residual hydroureter
  - Posterior urethral valves
    - Dilated bladder
    - "Keyhole" appearance due to dilated posterior urethra
    - Bilateral hydronephrosis
    - Look for urinoma/urinary ascites
  - Intrauterine growth restriction (IUGR)
    - Estimated fetal weight < 10th percentile
    - Decreased abdominal circumference
    - Use spectral Doppler to assess uteroplacental circulation/fetal response to adverse environment
- **Polyhydramnios of One Twin**
  - Idiopathic
  - Gastrointestinal obstruction
    - Look for dilated loops of bowel or "double bubble" sign
    - May not become apparent until 3rd trimester
  - CNS impairment → inability to swallow

- Obstructed swallowing: Goiter, oral mass, neck mass
  - Scan head and neck in axial plane
  - Look for splaying of carotid arteries with goiter
- High output state
  - Arteriovenous malformation: Use color Doppler to assess any apparent "cystic" structure
  - Tumor: Most fetal tumors are large/rapidly growing, use color Doppler to assess internal vascularity
  - Chorioangioma: Hypervascular placental mass
- Musculoskeletal anomaly
  - Skeletal dysplasia: Assess bone mineralization, measure all long bones
  - Arthrogryposis/akinesia sequence: Evaluate movement/swallowing in real time

Helpful Clues for Less Common Diagnoses

- **Premature Rupture of Membranes**
  - Premature membrane rupture of one sac results in oligohydramnios around that twin
  - Appropriate clinical history
  - Positive speculum examination with ferning
- **Myomatous Uterus**
  - Unable to support pregnancy due to poor placentation
  - Look for signs of placental insufficiency
    - Less than expected growth
    - Small abdominal circumference
    - Abnormal Doppler studies
  - Fibroids increase risk for abruption
    - Chronic abruption associated with growth restriction/oligohydramnios
    - Use color Doppler to look for avascular clot deep to vascular placenta

Other Essential Information

- Assessment of fluid volume
  - In singletons, amniotic fluid index (AFI) is used
    - Sum of four quadrants, deepest vertical pocket free of cord and fetal parts
  - In multiple gestations use maximum vertical pocket around each fetus
  - Show relative fluid distribution on either side of membrane
- TTTS may occur in monoamniotic twins
  - Lack of membrane precludes presentation with oligohydramnios/polyhydramnios
  - Look for absent bladder in smaller twin
  - Doppler may be abnormal depending on severity
- Renal agenesis in one of monoamniotic twins is easily missed
  - Normal twin produces urine, no inter-twin membrane therefore fluid may look normal
  - Check bladder carefully: Urine production → changing bladder volume over course of scan

Twin-Twin Transfusion Syndrome



Transabdominal ultrasound shows the "anti-gravity" appearance of a stuck twin. The membrane ➡ is closely applied to the stuck fetus. All of the visible fluid relates to the other twin.

Twin-Twin Transfusion Syndrome



Ultrasound shows the other twin ➡ who was freely mobile within large volumes of fluid. It is vital to demonstrate an inter-twin membrane in cases like this, which can be mistaken for monoamniotic twinning.

ASYMMETRIC FLUID DISTRIBUTION

Twin-Twin Transfusion Syndrome



Twin-Twin Transfusion Syndrome

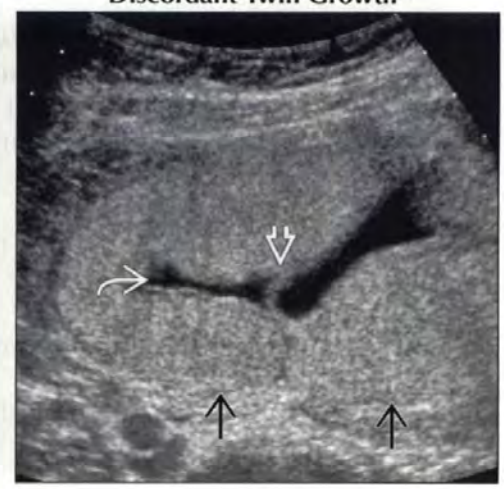


(Left) *Transabdominal ultrasound at presentation (18 weeks) shows measurement of the maximum vertical pocket (calipers) around the recipient twin. At 13 cm this indicates severe polyhydramnios.* ***(Right)*** *Color Doppler ultrasound in the same case as the previous image shows absence of urine in the bladder of the donor twin who has oligohydramnios. Normally the bladder is visible in the pelvis between the umbilical arteries.*

Discordant Twin Growth



Discordant Twin Growth

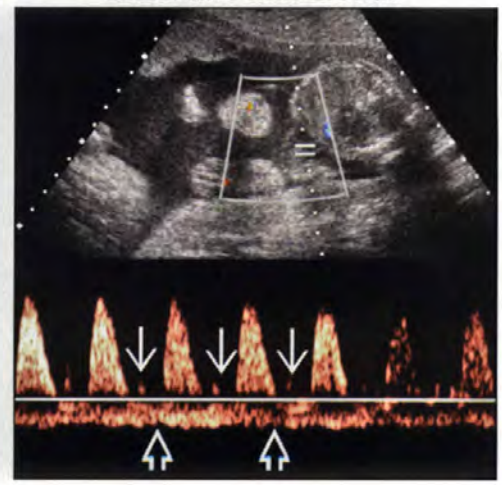


(Left) *Table shows biometric data for one of a dichorionic pair. Fetus A was concordant with dates in this assisted reproduction pregnancy. Fetus B was smaller with a lagging abdominal circumference.* ***(Right)*** *Transabdominal ultrasound in the same case shows two placentas and a thick membrane indicating dichorionicity. The fluid volume around the smaller twin was diminished. Follow-up at one month showed demise of this twin.*

Discordant Twin Growth



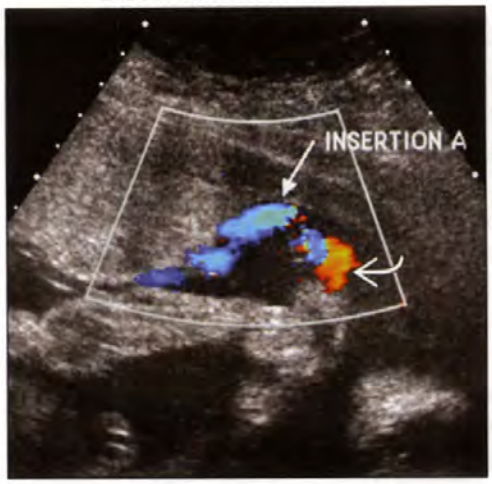
Discordant Twin Growth



(Left) *Transabdominal ultrasound shows oligohydramnios with membranes closely draped over the fetus. Other images showed normal kidneys and bladder.* ***(Right)*** *Pulsed Doppler ultrasound in the same case as previous image, shows absent end diastolic flow and pulsatile umbilical venous flow. These were dichorionic twins at 24 weeks gestation.*

ASYMMETRIC FLUID DISTRIBUTION

Discordant Twin Growth



Unequal Placental Sharing

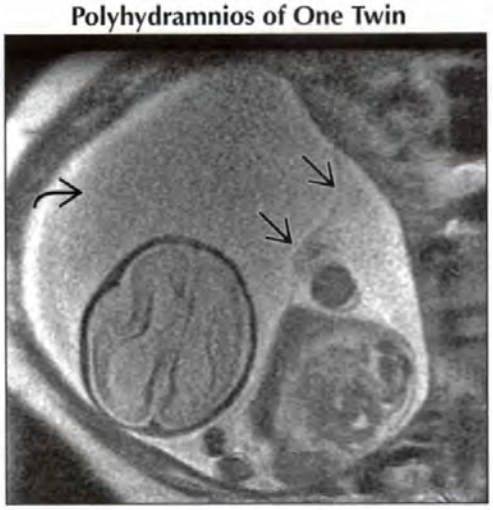


(Left) *Color Doppler ultrasound from the same case, shows a marginal cord insertion for the twin with oligohydramnios and 3 week growth lag.* ***(Right)*** *Color Doppler ultrasound in a different case, shows one cord inserting on the membranes rather than onto the placental disc proper (i.e., velamentous insertion). This twin was smaller with low fluid throughout the pregnancy. By convention, twins are not considered discordant unless one has IUGR.*

Oligohydramnios of One Twin

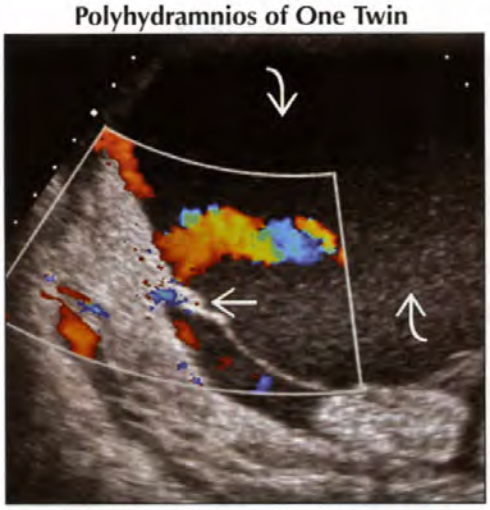


Polyhydramnios of One Twin



(Left) *Ultrasound of IVF twins at 16 weeks shows very little fluid around one twin which was also small. Although not visible on this image, fluid was normal around the co-twin. At 3 week follow-up, the twin with oligohydramnios had died.* ***(Right)*** *T2WI MR shows polyhydramnios around one twin. Note thin membrane. Encephalomalacia from co-twin demise caused impaired swallowing.*

Polyhydramnios of One Twin



Myomatous Uterus



(Left) *Image of membranes from the same case as prior image, confirms polyhydramnios. Despite the suggestion of a twin peak sign these were monochorionic twins with vascular connections. Encephalomalacia in the survivor compromised swallowing.* ***(Right)*** *Axial transabdominal ultrasound shows low fluid volume around a twin attributed to poor placentation in this myomatous uterus. Note multiple large transmural myomata.*

TWIN RELATED ANOMALIES

DIFFERENTIAL DIAGNOSIS

Common
- Fetal Demise
- Twin-Twin Transfusion Syndrome
- Conjoined Twins
- Twin Reversed Arterial Perfusion

Rare but Important
- Fetus-in-Fetu

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Most important step in evaluation of multiple gestation is determination of chorionicity
  - Certain problems only occur in monochorionic gestation
    - Twin-twin transfusion syndrome (TTTS)
    - Twin reversed arterial perfusion (TRAP)
    - Conjoined twins
- Assess fluid distribution
  - Show fluid on either side of inter-twin membrane
  - Measure maximum vertical pocket for each twin
  - If fluid is asymmetric, determine which side is abnormal
    - Decreased in one twin: Oligohydramnios differential
    - Increased in one twin: Polyhydramnios differential
    - Increased in one twin & decreased in the other: TTTS most likely diagnosis

Helpful Clues for Common Diagnoses
- **Fetal Demise**
  - Not uncommon in early pregnancy
  - Cardiac activity should always be seen in an embryo > 5 mm in crown rump length
  - If early, sac may completely resorb → "disappearing twin"
  - If later in monochorionic gestation, TRAP sequence may develop
  - Cord entanglement is a specific cause of demise in monoamniotic twins
- **Twin-Twin Transfusion Syndrome**
  - Monochorionic gestation
  - Asymmetric or discordant growth
  - Abnormal fluid distribution: One twin with oligohydramnios, one with polyhydramnios
    - Look for "stuck" twin with shrink-wrapped membrane in severe cases
  - TTTS staging
    - Stage 1: Donor bladder visible, normal Doppler
    - Stage 2: Donor bladder empty, normal Doppler
    - Stage 3: Donor bladder empty, abnormal Doppler
    - Stage 4: Hydrops in recipient
    - Stage 5: Demise of one or both
- **Conjoined Twins**
  - Monoamniotic gestation = no inter-twin membrane
  - Must have contiguous skin covering between fetuses
  - Nomenclature
    - Site of attachment & "pagus" (e.g., cephalopagus)
    - "Di" & site of duplication (e.g., dicephalus)
- **Twin Reversed Arterial Perfusion**
  - Monochorionic gestation with one normal "pump" twin and one anomalous twin
  - Anomalous twin
    - Absent cardiac activity or rudimentary heart
    - Often well-developed torso/lower extremities
    - Upper extremities/cranium poorly developed or absent
    - Marked skin edema ± effusions/ascites
  - Hallmark of this condition is umbilical artery flow toward the abnormal fetus rather than toward the placenta

Helpful Clues for Rare Diagnoses
- **Fetus-in-Fetu**
  - Apparent singleton: "Twin" is incorporated into the "singleton" as a mass
  - Always think about this with an unusual fetal mass
  - If vertebral column present ⇒ fetus-in-fetu more likely than teratoma

Other Essential Information
- Anomaly rate higher in twin than singleton pregnancies
- Any anomaly/aneuploidy can occur regardless of chorionicity
- Specific entities arise from vascular connections in monochorionic placentas

- TTTS arises secondary to artery to vein shunt within monochorionic placenta
  - Donor twin shunts arterial blood into recipient twin's venous circulation → donor oligemia → effective "hypotension"
  - ↓ Renal perfusion → ↓ urine output → oligohydramnios
  - ↓ Blood to placenta → ↓ return of oxygenated blood → chronic hypoxia → poor growth
  - Recipient twin receives all its own return from placenta & portion of co-twins → plethora/fluid overload
  - ↑ Volume status → ↑ cardiac output → eventual development of hydrops
- TRAP sequence arises secondary to artery to artery shunt within monochorionic placenta
  - Pump twin deoxygenated blood → co-twin umbilical arteries → preferential perfusion of lower extremities → lack of upper body/cranial development
  - Blood enters fetus via UA not UV → lack of normal circulation through heart → abnormal cardiac development

Alternative Differential Approaches

- Twin with hydrops
  - Anomalous twin in TRAP sequence: Massive skin edema/effusions common
  - Pump twin in TRAP sequence at risk for hydrops from cardiac compromise
  - Both twins in TTTS at risk for hydrops (greater in recipient due to volume overload)
  - May have hydrops from other causes unrelated to placental sharing
    - All cases need careful anatomic survey, infection work up ± chromosome evaluation
    - Perform formal fetal echocardiography for structure, function, rate and rhythm
- Twin with encephalomalacia
  - Any destructive process may end in encephalomalacia regardless of chorionicity
  - May occur secondary to chronic hypoxia in donor twin in TTTS
  - Monochorionic twin demise places surviving twin at risk for ischemic injury described as "twin embolization syndrome"
    - Shared placental circulation → vascular communications between fetuses
    - Death of one twin → sudden drop in peripheral resistance for survivor → effectively severe hypotensive episode
    - Ischemic injury seen in brain/myocardium/kidneys
  - Sonographic findings
    - ↑ Echogenicity in periventricular white matter, loss of grey-white matter differentiation
    - Eventual cystic encephalomalacia, porencephaly, microcephaly

Fetal Demise



Sagittal transabdominal ultrasound shows two sacs, one with poor decidual reaction ➡ which contained a dead embryo. At follow-up the other embryo died. Etiology for pregnancy failure unknown.

Fetal Demise



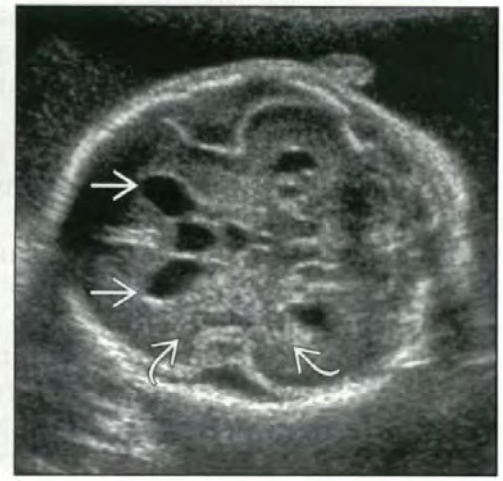
Ultrasound shows an inter-twin membrane ➡ and a twin peak sign ⇨. Fluid volume in the smaller sac is low due to twin demise. The dead twin and sac shrank over serial follow-up scans.

TWIN RELATED ANOMALIES

Fetal Demise



Fetal Demise

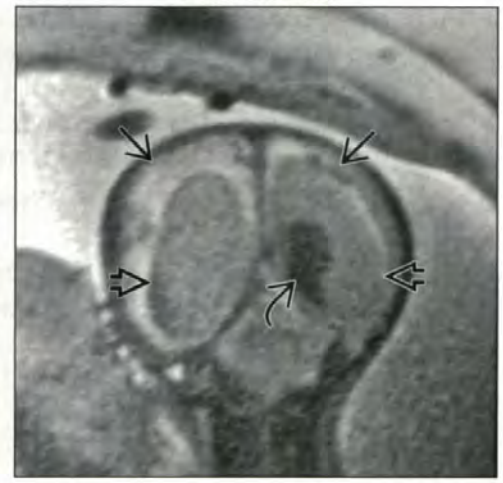


(Left) *Axial transabdominal ultrasound shows a normal appearance of this twin's brain at presentation.* ***(Right)*** *Axial transabdominal ultrasound 10 days following co-twin demise shows developing ventriculomegaly and loss of grey-white matter differentiation concerning for encephalomalacia. Monochorionic twin demise places the surviving twin at risk for ischemic injury ("twin embolization syndrome").*

Fetal Demise



Fetal Demise



(Left) *Axial oblique ultrasound at the same visit shows ventriculomegaly and intraventricular clot. The infant died within hours of delivery and autopsy confirmed encephalomalacia attributed to the hemodynamic effects of co-twin demise.* ***(Right)*** *Coronal T2WI MR in a different case shows diffuse abnormal signal of the cortex, ventriculomegaly and intraventricular clot consistent with ischemic encephalomalacia from co-twin demise.*

Twin-Twin Transfusion Syndrome



Twin-Twin Transfusion Syndrome

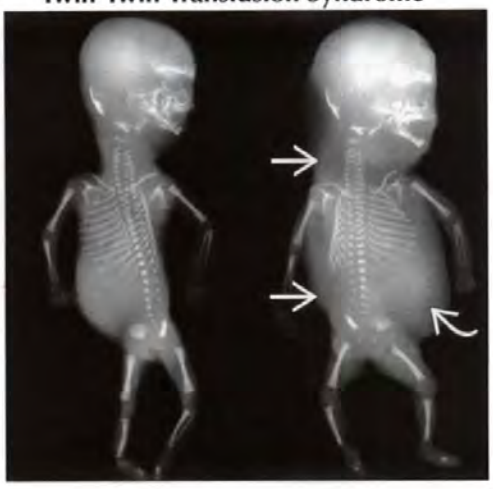


(Left) *Axial transabdominal ultrasound shows diffuse skin thickening over the scalp.* ***(Right)*** *Coronal transabdominal ultrasound in the same fetus as previous image, shows ascites, pleural effusion and more skin thickening indicating hydrops. This was the recipient twin in a monochorionic pair affected by twin-twin transfusion syndrome.*

Twin-Twin Transfusion Syndrome



Twin-Twin Transfusion Syndrome

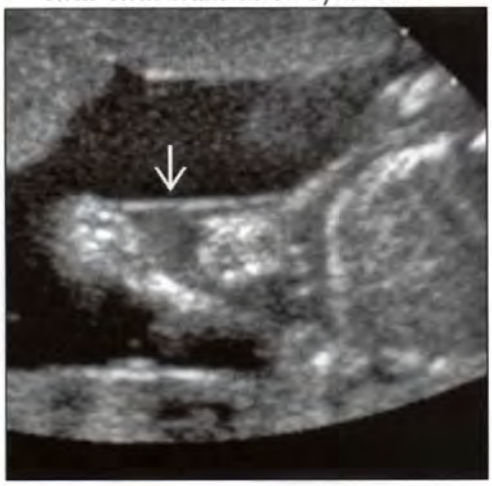


(Left) *Clinical photograph shows plethora, abdominal distension and diffuse edema secondary to hydrops in the recipient twin of a monochorionic pair with twin-twin transfusion syndrome.* ***(Right)*** *Radiograph as part of an autopsy shows skin thickening and abdominal distension secondary to ascites in the hydropic twin.*

Twin-Twin Transfusion Syndrome



Twin-Twin Transfusion Syndrome

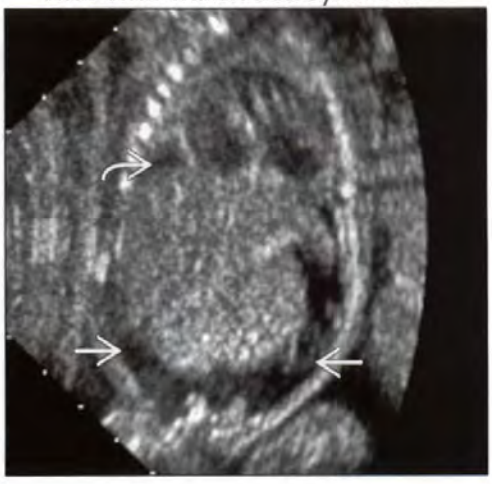


(Left) *Transabdominal ultrasound of a recipient twin, shows a full bladder and polyhydramnios reflecting volume overload.* ***(Right)*** *Transabdominal ultrasound shows oligohydramnios around a donor twin with the inter-twin membrane closely applied to the fetus.*

Twin-Twin Transfusion Syndrome



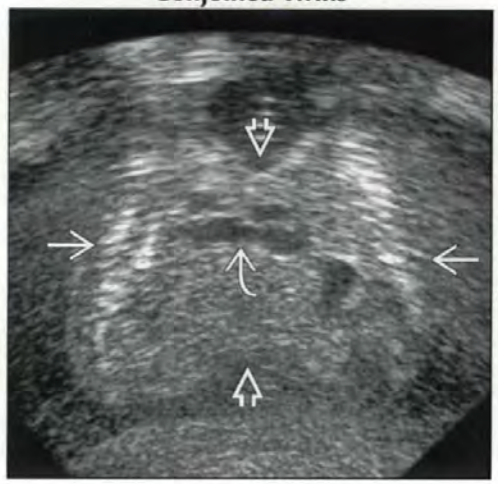
Twin-Twin Transfusion Syndrome



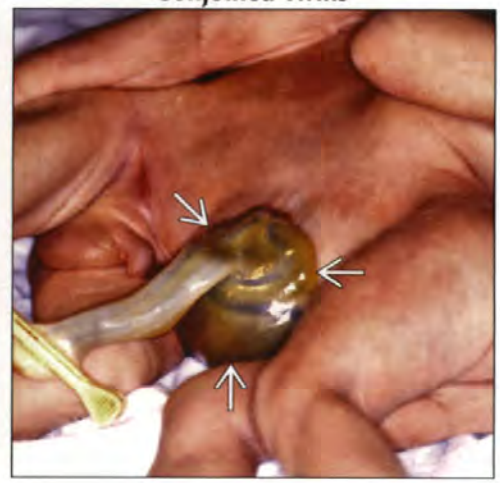
(Left) *Pulsed Doppler ultrasound shows absent end diastolic flow in a donor twin umbilical artery indicating stage 3 twin-twin transfusion syndrome.* ***(Right)*** *Coronal transabdominal ultrasound in a different case shows ascites and pleural effusion indicating hydrops in this recipient twin (i.e., stage 4 twin-twin transfusion syndrome).*

TWIN RELATED ANOMALIES

Conjoined Twins

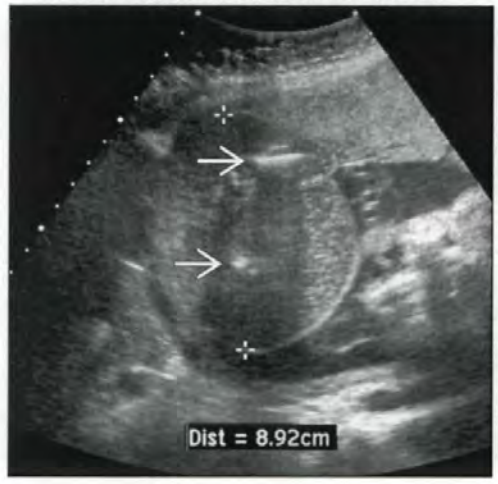


Conjoined Twins



(Left) *Coronal ultrasound shows thoracoomphalopagus fetuses ➡ facing each other. The heart ➡ is shared. Note that the contiguous skin covering ➡ extends to the level of the abdomen.*
(Right) *Clinical photograph confirms thoracoomphalopagus twins joined at the chest and abdomen. An omphalocele ➡ is often a component of omphalopagus conjoined twinning.*

Twin Reversed Arterial Perfusion

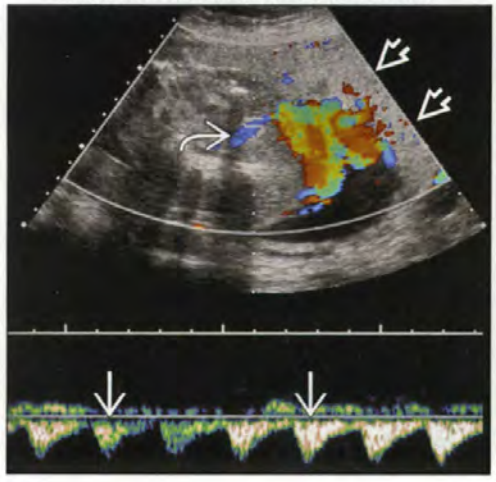


Twin Reversed Arterial Perfusion



(Left) *Transabdominal ultrasound shows an amorphous mass (calipers) with calcific elements ➡.*
(Right) *Transabdominal ultrasound in an orthogonal plane in the same case again shows the amorphous mass (calipers) adjacent to the limbs ➡ of the normal co-twin.*

Twin Reversed Arterial Perfusion



Twin Reversed Arterial Perfusion



(Left) *Color Doppler ultrasound shows umbilical arterial flow ➡ extending into this mass, away from the placenta ➡. Pulsed Doppler shows reversed arterial perfusion ➡ confirming the diagnosis of TRAP sequence.*
(Right) *Gross pathology shows the typical appearance of a TRAP fetus with marked soft tissue edema and no normal cranial/facial structures.*

Fetus-in-Fetu



Fetus-in-Fetu

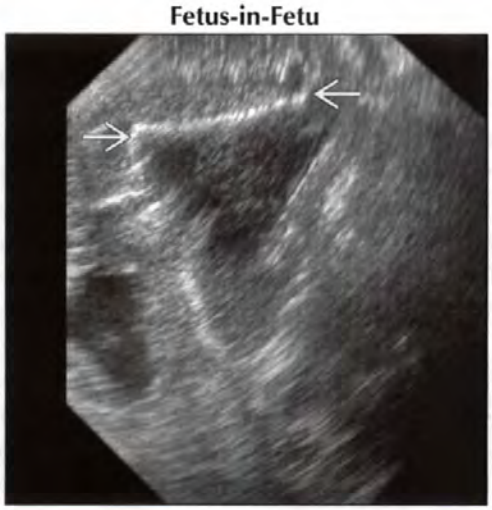


(Left) *Coronal transabdominal ultrasound shows an unusual intra-abdominal mass ➡ in a female fetus. The mass grew quite rapidly on serial evaluation.* ***(Right)*** *Axial oblique transabdominal ultrasound in the third trimester shows an apparent long bone ➡ within the abdominal mass.*

Fetus-in-Fetu

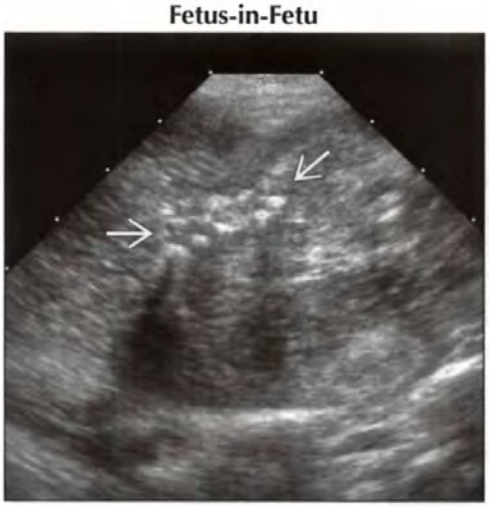


Fetus-in-Fetu

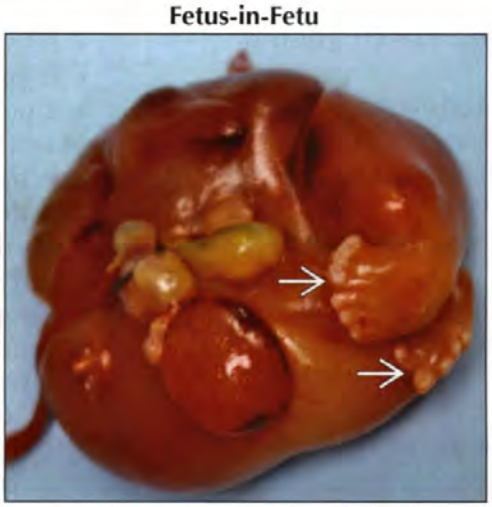


(Left) *Ultrasound of the abdomen after delivery shows the large, complex mass (calipers) with cystic components ➡ and bony elements ➡.* ***(Right)*** *Ultrasound confirms the presence of a long bone ➡ that was seen on the prenatal study.*

Fetus-in-Fetu



Fetus-in-Fetu



(Left) *Ultrasound through another section of the mass shows part of a vertebral column ➡ confirming the diagnosis of fetus-in-fetu rather than teratoma.* ***(Right)*** *Gross pathology of the resected specimen shows the feet ➡ of the fetus-in-fetu.*

CONJOINED TWINS

DIFFERENTIAL DIAGNOSIS

Common
- Thoracopagus
- Omphalopagus

Less Common
- Pygopagus
- Ischiopagus
- Cephalopagus
- Craniopagus

Rare but Important
- Dicephalus
- Fetus-in-Fetu
- Parasitic Twin

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Monochorionic
  - Single placenta
  - Twins same gender
- Monoamniotic
  - No inter-twin membrane
- Fetuses fused at some location
  - Contiguous skin covering at site of fusion is hallmark of this diagnosis
  - Conjoined twins with narrow or pliable area of fusion may vary in orientation to each other
- May see fused umbilical cord
  - More than three cord vessels
- Important to differentiate conjoined from monoamniotic twins given different prognosis
  - Monoamniotic twins are within the same sac but not fused
  - May have relatively fixed orientation if cord entanglement
  - Prognosis better for monoamniotic
    - No need for separation
    - Majority of conjoined twins die in utero or shortly after delivery
- Formal fetal echocardiography mandatory in all conjoined twins
  - Complex congenital heart disease may preclude separation attempt
  - Fetal echocardiography easier than postnatal echo
    - Multiple acoustic windows through amniotic fluid
    - No interference from aerated lung

Helpful Clues for Common Diagnoses
- **Thoracopagus**
  - Fetuses face each other
  - Fused from thoraces to umbilicus
  - Always some degree of cardiac fusion
    - Precludes separation in 75%
  - Common pericardial sac in 90%
  - Livers invariably fused
    - 25% share biliary system
  - 25% have common small intestine, usually duodenum
- **Omphalopagus**
  - Fetuses face each other
  - Fusion of ventral abdominal cavities from umbilicus ± lower thorax
  - 80% liver fusion, biliary anomalies common
  - 30% have shared small intestine at distal ileum at level of Meckel's diverticulum

Helpful Clues for Less Common Diagnoses
- **Pygopagus**
  - Fetuses face away from each other
  - Joined at sacrum/buttocks
  - 15% share urinary system with single bladder
  - One anus ± rectum, rest of bowel separate
  - May have spinal cord fusion
- **Ischiopagus**
  - Joined from umbilicus to single pelvis
  - Fetuses face-to-face or end-to-end
  - Variable number kidneys/bladder
  - Cloacal malformation/anal atresia/vesicocolic fistulas occur
  - Internal and external genitalia may be shared
  - Variable number of extremities
    - Tetrapus = 4
    - Tripus = 3
    - Bipus = 2
  - Major challenges to separation are genitourinary and orthopedic
  - Genitourinary
    - Continence, adequate bladder/bowel emptying/sexual function
  - Orthopedic
    - Number of lower limbs
    - Pelvic reconstruction for prosthetic limb attachment
- **Cephalopagus**
  - Joined from vertex to umbilicus with faces on opposite sides of common head

- Inseparable
- **Craniopagus**
  - Joined at skull anywhere except face or foramen magnum
  - Separability depends on degree of shared dural sinuses
  - Shared neural tissue precludes separation

Helpful Clues for Rare Diagnoses

- **Dicephalus**
  - Two heads
  - Single torso
  - Usually single set of extremities
  - Inseparable
- **Fetus-in-Fetu**
  - "Twin" forms a mass within an apparent singleton
  - Key difference between teratoma and fetus-in-fetu is presence of vertebral column
  - Mass is well-demarcated as contained within a membranous sac
  - Commonest in upper retroperitoneum
  - No malignant potential
  - Excision is curative
- **Parasitic Twin**
  - Vestigial fetal parts or incomplete twin attached to fully developed co-twin
    - Intact twin known as autosite
    - Any vestigial parts may be seen

Other Essential Information

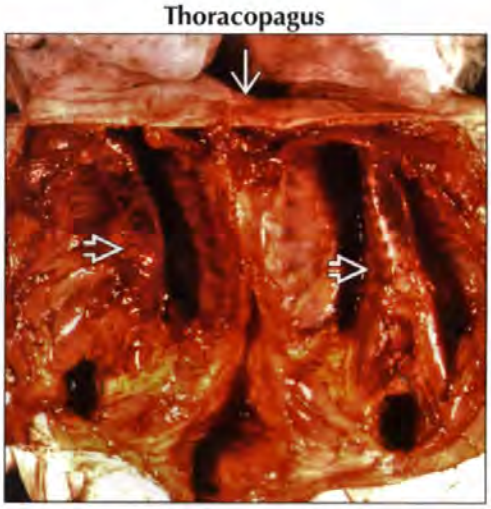
- Nomenclature
  - Site of attachment + "pagus" (e.g., thoracopagus = twins joined at chest)
  - "Di" + site of duplication (e.g., dicephalus = twins with two heads on a single torso)
    - Suffix may be used to describe number of extremities
    - Ischiopagus tripus = twins joined at lower body with single pelvis and three lower extremities
- Overall prognosis for conjoined twins is poor
  - 40-60% stillborn
  - 35% liveborn twins die in first 24 hours
- Each set of conjoined twins is unique
  - Associated malformations in some may constitute lethal condition
  - If lethal/inseparable, management focus is on maternal safety
    - Delivery mode should minimize morbidity
    - Aim to avoid classical cesarean section or bivalved uterus
    - Extensive uterine incision → risk accreta spectrum/uterine rupture in future pregnancies
- Evolution of surgical technique has led to increasing numbers of separation attempts
  - Ethical dilemma
  - Pre-operative surgical planning with multidisciplinary team
    - Skin expansion
    - Post-operative wound care
    - Psychological counseling after separation in older twins

Thoracopagus



Axial ultrasound shows two fetuses ⇄ facing each other with contiguous skin covering ⇨ and a shared heart ⇄.

Thoracopagus



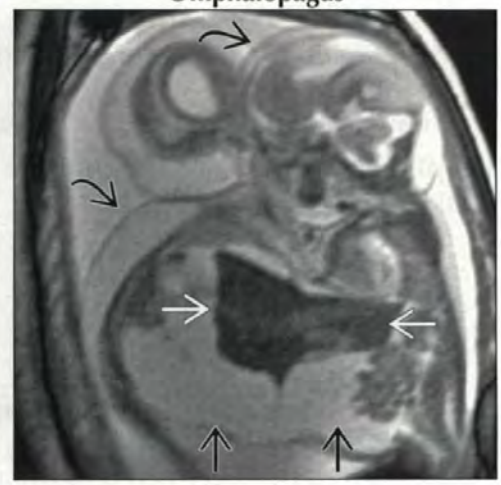
Gross pathology shows fused thoracic cavities splayed open at autopsy. Note the contiguous skin covering ⇨ and separate spines ⇄.

CONJOINED TWINS

Omphalopagus



Omphalopagus

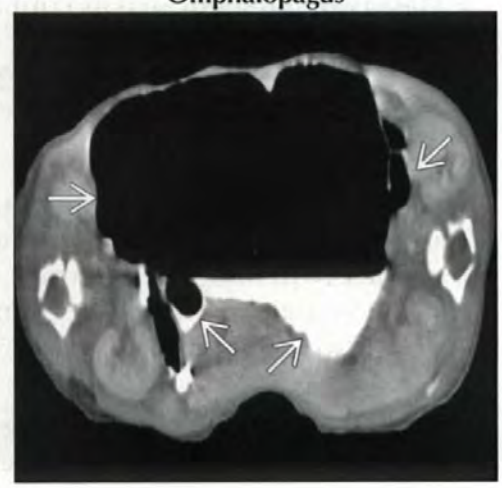


(Left) *Axial transabdominal ultrasound shows anterior fusion at the level of the abdomen with two separate stomachs but a shared liver. Note the contiguous skin cover, which proves conjoined twinning.* ***(Right)*** *Sagittal oblique T2WI MR shows a common liver in grossly abnormal conjoined twins, both of which are hydropic. Note ascites and skin edema. Both twins also had pleural effusions and pulmonary hypoplasia.*

Omphalopagus

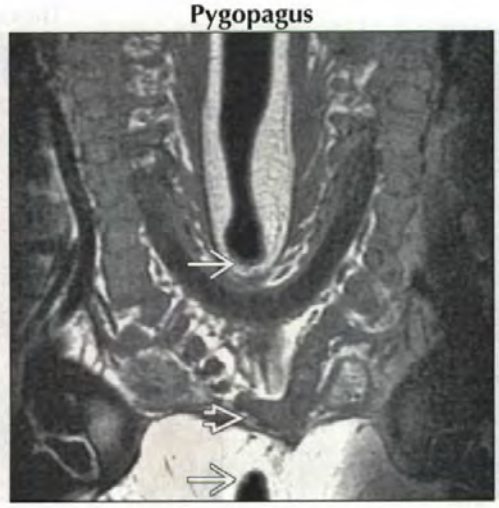


Omphalopagus



(Left) *Clinical photograph shows liveborn, conjoined twins with omphalocele rupture complicating delivery. This precipitated emergent separation, which was successful.* ***(Right)*** *Axial CECT in the same case as previous image shows a massively dilated common duodenum/proximal jejunum. This had been identified in utero as a cystic structure, but the exact nature was not clear until after delivery.*

Pygopagus



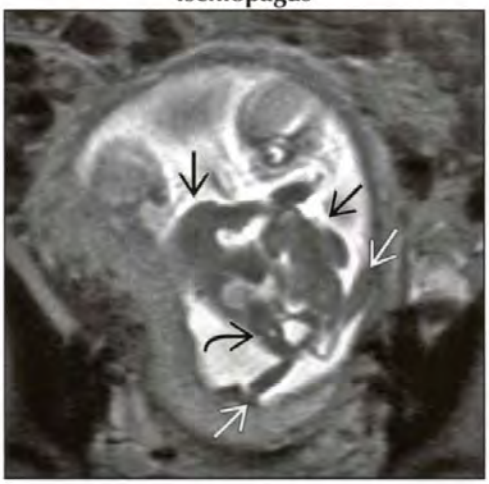
Pygopagus



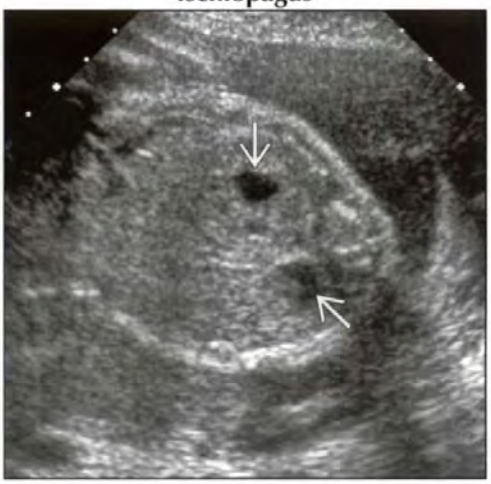
(Left) *Sagittal T1WI MR shows contiguous skin covering but a relatively small amount of bony fusion. (**Right**) Clinical photograph of the same case as previous image shows the typical site of fusion in pygopagus twins. The actual skin attachment was smaller than the areas touching, which made separation easier as the defect requiring closure was quite small.*

CONJOINED TWINS

Ischiopagus

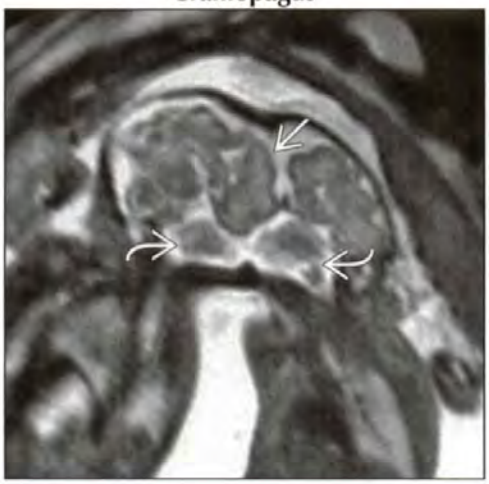


Ischiopagus



(Left) *Coronal T2WI MR shows two upper bodies, a single pelvis, and part of two lower extremities. There were in fact three lower extremities, but one was rudimentary.* ***(Right)*** *Axial transabdominal ultrasound shows bilateral hydronephrosis in one of the ischiopagus pair shown in the previous image. Hydronephrosis was secondary to obstruction as the twins had a common cloaca. Only one of four kidneys had adequate drainage and function.*

Craniopagus



Dicephalus



(Left) *Coronal oblique T2WI MR shows a common skull vault with separate cerebella but shared supratentorial brain.* ***(Right)*** *Coronal transabdominal ultrasound shows two separate heads with a single torso and two upper extremities. This pregnancy was terminated.*

Fetus-in-Fetu



Parasitic Twin



(Left) *Transabdominal ultrasound shows a long bone, thought to be a femur, within an intra-abdominal mass in a female fetus. Postnatal excision confirmed fetus-in-fetu.* ***(Right)*** *Gross pathology shows the head and upper extremities of a parasitic twin protruding from the epigastrium of the co-twin.*

SECTION 9
Placenta & Umbilical Cord

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Abnormal Umbilical Vessels	9-32

PLACENTAL SONOLUCENCIES

DIFFERENTIAL DIAGNOSIS

Common
- Placental Lake (PL)
- Placental Abruption (PA)
- Marginal Sinus Vessels
- Tornado-Shaped Vessels of Accreta

Less Common
- Placental Cord Insertion Cysts
- Complete Hydatidiform Mole
- Triploidy

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Obtain standard placenta views
  - Sagittal long views
    - Midsagittal to include lower uterine segment
    - Parasagittal
  - Axial views
    - Upper, mid, lower uterus
- Use color Doppler in evaluating placental lesions
  - Marginal sinus vessels have flow but abruption does not
    - Both can present with retroplacental hypoechoic region
  - Placental lakes often have no discernible flow
    - Use real time grayscale findings to see swirling flow
    - Obtain cine clip if documentation of flow is important
- Correlate placental findings with fetal findings
  - Fetal anomalies suggest triploidy
  - Intrauterine growth restriction suggests triploidy or placental insufficiency

Helpful Clues for Common Diagnoses
- **Placental Lake (PL)**
  - Enlarged intervillous vascular spaces
    - Contain maternal blood
    - May have fluid/fluid level
  - Variable size
  - Transient nature
    - Change size and shape with time
    - Can completely resolve
  - Location
    - Within placenta without appreciable border
    - Subchorionic towards fetal surface
  - Occasional PL are normal
    - Late second trimester
    - Third trimester
  - Numerous PL or early PL associated with placental insufficiency
    - Intrauterine growth restriction (IUGR)
    - Oligohydramnios
    - High resistive umbilical artery flow
  - Power Doppler shows no flow in most PL
    - Swirling flow seen with real time scanning
  - Can thrombose and fibrose
    - Increased echogenicity
    - Discernible wall
    - No swirling flow seen
- **Placental Abruption (PA)**
  - Marginal PA is most common
    - Hematoma has no blood flow
    - Placenta margin is lifted off uterus
    - Estimate how much placenta is detached
  - Retroplacental PA
    - May look like thick placenta when acute
    - Often presents with preterm labor not bleeding
  - Preplacental PA is rare
    - Blood in subchorionic/subamniotic space
    - May cause cord compromise if near cord insertion site
- **Marginal Sinus Vessels**
  - Retroplacental sonolucency
  - Normal maternal venous vessels
    - Mostly myometrial veins
  - Doppler will often show flow
- **Tornado-Shaped Vessels of Accreta**
  - Enlarged intraplacental lacunae
  - Triangle-shaped bizarre lucencies
    - Slow flow like with PL
  - Look for signs of accreta
    - Loss of myometrium
    - Invasion beyond uterine confines: Scan with full bladder to look for invasion
    - Use color Doppler to evaluate for vascular flow
  - MR may help with diagnosis

Helpful Clues for Less Common Diagnoses
- **Placental Cord Insertion Cysts**
  - Originate from cord or placental surface
  - Often multiple
  - ↑ Morbidity with large cysts (> 5 cm)
    - IUGR

- Hemorrhage
- Rare cord accident

- Can grow during pregnancy

- **Complete Hydatidiform Mole**
  - Gestational trophoblastic neoplasia
    - 100% paternal genetic makeup
    - 46 XX is most common karyotype
  - First trimester or early second trimester diagnosis
  - Ultrasound findings
    - Cystic placenta
    - Vascular mass
    - No fetus
    - Periplacental hemorrhage common
  - Clinical presentation
    - Bleeding
    - Hyperemesis
    - Elevated maternal serum hCG levels
- **Triploidy**
  - Karyotype reveals 69 chromosomes
    - Diandry if extra haploid is paternal (also called partial mole)
    - Digyny if extra haploid is maternal
  - Variable placenta appearance depending on source of extra chromosomes
    - Large and cystic placenta if diandry
    - Small placenta if digyny
  - Abnormal fetus
    - Severe IUGR
    - Oligohydramnios
    - Multiple anomalies, potentially involving all organ systems

Other Essential Information

- PLs rarely need follow-up
  - Common in the late second and early third trimesters
  - Transient
- If numerous PLs then look very carefully at fetus
  - Triploidy
  - IUGR from placental insufficiency
- Abruption symptoms related to location of PA
  - Marginal abruption present with bleeding
  - Retroplacental PA present with pain, fetal distress and preterm labor
  - Preplacental PA often asymptomatic unless cord compression
- Have high index of suspicion for accreta
  - Patients often at high risk secondary to prior cesarean sections
  - Suspect placenta accreta if placenta previa and tornado-shaped vessels both present
  - Look for loss of myometrium and vessels beyond myometrium
- Evaluate location and pattern of sonolucencies
  - Placental lakes are infrequent and scattered
  - Abruptions are most often marginal
  - Marginal sinus vessels are retroplacental but not mass-like

Placental Lake (PL)



Axial ultrasound shows a large venous lake ➡ in the placenta, near the cord insertion site ➡. A follow-up study was recommended because the lucency was large and persistent.

Placental Lake (PL)



Axial ultrasound of the same placental region several weeks later shows that the venous lake ➡ is much smaller. The cord insertion site is once again shown ➡.

PLACENTAL SONOLUCENCIES

Placental Abruption (PA)



Placental Abruption (PA)



(Left) *Sagittal ultrasound shows a hypoechoic collection of blood at the placental margin ➡. The patient had a history of trauma, and the finding is typical for a subacute placental abruption.* ***(Right)*** *Sagittal ultrasound shows a large retroplacental and marginal abruption. The hematoma morphology is complex and cystic ➡, and the abruption lifts the placenta ➡ off the myometrium.*

Marginal Sinus Vessels

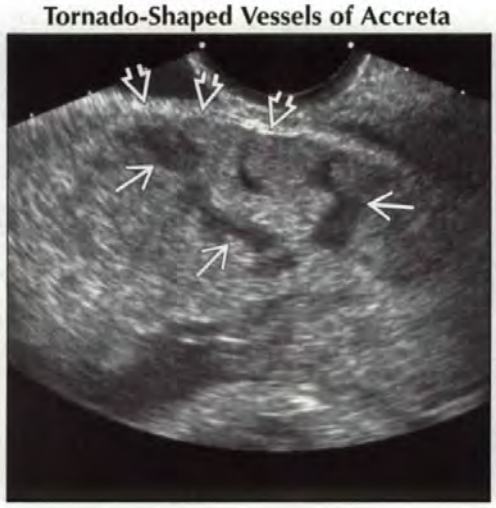


Marginal Sinus Vessels

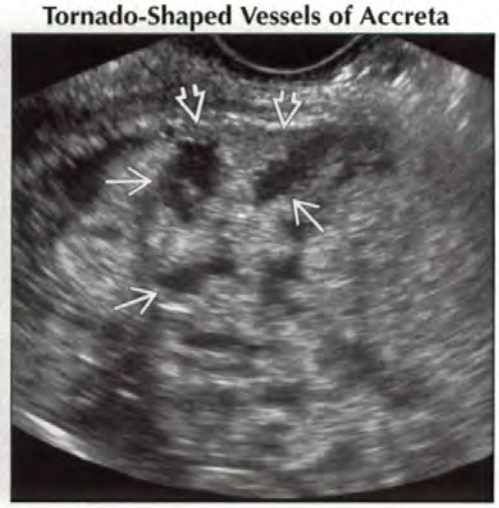


(Left) *Axial ultrasound shows typical normal subplacental as well as marginal placental and myometrial veins ➡. These vessels should not be mistaken for abruption.* ***(Right)*** *Axial ultrasound shows an abundance of marginal sinus and myometrial veins ➡. Although the size of the vessels was extreme in this case, the outcome was still normal.*

Tornado-Shaped Vessels of Accreta



Tornado-Shaped Vessels of Accreta



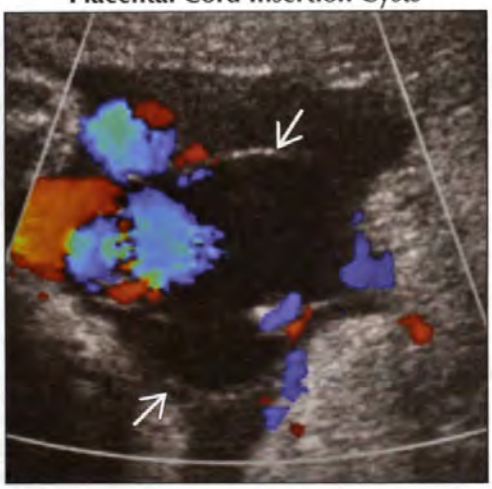
(Left) *Axial ultrasound shows enlarged vascular lacunae ➡ associated with placenta accreta in this patient with two prior c-sections and placenta previa. No appreciable myometrium is seen ➡.* ***(Right)*** *Sagittal transvaginal ultrasound shows the typical appearance of vascular lacunae ➡ associated with placenta accreta. Note the area of myometrial absence ➡.*

PLACENTAL SONOLUCENCIES

Placental Cord Insertion Cysts

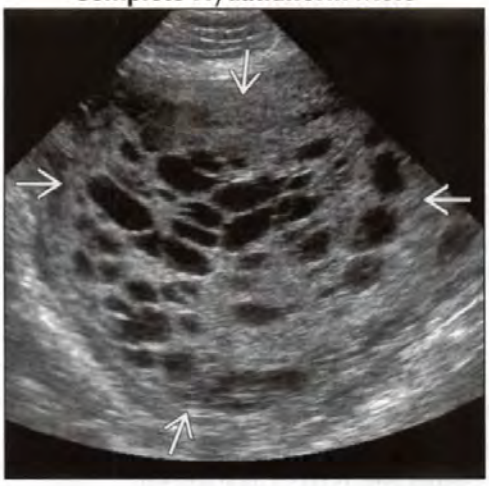


Placental Cord Insertion Cysts

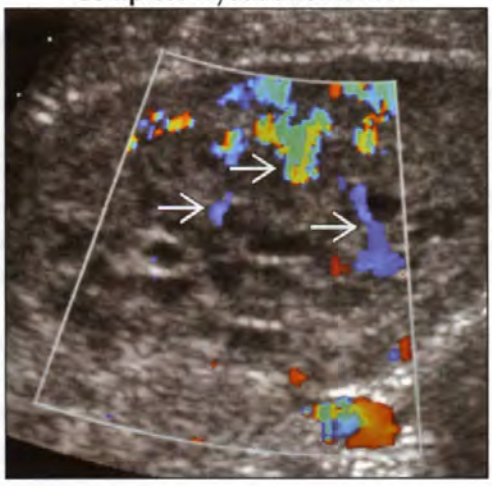


(Left) *Axial ultrasound shows a large cyst ➡ at the cord insertion site of the placenta. Umbilical vessels are seen within the cyst ➡.* ***(Right)*** *Axial color Doppler ultrasound of the same case as previous image confirms that the cyst ➡ is associated with the umbilical cord insertion site. Pregnancy and fetal outcome were normal.*

Complete Hydatidiform Mole



Complete Hydatidiform Mole

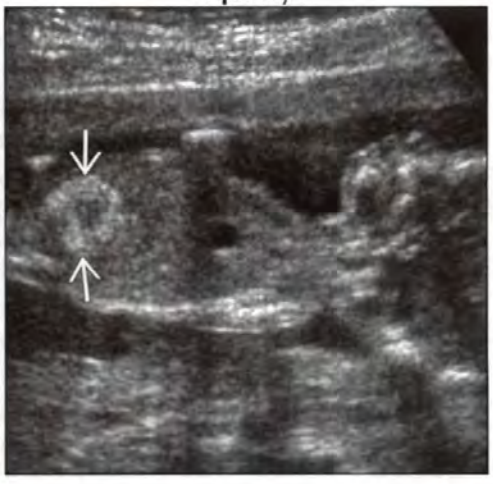


(Left) *Axial transabdominal ultrasound shows the placenta is completely replaced with cysts ➡. There was no associated fetus.* ***(Right)*** *Axial color Doppler ultrasound shows another molar pregnancy. Note that the cystic placental mass fills the uterus and is markedly vascular ➡.*

Triploidy



Triploidy



(Left) *Sagittal ultrasound shows a cystic placenta ➡ and a fetus ➡. The fetus had multiple mild anomalies and intrauterine growth restriction. Amniocentesis results revealed triploidy.* ***(Right)*** *Sagittal ultrasound of the same fetus with triploidy shows echogenic bowel ➡ and oligohydramnios.*

ABNORMAL PLACENTAL LOCATION

DIFFERENTIAL DIAGNOSIS

Common
- Placenta Previa
- Marginal Sinus Previa
- Placenta Accreta Spectrum

Less Common
- Succenturiate Lobe

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Rule out lower uterine segment (LUS) placentation in 2nd and 3rd trimesters
  - Transabdominal routine images
    - Midsagittal view
    - Parasagittal views
  - Perform transvaginal ultrasound (TVUS) if LUS not seen with routine views
    - Bleeding is not a contraindication
    - Use careful technique
    - Watch screen while inserting probe
  - Perform translabial/transperineal ultrasound if TVUS not possible
    - Collapsed vagina is acoustic window
    - Elevate maternal hips to minimize bowel artifact
    - Place probe on perineum (labia minora)
- Is fetus or fluid in direct contact with cervix?
  - Placenta may block direct contact
  - Floating fetus in 3rd trimester
  - Transverse presentation
- Does the uterus look asymmetrically thick?
  - Placenta & myometrium vs. area with only myometrium
- Succenturiate lobes often missed
  - Placenta location assigned before uterus completely evaluated
- Have a high index of suspicion for accreta
  - Suspect accreta if previa & prior cesarean section
    - ↑ Risk with ↑ number of cesarean sections
- Use color and pulse Doppler
  - Placenta accreta
    - Vessels may extend beyond myometrium
  - Succenturiate lobe
    - Vessels connect placentae
    - Rule out vasa previa
  - Pulse Doppler can help differentiate maternal from fetal vessels
    - Fetal vs. maternal heart rate

Helpful Clues for Common Diagnoses
- **Placenta Previa**
  - Subtypes of PP are based on the distance between placenta margin and cervix internal os (IO)
    - Complete PP completely covers IO
    - Partial PP partially covers IO
    - Marginal PP within 2 cm of IO
  - Second trimester PP often resolves
    - Most PP seen < 20 wks resolve by 34 wks
    - 5% PP incidence at 15-16 wks
    - 0.5% PP incidence at term
    - Placental "migration" (trophotropism): Areas of placenta atrophy as others grow
    - LUS "stretches" later in pregnancy
  - Associated with placental abruption
    - Patient presents with bleeding
    - Placental edge lifted by hematoma
    - Mass-like hematoma seen by cervix
  - Associated with preterm labor
    - More common if also bleeding
    - Assess cervical length
    - Assess for IO distention/funneling
    - Cervical canal may be distended with blood
  - 5% of PP will have associated accreta
- **Marginal Sinus Previa**
  - Marginal sinus PP is a subtype of marginal PP
  - Evaluate marginal placental vein distance to IO
    - Placental vessels < 2 cm from IO
    - Veins are maternal, not fetal
    - Do not confuse with vasa previa
- **Placenta Accreta Spectrum**
  - PA: Pathologic nomenclature based on depth of placental invasion
    - Accreta means myometrial attachment without muscle invasion
    - Increta means myometrial invasion
    - Percreta means invasion through uterus
    - Imaging does not differentiate between subtypes well
  - PP & invasion of cesarean section scar
    - ↓ Subplacental hypoechoic myometrial zone (< 2 mm)
    - Vessels extending through myometrium
  - Distended placental lacunae
    - Bizarre large sonolucencies
    - "Tornado-shaped"
    - Often near PA site

- MR findings
  - Loss of normal low signal myometrium with T2WI
  - Avoid gadolinium

Helpful Clues for Less Common Diagnoses

- **Succenturiate Lobe**
  - SL: Accessory placental lobe or lobes
    - Often smaller than main lobe
  - Identify placental cord insertion site
    - Most often on main lobe
    - May be velamentous (between lobes)
  - Rule out vasa previa
    - Low SL
    - Crossing vessels cover IO
    - At risk for fetal hemorrhage
  - SL is often missed if the entire uterus not imaged
    - 5% of all deliveries have SL
    - Most often asymptomatic and discovered at delivery
    - May present with bleeding if SL previa
    - May present as retained product of conception

Other Essential Information

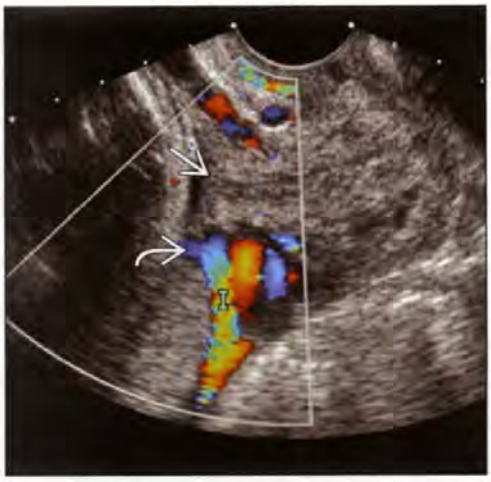
- High risk patients for abnormal placental implantation
  - Prior placenta previa
  - Prior cesarean section
  - Prior suction curettage
  - Prior uterine surgery
  - Advanced maternal age
  - Multiparity
  - Smoking
  - Cocaine use
- Symptoms at presentation
  - Incidental finding
  - Painless bleeding
  - Fetal distress
  - Preterm labor
- Multiple diagnoses often seen together
  - Previa associated with accreta
  - Succenturiate lobe may cause previa
- Rule out vasa previa in every case of low placentation
  - Use color Doppler to find vessels in close proximity to IO
  - Use pulse Doppler to prove vessels are fetal and not maternal
  - Vasa previa and succenturiate lobe
    - Vessels between placentae cross IO
  - Vasa previa and PP/PA
    - Low placenta with velamentous cord insertion near IO
- Prognosis
  - Placenta previa prognosis excellent with appropriate management
    - < 1% mortality
  - Placenta accreta prognosis related to predelivery planning
    - 7% mortality
  - Succenturiate lobe prognosis is excellent if no vasa previa
    - 60-80% fetal mortality if vasa previa missed prenatally

Placenta Previa



Sagittal transabdominal ultrasound of a second trimester pregnancy shows a complete placenta previa. The inferior edge of the posterior placenta ➡ completely covers the internal cervical os ➡.

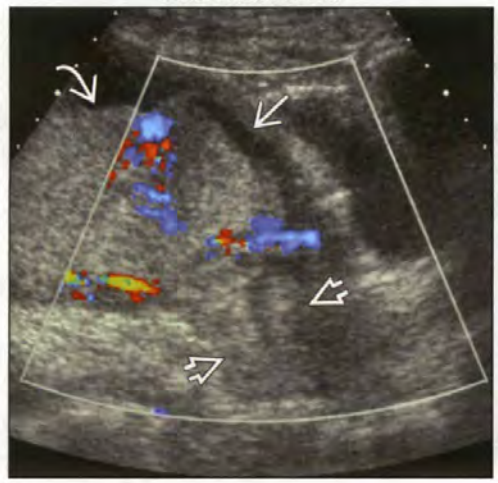
Placenta Previa



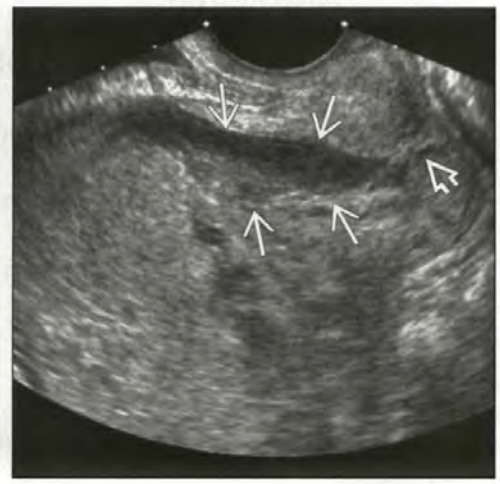
Sagittal color Doppler ultrasound in the third trimester of the same pregnancy shows a marginal placenta previa. The placental edge ➡ is close to but does not cover the internal os ➡.

ABNORMAL PLACENTAL LOCATION

Placenta Previa

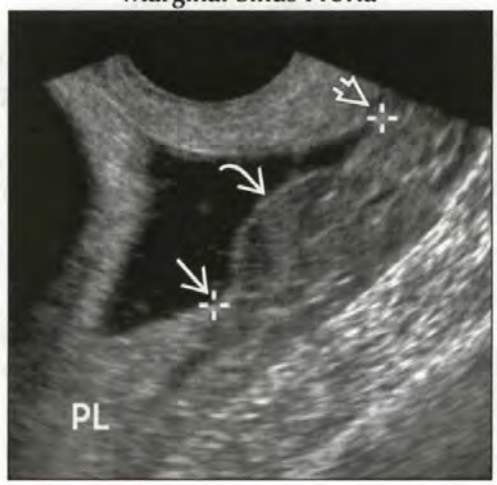


Placenta Previa

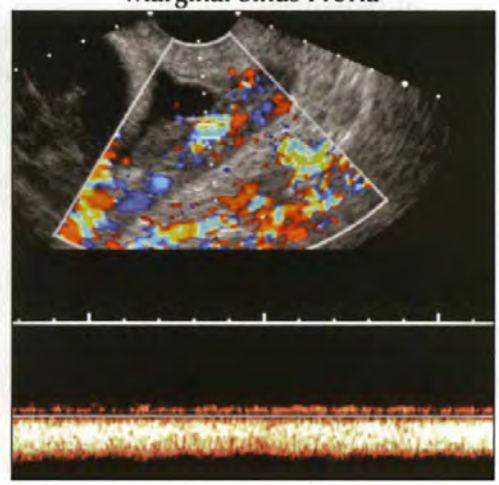


(Left) *Sagittal color Doppler ultrasound in a patient with complete placenta previa and bleeding shows the placenta covering the cervix. In addition, there is a subtle subplacental abruption anteriorly.* ***(Right)*** *Sagittal transvaginal ultrasound shows the cervical canal distended with blood. The closed portion of the cervix is short.*

Marginal Sinus Previa

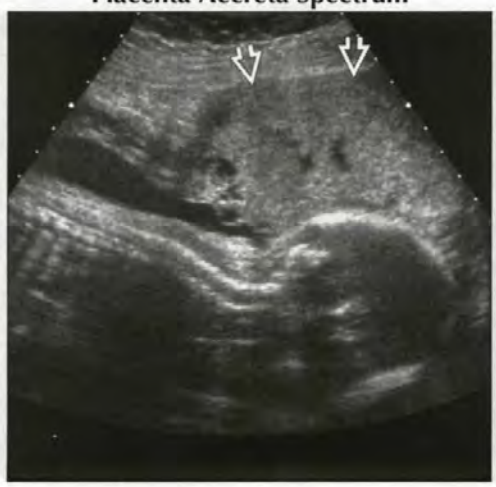


Marginal Sinus Previa



(Left) *Sagittal transvaginal ultrasound shows that the placental edge is more than 2 cm from the internal cervical os. However, the marginal placental vessels are close to the cervical os.* ***(Right)*** *Sagittal color Doppler ultrasound confirms that the vessels are venous and are located very close to the internal os of the cervix. The diagnosis of marginal sinus placenta previa was made in this patient with second trimester bleeding.*

Placenta Accreta Spectrum



Placenta Accreta Spectrum



(Left) *Sagittal ultrasound shows an exophytic bulge of the lower uterine segment in the region of placentation and a prior cesarean section scar. In addition, the normal hypoechoic myometrium is lost in this area.* ***(Right)*** *Sagittal T2WI MR confirms loss of myometrium in this case of a placenta percreta, which involved a large portion of the placenta. The points to regions of intact myometrium.*

ABNORMAL PLACENTAL LOCATION

Placenta Accreta Spectrum



Placenta Accreta Spectrum

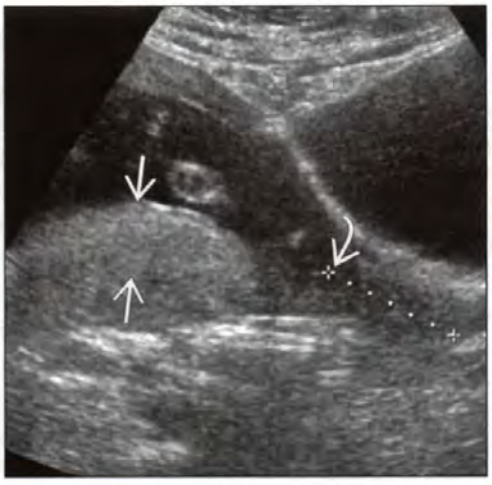


(Left) *Sagittal color Doppler ultrasound shows multiple large placental vessels ➔ extending beyond the confines of the myometrium in this case of placenta percreta.* ***(Right)*** *Sagittal transvaginal ultrasound shows loss of the subplacental hypoechoic myometrium ➔ and a distended "tornado-shaped" placental lacuna ➔. The pathologic final diagnosis in this case was placenta increta.*

Succenturiate Lobe



Succenturiate Lobe

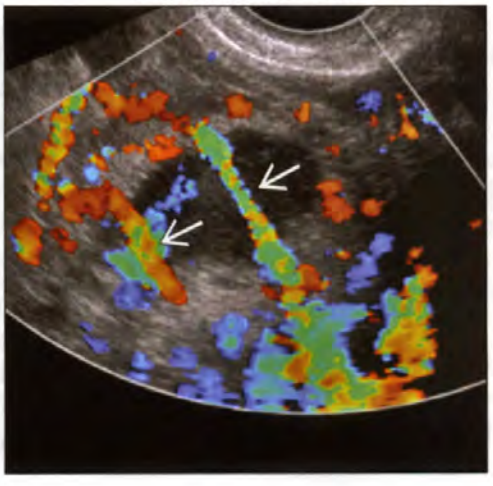


(Left) *Sagittal ultrasound shows an anterior placenta ➔. However, asymmetric posterior uterine wall thickening ➔ suggests a possible second placenta.* ***(Right)*** *Sagittal ultrasound of the lower uterine segment in the same case as previous image, shows a second, smaller, posterior placenta ➔. The inferior edge of the succenturiate lobe lies near the cervical internal os ➔.*

Succenturiate Lobe



Succenturiate Lobe



(Left) *Sagittal ultrasound of the lower uterine segment shows placental tissue anterior ➔ and posterior ➔ to the cervix ➔. It is difficult to tell if there is connecting tissue between the two placental "masses".* ***(Right)*** *Endovaginal color Doppler ultrasound in the same case as previous image. The umbilical cord inserted eccentrically upon the larger posterior placenta and crossing fetal vessels ➔ are identified between the main placenta and the succenturiate lobe.*

ABNORMAL PLACENTAL MARGIN

DIFFERENTIAL DIAGNOSIS

Common
- Marginal Placental Abruption
- Circumvallate Placenta
- Synechiae
- Marginal Cord Insertion

Less Common
- Chorioamniotic Separation
- Placenta Accreta Spectrum

Rare but Important
- Vasa Previa

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Always scan entire placental surface
- Doppler essential for evaluating placental pathology
  - Document cord insertion site
  - Marginal cord insertion versus velamentous cord insertion
    - Are branching vessels on surface of placenta or submembranous?
    - Marginal insertion if all vessels on surface of placenta
    - Velamentous cord insertion if any fetal vessel is submembranous
    - Vasa previa if fetal vessels are near cervix
  - Vessels beyond uterine wall with accreta
    - Look for invasion of bladder
  - Synechiae may show flow
  - Pulsed Doppler differentiates fetal vessels from maternal
    - Fetal arterial flow similar to cord Doppler waveform

Helpful Clues for Common Diagnoses
- **Marginal Placental Abruption**
  - Most common type of placental abruption
  - Detachment of edge of placenta
    - Raised placental margin seen in 50%
    - Submembranous blood
  - Appearance of hematoma related to age
    - Acute blood can be isoechoic to placenta
    - Becomes hypoechoic with time
    - Sonolucent if old
- **Circumvallate Placenta**
  - Membranes attach to fetal surface of placenta instead of villous margin
    - Caused by discrepant size between chorion and basal plates
  - Placental marginal "shelf" seen with ultrasound
    - Elevated margin of placenta
    - Placenta lifted towards cord insertion site
    - Placenta edge appears to be floating free in fluid
  - Short bands of tissue
    - Extend from placental margin to placental margin
    - Thicker earlier in pregnancy
    - Tissue may become fibrosed
- **Synechiae**
  - Fibrous band in uterus
    - Secondary to uterine scar
    - Amnion and chorion drape over synechiae
  - Long bands of tissue
    - Extend from uterine wall to uterine wall
    - Bands are thicker earlier in pregnancy
    - Stretch thin as uterus grows
  - Fetus moves freely around synechia
    - Differentiates from amniotic bands
  - Placental implantation common
    - Edge of placenta lifted onto synechia
    - Follow course of synechia to show uterine attachment
  - Doppler may show flow in synechia
- **Marginal Cord Insertion**
  - Eccentric placental cord insertion
    - Within 2 cm of placental margin
  - Battledore placenta is a subtype
    - Thick placenta
    - Small surface area of attachment
    - Higher risk for abruption
  - At risk for becoming velamentous cord insertion
    - Cord insertions < 5 mm from placental margin
    - Placenta may involute and leave vessels subplacental
    - At risk for vasa previa if vessels near cervix

Helpful Clues for Less Common Diagnoses
- **Chorioamniotic Separation**
  - Persistent unfused amnion after 16 wks
    - Primary nonfusion
    - Post amniocentesis
  - Ultrasound findings

- Thin amniotic membrane separate from uterine wall
- Associations with nonfusion
  - Trisomy 21
  - Genitourinary anomalies
  - Oligohydramnios

- **Placenta Accreta Spectrum**
  - Placenta grows beyond endometrial lining
  - Associated with multiple prior cesarean sections
  - Ultrasound and MR findings
    - Loss of subplacental myometrium
    - Vessels or placental tissue beyond uterus
    - Distended vascular lacunae

Helpful Clues for Rare Diagnoses

- **Vasa Previa**
  - Submembranous fetal vessels within 2 cm of internal cervical os
  - Clinical scenarios
    - Succenturiate lobe crossing vessels
    - Velamentous cord insertion near cervix
  - Transvaginal Doppler essential for diagnosis
    - Color Doppler shows crossing vessels
    - Pulsed Doppler to prove fetal vascularity

Other Essential Information

- Size of abruption is associated with outcome
  - Excellent prognosis if < 30% detached
  - Poor prognosis if > 50% detached
  - Look for other areas of abruption once one abruption is seen
    - Retroplacental, preplacental, multiple margins
- Rare complications of significant circumvallate placenta
  - Abruption
  - Intrauterine growth restriction
  - ↑ Risk if > 2/3 of margin involved
- Look for fetal anomalies if chorioamniotic separation seen
  - Markers for trisomy 21
  - Maternal serum screen results
- Pathologic classification of accreta
  - Accreta vera
    - Invasion through endometrium
    - No muscle invasion
  - Increta
    - Myometrial invasion
  - Percreta
    - Invasion beyond uterus
  - Imaging with only 33-38% sensitivity for all types of accreta
    - Must have high clinical suspicion for diagnosis

Alternative Differential Approaches

- Placenta implanted on a band of tissue
  - Synechia versus circumvallate placenta
    - Follow band to show insertion sites
    - Synechia extends from uterine wall to uterine wall
    - Circumvallate extends from placenta to placenta
  - Rule out amniotic bands
    - Thinner than synechia or placental shelf
    - Fetus becomes entrapped in bands
    - Fetal defects and amputations

Marginal Placental Abruption



Sagittal transabdominal ultrasound shows the inferior margin of the placenta ➡ lifted off the uterus by a hypoechoic wedge-shaped hematoma ➡. The blood clot is near the cervix (cx).

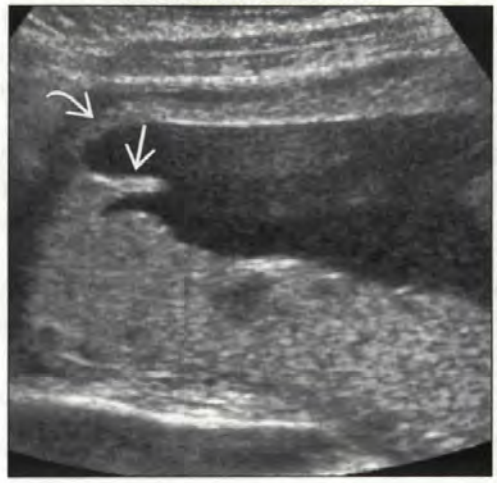
Marginal Placental Abruption



Long axis ultrasound shows the superior margin of the placenta ⇨ is flattened and displaced. A marginal hypoechoic collection of blood ➡ is causing this mass effect.

ABNORMAL PLACENTAL MARGIN

Circumvallate Placenta





(Left) *Sagittal ultrasound shows the edge of the placenta ➡ lifted off the uterine wall ➡. There is no associated hematoma.* ***(Right)*** *Sagittal oblique ultrasound of the same placenta as previous image shows a marginal shelf ➡ that extends from one margin of the placenta to the other ➡. This finding differentiates circumvallate placenta from uterine synechia.*

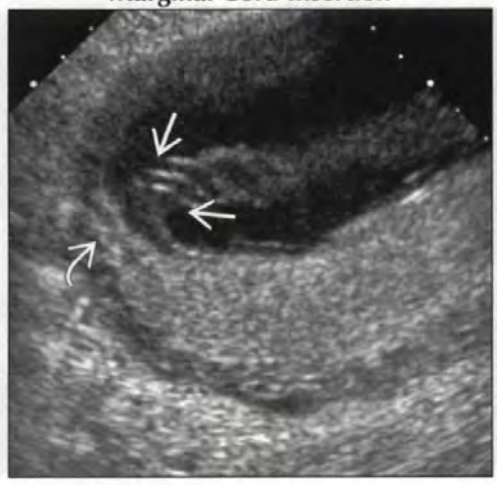
Synechiae

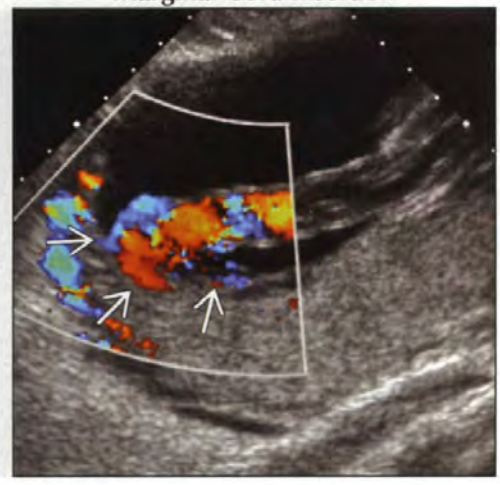




(Left) *Axial ultrasound shows the edge of the placenta ➡ lifted by a uterine band ➡.* ***(Right)*** *Axial oblique ultrasound of the same case as previous image shows that the band inserts on the uterine wall ➡ and is therefore a uterine synechia. A significant amount of placenta ➡ is implanted on this synechia.*

Marginal Cord Insertion





(Left) *Axial ultrasound shows the umbilical cord attachment ➡ on the lateral edge of the posterior placenta ➡.* ***(Right)*** *Axial color Doppler ultrasound shows that the umbilical vessels from the cord branch on the fetal surface of the placenta ➡. No velamentous vessels were seen; however, this extremely marginal cord insertion is at risk for becoming a velamentous cord insertion.*

ABNORMAL PLACENTAL MARGIN

Chorioamniotic Separation



Chorioamniotic Separation

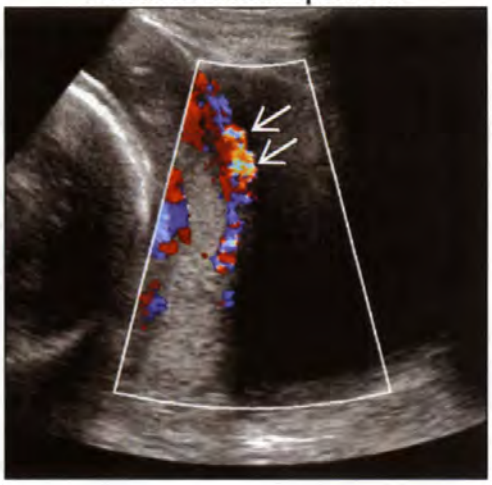


(Left) *Sagittal ultrasound shows the placental edge and the amnion extending from the margin.* ***(Right)*** *Coronal ultrasound confirms significant amniotic membrane separation from the placenta and uterine wall. The fetus had multiple anomalies and trisomy 18, one of the causes of delayed chorioamniotic fusion.*

Placenta Accreta Spectrum



Placenta Accreta Spectrum

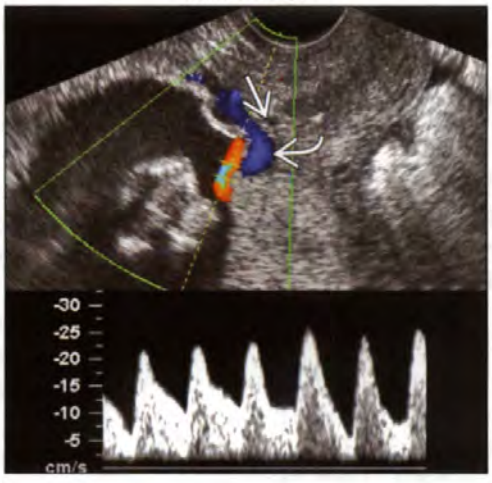


(Left) *Sagittal transabdominal ultrasound shows uterine wall irregularity and loss of the normal myometrial hypoechoic zone.* ***(Right)*** *Sagittal color Doppler ultrasound of the same case as previous image shows large subplacental vessels extending beyond the expected confines of the uterus.*

Vasa Previa



Vasa Previa



(Left) *Sagittal transvaginal ultrasound shows an anterior placenta and a posterior placenta with an intervening placental sonolucency overlying the internal os of the cervix.* ***(Right)*** *Sagittal pulsed Doppler ultrasound shows fetal arterial flow between the two placentas. The sonolucency was a vein that changed caliber during scanning. Here it is quite small. The fetal vessels cover the internal os.*

PLACENTAL MASS-LIKE LESIONS

DIFFERENTIAL DIAGNOSIS

Common
- Acute Placental Abruption
- Placental Implantation on Myoma
- Focal Myometrial Contraction (FMC)
- Chorioangioma
- Complete Hydatidiform Mole

Less Common
- Placental Teratoma

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Routinely evaluate the whole placenta
  - Sagittal views
    - Midsagittal (with lower uterine segment)
    - Right and left parasagittal
  - Axial views
    - Upper, mid, lower
- Identify location of mass
  - In the placenta
    - Chorioangioma
    - Extension of abruption
    - Teratoma (rare)
  - Behind the placenta
    - Abruption
    - Myoma
    - FMC
  - Replaces the placenta
    - Complete hydatidiform mole
- Interrogate mass with Doppler
  - Masses with flow
    - Chorioangioma
    - Complete hydatidiform mole
  - Masses with little or no flow
    - Abruption
    - FMC
  - Characterize pattern of flow
    - Peripheral flow with myoma
    - Linear contiguous flow with FMC

Helpful Clues for Common Diagnoses
- **Acute Placental Abruption**
  - Identify abruption location
    - Marginal (most common)
    - Retroplacental
    - Preplacental (most rare)
  - Retroplacental abruption can mimic mass
    - Thick placenta may be only finding
    - Acute blood isoechoic to placenta
    - Blood becomes hypoechoic with time
  - Doppler shows no flow in hematoma
  - Look for signs of fetal distress
    - Assess fetal heart rate
    - Fetal tone and movement
    - Cord Doppler evaluation
  - Assess amount of placenta detached
    - < 30% associated with good prognosis
    - > 50% associated with > 50% fetal death
- **Placental Implantation on Myoma**
  - Myoma appearance
    - Hypoechoic to uterus and placenta
    - Calcifications are common
  - Degenerating myomas
    - Central cystic change
    - Decreased blood flow
  - Retroplacental myomas are associated with abruption
    - Use Doppler to differentiate blood from myoma
  - Rarely associated with intrauterine growth restriction
    - Large amount of placenta implanted on myomatous uterus
- **Focal Myometrial Contraction (FMC)**
  - Normal finding throughout pregnancy
  - Uterine wall contraction
  - Inner contour affected most
    - Inner uterine bulge
    - Outer contour relatively preserved
  - FMC tends to be isoechoic to uterine wall
  - Will resolve or change with time
    - May take more than 30 minutes
    - May need to reassess on follow-up exams
- **Chorioangioma**
  - Benign, vascular placental tumor
    - Most < 5 cm
  - Common location is on fetal side of placenta, near cord insertion site
  - Ultrasound features
    - Well-defined mass
    - Generally hypoechoic
    - Heterogeneous if hemorrhage, infarction or degenerating
    - Variable amount of blood flow
- **Complete Hydatidiform Mole**
  - Most common type of gestational trophoblastic neoplasia
    - 100% paternal genetic makeup
  - Variable ultrasound appearance
    - Placental cysts
    - Anembryonic gestational sac

        - Associated perigestational hemorrhage
    - Doppler findings
        - ↑ Flow between cysts
        - High-velocity, low-impedance flow
    - Associated theca lutein cysts
        - Bilateral, multiseptated ovarian cysts
        - Seen in 50% of cases

Helpful Clues for Less Common Diagnoses

- **Placental Teratoma**
    - Extremely rare
    - Benign mature teratoma
    - Calcifications suggest diagnosis
    - Differentiate from demised twin next to placenta
    - Histogenesis theories
        - Twin incorporated in placenta
        - Primitive gut tissue grows in placenta

Other Essential Information

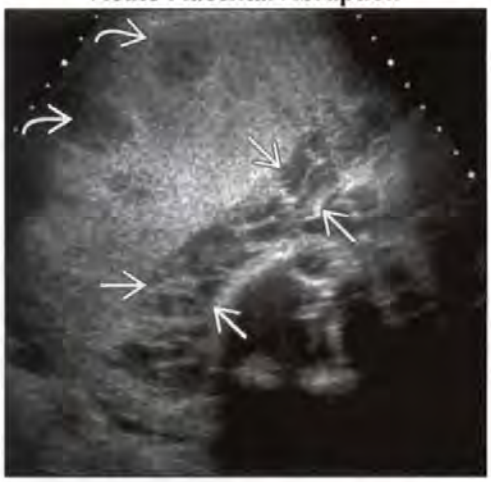
- Placental masses may be either incidental at time of exam or symptomatic
- Symptoms associated with abruption
    - Retroplacental abruption
        - Preterm labor
        - Pain
        - Fetal distress
    - Marginal abruption
        - Bleeding with or without contractions
    - Preplacental
        - Asymptomatic
        - Fetal distress
    - Large abruptions can be multifocal
        - Can bleed directly into placenta
    - Evaluate placenta first and quickly if fetal distress
        - If large abruption seen then curtail exam
        - Viable fetus may need emergent delivery
    - Patients at high risk for abruption
        - Prior history of abruption
        - Trauma
        - Hypertension
        - Cocaine use
        - Smoking
        - Increased parity
        - Advanced maternal age
        - Placenta implanted on myoma
- Signs and symptoms of molar pregnancy
    - Bleeding
    - Rapid uterine enlargement
    - Hyperemesis
        - ↑ Human chorionic gonadotropin levels
    - Preeclampsia
- Signs and symptoms associated with large chorioangioma
    - > 5 cm considered large
    - Elevated maternal serum alpha-fetoprotein
    - Hydrops fetalis
        - ↑ Arterial flow leads to high output cardiac failure
        - Fetal anemia
    - Polyhydramnios
    - Preterm labor
    - Preeclampsia

Acute Placental Abruption



Sagittal ultrasound shows a complex hypoechoic blood collection ➡ that has lifted the placenta ➡ off the myometrium. This large abruption was retroplacental and marginal.

Acute Placental Abruption



Sagittal ultrasound of the placenta in a patient with preterm labor but no bleeding shows both a retroplacental ➡ and a preplacental ➡ abruption.

PLACENTAL MASS-LIKE LESIONS

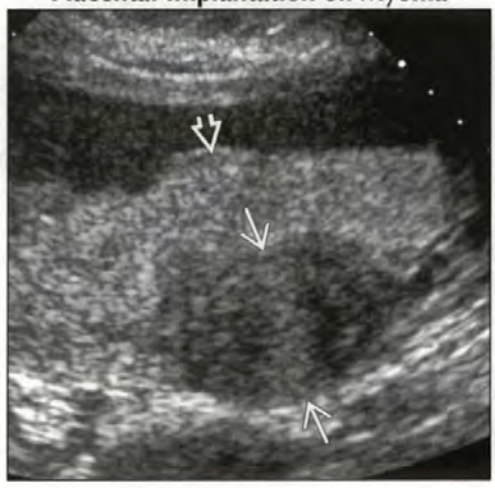
Acute Placental Abruption

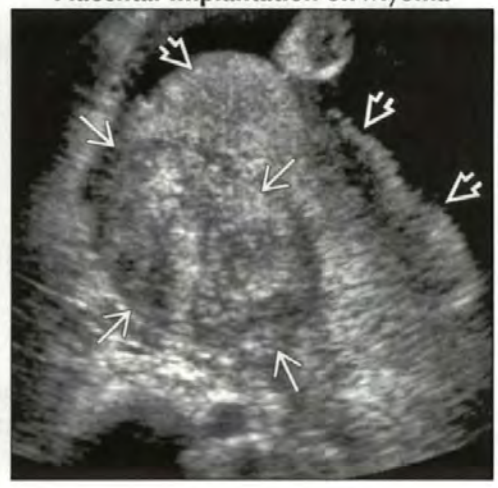




(Left) *Sagittal ultrasound shows a large retroplacental abruption, which mimics a placental mass. The placenta is lifted off the uterine wall, and a large portion is detected.* ***(Right)*** *Axial ultrasound of the same case as previous image shows the different echogenicities of the myometrium, hematoma, and placenta. The hematoma is hypoechoic.*

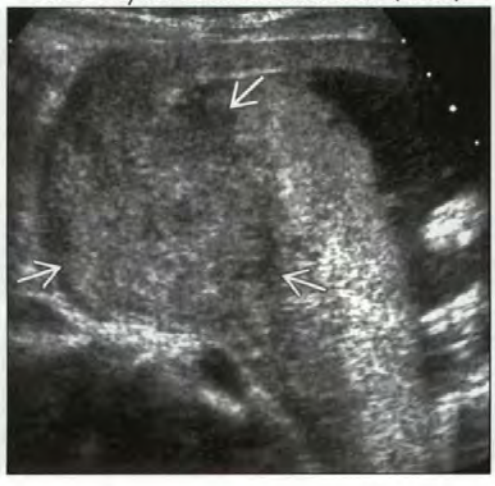
Placental Implantation on Myoma





(Left) *Sagittal ultrasound shows placental implantation on a leiomyoma. The fibroid is hypoechoic, and the placenta implants directly upon it.* ***(Right)*** *Sagittal ultrasound in another case shows a more heterogeneous myoma. The placenta is again seen implanting directly upon the myoma.*

Focal Myometrial Contraction (FMC)

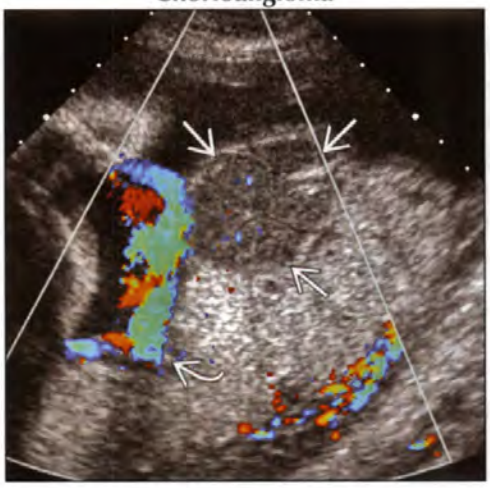




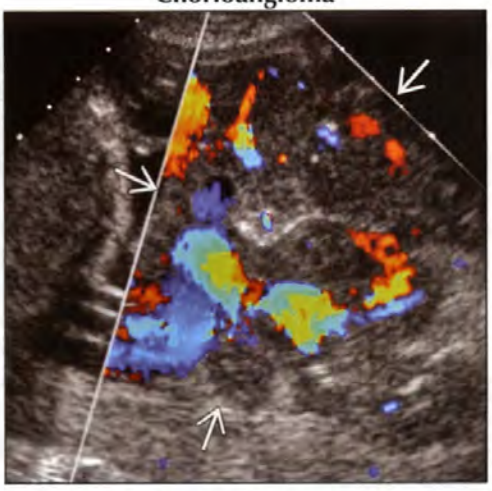
(Left) *Sagittal ultrasound shows a retroplacental focal myometrial contraction, which mimics a retroplacental myoma. Follow-up ultrasound was necessary to prove this was a contraction.* ***(Right)*** *Axial ultrasound shows that the myometrium is thick at the site of the FMC. This more typical appearance shows how the inner myometrium is preferentially thickened.*

PLACENTAL MASS-LIKE LESIONS

Chorioangioma

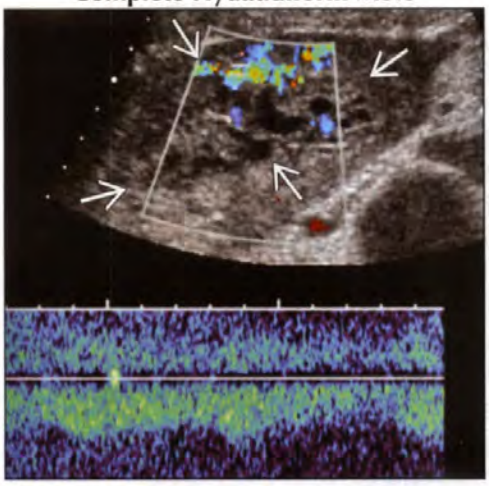


Chorioangioma



(Left) *Axial color Doppler ultrasound shows a typical chorioangioma. The mass is hypoechoic with blood flow. It is located on the fetal side, near the umbilical cord insertion site.* ***(Right)*** *Axial color Doppler ultrasound shows a large heterogeneous placental mass with vascular flow. Large chorioangiomas are uncommon and associated with fetal cardiac failure.*

Complete Hydatidiform Mole



Complete Hydatidiform Mole



(Left) *Sagittal pulsed Doppler ultrasound of an early second trimester pregnancy shows that the uterus is filled by a cystic, vascular mass and that there is no fetus.* ***(Right)*** *Axial transabdominal ultrasound in another case shows a molar pregnancy and a coexistent twin. The mole is cystic and is separated from the normal placenta by a thick dividing membrane.*

Placental Teratoma



Placental Teratoma



(Left) *Axial ultrasound shows a well-marginated heterogeneous mass arising from the placenta. The mass has diffuse medium level echoes with floating debris.* ***(Right)*** *Sagittal ultrasound shows a focal echogenicity with shadowing, consistent with a large calcification, in the mass. Calcification is the most specific finding for this rare mass.*

PLACENTOMEGALY

DIFFERENTIAL DIAGNOSIS

Common
- Hydrops
- Acute Placental Abruption
- Macrosomia
- Twins

Less Common
- Battledore Placenta
- Intrauterine Growth Restriction (IUGR)

Rare but Important
- Triploidy

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Normal placenta is < 40 mm thick
- Placenta grows approximately 1 mm/wk
  - 10 wk placenta = 10 mm
  - 20 wk placenta = 20 mm
  - 30 wk placenta = 30 mm
- Evaluate placental attachment
  - Normal attachment
    - Hydrops
    - Macrosomia
    - Diabetes
    - IUGR
  - Small attachment
    - Battledore placenta
  - Heterogeneous attachment
    - Abruption
- Evaluate placental morphology
  - Increased sonolucencies
    - Triploidy
    - IUGR
  - Focal thickening
    - Dichorionic twins
    - Focal abruption
  - Large thin placenta
    - Monochorionic twins
  - Heterogeneous thick placenta
    - Abruption
    - IUGR
- Use Doppler to assess placental function
  - Uterine artery Doppler
    - Low resistive flow in second trimester
    - Post systolic notch abnormal > 18 wks
  - Umbilical artery (UA) Doppler
    - Low resistive flow after 2nd trimester
    - Absent diastolic flow abnormal > 16 wks
    - Systolic/diastolic ratio < 3.0 if > 30 wks
    - Reversed diastolic flow always abnormal
  - Fetal middle cerebral artery (MCA) Doppler
    - Higher resistive flow than UA
    - Severe IUGR leads to ↓ resistance
- Use MCA peak systolic velocity to assess for fetal anemia
  - Insonation angle is important
    - 0° angle on MCA
  - Use normogram to compare with gestational age
    - ↑ Velocity suggests anemia

Helpful Clues for Common Diagnoses
- **Hydrops**
  - Excessive fetal body fluid
    - Skin edema
    - Pleural effusion
    - Ascites
    - Pericardial effusion
  - Hydrops if ↑ fluid in 2 areas
  - Amniotic fluid findings variable
    - Polyhydramnios
    - Oligohydramnios
  - Placenta edema
    - More likely if immune hydrops
  - Common causes of nonimmune hydrops
    - Infection
    - Aneuploidy (Turner most common)
    - Heart failure
    - Fetal anemia of any etiology
- **Acute Placental Abruption**
  - Acute blood has same echogenicity as placenta
    - Mimics thick placenta
    - Becomes hypoechoic with time
  - Direct hemorrhage into placenta can occur
    - Most common with retroplacental abruption
    - Arterial hemorrhage
  - Use power Doppler
    - No flow in hematoma
  - > 50% detachment associated with > 50% fetal death
- **Macrosomia**
  - Definition
    - Estimated fetal weight > 90th percentile
    - Birthweight > 4500 g
  - Causes
    - Hereditary
    - Diabetes
    - Beckwith-Wiedemann (BW)
  - Polyhydramnios

- Both hereditary causes and diabetes associated with ↑ fluid
- Impaired swallowing if macroglossia and BW

- **Twins**
  - Dichorionic twins may have appearance of single large placental mass
    - "Twin peak" or "delta" signs are focal point where 2 placentas meet
    - Thick membrane (4 layers) extends from "peak" or "delta"
  - Large thin placenta of monochorionic twinning
    - No triangular peak
    - Thin membrane (two layers)

Helpful Clues for Less Common Diagnoses

- **Battledore Placenta**
  - Thick placenta with small attachment
    - ↑ Risk for abruption
  - Marginal cord insertion site
    - Within 2 cm of placental margin
    - At risk for becoming velamentous
- **Intrauterine Growth Restriction (IUGR)**
  - Estimated fetal weight < 10th percentile
  - Variable placenta appearance
    - ↑ Sonolucencies
    - Calcifications
    - Thin or thick placenta
  - Doppler findings
    - Uterine artery with post systolic notch
    - High resistive UA waveform
    - Low resistive MCA waveform
  - Oligohydramnios

Helpful Clues for Rare Diagnoses

- **Triploidy**
  - Karyotype is 69 XXY or 69 XYY
    - Diandry is 69 XYY
    - Digyny is 69 XXY
  - Partial mole if diandry
  - Placental findings with triploidy
    - Thick cystic placenta more common if partial mole
    - Thin placenta more likely if digyny
  - Fetal findings
    - Severe IUGR
    - Multiple anomalies
  - Theca-lutein cysts in maternal ovaries
    - Enlarged ovaries with multiple cysts
    - Seen more often with partial mole
  - Oligohydramnios

Other Essential Information

- Look for treatable causes of hydrops
  - Fetal anemia
    - Use MCA peak systolic velocity values
    - Treated with in-utero transfusion
  - Fetal tachyarrhythmia
    - Treated with medication
- Severe IUGR can mimic triploidy
  - Placenta with sonolucencies
  - Oligohydramnios
  - Preeclampsia presentation
  - Need amniocentesis to differentiate

Hydrops



Sagittal ultrasound shows placentomegaly from immune hydrops. The placenta measures 7.3 cm thick. In addition, there is fetal anasarca, pleural effusion, and ascites.

Hydrops



Axial ultrasound of the same case of immune hydrops shows polyhydramnios. Once again the thick placenta is seen anteriorly, as is the torso of the hydropic fetus.

PLACENTOMEGALY

Acute Placental Abruption





(Left) *Axial ultrasound shows a markedly thick and heterogeneous placenta. In this case there was a large acute retroplacental and intraplacental hemorrhage.* ***(Right)*** *Axial ultrasound in another case shows what initially appears to be a thick heterogeneous placenta. However, a large hypoechoic subacute retroplacental abruption has lifted the placenta off the uterine wall, and the two together mimic a thick placenta.*

Twins





(Left) *Axial ultrasound shows the "twin peak" or "delta" sign of a dichorionic, diamniotic gestation. The two placentae meet at the area of focal thickening, and the thick 4 layer membrane arises from the peak.* ***(Right)*** *Sagittal T2WI MR shows a long, thin placenta in a case of monochorionic twinning. Notice the lack of any focal placental thickening.*

Battledore Placenta



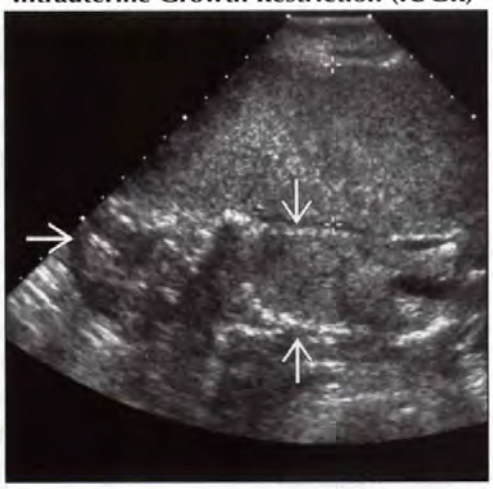


(Left) *Axial ultrasound shows a thick placenta with a relatively small attachment surface area to the uterus.* ***(Right)*** *Axial oblique ultrasound of the same placenta shows that the cord insertion site is near the placental margin. This pregnancy was complicated by placental insufficiency and IUGR.*

Intrauterine Growth Restriction (IUGR)

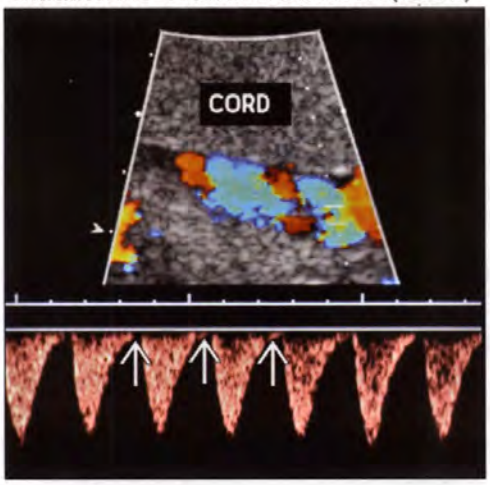


Intrauterine Growth Restriction (IUGR)

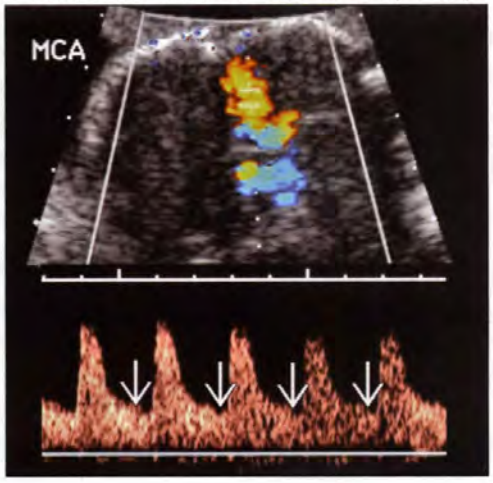


(Left) *Sagittal ultrasound shows a thickened placenta* ➡ *in a pregnancy complicated by hypertension, oligohydramnios, and IUGR.* ***(Right)*** *Sagittal ultrasound of the same case as previous image once again shows a thick placenta (calipers) as well as oligohydramnios. Note the lack of fluid surrounding the fetus* ➡.

Intrauterine Growth Restriction (IUGR)



Intrauterine Growth Restriction (IUGR)

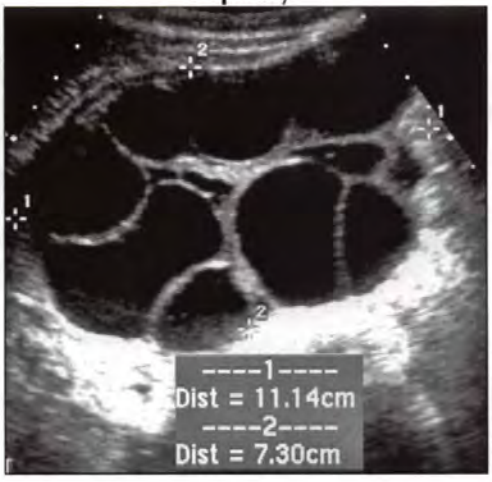


(Left) *Sagittal pulsed Doppler ultrasound of the UA in the same pregnancy as previous image shows absent diastolic flow* ➡. ***(Right)*** *Axial pulsed Doppler ultrasound of the MCA shows lower resistive flow than the UA. There is more diastolic flow* ➡ *in the MCA than in the UA. This reversal of the normal relationship between the two vessels suggests the fetus is in "brain sparing" physiology secondary to severe placental insufficiency.*

Triploidy



Triploidy



(Left) *Sagittal ultrasound shows a thick placenta* ➡, *which contains multiple cysts* ➡. *The fetus had severe IUGR and multiple anomalies.* ***(Right)*** *Axial ultrasound of one of the ovaries, in the same case as previous image, shows enlargement secondary to multiple theca lutein cysts. The fetus had triploidy.*

ABNORMAL PLACENTAL CORD INSERTION

DIFFERENTIAL DIAGNOSIS

Common
- Marginal Cord Insertion
- Velamentous Cord Insertion (VCI)

Less Common
- Umbilical Cord Cyst
- Vasa Previa (VP)

Rare but Important
- Umbilical Cord Aneurysms

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Placental cord insertion (PCI) identification
  - Easily achievable
    - Seen in almost 100% 2nd trimester cases
    - More difficult if posterior placenta and 3rd trimester fetus
  - Use grayscale first
    - Find general area of PCI
    - Less motion artifact than Doppler
  - Use color Doppler to confirm
    - Rule out adjacent cord
    - Show insertion and branching vessels
    - All vessels should be on fetal surface of placenta
    - Rule out submembranous vessels
  - Use pulsed Doppler to show fetal flow
    - Low resistive arterial flow
    - Document fetal heart rate
- Look for PCI routinely in high risk cases
  - Monochorionic twins
  - Placenta previa
  - Succenturiate lobe
  - Abnormally large or small placenta
  - Intrauterine growth restriction
  - Anomalous fetus
- Use Doppler to make a specific diagnosis
  - Look for submembranous vessels
    - Velamentous cord
    - Vasa previa
  - Cyst versus aneurysm
    - Identical appearance without color Doppler
  - Use pulse Doppler
    - Differentiate maternal from fetal vessels

Helpful Clues for Common Diagnoses
- **Marginal Cord Insertion**
  - PCI is within 2 cm of placental edge
  - Good prognosis if isolated finding
  - Sometimes associated with other placental abnormalities
    - Small placenta
    - Unusually thick placenta (Battledore)
    - Monochorionic twinning
    - Abruption
    - Intrauterine growth restriction
  - Progression to velamentous cord insertion
    - Rare complication
    - More likely if PCI is < 5 mm from margin
    - Follow-up scans indicated
- **Velamentous Cord Insertion (VCI)**
  - Membranous cord insertion
    - VCI is often adjacent to placenta
    - Some or all vessels are submembranous
    - VCI may be seen between two placental lobes
  - Atypical vessel appearance from lack of placental support
    - Dilated vessels
    - Excessively separated vessels
  - Doppler essential for diagnosis
    - Helps identify VCI
    - Shows vessels extending from VCI to placenta
    - Pulsed Doppler proves vessels are fetal

Helpful Clues for Less Common Diagnoses
- **Umbilical Cord Cyst**
  - 33% of all UC cysts are at PCI
    - Other 2/3 are mid cord or near fetus
  - Often multiple and clustered at PCI
  - PCI cysts are less likely to resolve
    - First trimester UC cysts often resolve
  - Rare complication includes intracystic hemorrhage
    - May lead to cord compromise
  - Rarely associated with fetal anomalies and aneuploidy
    - Genitourinary anomalies
    - Trisomy 18
    - Amniocentesis probably not necessary if isolated finding in low risk patient
- **Vasa Previa (VP)**
  - Submembranous fetal vessels near internal cervical os
    - Within 2 cm of os considered VP
  - VP from succenturiate lobe
    - Most common etiology

- Communicating vessels between main lobe and succenturiate lobe located near internal cervical os
- Associated with low lying placenta (primary or succenturiate)

- Vasa previa from VCI
  - Low lying placenta with VCI
  - Velamentous vessels near internal cervical os
- Prenatal diagnosis is imperative
  - 60-80% fetal mortality associated with missed diagnosis
  - Fetal exsanguination with cervical dilatation

Helpful Clues for Rare Diagnoses

- **Umbilical Cord Aneurysms**
  - Umbilical artery (UA) aneurysm
    - Most common location is at PCI
    - Saccular dilatation of UA
    - May have arteriovenous fistula to umbilical vein
    - Associated with single umbilical artery, fetal anomalies and trisomy 18
    - Best prognosis if isolated finding
  - Umbilical vein (UV) varix
    - Most common location is intraabdominal in fetus
    - Rarely in free floating loops of cord
    - Associated with increased venous pressure and hydrops
    - Rarely can thrombose or rupture

Other Essential Information

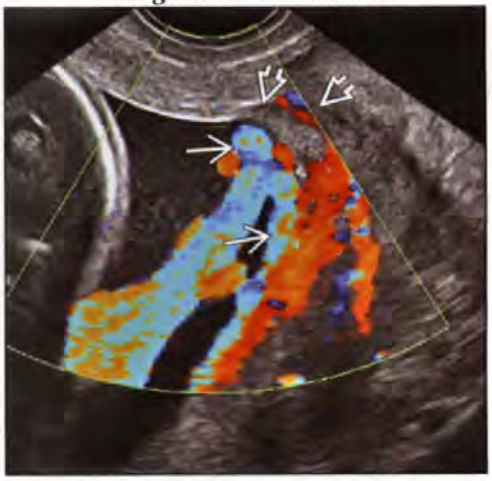
- Do not confuse VP with marginal sinus previa (MSP)
  - MSP definition
    - Low lying placenta
    - Marginal placental vessels < 2 cm from internal cervical os
  - MSP vessels are maternal
    - Pulse Doppler shows placental venous flow
    - No fetal arterial flow detectable
    - Bleeding from MSP is maternal blood
  - VP vessels are fetal
  - Vasa previa vs. marginal sinus previa management
    - MSP managed as marginal placenta previa
    - MSP may resolve with advancing pregnancy
    - VP managed with elective cesarean section delivery
    - 3rd trimester VP often managed as an inpatient
- Look for PCI in multiple gestation cases
  - Not all discordant twin growth is twin-twin transfusion
    - Suspect diagnosis if no fluid discordance
    - Monochorionic twinning at higher risk for VCI
    - Unequal sharing of placenta is possible cause of discordant growth

Marginal Cord Insertion



Sagittal transvaginal ultrasound shows the placental cord insertion ➡ is located within 5 mm of the thin placental margin ⇨. There is risk of progression to a velamentous insertion.

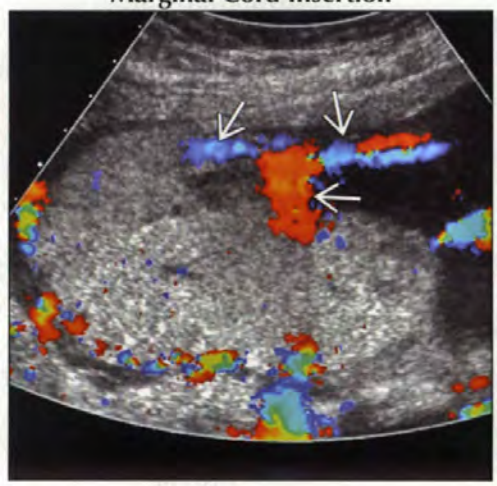
Marginal Cord Insertion



Sagittal color Doppler ultrasound confirms that the umbilical cord inserts on the margin of the placenta ⇨. All the vessels are attached to the placenta ➡ and no velamentous vessels are seen.

ABNORMAL PLACENTAL CORD INSERTION

Marginal Cord Insertion

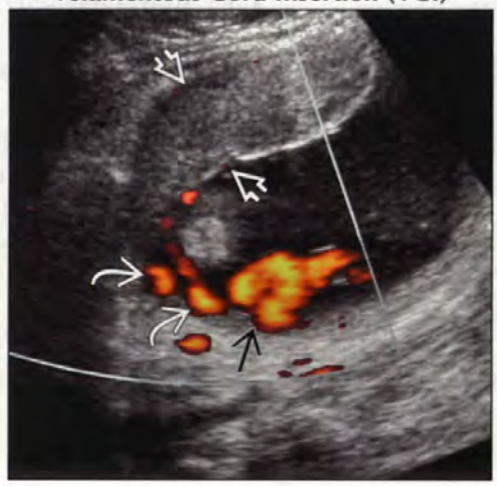


Marginal Cord Insertion



(Left) *Sagittal ultrasound shows the umbilical cord insertion site ➡ along the margin of a thick placenta with a small uterine implantation surface area (battledore placenta). Note the dividing vessels at the insertion site.* ***(Right)*** *Sagittal ultrasound from the same patient as previous image, shows an associated marginal abruption ➡ extending from the inferior placental margin ➡ to the cervix ➡. This pregnancy was complicated by placental insufficiency.*

Velamentous Cord Insertion (VCI)

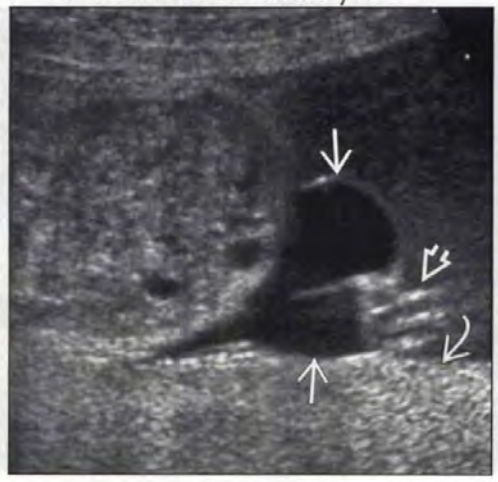


Velamentous Cord Insertion (VCI)



(Left) *Transverse power Doppler ultrasound in a case of monochorionic twinning and discordant growth shows the small twin's cord inserts upon the posterior uterus ➡. The cord vessels travel beneath the membranes ➡ to reach the anterior placenta ➡.* ***(Right)*** *Sagittal color Doppler ultrasound shows another case with velamentous cord insertion ➡ between an anterior placenta ➡ and a posterior succenturiate lobe ➡.*

Umbilical Cord Cyst



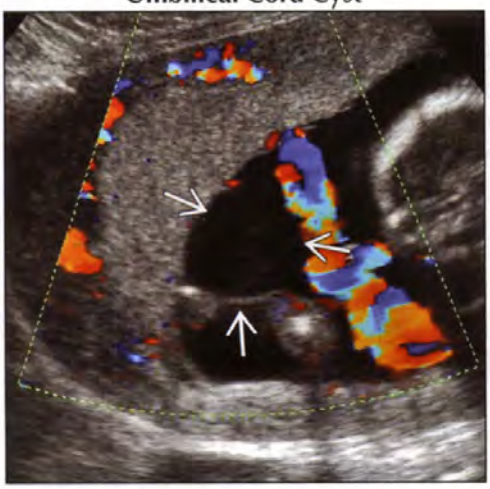
Umbilical Cord Cyst



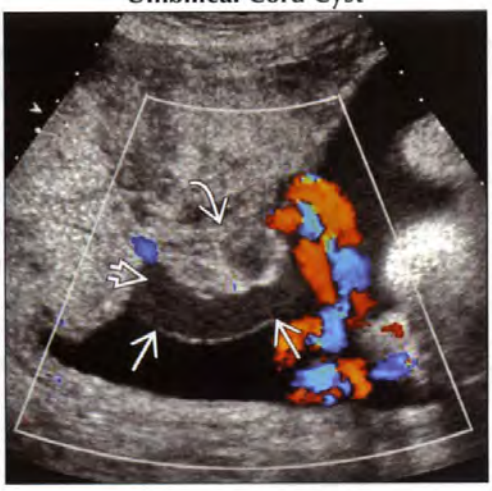
(Left) *Axial ultrasound shows two anechoic cysts ➡ adjacent to the umbilical cord ➡ and placenta ➡.* ***(Right)*** *Axial color Doppler ultrasound shows the umbilical cord drapes over the cyst ➡ which is located near the insertion site ➡.*

ABNORMAL PLACENTAL CORD INSERTION

Umbilical Cord Cyst

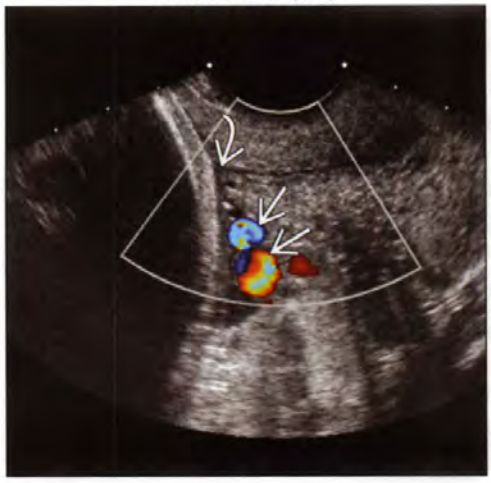


Umbilical Cord Cyst

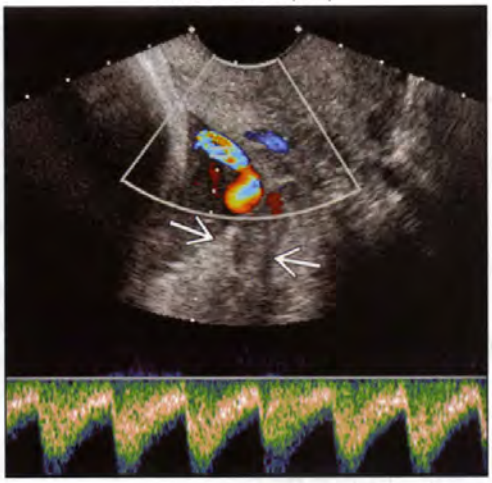


(Left) *Sagittal color Doppler ultrasound shows a large anechoic cord insertion cyst* ➡ *incidentally noted in a second trimester case.* ***(Right)*** *Axial color Doppler ultrasound in the third trimester, shows the cyst* ➡ *is no longer anechoic. It now contains diffuse low level echoes* ➡ *and echogenic material* ➡. *At delivery, a hemorrhagic cyst was found at the cord insertion site.*

Vasa Previa (VP)



Vasa Previa (VP)

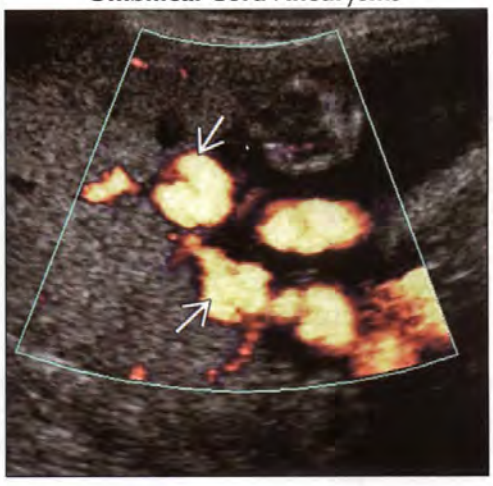


(Left) *Sagittal color Doppler ultrasound shows prominent vessels* ➡ *within 2 cm of the internal cervical os* ➡. ***(Right)*** *Sagittal pulsed Doppler ultrasound shows fetal arterial flow within the vessels. The placental edge* ➡ *is also seen on this image. The patient has a velamentous cord insertion with fetal vessels in close proximity of the internal cervical os.*

Umbilical Cord Aneurysms



Umbilical Cord Aneurysms



(Left) *Sagittal ultrasound shows two cyst-like lesions* ➡ *at the placenta cord insertion site.* ***(Right)*** *Sagittal power Doppler ultrasound shows blood flow in these lesions* ➡. *Pulsed Doppler showed arterial flow in these umbilical artery aneurysms. This fetus had trisomy 18.*

ABNORMAL UMBILICAL CORD

DIFFERENTIAL DIAGNOSIS

Common

- Umbilical Cord Cyst
  - Allantoic Cyst with Patent Urachus
  - Omphalomesenteric Duct Cyst
  - Pseudocyst
- Cystic Wharton Jelly
- Omphalocele (Mimic)
- Physiologic Gut Herniation (Mimic)

Less Common

- Cord Knot
- Short Cord
- Abnormal Cord Coiling
- Cord Hematoma
- Cord Thrombosis

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Cord assessment is an important part of all OB scans
  - Look at abdominal cord insertion site
  - Look at placental cord insertion site
  - Evaluate cord structure
    - Are the vessels normal?
    - Is the cord length normal?
    - Is there an appropriate degree of "twist" to the vessels?

Helpful Clues for Common Diagnoses

- **Umbilical Cord Cyst**
  - Equally common at fetal & placental ends and in free loops of cord
  - May be paraxial (eccentric, do not displace vessels) or axial (centrally located and splay vessels)
  - Generally thin-walled, anechoic, often multiple
    - If echogenic content, consider intracystic hemorrhage, which may lead to cord compromise
  - May be true cysts (allantoic, omphalomesenteric duct cysts) or pseudocysts
  - Isolated cord cysts may spontaneously resolve with etiology never determined
  - **Allantoic Cyst with Patent Urachus**
    - Always near fetal insertion
    - May grow and compress cord
    - Allantoic cysts may be isolated or communicate with the urachus
    - Patent urachus: Cystic mass superior to, and communicating with, bladder
    - Obstructed bladder decompresses into urachus and base of cord
  - **Omphalomesenteric Duct Cyst**
    - 2° to omphalomesenteric duct remnant
    - + Abdominal wall anomalies
    - + Intraabdominal mesenteric cysts
    - + Other severe anomalies
  - **Pseudocyst**
    - Often associated with cystic Wharton jelly
    - May also be sequela of cord hematoma
- **Cystic Wharton Jelly**
  - Mucoid degeneration of abnormal Wharton jelly
  - Innumerable small pseudocysts develop surrounding cord vessels
  - Likely to be associated with aneuploidy and syndromes
- **Omphalocele (Mimic)**
  - Smooth mass protruding from central anterior abdominal wall with covering membrane
  - Umbilical cord inserts onto membrane, usually central but may be eccentric
  - Liver and small bowel most common contents (those with small bowel most likely to be confused with abnormal cord)
- **Physiologic Gut Herniation (Mimic)**
  - Normal embryological developmental phenomenon
  - Bowel elongates, herniates into base of cord, rotates 270°, then returns to peritoneal cavity
  - Bowel returns to abdomen by 11.2 weeks
  - Should not extend more than 1 cm into base of cord
  - Never contains liver

Helpful Clues for Less Common Diagnoses

- **Cord Knot**
  - True knot
    - Most common in monoamniotic twins
    - Rarely also seen in singletons
    - Risk factors include advanced maternal age, multiparity, long umbilical cords
    - May restrict flow → hypoxia, growth restriction
    - May occlude cord → fetal demise
    - Reported to lead to a 4-fold increase in fetal loss

- False knot
  - Due to kinks in vessels, not a true knot
  - No known clinical significance

- **Short Cord**
  - Average cord is 55 cm (range 35-80 cm)
  - Not possible to measure length prenatally, but short cord subjectively associated with fetus being "tethered"
  - Watch fetal movement in real time to assess for akinesia/arthrogryposis sequence
  - Associated with abruption/cord rupture
- **Abnormal Cord Coiling**
  - Normal cord is helical, with up to 380 helices
  - Coiling is well established by 9 weeks and is thought to strengthen cord
  - Lack of normal coiling and length associated with fetal akinesia
    - Look at movements in real time
    - Assess joints for abnormal posture
- **Cord Hematoma**
  - True cord hematoma is due to extravasation of blood into Wharton jelly surrounding cord vessels
  - Use Doppler to look for increased vascular resistance if large hematoma
  - May occur following invasive prenatal procedures
  - May also be seen adherent to cord secondary to intra-amniotic bleeding from any cause
- **Cord Thrombosis**
  - Look for hypoechoic material distending vessels on grayscale images
  - Lack of flow on color or power Doppler
  - Venous thrombosis is a cause of sudden fetal demise
  - Most cases with surviving fetuses are reported as pathological finding after emergency delivery for distress in labor
  - Umbilical vein varix is a risk factor
  - May occur following invasive prenatal procedures, especially if large hematoma compresses vessels
  - May occur in association with large cord cysts, particularly at placental end of cord

Other Essential Information

- Cord embryology
  - Early connecting stalk connects the embryo to the chorion
  - Allantois forms from caudal end of yolk sac
  - Cord formed from fusion of allantois and connecting stalk
  - Allantois functions as primitive bladder and early blood forming organ
    - Persistent segments of allantois are termed urachal remnants
    - Urachus serves as "pop-off valve" to decompress bladder if outlet obstruction
    - Allantois involutes to become median umbilical ligament
- Multiple umbilical cord cysts associated with 7.6x increased risk of poor outcome
- Straight cords with few or absent helices are associated with adverse fetal outcomes

Umbilical Cord Cyst



Transvaginal ultrasound shows the yolk sac ⇒ outside the amnion ⇒ that contains the embryo ⇒ and developing cord. A cord cyst ⇒ is seen. This resolved spontaneously, and the infant was normal at birth.

Umbilical Cord Cyst



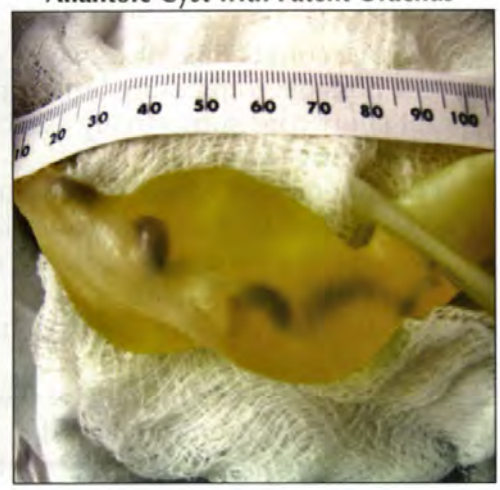
Axial oblique color Doppler ultrasound shows an axial simple cyst ⇒ within the cord, splaying the vessels, close to the abdominal insertion. This was an isolated finding in an otherwise normal fetus.

ABNORMAL UMBILICAL CORD

Allantoic Cyst with Patent Urachus



Allantoic Cyst with Patent Urachus



(Left) *Sagittal ultrasound shows a large allantoic cyst ➡ at the fetal end of the umbilical cord. A patent urachus was not seen prenatally but was suspected, given the size and location of this umbilical cord cyst.* ***(Right)*** *Clinical photograph of a newborn in a similar case shows a urine-filled umbilical cord cyst. A patent urachus was diagnosed on voiding cystourethrogram and was surgically treated.*

Allantoic Cyst with Patent Urachus

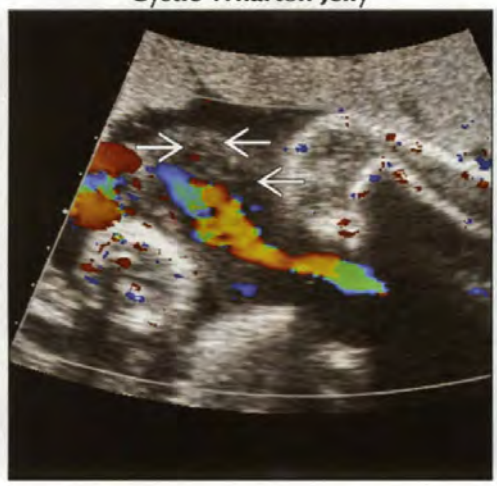


Omphalomesenteric Duct Cyst

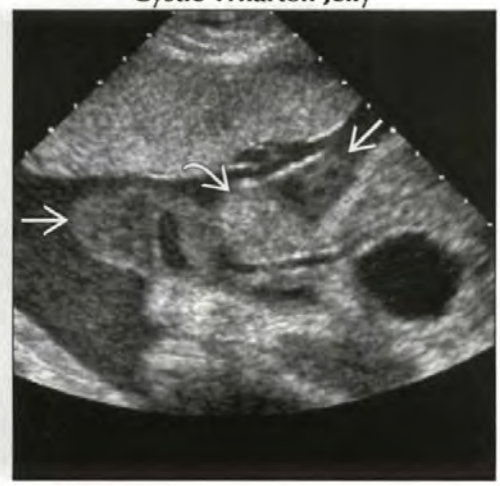


(Left) *Gross pathology of a fetus with prune belly syndrome shows fluid ➡ within the cord, which was attributed to communication with the bladder through a patent urachus.* ***(Right)*** *Ultrasound of the umbilical cord shows a cyst ➡ and cystic Wharton jelly ➡. There were multiple other anomalies including an omphalocele, which is a common associated finding with omphalomesenteric duct cyst.*

Cystic Wharton Jelly



Cystic Wharton Jelly



(Left) *Color doppler ultrasound shows flow in the umbilical vessels that are encased in thickened Wharton jelly. There are tiny cystic areas ➡ indicating mucoid degeneration.* ***(Right)*** *Axial oblique ultrasound shows a bowel-only omphalocele ➡ associated with mucoid degeneration of Wharton jelly ➡. Amniocentesis was normal. After clinical evaluation, the infant was diagnosed with Beckwith-Wiedemann syndrome.*

ABNORMAL UMBILICAL CORD

Omphalocele (Mimic)



Physiologic Gut Herniation (Mimic)



(Left) *Axial ultrasound in the second trimester shows a bowel-only* ➡ *omphalocele in the base of the cord* ➡. *In this case chromosomes were normal, and surgical repair was relatively simple.* ***(Right)*** *Axial color Doppler ultrasound in the first trimester shows physiological herniation of bowel* ➡ *into the base of the cord* ➡. *This normal embryological process should not be mistaken for an omphalocele or cord mass.*

Cord Knot



Cord Knot

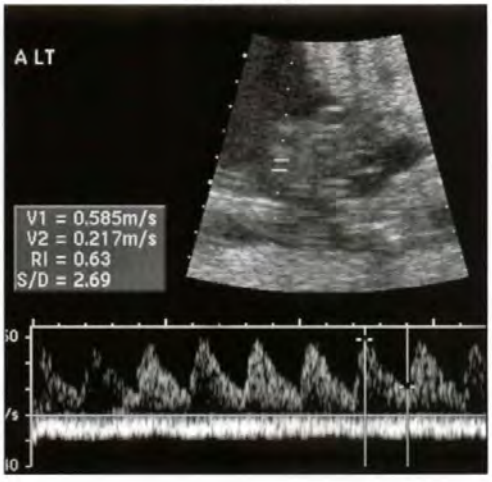


(Left) *Sagittal ultrasound shows several inseparable cord loops* ➡ *between monoamniotic twins. The orientation of the loops did not change during the scan concerning for entanglement.* ***(Right)*** *Color Doppler ultrasound in the same case shows flow in all cord loops* ➡ *and both fetuses* ➡, *indicating no evidence of compromise secondary to cord entanglement at this time.*

Cord Knot



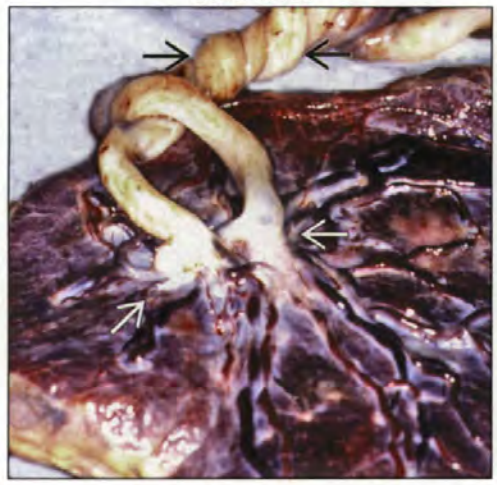
Cord Knot



(Left) *Axial oblique ultrasound in the same pregnancy three weeks later shows skin edema* ➡ *and overlapping skull bones* ➡, *indicating the demise of twin B. Cord entanglement can result in demise of one or both twins.* ***(Right)*** *Pulsed Doppler ultrasound in the same case shows normal umbilical arterial and venous flow to twin A despite the cord knot. In this case twin A suffered no apparent adverse consequences of co-twin demise and was delivered close to term.*

ABNORMAL UMBILICAL CORD

Cord Knot





(Left) *Gross pathology shows cord entanglement in monoamniotic twins. Note the close proximity of the placental cord insertion sites.* ***(Right)*** *Color Doppler ultrasound shows an apparent cord knot close to the abdominal wall insertion site in a singleton pregnancy. There were no other findings. This was a false knot due to kinked vessels rather than a true knot.*

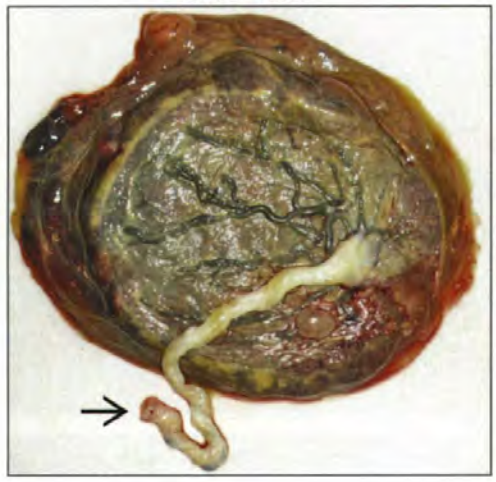
Short Cord





(Left) *Color Doppler ultrasound shows a short segment of cord at the placental insertion site. A fixed position lower extremity is also seen.* ***(Right)*** *Sagittal oblique transvaginal ultrasound in the same case (at 15 weeks gestation) shows extensive edema of the torso. Note that the fetus is in a fixed "pike" position. No movement was observed during the examination indicating fetal akinesia sequence and short umbilical cord.*

Short Cord

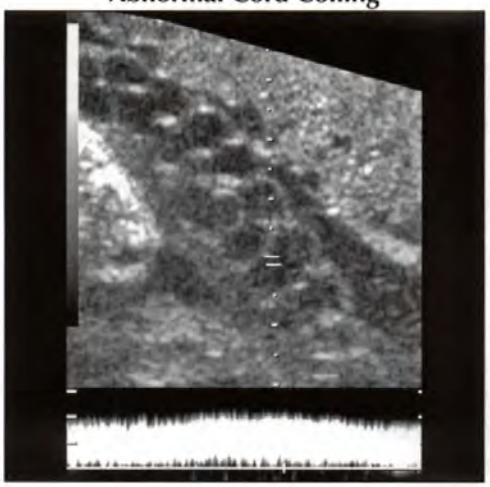




(Left) *Gross pathology shows a short cord with diminished coiling from a dizygotic twin with fetal akinesia sequence. The cord measured 38 cm in length. The other fetus was normal.* ***(Right)*** *Gross pathology in the same case shows the abnormal extremity posturing in the twin who died immediately after delivery. There were fixed flexion deformities of the hips and knees, camptodactyly of the fingers, and rocker bottom feet.*

ABNORMAL UMBILICAL CORD

Abnormal Cord Coiling



Abnormal Cord Coiling

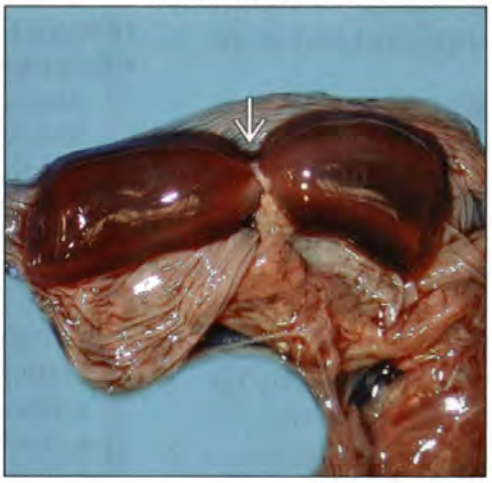


(Left) *Pulsed Doppler ultrasound shows hypercoiled cord with normal venous flow. Amniotic fluid volume and cord Doppler were normal, as was the infant at birth.* ***(Right)*** *Ultrasound of a third trimester fetus with a two vessel cord shows diminished cord coiling. Fetal movement and growth were normal in this case.*

Cord Hematoma



Cord Thrombosis



(Left) *Ultrasound shows "ragged", amorphous material surrounding the cord vessels. The patient had severe vaginal bleeding and had an emergency C-section for abruption. This is clot adherent to the surface of the cord.* ***(Right)*** *Gross pathology shows an amniotic band wrapped tightly around the cord, resulting in thrombosis and fetal demise. This was particularly unfortunate as the only fetal anomaly involved the fingers of one hand.*

Cord Thrombosis



Cord Thrombosis



(Left) *Color Doppler ultrasound shows thrombosis of one umbilical artery. This was the pump twin in a TRAP pregnancy treated with radiofrequency ablation. Cord thrombosis is not a generally recognized complication.* ***(Right)*** *Pulsed Doppler ultrasound in the same case shows normal umbilical vein flow. The fetus was normally grown and active; therefore no intervention occurred. Delivery was close to term with no adverse consequences detected.*

ABNORMAL UMBILICAL VESSELS

DIFFERENTIAL DIAGNOSIS

Common
- Single Umbilical Artery
- Hypoplastic Umbilical Artery
- Velamentous Cord

Less Common
- Persistent Right Umbilical Vein
- Fused Umbilical Cords
- Conjoined Twins
- Twin Reversed Arterial Perfusion

Rare but Important
- Body Stalk Anomaly
- Umbilical Cord Aneurysms
  - Umbilical Vein Varix
  - Umbilical Artery Aneurysm

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Cord assessment is an important part of all obstetric scans
  - Look at abdominal cord insertion site
  - Look at placental cord insertion site
  - Evaluate cord structure
    - How many vessels are there?
    - Is the cord length normal?
    - Is there an appropriate amount of "twist" to the vessels?
  - Follow umbilical vein
    - Normal course of umbilical vein (UV) is to enter left lobe of liver medial to gallbladder
    - UV connects with left portal vein (LPV)
    - LPV connects with inferior vena cava via ductus venosus

Helpful Clues for Common Diagnoses
- **Single Umbilical Artery**
  - Seen best on free loop of cord cross-section
  - Only 1 artery adjacent to fetal bladder
  - Single umbilical artery (SUA) is larger than normal UA (i.e., in a 3-vessel cord)
    - Carries twice the blood volume
  - 15% develop intrauterine growth restriction (IUGR)
  - Look for additional fetal anomalies
    - 50% risk of aneuploidy if other anomalies in addition to SUA
- **Hypoplastic Umbilical Artery**
  - Within spectrum of SUA
  - Asymmetry in size of umbilical arteries
  - One artery smaller than the other adjacent to bladder
- **Velamentous Cord**
  - Submembranous cord insertion (i.e., umbilical cord inserts onto membranes not placental disc)
  - Often adjacent to placenta
  - Cord vessels are dilated due to lack of support from surrounding tissue
  - Submembranous vessels are extremely fragile
  - Associated with succenturiate lobe of placenta, placenta previa, twin gestations
  - Vasa previa: Submembranous fetal vessels cross cervical os
    - If membranes rupture fetus can exsanguinate
    - 60-80% fetal mortality if diagnosis missed

Helpful Clues for Less Common Diagnoses
- **Persistent Right Umbilical Vein**
  - Associated with SUA in most cases
  - May be either intrahepatic or extrahepatic
  - Intrahepatic: UV passes to right (lateral) of gallbladder (GB) curving toward stomach
    - GB medially displaced
    - GB transversely oriented
    - UV fuses with left portal vein
  - Extrahepatic: UV bypasses liver and portal system running anterior to liver
    - Drains into systemic veins
    - Associated with aneuploidy
    - Associated with multiple anomalies
- **Fused Umbilical Cords**
  - Abnormal number of cord vessels in excess of the usual 3
    - Most commonly seen with conjoined twins
    - Described in monoamniotic twins where cords fuse proximal to placental insertion site
  - Differentiate from cord knot in monoamniotic twins
    - Cord vessels appear to "branch" within the knot
    - In fused cords, the vessels are tubular with the usual helical twist but no entanglement
    - Fetuses may lie close to each other but do not have contiguous skin covering

ABNORMAL UMBILICAL VESSELS

- **Conjoined Twins**
  - Monochorionic twin gestation
  - Contiguous skin covering between fetuses
  - Variable cord vascular anomalies described
  - Most common is fused cord with 6 vessels (2 arteries and 1 vein from each fetus)
- **Twin Reversed Arterial Perfusion**
  - Monochorionic twin gestation
  - Pump twin structurally normal
  - "Acardiac" twin dysmorphic with extensive soft tissue edema
  - Single umbilical artery in 66% of acardiac twins
  - Hallmark of diagnosis is abnormal direction of flow in UA
    - Normal UA flow is toward placenta, away from fetus
    - In TRAP sequence UA flow is away from placenta, into anomalous fetus

Helpful Clues for Rare Diagnoses

- **Body Stalk Anomaly**
  - Absent or very short umbilical cord
    - Vessels seen running between placental surface and fetal torso
  - Large thoraco-abdominal wall defect without covering membrane
  - Scoliosis is a prominent feature
  - Fixed fetal/placental relationship essential for this diagnosis
- **Umbilical Cord Aneurysms**
  - **Umbilical Vein Varix**
    - Focal dilatation of UV > 9 mm diameter or varix diameter 50% > intrahepatic portion of UV
    - Cyst-like space in upper abdomen with venous flow on Doppler
    - Rarely seen in free-floating loops of cord
    - Evaluate with color and spectral Doppler
    - Increasing turbulence on spectral or incomplete filling on color concerning for thrombus
    - Associated with increased venous pressure and hydrops
  - **Umbilical Artery Aneurysm**
    - Saccular dilatation of umbilical artery
    - Usually near placental end of cord
    - Spectral Doppler shows arterial waveform
    - May have arteriovenous fistula to umbilical vein
    - Look for associated anomalies (associated with trisomy 18)

Other Essential Information

- SUA may be an incidental finding but may be associated with multiple anomalies
  - Careful fetal assessment required for structural malformation
  - If additional malformations seen, risk of aneuploidy up to 50%
  - Even if no other findings fetus at risk for IUGR
    - Follow up growth in 3rd trimester
  - Consider Doppler studies of cord vessels
    - Increased systolic to diastolic ratio associated with increased risk of IUGR

Single Umbilical Artery



Ultrasound shows only 2 vessels in the free-floating loops of the cord. The larger vessel is the UV ➡, and the smaller is the UA ➡. The fetus was otherwise normal.

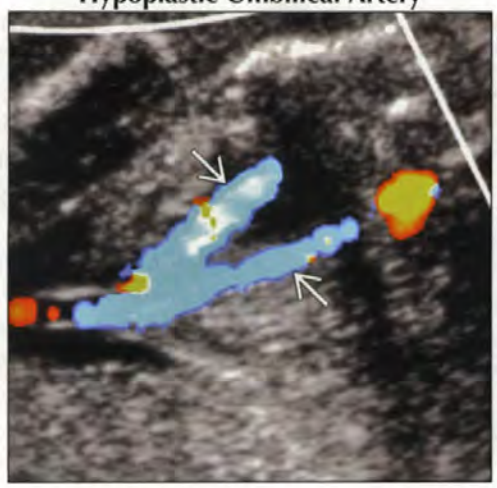
Single Umbilical Artery



Ultrasound, with clinical correlation, shows a 2-vessel cord with a single umbilical artery ➡ wrapping around the vein ➡.

ABNORMAL UMBILICAL VESSELS

Hypoplastic Umbilical Artery



Hypoplastic Umbilical Artery

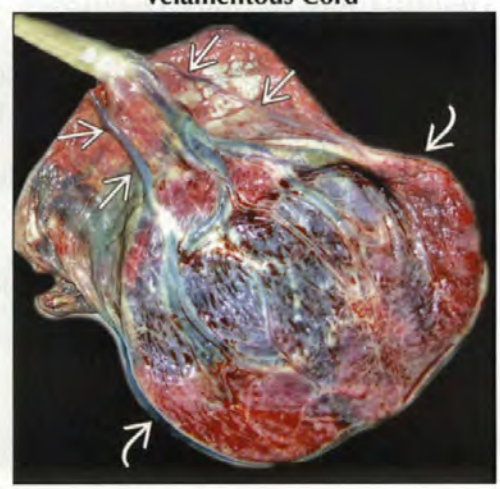


(Left) *Axial color Doppler ultrasound at the fetal bladder shows asymmetric size of the umbilical arteries. Three vessels were present in the cord, with one artery significantly smaller than the other.* ***(Right)*** *Axial oblique color Doppler ultrasound shows 2 umbilical arteries adjacent to the bladder. The left is smaller than the right (Sp = spine). The left artery is more often hypoplastic or absent than the right.*

Velamentous Cord

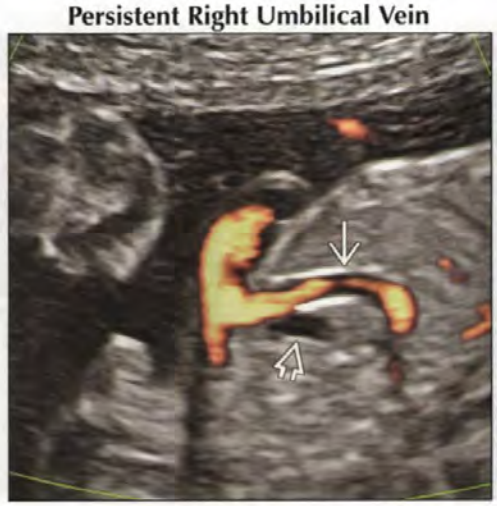


Velamentous Cord

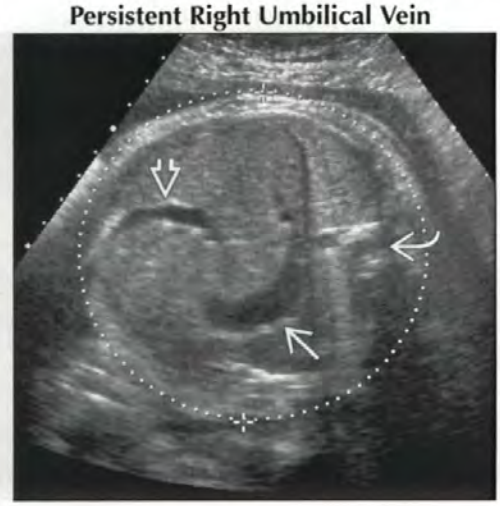


(Left) *Sagittal color Doppler ultrasound shows cord vessels that insert on the membranes and run across the cervix to the placenta. Vasa previa was confirmed by transvaginal scans.* ***(Right)*** *Gross pathology shows a velamentous cord insertion with umbilical vessels running along the membranes before entering the placenta disc.*

Persistent Right Umbilical Vein



Persistent Right Umbilical Vein



(Left) *Axial oblique power Doppler ultrasound shows a persistent right umbilical vein. The gallbladder is located to the left of the umbilical vein and is displaced medially. No other anomalies were detected.* ***(Right)*** *Axial ultrasound shows the characteristic curve of a persistent right umbilical vein. As it enters the abdomen and fuses with the left portal vein it forms a curve, apex to the right, convex away from the stomach (spine).*

ABNORMAL UMBILICAL VESSELS

Fused Umbilical Cords



Fused Umbilical Cords

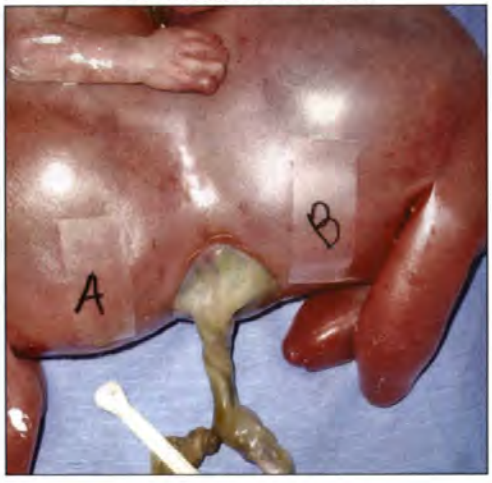


(Left) *Ultrasound in a case of omphalopagus conjoined twins shows 2 placental cord insertion sites ➡ quite close to each other.* ***(Right)*** *Ultrasound of a free floating loop in the same case shows fusion of the cords to form a single 6-vessel cord with 2 veins ➡ and 4 arteries ➡.*

Fused Umbilical Cords



Fused Umbilical Cords

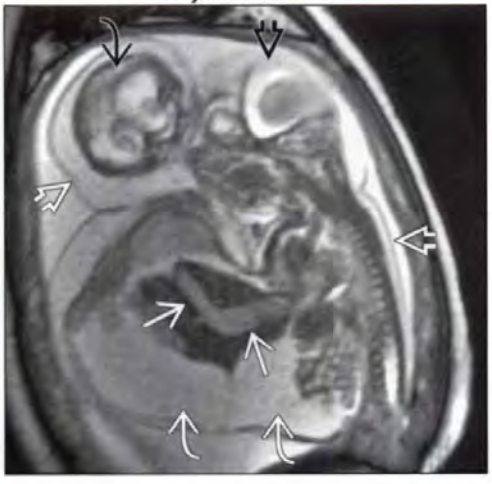


(Left) *Color Doppler ultrasound in the same case shows that the vessels branch immediately after insertion onto an omphalocele, extending into both abdomens (A, B). Both twins had normal portal veins, but there was only one gallbladder and common bile duct.* ***(Right)*** *Gross pathology in a different case of omphalopagus conjoined twins shows the cord insertion with a 5-vessel fused cord (3 arteries, 2 veins) inserting on the apex of an omphalocele.*

Conjoined Twins



Conjoined Twins



(Left) *Axial oblique color Doppler ultrasound shows an unusual large vessel ➡ traversing the livers of conjoined twins, both of whom have ascites ➡.* ***(Right)*** *Sagittal T2WI MR in the same case shows the same large vessel ➡, thought to be an abnormal ductus venosus, shunting placental return (via an anomalous umbilical vein) from the anterior twin ➡ to the posterior twin ➡. Hydrops with skin edema ➡ and ascites ➡ is evident.*

ABNORMAL UMBILICAL VESSELS

Twin Reversed Arterial Perfusion

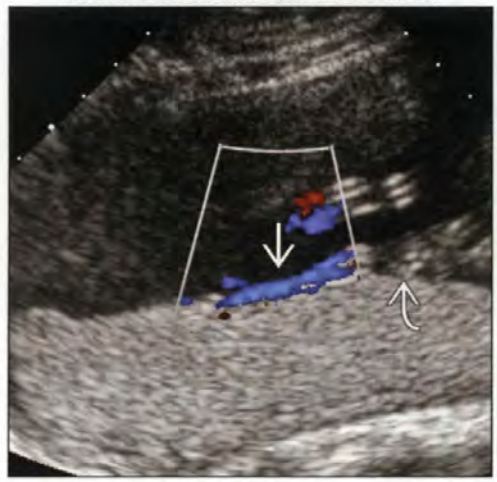


Twin Reversed Arterial Perfusion

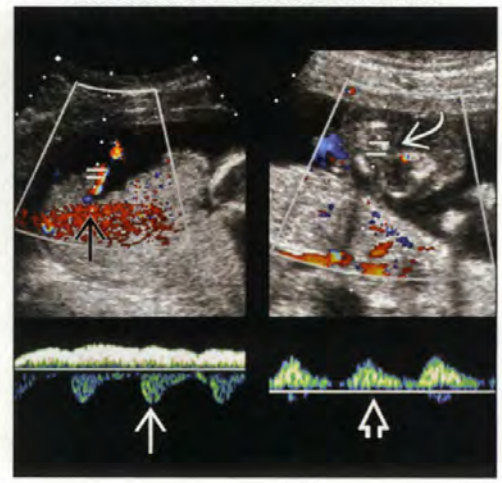


(Left) *Ultrasound at 16 weeks shows the normal pump twin's abdominal cord insertion site. Note the grossly edematous acardiac twin.* ***(Right)*** *Color Doppler ultrasound shows the abnormal fetus with few recognizable structures, although there were 2 flipper-like lower extremities. Umbilical cord flow is present.*

Twin Reversed Arterial Perfusion



Twin Reversed Arterial Perfusion



(Left) *Color Doppler ultrasound shows a shunt vessel on the placental surface, extending from the pump twin's cord insertion site toward the anomalous twin's cord insertion.* ***(Right)*** *Pulsed Doppler ultrasound shows normal UA flow from the pump twin entering the placenta (i.e., away from the fetus). Contrast this with the reversed flow in the UA of the acardiac twin (i.e., away from the placenta, toward the anomalous fetus).*

Twin Reversed Arterial Perfusion



Twin Reversed Arterial Perfusion



(Left) *Gross pathology shows the shunt vessel extending from the pump twin's cord to the acardiac twin's cord. The acardiac twin was small, but cord entanglement resulted in pump twin demise (note the dark thrombosed cord vessels).* ***(Right)*** *Gross pathology from a different case, treated with radiofrequency ablation, shows the post-treatment defect in the acardiac twin. This treatment obliterates the shunt and prevents cord entanglement.*

ABNORMAL UMBILICAL VESSELS

Body Stalk Anomaly



Body Stalk Anomaly



(Left) *Ultrasound shows that the umbilical vessels run directly from the fetus (F) into the placenta (P), as there is no normal cord formed in this condition.* ***(Right)*** *Gross pathology shows the typical findings in body stalk anomaly. The peritoneal cavity is in continuity with the amnion, and the fetus was adherent to the placenta. There is no normal cord insertion site, and the liver and bowel are extruded.*

Umbilical Vein Varix

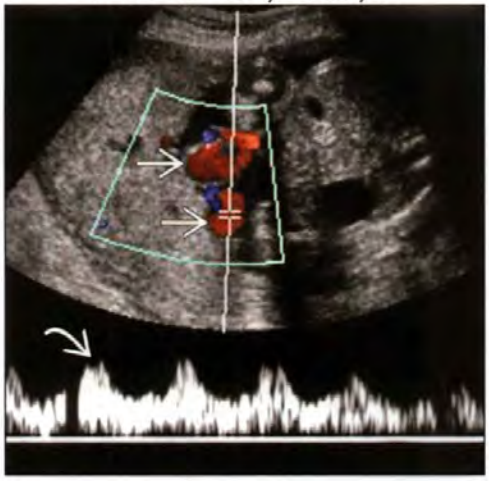


Umbilical Vein Varix

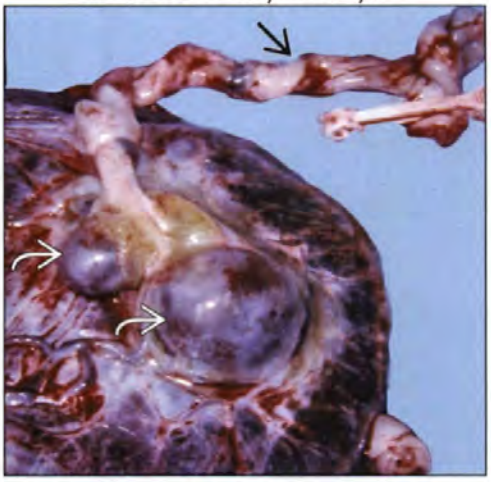


(Left) *Axial ultrasound shows the typical appearance of an umbilical vein varix as an apparently cystic structure in the abdomen.* ***(Right)*** *Axial color Doppler ultrasound in the same case shows blood swirling in the varix. Note that color fills the entire varix. Lack of complete filling is concerning for peripheral thrombus.*

Umbilical Artery Aneurysm



Umbilical Artery Aneurysm



(Left) *Pulsed Doppler ultrasound shows vascular spaces adjacent to the placental cord insertion site. Note the arterial waveform.* ***(Right)*** *Gross pathology confirms 2 umbilical artery aneurysms and a single umbilical artery. The fetus had multiple other anomalies, and amniocentesis revealed trisomy 18.*

SECTION 10
Fluid

Oligohydramnios	10-2
Polyhydramnios	10-6
Echogenic Amniotic Fluid	10-10
Linear Echoes in Amniotic Fluid	10-12

OLIGOHYDRAMNIOS

DIFFERENTIAL DIAGNOSIS

Common

- Preterm Premature Rupture of Membranes (PPROM)
- Intrauterine Growth Restriction (IUGR)
- Bilateral Renal Anomaly
  - Renal Agenesis
  - Ureteropelvic Junction Obstruction (UPJ)
  - Multicystic Dysplastic Kidney (MCDK)
  - Autosomal Recessive Polycystic Kidney Disease
- Bladder Outlet Obstruction
  - Posterior Urethral Valves
  - Urethral Atresia

Less Common

- Twin-Twin Transfusion Syndrome

Rare but Important

- Prune Belly Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Diagnosis of oligohydramnios
  - Subjective
    - ↓ Pockets of fluid
    - Fetus:fluid ratio > 1:1
  - Objective
    - Maximum vertical pocket is ↓ 2 cm
    - Amniotic fluid index < 5 cm

Helpful Clues for Common Diagnoses

- **Preterm Premature Rupture of Membranes (PPROM)**
  - Clinical diagnosis
  - Etiology
    - Spontaneous
    - Infection
    - Post-amniocentesis
  - Worse prognosis if PPROM early or prolonged
    - Early: PPROM < 25 weeks
    - Prolonged: PPROM > 14 days
- **Intrauterine Growth Restriction (IUGR)**
  - Estimated fetal weight < 10th percentile
  - Oligohydramnios often earliest finding
  - Causes
    - Placental insufficiency
    - Fetal aneuploidy/syndrome
  - Doppler assessment
    - ↑ Umbilical artery resistance
    - ↓ Middle cerebral artery resistance
- **Renal Agenesis**
  - Ultrasound findings
    - Anhydramnios (no fluid)
    - Absent kidneys and bladder
    - No renal arteries (color Doppler)
    - Clubfeet, other joint contractures
  - Pitfalls
    - May have normal fluid early (< 17 weeks)
    - Bladder secretions may mimic urine
    - Adrenal gland may mimic kidney
    - Bowel in renal fossa may mimic kidney
  - Fatal prognosis
    - Pulmonary hypoplasia
- **Ureteropelvic Junction Obstruction (UPJ)**
  - Ultrasound findings
    - ↑ Renal pelvis is hallmark finding
    - Renal pelvis ≥ 7 mm after 33 weeks
    - Associated caliectasis
    - No distended ureter or bladder
  - Evaluate contralateral kidney
    - Bilateral UPJ in 10%
    - UPJ + contralateral renal anomaly in 25%
  - Prognosis
    - Depends on severity of obstruction
    - Early oligohydramnios ⇒ pulmonary hypoplasia
    - Post-obstructive cystic dysplasia
- **Multicystic Dysplastic Kidney (MCDK)**
  - Renal tissue replaced by cysts
  - Ultrasound findings
    - Multiple variable-sized cysts
    - Kidney may lose reniform shape
    - ↑ Renal size initially, then ↓
    - Severe oligohydramnios if bilateral
  - 20% of MCDK are bilateral (anhydramnios)
  - 40% have contralateral renal anomaly
  - Poor prognosis if bilateral anomalies
- **Autosomal Recessive Polycystic Kidney Disease**
  - Single gene disorder
    - Distal tubule/collecting duct dilatation
  - Ultrasound findings
    - Enlarging echogenic kidneys
    - Majority detected > 24 weeks
    - Macrocysts rarely seen
    - Variable oligohydramnios
  - Perinatal, neonatal, infantile and juvenile presentations

- Perinatal form with 30-50% mortality
- Severe oligohydramnios ⇒ pulmonary hypoplasia
- Hepatic fibrosis (rarely in utero)

• **Posterior Urethral Valves**
- Partial or complete obstruction
- "Keyhole" appearance
  - ↑ Bladder + ↑ posterior urethra
- Variable hydronephrosis/hydroureter
  - ± Post-obstructive renal cystic dysplasia
- Spontaneous decompression may occur
  - Bladder rupture ⇒ urinary ascites
  - Renal fornix rupture ⇒ urinoma
- In-utero treatment if severe oligohydramnios
  - Serial bladder drainage
  - Vesicoamniotic shunt
- In female fetuses, consider urethral atresia

Helpful Clues for Less Common Diagnoses

• **Twin-Twin Transfusion Syndrome**
- Complication of monochorionic twinning
  - Artery to vein anastomosis in placenta
  - Donor twin partly perfuses recipient twin
- Donor twin with oligohydramnios
  - "Stuck" twin if severe
  - IUGR
  - ↑ Resistive flow in umbilical artery
- Recipient twin with polyhydramnios
  - Larger twin

Helpful Clues for Rare Diagnoses

• **Prune Belly Syndrome**
- Deficient abdominal musculature ⇒ collecting system dilatation
  - Associated cryptorchidism
- Gross dilatation of collecting system is hallmark finding
  - Large, thin-walled bladder
  - Bilateral hydroureter
  - Bilateral hydronephrosis
- Variable oligohydramnios
- Difficult to differentiate from posterior urethral valves
  - No "keyhole" bladder with prune belly
  - Entire urethra may be dilated

Other Essential Information

• Worsening oligohydramnios ⇒ worse prognosis
- Associated with fetal acidosis and death
- Further fetal assessment indicated
  - Fetal growth and Doppler
  - Nonstress test
  - Biophysical profile
  - Reassess fetal anatomy carefully

• Biophysical profile fluid assessment
- Score of 0 or 2 for fluid
  - 2 = at least 1 pocket of fluid measures ≥ 2 x 2 cm
  - 0 = no fluid pocket measures > 2 x 2 cm
- BPP not same as amniotic fluid index
  - Can have BPP score of 2 for fluid and still have oligohydramnios

• Pay close attention to contralateral kidney when renal anomaly seen
- Bilateral/contralateral anomalies common
- Unilateral renal anomaly usually not associated with oligohydramnios

Preterm Premature Rupture of Membranes (PPROM)



Axial ultrasound of a 22 week pregnancy shows severe oligohydramnios. The presence of fluid in the fetal bladder ➡ rules out renal agenesis. PPROM was suspected clinically.

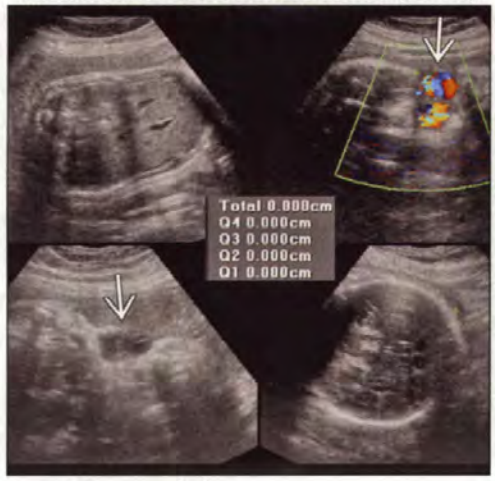
Preterm Premature Rupture of Membranes (PPROM)



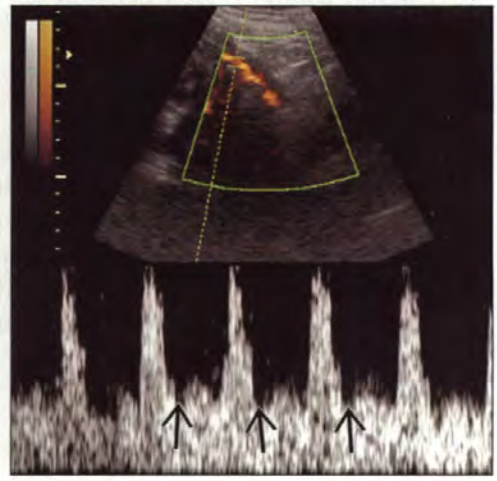
Axial ultrasound of a pregnancy with PPROM after amniocentesis shows the amnion ➡ displaced away from the uterine wall. The presence of fluid on both sides of the amnion is from a defect in the membrane.

OLIGOHYDRAMNIOS

Intrauterine Growth Restriction (IUGR)

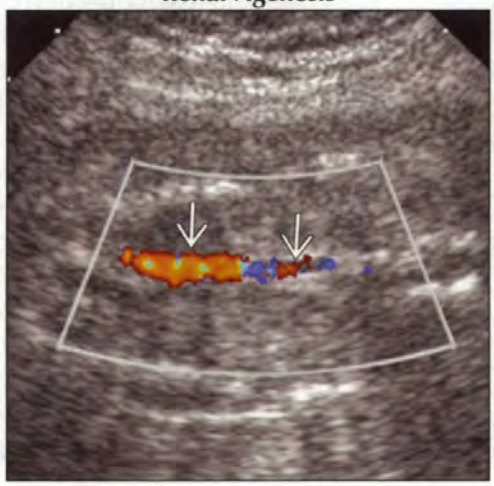


Intrauterine Growth Restriction (IUGR)



(Left) *Four quadrant US shows no measurable pockets of fluid in a pregnancy complicated by hypertension and IUGR. All the hypoechoic "pockets" contain umbilical cord* ➡. ***(Right)*** *Sagittal pulsed Doppler ultrasound of the left uterine artery in the same case shows high resistive flow and a post-systolic notch* ➡. *This is abnormal in the third trimester and is highly suggestive of placental insufficiency. The 28 week fetus was delivered immediately.*

Renal Agenesis

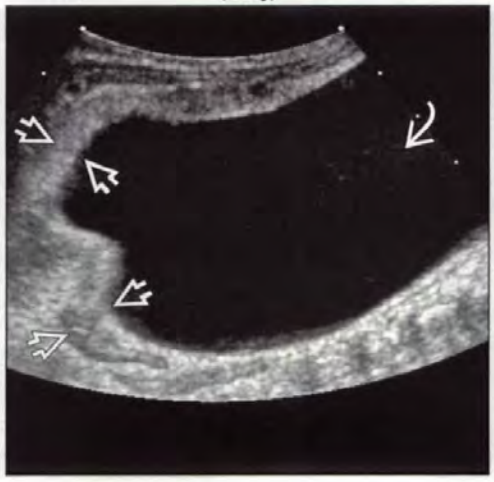


Renal Agenesis

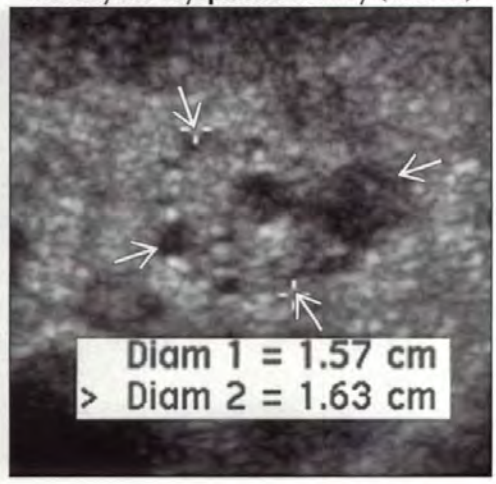


(Left) *Coronal color Doppler ultrasound of the aorta* ➡ *shows absent renal arteries. There was no amniotic fluid (anhydramnios), and the bladder was empty.* ***(Right)*** *Axial ultrasound through the fetal abdomen in a different case shows bowel* ➡ *in the renal fossa. Fetal bowel and adrenal glands can mimic kidneys.*

Ureteropelvic Junction Obstruction (UPJ)



Multicystic Dysplastic Kidney (MCDK)



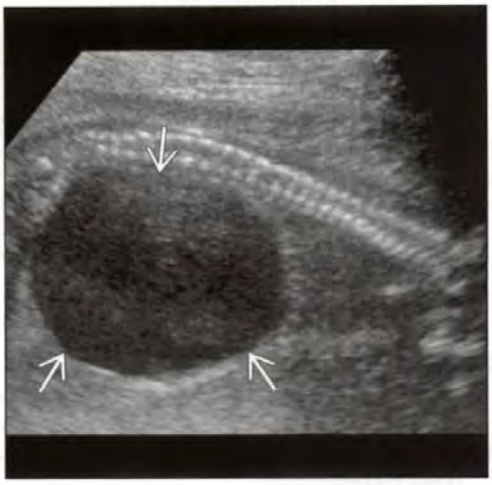
(Left) *Axial ultrasound shows a massively distended renal pelvis* ➡ *and compressed renal tissue* ➡. *There was no amniotic fluid surrounding the fetus, and therefore a contralateral renal anomaly was suspected.* ***(Right)*** *Axial ultrasound in the same fetus shows that the contralateral kidney is small and cystic* ➡. *In this case, both kidneys lost their normal reniform shape. The neonate had severe pulmonary hypoplasia and died.*

OLIGOHYDRAMNIOS

Autosomal Recessive Polycystic Kidney Disease



Posterior Urethral Valves



(Left) *Coronal ultrasound shows bilateral enlarged echogenic kidneys and a small bell-shaped chest. There is no amniotic fluid in this case, but ARPKD can have variable degrees of oligohydramnios depending on the severity of renal disease.* ***(Right)*** *Sagittal ultrasound of a fetus with posterior urethral valves shows a massively distended fetal bladder and severe oligohydramnios. Posterior urethral valves can be partially or completely obstructive.*

Twin-Twin Transfusion Syndrome

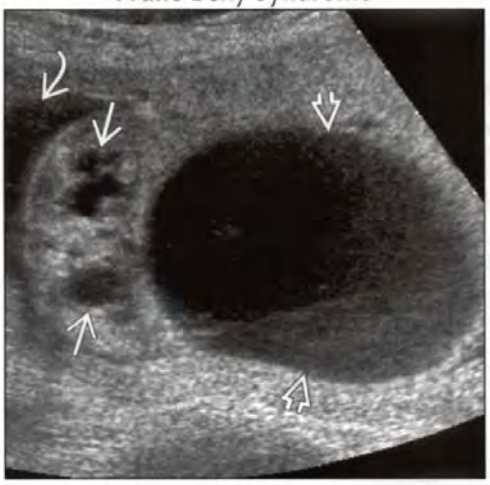


Twin-Twin Transfusion Syndrome



(Left) *Axial ultrasound shows a "stuck" twin. A very small pocket of fluid surrounds the fetus, and the membranes entrap it along the uterine wall. The other twin is surrounded by excessive fluid.* ***(Right)*** *Axial ultrasound of another pregnancy shows a thin separating membrane with a small pocket of fluid around the small, donor twin. There is polyhydramnios in the sac of the the larger, recipient twin.*

Prune Belly Syndrome



Prune Belly Syndrome



(Left) *Axial ultrasound shows severe bladder distention and bilateral hydronephrosis. Although decreased, amniotic fluid is present. Since bladder outlet obstruction was suspected, vesicocentesis was performed in this case.* ***(Right)*** *Axial ultrasound of the same fetus after vesicocentesis shows a markedly redundant and lax abdominal wall, more consistent with the diagnosis of prune belly syndrome than bladder outlet obstruction.*

POLYHYDRAMNIOS

DIFFERENTIAL DIAGNOSIS

Common
- Idiopathic
- Macrosomia
- Diabetes
- Hydrops
- Twin-Twin Transfusion Syndrome
- Fetal Bowel Obstruction

Less Common
- Arthrogryposis, Akinesia Sequence
- Skeletal Dysplasia, Severe
- Chorioangioma
- Cystic Adenomatoid Malformation

Rare but Important
- Mesoblastic Nephroma
- Unilateral Ureteropelvic Junction Obstruction

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Amniotic fluid index (AFI)
  - Divide uterus into 4 equal quadrants
  - Measure maximum vertical pocket (MVP)
    - Avoid fetal parts/cord in MVP
  - Add the 4 MVPs to calculate AFI
  - AFI/gestational age normogram tables available
    - > 24 cm AFI = polyhydramnios
- Twin fluid assessment
  - Measure MVP for each fetus
  - ≥ 8 cm = polyhydramnios

Helpful Clues for Common Diagnoses
- **Idiopathic**
  - Most common cause (2/3)
    - Mild, stable finding
  - Normal patient and fetus
    - No diabetes or fetal anomalies
  - 28% with fetal macrosomia
- **Macrosomia**
  - Estimated fetal weight > 90th percentile
  - Large abdominal circumference is a hallmark finding
    - ↑ Truncal echogenic fat
  - Causes
    - Hereditary characteristics
    - Maternal obesity
    - Diabetes
    - Beckwith-Wiedemann syndrome
- **Diabetes**
  - Gestational diabetes (most common)
    - Glucose intolerance during pregnancy
    - Oral glucose tolerance test given between 24-28 wks
  - Pregestational diabetes (type 1 or 2)
  - Polyhydramnios associated with poor glucose control
    - May be first finding in pregnancy
    - Associated with macrosomia
- **Hydrops**
  - Excessive fetal fluid accumulation (2 or more body cavities)
    - Anasarca
    - Pleural effusion (± pericardial)
    - Ascites
  - Amniotic fluid may be ↑ or ↓
  - Immune hydrops (10%)
    - Hemolytic disease ⇒ fetal anemia
  - Non-immune (90%)
    - Infection
    - Any cause of fetal anemia
    - Any cause of fetal heart failure
    - Lymphatic obstruction
  - Aneuploidy associations
    - Turner syndrome (cystic hygroma)
    - Trisomy 21
    - Trisomy 18
- **Twin-Twin Transfusion Syndrome**
  - Monochorionic twinning
    - Artery-to-vein anastomoses in shared placenta
    - Donor twin partly perfuses recipient twin
  - Fluid discrepancy may be 1st sign of developing twin-twin transfusion syndrome (need to follow carefully)
  - Recipient twin with polyhydramnios
    - Larger twin
  - Donor twin with oligohydramnios
    - Smaller "stuck" twin
- **Fetal Bowel Obstruction**
  - Late polyhydramnios (> 24 wks)
  - Esophageal atresia
    - Absent or small stomach
    - 1/3 with trisomy 18 or 21
    - Associated with VACTERL syndrome
  - Duodenal atresia
    - "Double-bubble" appearance
    - 1/3 with trisomy 21
  - Jejunal, ileal atresia
    - "Sausage-shaped" bowel loops

POLYHYDRAMNIOS

Helpful Clues for Less Common Diagnoses

- **Arthrogryposis, Akinesia Sequence**
  - Heterogeneous group of disorders
    - Lack of extremity motion
    - Contractures
  - Polyhydramnios from ↓ fetal movement/swallowing
  - Associations
    - Trisomy 18
    - Autosomal dominant and recessive syndromes
- **Skeletal Dysplasia, Severe**
  - Common dysplasias
    - Thanatophoric
    - Achondroplasia
    - Achondrogenesis
    - Osteogenesis imperfecta
  - Common skeletal findings
    - Short limbs
    - Poor ossification
    - Bowed or broken bones
    - Craniosynostosis
  - Polyhydramnios common in 3rd trimester
- **Chorioangioma**
  - Benign, vascular placental tumor
  - Large masses (> 5 cm): ↑ Complications
  - Polyhydramnios
    - Transudate from leaky vessels
  - Hydrops
    - Arteriovenous shunting in mass
    - Fetal anemia from hemolysis
- **Cystic Adenomatoid Malformation**
  - Lung hamartoma
  - Microcystic and macrocystic types
  - Polyhydramnios etiology
    - Esophagus compression
    - Associated with hydrops
    - Mass may make fluid

Helpful Clues for Rare Diagnoses

- **Mesoblastic Nephroma**
  - Benign solid mesenchymal tumor
  - 70% with polyhydramnios
    - Often progressive and severe
  - Proposed etiologies for polyhydramnios
    - Hypercalcemia ⇒ polyuria
    - Renal hyperemia ⇒ ↑ urine output
    - Bowel obstruction (large mass)
- **Unilateral Ureteropelvic Junction Obstruction**
  - Dilated renal pelvis is hallmark finding
    - Distention ends abruptly at UPJ
    - Normal ureters and bladder
  - Polyhydramnios in 1/3 of cases
    - Apparent paradox that obstruction would lead to polyhydramnios but etiology thought to be from impaired renal concentrating ability which causes ↑ urine output
  - Contralateral renal anomaly in 25%
    - ↓ Fluid if severe

Other Essential Information

- Amniocentesis for polyhydramnios
  - Not indicated if idiopathic etiology
  - Polyhydramnios + growth restriction has ↑ risk for aneuploidy/syndromes
    - Trisomy 18 most common

Macrosomia



Axial ultrasound shows moderate polyhydramnios. The four quadrant fluid measurement is 38.9 cm. Fetal anatomic survey was normal, however the fetus was large for gestational age (macrosomia).

Diabetes



Axial ultrasound shows polyhydramnios in a pregnancy complicated by uncontrolled diabetes. There was also fetal macrosomia (excessive subcutaneous truncal fat).

POLYHYDRAMNIOS

Hydrops





(Left) *Sagittal ultrasound of a pregnancy complicated by maternal Rh sensitization shows features of hydrops fetalis. There is polyhydramnios and skin edema, particularly involving the scalp. Pleural effusions are also present.* ***(Right)*** *Axial ultrasound through the fetal abdomen and placenta in the same case shows placentomegaly, anasarca, and ascites. Polyhydramnios is more often associated with immune hydrops than non-immune.*

Twin-Twin Transfusion Syndrome





(Left) *Axial ultrasound shows a large amount of fluid surrounding fetus A (calipers) and a barely visible thin separating membrane between the monochorionic twins.* ***(Right)*** *Another image of the same case shows calipers measuring the largest pocket of fluid surrounding twin B. The severe polyhydramnios associated with twin A is again seen. Twin B is the "stuck" donor twin with oligohydramnios. Twin A is the recipient twin with polyhydramnios.*

Fetal Bowel Obstruction





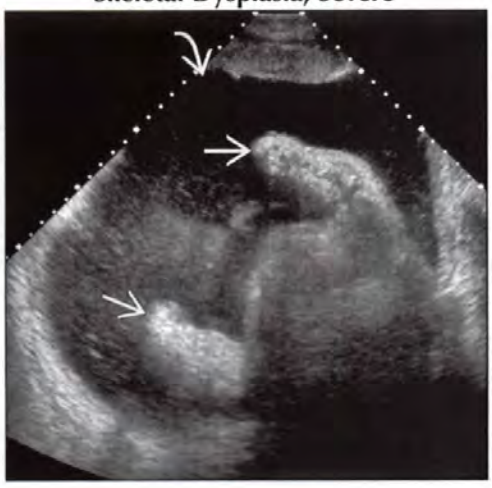
(Left) *Axial ultrasound of the abdomen in a fetus with esophageal atresia shows polyhydramnios and lack of a fluid-filled fetal stomach (fetal spine).* ***(Right)*** *Axial ultrasound of the abdomen in a different fetus shows a "double bubble", the classic appearance of duodenal atresia. There is associated polyhydramnios. Both esophageal atresia and duodenal atresia are associated with aneuploidy.*

POLYHYDRAMNIOS

Arthrogryposis, Akinesia Sequence

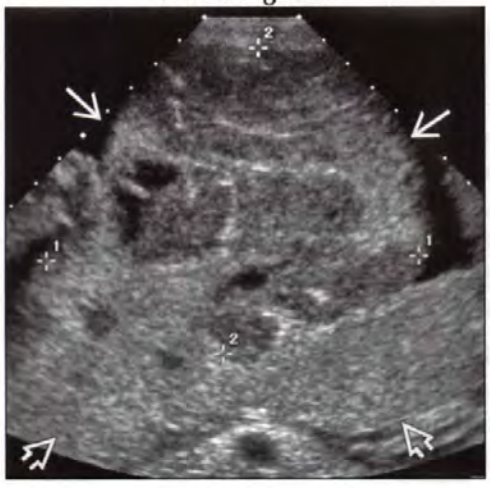


Skeletal Dysplasia, Severe



(Left) *Sagittal ultrasound shows severe polyhydramnios and a gravity dependent fetus. The fetus lay motionless in the posterior uterus throughout the exam.* ***(Right)*** *Axial ultrasound of a fetus with thanatophoric dysplasia type II shows polyhydramnios and very short "flipper-like" upper extremities. All the long bones were severely shortened and the skull had a "clover-leaf" morphology.*

Chorioangioma

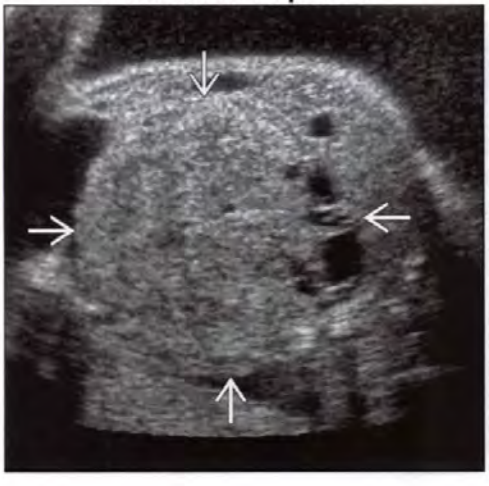


Cystic Adenomatoid Malformation



(Left) *Axial ultrasound shows a large chorioangioma extending from the anterior surface of the placenta. Although rare, large chorioangiomas are associated with polyhydramnios and hydrops fetalis.* ***(Right)*** *Axial ultrasound shows a unilocular medial cystic chest mass, which is causing mass effect upon the aorta. Polyhydramnios can occur from esophageal compression or hydrops.*

Mesoblastic Nephroma



Unilateral Ureteropelvic Junction Obstruction



(Left) *Axial ultrasound shows a large, predominantly solid, renal mass. The contralateral kidney and bladder were normal. There was severe polyhydramnios necessitating therapeutic amniocentesis.* ***(Right)*** *Axial ultrasound shows a massively distended renal pelvis. The pregnancy was also complicated by mild polyhydramnios. Unilateral renal anomalies are sometimes associated with polyhydramnios secondary to impaired renal concentrating ability.*

ECHOGENIC AMNIOTIC FLUID

DIFFERENTIAL DIAGNOSIS

Common
- Normal Variant, Third Trimester
- Placental Abruption
- Bowel Anomalies
  - Jejunal, Ileal Atresia
  - Gastroschisis

Less Common
- Exencephaly, Anencephaly
- Umbilical Vessel Disruption

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Echoes in amniotic fluid may be normal, particularly with high resolution transducers
- Look for anomalies, especially of gastrointestinal tract
- Ask about vaginal bleeding
  - 50% of abruption cases have no sonographic findings acutely
- Ask about previous procedures
  - Patient may be asymptomatic despite intraamniotic, subchorionic bleeding secondary to intrauterine procedure

Helpful Clues for Common Diagnoses
- **Normal Variant, 3rd Trimester**
  - Vernix may layer over cervix
- **Placental Abruption**
  - Initial subchorionic bleed → small breach in membranes → blood mixes with amniotic fluid → diffuse ↑ echogenicity
    - May see clot adherent to cord or fetus
  - Clot decreases in echogenicity as it ages → linear echo from chorion with amniotic fluid on one side, old hypoechoic clot on other side
- **Jejunal, Ileal Atresia**
  - Fetal regurgitation secondary to small bowel obstruction
  - Multiple dilated loops of bowel within abdomen
- **Gastroschisis**
  - Non membrane-bound abdominal wall defect
  - Cord inserted adjacent to defect
  - Bowel loops float freely in amniotic fluid
  - Meconium stained fluid typical at delivery of gastroschisis infants

Helpful Clues for Less Common Diagnoses
- **Exencephaly, Anencephaly**
  - Skull vault bones absent
  - Proptotic orbits create "frog eye" appearance
  - Trauma to exposed brain results in fragmentation
  - Look for linear amniotic bands as etiology
- **Umbilical Vessel Disruption**
  - Umbilical vessel bleeds into amniotic fluid
  - Risk factors include intrauterine procedures, rupture of varix in free loops

Other Essential Information
- Check fetal cardiac activity and look for bleeding at the end of every invasive procedure

Normal Variant, Third Trimester



Coronal ultrasound shows diffuse low-level echoes in the amniotic fluid around the nose and lips of this normal third trimester fetus.

Placental Abruption



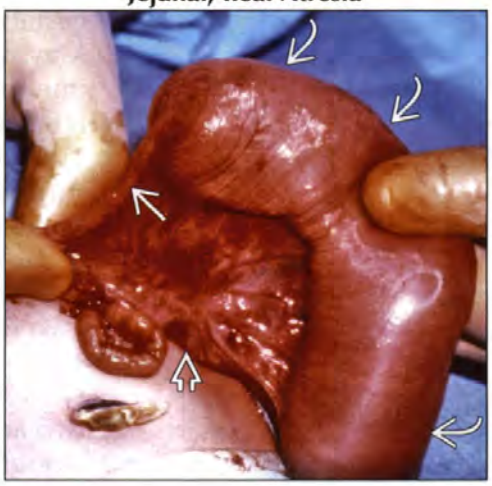
Ultrasound shows hypoechoic blood adjacent to the placenta lifting the membranes off the uterine wall to create linear echoes in the fluid, as well as creating intermediate echoes in the subacute clot.

ECHOGENIC AMNIOTIC FLUID

Jejunal, Ileal Atresia



Jejunal, Ileal Atresia



(Left) *Axial ultrasound shows polyhydramnios (calipers) with echogenic debris in the amniotic fluid. Multiple loops of distended fluid-filled bowel were seen as well as a "triple bubble sign" created by distended stomach, duodenum and jejunum.* ***(Right)*** *Intra-operative photograph in a similar infant shows the very dilated proximal jejunum ➔ terminating at a fibrous cord ➔. The mesentery ➔ is intact.*

Gastroschisis



Gastroschisis



(Left) *Ultrasound shows multiple mildly dilated, thick-walled loops of externalized bowel in a fetus with gastroschisis.* ***(Right)*** *Another image in the same fetus shows marked increase in echogenicity of the amniotic fluid with a lot of suspended particulate material. The fetus had intrauterine growth restriction and poor biophysical scores, which precipitated early delivery. There was severe meconium staining of the amniotic fluid at birth.*

Exencephaly, Anencephaly



Umbilical Vessel Disruption



(Left) *Ultrasound in a fetus with exencephaly due to amniotic band syndrome shows amorphous residual brain tissue ➔ above the orbital ridge ➔. The fluid is echogenic & there is layering debris ➔ within the amniotic cavity.* ***(Right)*** *Ultrasound shows new development of highly echogenic fluid ➔ & polyhydramnios. Biophysical score was zero. The infant was delivered alive by emergency c-section & was found to have a ruptured UV varix in free loops of cord.*

LINEAR ECHOES IN AMNIOTIC FLUID

DIFFERENTIAL DIAGNOSIS

Common
- Synechiae
- Dichorionic Diamniotic Twins
- Monochorionic Diamniotic Twins
- Chorioamniotic Separation
- Placental Abruption, Old
- Uterine Septum
- Placental Cysts

Less Common
- Amniotic Band Syndrome
- Circumvallate Placenta

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Single or multiple gestation?
  - If multiple, inter-twin membrane is most likely cause of a linear echo in the amniotic fluid
- Does linear echo cross cavity from side to side?
  - Placental edge to placental edge → circumvallate placenta
  - Uterine wall to uterine wall → synechia
- Does linear echo parallel wall of uterine cavity?
- How do linear echoes relate to placenta?
- How do linear echoes relate to fetus?
- Is there a history of intervention?
  - Prior uterine instrumentation
    - D&C, myomectomy, metroplasty
  - Procedure performed during current pregnancy?
    - Amniocentesis, amnioreduction, intrauterine transfusion, laser therapy or radiofrequency ablation

Helpful Clues for Common Diagnoses
- **Synechiae**
  - Shelf or band-like structure which does not restrict fetal movement
  - Extra-amniotic: Fetal membranes wrap over synechiae
  - Straight, bulbous free edge with thinner sheet extending to endometrial surface
  - Y-shaped notch at endometrial base, created by membranes separating at endometrial margin
  - Placenta can abut or even wrap around synechia
  - Color Doppler may demonstrate flow within synechiae
  - In first trimester may cause distortion of gestational sac shape
  - May no longer be visible in 3rd trimester due to rupture or compression
- **Dichorionic Diamniotic Twins**
  - Thick echogenic chorion completely surrounds each embryo
  - "Twin peak" sign: Wedge of chorionic tissue extending into base of inter-twin membrane
  - Two fetuses in separate chorionic sacs
    - Two amniotic sacs with two yolk sacs
  - Dichorionic membrane actually separates two amniotic cavities (i.e., extra-amniotic)
- **Monochorionic Diamniotic Twins**
  - Two fetuses in single chorionic sac containing two amniotic sacs
  - Thin inter-twin membrane formed by two layers of amnion without interposed chorion
  - No "twin peak"
  - Twins must be same gender
- **Chorioamniotic Separation**
  - Persistent unfused amnion and chorion > 16 wks
  - Amniotic membrane separate from uterine wall
    - Complete: Attached only at placental cord insertion site
    - Incomplete: Unattached around part of the uterine cavity, the commonest form
  - May be primary non-fusion
    - Look for signs of aneuploidy
  - May occur secondary to amniocentesis or fetal intervention
    - Increases risk of membrane rupture in twins → functional monoamniotic state → ↑ risk of cord entanglement
- **Placental Abruption, Old**
  - Hypoechoic blood clot near or behind placenta
  - Marginal (most common), retroplacental or preplacental
    - Marginal: Bleed at edge of placental disc, dissects between chorion and uterine wall

- Retroplacental does not cause confusion for linear echoes in amniotic fluid as located between placenta and myometrium
- Preplacental abruption is rare
  - Hematoma on fetal surface of placenta
  - Clot may compress cord if close to insertion site
- Subacute
  - May contain fluid-fluid level, septations common
- Old
  - Liquefying blood, eventually sonolucent and may mimic amniotic fluid
- Hemorrhage can dissect under chorionic membrane
  - Clot seen at a distance from placenta
  - Look in front of cervical os
- Intraamniotic blood common → echogenic fluid → echogenic fetal bowel from swallowed blood
- In twins, rarely, hematoma dissects between membranes
  - When old may appear as fluid-filled mass between membranes → increased linear interfaces in amniotic fluid

- **Uterine Septum**
  - Midline, arising from fundus
    - In first trimester use 3D to create coronal images and confirm location, assess fundal contour
  - May be fibrous or composed of myometrium
  - Thicker than synechiae
  - Creates two distinct endometrial cavities
- **Placental Cysts**
  - Chorionic cysts are simple cysts on fetal placental surface
  - Often near cord insertion site
  - If multiple may appear to create linear echoes in amniotic fluid
  - Curvilinear shape and relationship to placenta indicate etiology
  - If large, or if hemorrhage occurs, may compress cord

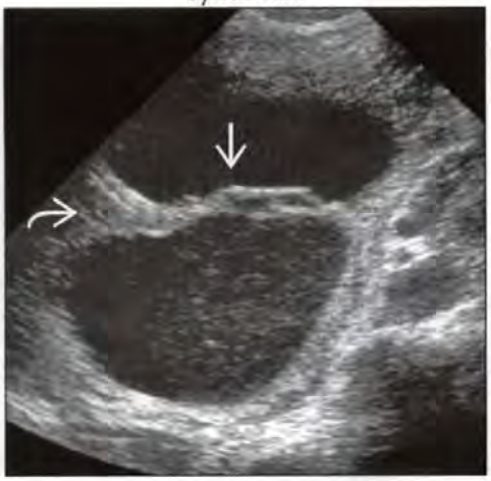
Helpful Clues for Less Common Diagnoses

- **Amniotic Band Syndrome**
  - Entrapment of fetal parts by disrupted amnion
  - Amniotic band in contact with deformity, extends to uterine wall
  - Bands in amniotic fluid appear as multiple thin membranes
  - No flow in band on Doppler evaluation
- **Circumvallate Placenta**
  - Placental margin elevated off uterine wall
  - Scanning parallel to edge → "marginal shelf"
  - Scanning longitudinally → "curled lip" of placental margin

Other Essential Information

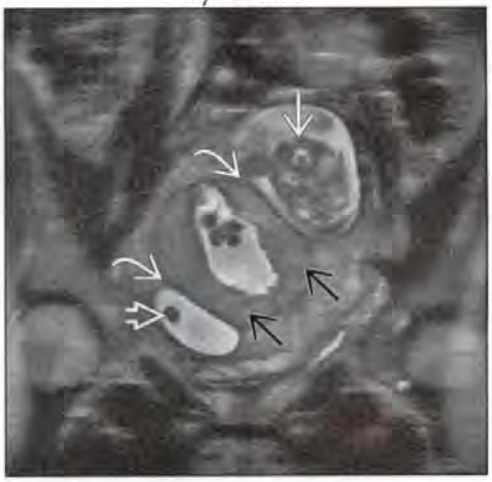
- Most linear echoes in amniotic fluid are of little clinical significance
- Amniotic band syndrome can be lethal depending on extent of band-related damage

Synechiae



Ultrasound shows linear echoes ➡ crossing the amniotic cavity due to synechia. Note Y-shaped base ➡. The patient had a history of multiple D&C procedures for recurrent abortions.

Synechiae



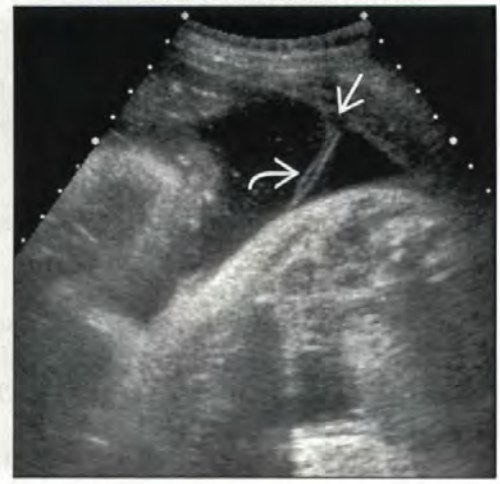
Coronal T2WI MR shows a midtrimester pregnancy in a patient with synechiae ➡. Fetal parts ➡ and cord ➡ are seen in various "compartments" and the placenta ➡ is partially implanted on the synechiae.

LINEAR ECHOES IN AMNIOTIC FLUID

Dichorionic Diamniotic Twins

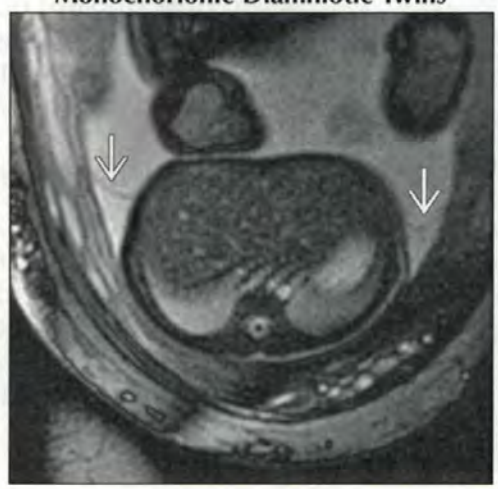


Dichorionic Diamniotic Twins

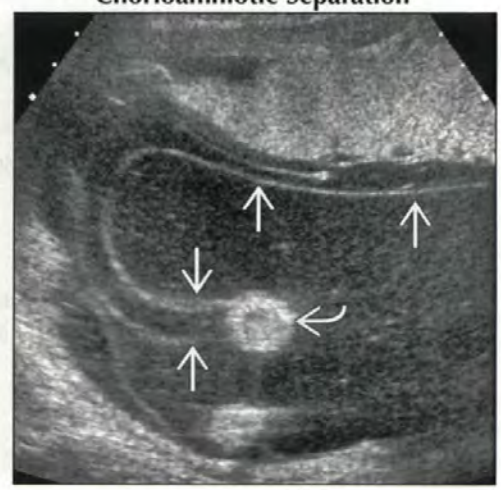


(Left) *Ultrasound shows the thick inter-twin membrane ➡ of a dichorionic twin pregnancy in the first trimester. The amniotic cavities ➡ are completely separate.* ***(Right)*** *Ultrasound shows how the membrane ➡ becomes thinner with advancing gestational age. These twins are dizygotic but the membranes look thin and there is no "twin peak" ➡. The presence of two fetuses makes inter-twin membrane the most likely cause for a linear structure in the amniotic fluid.*

Monochorionic Diamniotic Twins

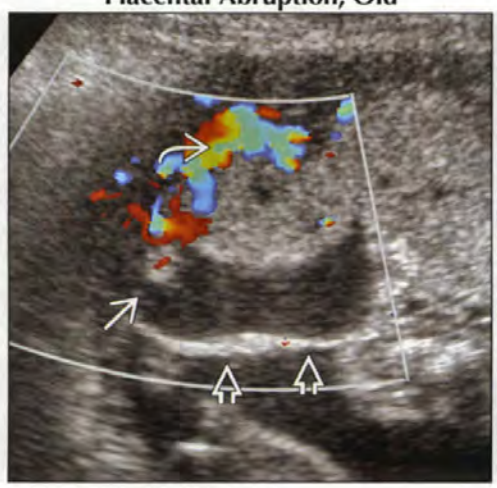


Chorioamniotic Separation

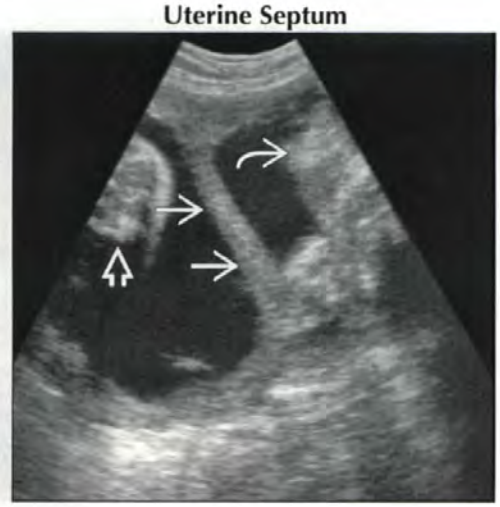


(Left) *T2WI MR shows the thin membrane ➡ in monochorionic diamniotic twins.* ***(Right)*** *Ultrasound shows chorioamniotic separation creating linear echoes ➡ in the amniotic fluid. This was a complication of serial amnioreduction in a case of twin-twin transfusion syndrome. Although the membranes appeared tangled around the fetal extremities ➡ there were no constriction defects at delivery.*

Placental Abruption, Old



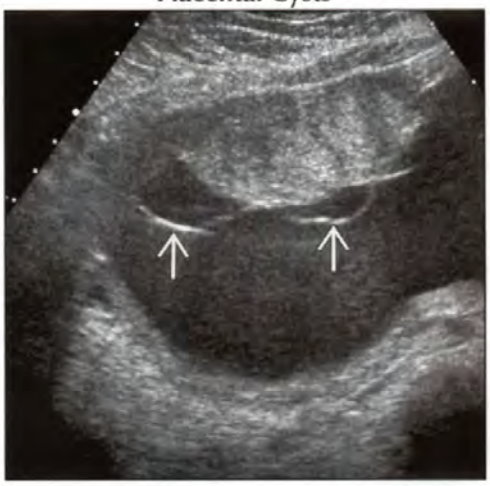
Uterine Septum



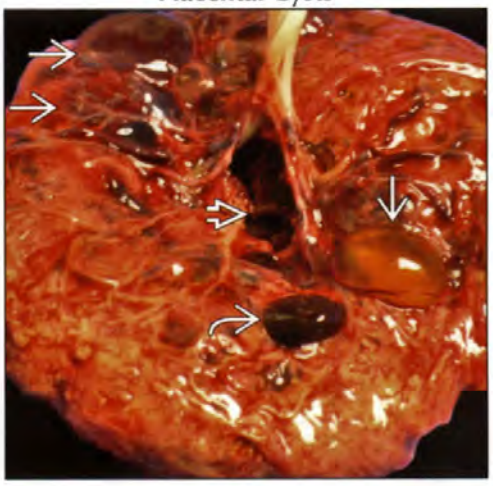
(Left) *Color Doppler ultrasound shows myometrial vascularity ➡ without active flow into the subacute abruption ➡. Elevation of the membranes ➡ produces a linear interface in the amniotic fluid.* ***(Right)*** *Ultrasound shows a uterine septum ➡ creating two "compartments" in the uterus. The placenta ➡ is seen on one side of the septum and the fetus ➡ on the other. The fetus was freely mobile.*

LINEAR ECHOES IN AMNIOTIC FLUID

Placental Cysts



Placental Cysts



(Left) *Ultrasound shows two of multiple curvilinear echoes ➡ in the amniotic fluid. These are all arising from the placental surface.* ***(Right)*** *Gross pathology shows the placenta in the same case. There are multiple chorionic cysts ➡, some of which are hemorrhagic ➡. Note the central area of infarction ➡ at the placental cord insertion site. Delivery was precipitated by abnormal biophysical profile & reversed end diastolic flow in the umbilical artery.*

Amniotic Band Syndrome



Amniotic Band Syndrome

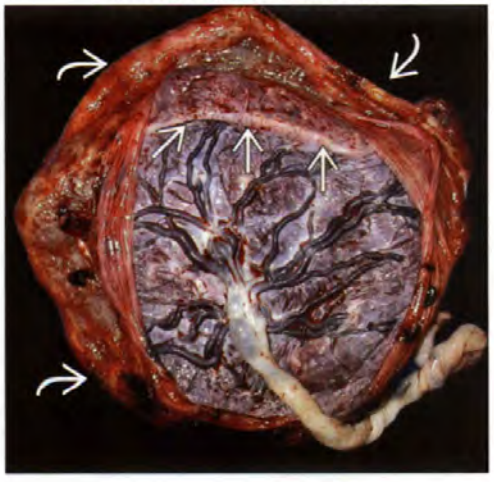


(Left) *Ultrasound shows abdominoschisis secondary to amniotic bands ➡. Liver ➡ and loops of bowel ➡ are outside the abdominal cavity.* ***(Right)*** *Clinical photograph in the same case shows a fine linear band ➡ wrapped around the fetal arm. Isolated constriction defects are not lethal, but extensive abdominoschisis and exencephaly are. The bands themselves create fine linear echoes within the amniotic fluid.*

Circumvallate Placenta



Circumvallate Placenta



(Left) *Ultrasound shows the placental "shelf" ➡ as the edge of the placenta is lifted off the uterine wall ➡. The "shelf" looks like a band on longitudinal views along the edge of the placenta. The band connects to the placenta not the uterine wall, differentiating it from a synechia.* ***(Right)*** *Gross pathology shows how the membranes attach to the fetal surface of the placenta ➡, instead of the edge ➡. This curled edge creates a "shelf-like" linear echo.*

SECTION 11
Growth and Well Being

Intrauterine Growth Restriction	11-2
Macrosomia	11-8
Hydrops	11-10
Fetal Anemia	11-16
Abnormal Fetal Presentation	11-20

INTRAUTERINE GROWTH RESTRICTION

DIFFERENTIAL DIAGNOSIS

Common
- Placental Insufficiency

Less Common
- Chromosome Abnormality
  - Trisomy 18 (T18)
  - Trisomy 13 (T13)
  - Triploidy
- Twin-Twin Transfusion Syndrome
- Isolated Anomalies with IUGR
  - Gastroschisis
  - Single Umbilical Artery

Rare but Important
- Infection

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Intrauterine growth restriction (IUGR) defined as estimated fetal weight (EFW) < 10th percentile for gestational age (GA)
  - Accurate GA essential for diagnosis
- IUGR vs. small for gestational age (SGA)
  - IUGR: Fetus not reached growth potential
  - SGA: Fetus is small but normally grown
  - Difficult to differentiate prenatally
    - Look at parents and siblings
- Symmetric vs. asymmetric IUGR
  - Symmetric: All biometry equally affected
    - Often early and severe IUGR
    - Suggests fetal problem
    - Possible early placental dysfunction
  - Asymmetric: "Head sparing" with abdomen, extremities more severely affected
    - Often presents later in pregnancy
    - Suggests placental cause
    - Better prognosis if not severe
- Early IUGR vs. late IUGR
  - Early IUGR more likely fetal cause
    - Look for anomalies
    - Consider amniocentesis
  - Late IUGR more likely placental cause
- IUGR differential diagnosis approach
  - Rule out fetal anomaly as cause for IUGR
    - Amniocentesis if fetal anomaly suspected
  - Consider maternal medical history
  - Assess amniotic fluid
  - Assess fetal/placental circulation
    - Doppler
    - Biophysical profile (BPP)

Helpful Clues for Common Diagnoses
- **Placental Insufficiency**
  - Maternal causes
    - Hypertension (acute or chronic)
    - Uncontrolled diabetes mellitus
    - Thrombophilia
    - Collagen vascular disease
    - Drugs/alcohol/smoking
    - Malnutrition
  - Uterine-placental causes
    - Chronic abruption
    - Infarction
    - Confined placental mosaicism
    - Marginal or velamentous cord insertion
  - Doppler findings
    - ↑ Uterine artery (UtA) resistance with post-systolic notch
    - ↑ Umbilical artery (UA) resistance
    - ↑ Ductus venosus (DV) resistance
    - ↓ Middle cerebral artery (MCA) resistance
  - Findings in addition to IUGR
    - Oligohydramnios
    - Placental sonolucencies
    - Poor BPP score
  - Management/treatment
    - Manage maternal condition
    - Increased surveillance
    - Abnormal Doppler, fluid, BPP in 3rd trimester ⇒ consider delivery

Helpful Clues for Less Common Diagnoses
- **Trisomy 18 (T18)**
  - IUGR in 51% (rarely isolated)
    - Early onset, symmetric IUGR
  - Anomalies associated with T18
    - Cardiac defects
    - Dandy-Walker continuum
    - Spina bifida
    - Omphalocele
    - Clenched hands + overlapping index finger, rockerbottom feet
  - Markers associated with T18
    - Choroid plexus cyst
    - Single umbilical artery
    - Umbilical cord cyst
    - Nuchal thickening
- **Trisomy 13 (T13)**
  - IUGR in 50% (rarely isolated)
    - Early onset, with microcephaly
  - Anomalies associated with T13
    - Holoprosencephaly, microcephaly

- Hypotelorism, cyclopia, proboscis
- Dandy-Walker continuum
- Polydactyly
- Cardiac defects
- Gastrointestinal anomalies

- Markers associated with T13
  - Echogenic cardiac focus
  - Single umbilical artery
  - Nuchal thickening

- **Triploidy**
  - 69 chromosomes (extra haploid set)
    - Maternal or paternal extra set
  - Early severe IUGR is hallmark finding
    - Asymmetric if maternal extra set
  - Variable placenta findings according to source of extra set
    - Thick and cystic (paternal)
    - Small or normal (maternal)
  - Ovarian theca lutein cysts
  - Fetal anomalies often severe but difficult to completely characterize prenatally
    - Small fetus
    - Oligohydramnios
    - Thick cystic placenta displaces fetus
- **Twin-Twin Transfusion Syndrome**
  - Monochorionic twinning with artery-to-vein anastomoses in placenta
    - Donor twin partly perfuses recipient twin
  - Donor twin with IUGR
    - Oligohydramnios
    - Abnormal Doppler
- **Gastroschisis**
  - Bowel herniation through right paramedial abdominal wall defect
  - 50% develop IUGR
    - Often leads to early delivery
  - Bowel complications may develop during pregnancy
    - Dilatation, ischemia, rupture
- **Single Umbilical Artery**
  - 15% of fetuses with an isolated single umbilical artery (SUA) have IUGR
    - Follow-up for growth into 3rd trimester
  - Non-isolated SUA
    - 50% aneuploidy rate
    - T18 most common

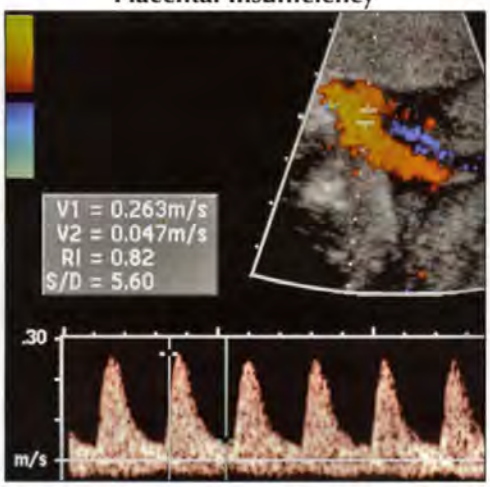
Helpful Clues for Rare Diagnoses

- **Infection**
  - IUGR and hydrops are early findings
  - Common infections: Parvovirus, cytomegalovirus, toxoplasmosis, varicella
  - Other findings
    - Echogenic bowel
    - Brain, liver, spleen calcifications

Other Essential Information

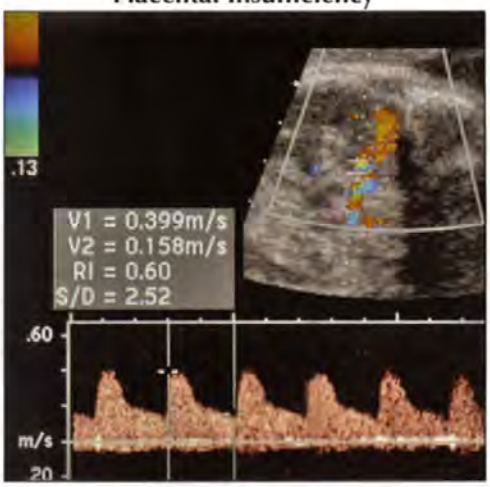
- Late presentation case: Is fetus small or are dates wrong?
  - Look for lower extremity ossification centers to verify dating
    - Distal femoral epiphyseal ossification ≥ 32 weeks
    - Proximal tibial epiphyseal ossification ≥ 35 weeks
  - Look at fluid and Doppler values
- IUGR + polyhydramnios ⇒ bad prognosis
  - Associated with aneuploidy, syndromes
  - Amniocentesis warranted

Placental Insufficiency



Pulsed Doppler ultrasound of the umbilical artery shows elevated UA resistance in a fetus with third trimester IUGR and oligohydramnios. The systolic/diastolic ratio (S/D) is 5.6 and should be < 3.0.

Placental Insufficiency

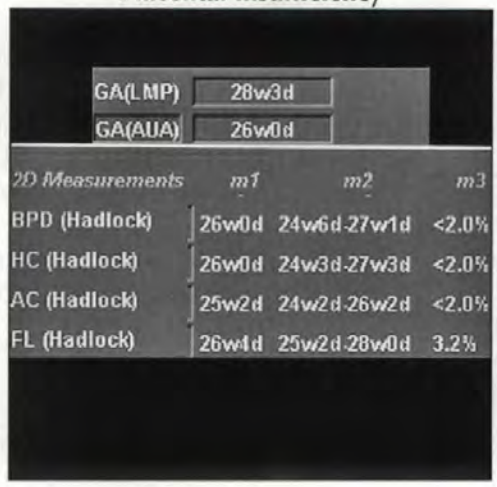


Axial pulsed Doppler ultrasound of the MCA in the same fetus shows an S/D ratio of 2.5. MCA resistance is normally higher than the UA, and the finding suggests "head sparing" compensatory physiology.

INTRAUTERINE GROWTH RESTRICTION

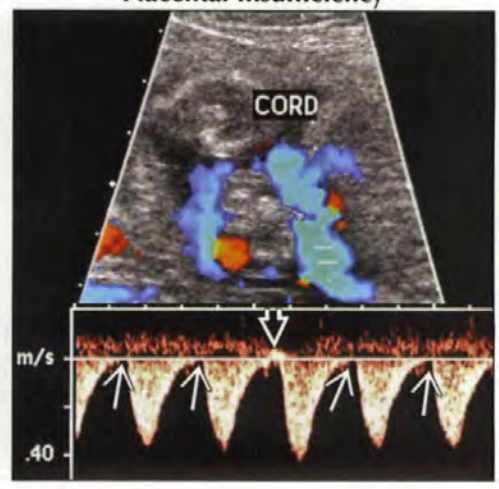
(Left) *Table of a 28 week 3 day fetus with late care shows IUGR, but only if the LMP dates are accurate. Oligohydramnios was also present, and Doppler assessment was undertaken to assess for IUGR.* ***(Right)*** *Pulsed Doppler ultrasound of the umbilical artery, in the same case, shows absent diastolic flow ➡ and occasional reversal of diastolic flow ➡, suggesting markedly increased placental resistance. This finding is highly suggestive of placental insufficiency and IUGR.*

Placental Insufficiency



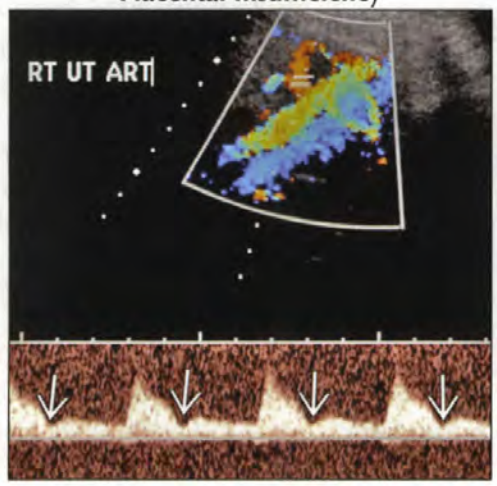
2D Measurements	m1	m2	m3
BPD (Hadlock)	26w0d	24w6d-27w1d	<2.0%
HC (Hadlock)	26w0d	24w3d-27w3d	<2.0%
AC (Hadlock)	25w2d	24w2d-26w2d	<2.0%
FL (Hadlock)	26w1d	25w2d-28w0d	3.2%

Placental Insufficiency

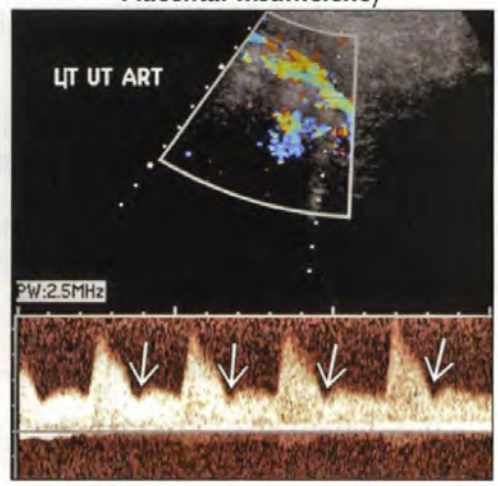


(Left) *Sagittal oblique ultrasound of the right uterine artery, in a continuation of the same case, shows high resistive flow with post-systolic notching ➡.* ***(Right)*** *Sagittal oblique ultrasound of the left uterine artery also shows post-systolic notching ➡. This pattern is abnormal in the second trimester (normal in first trimester). Abnormal UtA waveform carries a 71% positive predictive value for adverse outcome.*

Placental Insufficiency

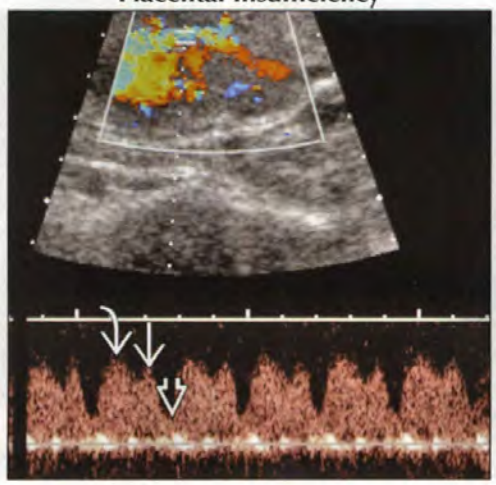


Placental Insufficiency

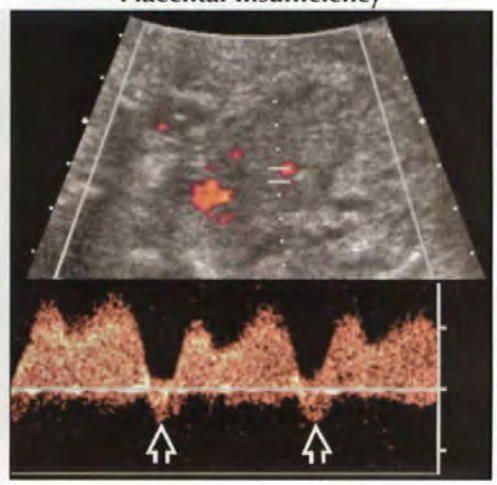


(Left) *Pulsed Doppler ultrasound of the ductus venosus, in the same case, shows that normal flow is still maintained despite the severe IUGR. The S ➡, D ➡, and A ➡ waves reflect systole, diastole, and atrial contraction, respectively. Flow should be continuously towards the heart.* ***(Right)*** *Pulsed Doppler ultrasound in another fetus with severe placental insufficiency shows reversal of the A wave ➡. The finding is associated with fetal hypoxia and cardiac decompensation.*

Placental Insufficiency



Placental Insufficiency



Placental Insufficiency

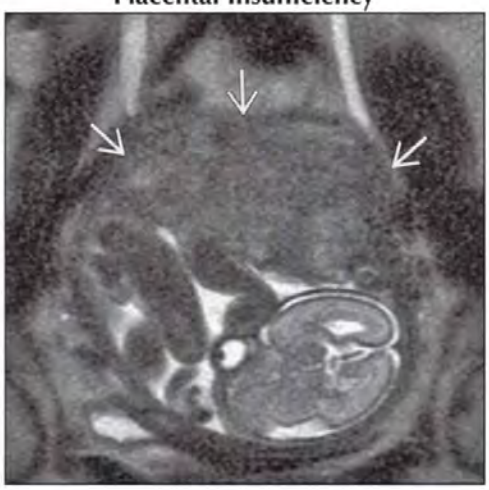


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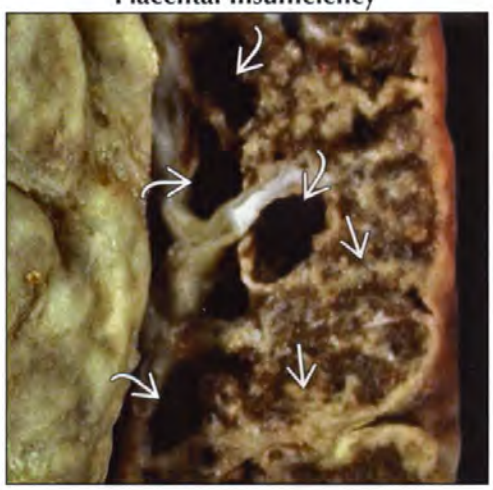


(Left) *Axial ultrasound shows a thickened, cystic placenta and oligohydramnios. The findings were highly suspicious for triploidy, but amniocentesis results were normal.* ***(Right)*** *Coronal ultrasound on follow-up in the same case shows fetal demise with overlapping calvarial sutures, the Spalding sign. Placental pathology showed placental fibrin deposition and infarction.*

Placental Insufficiency

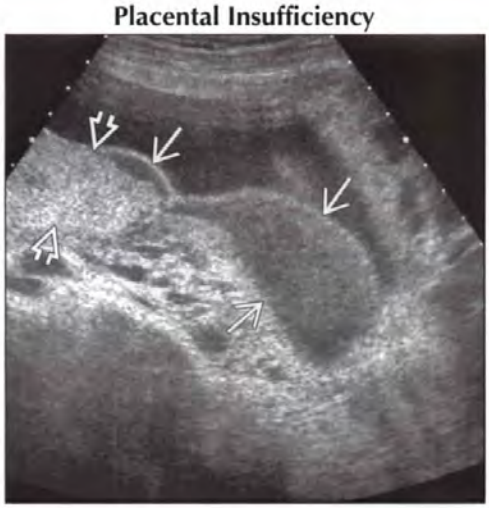


Placental Insufficiency

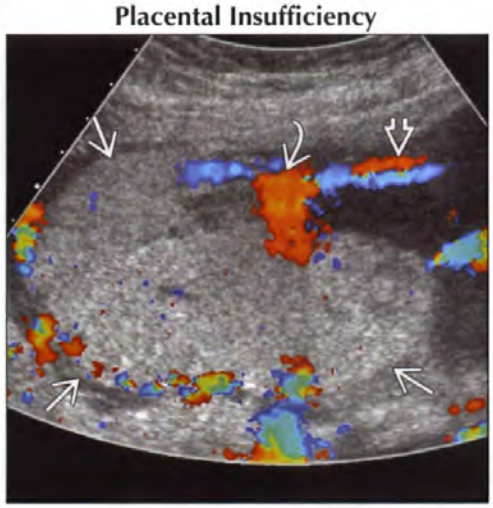


(Left) *T2WI MR shows oligohydramnios, IUGR, and a heterogeneous placenta. The MR confirmed the absence of fetal anomalies. IUGR was the result of placental insufficiency.* ***(Right)*** *Cross section of a placenta from a pregnancy complicated by severe early IUGR shows extensive tannish fibrin deposits and diffuse thrombosis. Several lakes without thrombosis are distended to compensate for the diffuse thrombosis.*

Placental Insufficiency



Placental Insufficiency



(Left) *Sagittal ultrasound shows a subacute abruption from the margin of a posterior placenta.* ***(Right)*** *Axial color Doppler ultrasound of the same placenta shows a marginal cord insertion, a single umbilical artery, and a small "globular" placenta, also known as a battledore placenta. The pregnancy was complicated by chronic bleeding and IUGR. The fetus was delivered at 30 weeks and did well.*

INTRAUTERINE GROWTH RESTRICTION

(Left) *Table shows early, symmetric IUGR in a fetus with a small ventricular septal defect ➔. These findings led to amniocentesis and the diagnosis of trisomy 18.* ***(Right)*** *Clinical photograph of a full-term newborn with trisomy 18 who had in-utero symmetric IUGR. There are minor external anomalies, such as overlapping fingers ➔ and poor tone. In-utero findings also included a ventricular septal defect. The baby died at 2 months of age.*

Trisomy 18 (T18)

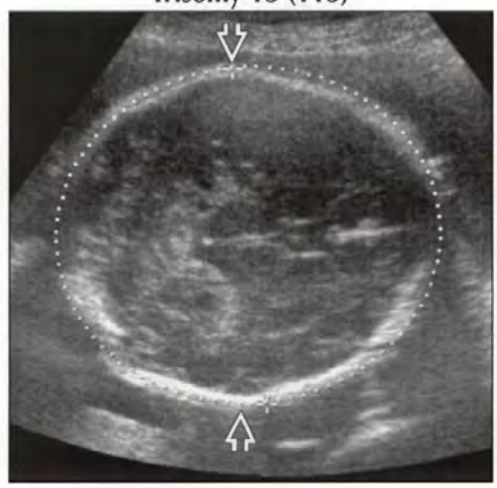


Trisomy 18 (T18)

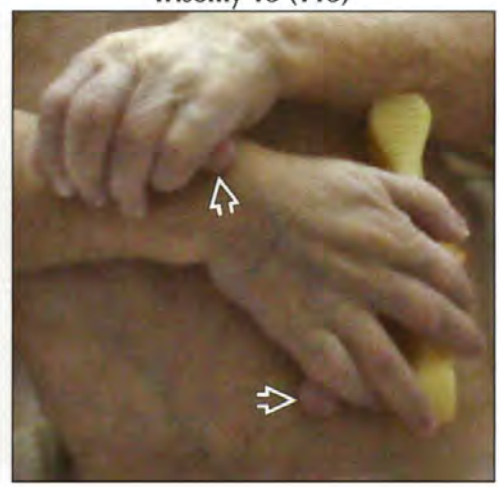


(Left) *Head measurement of a fetus with trisomy 13 shows a normal biparietal diameter, but that is secondary to a very round ➔, brachycephalic shape. The head circumference was actually 2.5 weeks behind. Microcephaly is a feature of trisomy 13.* ***(Right)*** *Autopsy photograph in the same case shows polydactyly ➔, a minor anomaly associated with trisomy 13. IUGR was an important finding in the prenatal diagnosis in this case.*

Trisomy 13 (T13)



Trisomy 13 (T13)



(Left) *Coronal ultrasound shows severe, early, asymmetric IUGR, with the head ➔ larger than the body ➔. Amniocentesis results revealed triploidy.* ***(Right)*** *Clinical photograph shows asymmetric IUGR in a case of triploidy. Late asymmetric IUGR is associated with placental insufficiency, but early IUGR is more suspicious for aneuploidy. Triploidy with an extra maternal set of chromosomes classically presents with early severe asymmetric IUGR.*

Triploidy



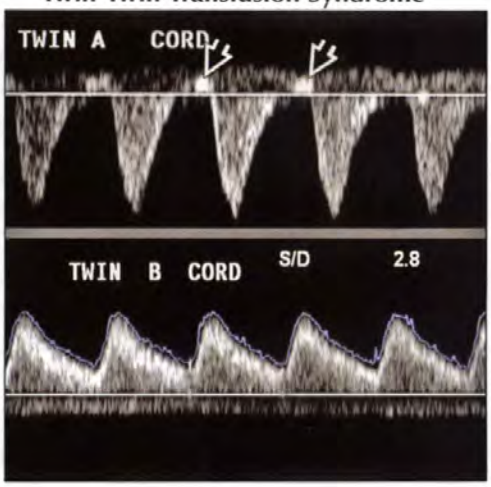
Triploidy



Twin-Twin Transfusion Syndrome



Twin-Twin Transfusion Syndrome

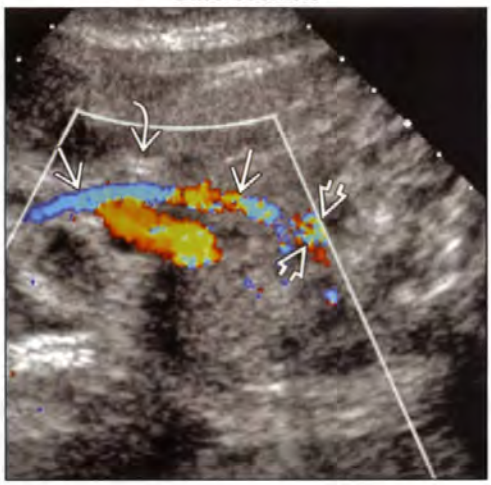


(Left) *Axial ultrasound shows monochorionic twins with discordant abdominal circumferences. Twin A is clearly smaller than twin B. The difference in EFW was > 20%, and twin A had oligohydramnios.* ***(Right)*** *Pulsed Doppler ultrasound of UA flow for twin A shows reversal of diastolic flow ➪, while twin B's UA waveform is normal. Twin A is the pump twin in this pregnancy complicated by twin-twin transfusion.*

Gastroschisis



Gastroschisis

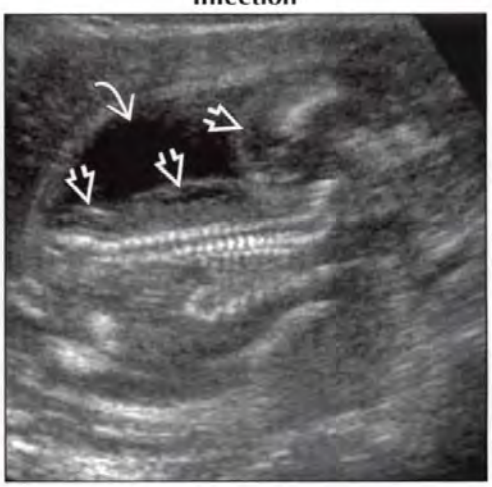


(Left) *Axial ultrasound of a third trimester pregnancy with gastroschisis shows extracorporeal ➪ and intracorporeal ➪ bowel distention.* ***(Right)*** *Color Doppler ultrasound of the abdomen shows the superior mesenteric artery ➪ extending from the aorta ➪, through the abdominal defect ➪ to the extruded bowel. Fetuses with gastroschisis are at risk for IUGR, oligohydramnios, and bowel complications in the third trimester, often leading to early delivery.*

Single Umbilical Artery



Infection



(Left) *Axial ultrasound of the umbilical cord shows a single umbilical artery ➪ and its corresponding umbilical vein ➪. The SUA is larger than expected because it carries 100% of the blood volume, instead of 50%. IUGR complicates 15% of pregnancies with isolated SUA.* ***(Right)*** *Sagittal oblique ultrasound shows oligohydramnios ➪ and diffuse skin edema ➪ in a fetus with IUGR. Amniocentesis confirmed parvovirus infection.*

MACROSOMIA

DIFFERENTIAL DIAGNOSIS

Common
- Idiopathic Macrosomia
- Diabetes

Less Common
- Hydrops

Rare but Important
- Beckwith-Wiedemann Syndrome

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Large for gestational age fetus
  - ↑ Estimated fetal weight (EFW)
    - > 90th or 95th percentile
- Birth weight > 4,000-4,500 g
- AC alone can predict macrosomia
  - 37% risk of macrosomia if AC > 37 cm
  - < 1% risk of macrosomia if AC < 35 cm
  - AC often 1st measurement to ↑
    - ↑ Glycogen stores in liver
    - Truncal obesity
- Prevalence
  - 16-18% in diabetics
  - 6-8% in non-diabetics

Helpful Clues for Common Diagnoses
- **Idiopathic Macrosomia**
  - No fetal anomalies
  - Hereditary factors (↑ risk if mom is large)
  - 1/3 with polyhydramnios
- **Diabetes**
  - Polyhydramnios common
  - Associated anomalies
    - Caudal dysplasia/regression
    - Open neural tube defect
    - Cardiac anomalies (5x ↑ risk)
    - Polydactyly
    - GU/GI anomalies
    - Single umbilical artery

Helpful Clues for Less Common Diagnoses
- **Hydrops**
  - Immune and nonimmune causes
  - Excessive body fluid ⇒ large fetus
    - Anasarca ± cystic hygroma
    - Pleural effusion
    - Ascites

Helpful Clues for Rare Diagnoses
- **Beckwith-Wiedemann Syndrome**
  - Signs/symptoms
    - Macroglossia
    - Enlarged internal organs (especially kidneys)
    - Hemihypertrophy
    - Omphalocele
  - Prognosis
    - 20% infant mortality
    - 7-10% tumor risk (Wilms tumor most common)

Other Essential Information
- Fetal complications of macrosomia
  - Shoulder dystocia (10%)
  - Asphyxia
  - Hypoglycemia, hypocalcemia
- Macrosomia associated with polyhydramnios

Idiopathic Macrosomia



Axial ultrasound of the fetal abdomen shows increased truncal subcutaneous fat. The abdominal circumference measurement was 5 wks greater than the other fetal biometric measurements.

Idiopathic Macrosomia



Anteroposterior radiograph shows newborn complications of macrosomia. The baby is intubated because of meconium aspiration. Also, there is a left clavicular fracture from birth trauma.

MACROSOMIA

Diabetes



Diabetes



(Left) *Axial ultrasound shows polyhydramnios and an enlarged abdominal circumference with excessive subcutaneous fat. The patient presented in the third trimester with uncontrolled diabetes. Multiple fetal anomalies were also present.* ***(Right)*** *Clinical photograph of the same neonate shows the large abdomen, polydactyly & internally rotated, flexed feet. Caudal regression, abnormal leg posturing, & polydactyly are anomalies associated with diabetic embryopathy.*

Hydrops

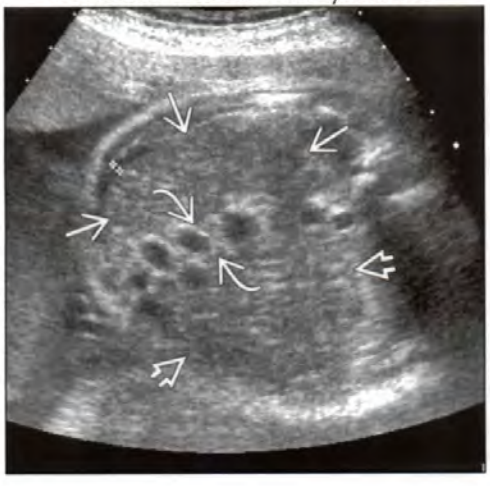


Hydrops

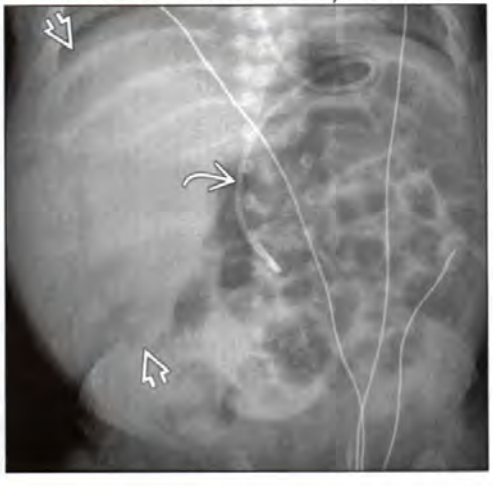


(Left) *Axial ultrasound through the abdomen shows massive ascites. The fetal bowel is displaced posteriorly towards the spine. The fetus is large because of anasarca and ascites.* ***(Right)*** *Clinical photograph in a similar but different case of a stillborn with macrosomia from hydrops. There is a massively distended abdomen from ascites and subcutaneous edema involving the extremities, face, and scalp.*

Beckwith-Wiedemann Syndrome



Beckwith-Wiedemann Syndrome



(Left) *Axial ultrasound through the fetal abdomen shows an enlarged liver and spleen in a fetus with Beckwith-Wiedemann syndrome. The bowel is displaced centrally. The fetus also had macroglossia and polyhydramnios.* ***(Right)*** *Frontal radiograph of a newborn with Beckwith-Wiedemann syndrome shows hepatomegaly. The fetal bowel and feeding tube are displaced to the left by the large liver.*

HYDROPS

DIFFERENTIAL DIAGNOSIS

Common

- Nonimmune Hydrops
  - Idiopathic
  - Cardiac
    - Structural Cardiac Defect
    - Tachyarrhythmia
    - Bradyarrhythmia
  - Fetal Masses
    - Hemangioendothelioma
    - Teratoma
    - Vascular Malformations
  - Placental Chorioangioma
  - Chromosome Abnormalities
    - Turner Syndrome (XO)
    - Trisomy 21
  - Twin-Twin Transfusion Syndrome
  - Infection
- Immune Hydrops
  - Rh Incompatibility
  - Other Antibodies

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Defined as fluid accumulation in 2 or more body cavities
  - Skin/subcutaneous edema
    - Scalp edema often first sign
  - Ascites
  - Bilateral pleural effusions
  - Pericardial effusion
  - Other findings
    - Placentomegaly (placenta thickness > 40 mm)
    - Polyhydramnios
    - Hepatosplenomegaly
- Broadly classified as immune (hemolytic disease → fetal anemia) and nonimmune (all others)
  - 90% are nonimmune hydrops
  - 10% immune

Helpful Clues for Common Diagnoses

- **Idiopathic**
  - Over 50% of cases will not have an identifiable cause
- **Structural Cardiac Defect**
  - Poor contractility → heart failure → hydrops
  - May be accompanied by bradycardia
- **Tachyarrhythmia**
  - Sustained heart rate > 200 bpm
  - Supraventricular tachycardia (SVT) most common cause
  - Hydrops develops in 50-75% fetuses with sustained tachycardia
  - Increased risk of ischemic brain injury when hydrops is present
- **Bradyarrhythmia**
  - 50% associated with cardiac malformation, particularly atrioventricular septal defects
  - 50% of cases seen in mothers with connective tissue disease
  - Increased mortality with heart rate < 50 bpm
- **Fetal Masses**
  - Any mass causing increased cardiac output may lead to failure and hydrops
    - Teratomas and vascular malformations most common
    - Hemangioendothelioma may cause hemolytic anemia in addition to arteriovenous shunting
  - Chest masses may also impede cardiac return
- **Placental Chorioangioma**
  - Benign, vascular placental tumor
  - Fetal hydrops from arteriovenous shunting or from fetal anemia secondary to hemolysis
    - Hydrops uncommon if mass is < 5 cm
  - Polyhydramnios common with large masses
- **Turner Syndrome (XO)**
  - Female fetus with large, septated cystic hygroma
    - Failed or delayed connection between internal jugular veins and nuchal lymph sacs
  - Hydrops secondary to fluid overload from lymphatic obstruction
    - Edema is diffuse and may be dramatic
    - Dorsal pedal edema prominent feature
  - Hydrops can be seen in first trimester
  - Prognosis with hydrops is dismal
- **Trisomy 21**
  - Small cystic hygroma (increased nuchal translucency in 1st trimester) becomes nuchal thickening in 2nd trimester
  - May present with hydrops
  - Other markers often seen

- **Twin-Twin Transfusion Syndrome**
  - Monochorionic twins with artery-to-vein anastomoses in the placenta
  - Recipient at risk for hydrops
    - Larger twin with polyhydramnios
  - Donor at risk for growth restriction
    - Smaller twin with oligohydramnios
  - Twin-twin transfusion syndrome (TTTS) staging
    - Stage 1: Donor bladder visible, normal Doppler
    - Stage 2: Donor bladder empty, normal Doppler
    - Stage 3: Donor bladder empty, abnormal Doppler
    - Stage 4: Hydrops in recipient
    - Stage 5: Demise of one or both
- **Infection**
  - Parvovirus most common but can occur with any severe infection
  - Infection → anemia, myocarditis
  - Look for other signs of infection
    - Intracranial and liver calcifications, ventriculomegaly, hepatosplenomegaly, echogenic bowel, growth restriction
- **Immune Hydrops**
  - Maternal antibodies cross placenta and cause lysis of fetal red blood cells, leading to fetal anemia
  - Anemia causes an elevated middle cerebral artery (MCA) peak systolic velocity (PSV)
  - Need for intervention (transfusion) generally based on relationship of MCA PSV to gestational age
  - **Rh Incompatibility**
    - Maternal lack of D antigen on erythrocyte membrane (Rh -)
    - Sensitization 2° to fetal-maternal hemorrhage
    - Fetal D antigen causes maternal antibody response (< 1 cc fetal cells can lead to anti-D antibody response)
    - With subsequent pregnancy, maternal antibodies attack fetal red blood cells
    - Leads to lysis of fetal erythrocytes
    - Causes anemia and may progress to hydrops if left untreated
  - **Other Antibodies**
    - Non-D antigen causes alloimmunization (usually from incompatible blood transfusion)
    - Kell, Duffy, Kidd, E, C, c, and others
    - Most are variably present in different ethnic populations

Other Essential Information

- First trimester hydrops highly associated with aneuploidy
  - Turner, trisomy 21 most common
- Nonimmune hydrops
  - Over 50% have no unifying diagnosis or directly identifiable cause
  - 22% have a cardiac defect
  - 16% have aneuploidy
    - Turner syndrome > trisomy 21
    - Trisomy 18 and 13 less likely to present with hydrops (growth restriction more common)

Idiopathic



Axial color Doppler ultrasound of the pelvis shows ascites on either side of the bladder, which is flanked by the umbilical arteries (skin edema). No obvious etiology was found for the hydrops.

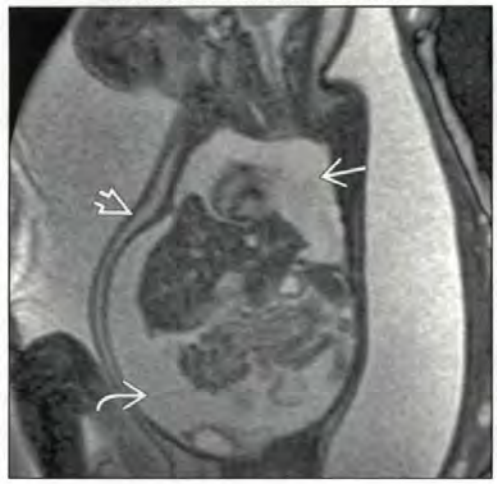
Idiopathic



Sagittal ultrasound in the same case shows marked scalp and face edema. The scalp is one of the first places to see skin edema. This fetus died in utero with no unifying diagnosis found at autopsy.

HYDROPS

Structural Cardiac Defect

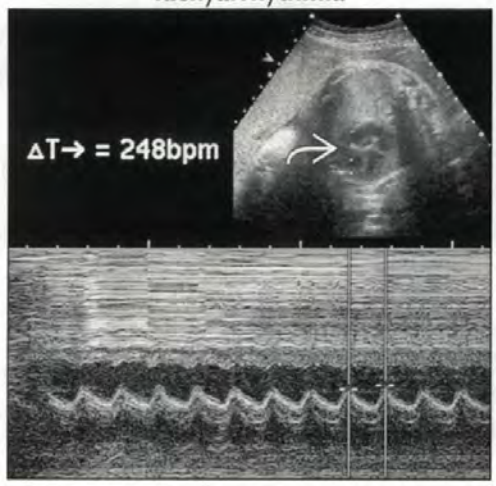


Structural Cardiac Defect



(Left) *Coronal T2WI MR shows the typical MR findings of hydrops including high-signal skin edema, pleural effusions, and ascites. Hydrops resulted from poor cardiac return secondary to ectopia cordis (seen on other images).* ***(Right)*** *Four chamber view shows both a ventricular septal defect and absence of the atrial septum. The ventricular rate was 53 beats per minute. 50% of fetuses with sustained bradycardia will have a cardiac malformation.*

Tachyarrhythmia

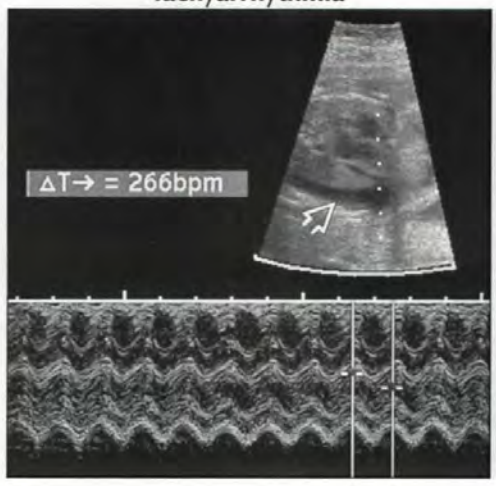


Tachyarrhythmia



(Left) *M-mode ultrasound at initial presentation shows a fetal ventricular rate was 248 beats per minute. The heart size was normal, and there was no evidence of hydrops. Despite immediate instigation of treatment, the fetus remained in SVT.* ***(Right)*** *Coronal ultrasound of the same fetus shows the development of hydrops with pleural effusions, ascites (not shown), and hepatomegaly. The infant was delivered by emergency C-section at 29 weeks gestation.*

Tachyarrhythmia



Tachyarrhythmia



(Left) *M-mode ultrasound shows a sustained ventricular rate of 266 bpm. The fetus was hydropic at presentation (pleural effusion).* ***(Right)*** *Radiograph immediately after delivery shows features of congestive heart failure, including cardiomegaly, ascites causing abdominal distention, and diffuse skin edema.*

HYDROPS

Bradyarrhythmia

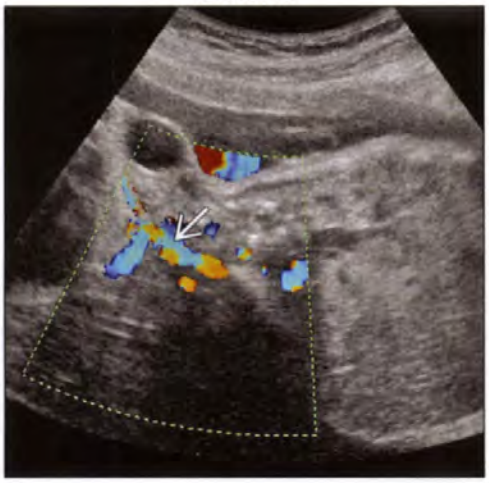


Teratoma



(Left) *Coronal ultrasound of a hydropic fetus shows cardiomegaly, ascites, and skin edema. Mortality approximates 75% when hydrops occurs with bradycardia.* ***(Right)*** *3D ultrasound of a large sacrococcygeal teratoma (sacrum, leg). Masses of this size put a large strain on cardiac output.*

Teratoma



Teratoma



(Left) *Coronal color Doppler ultrasound from the previous case shows a large vessel supplying the mass. The vessel caliber was larger than that of the aorta.* ***(Right)*** *Coronal ultrasound higher in the abdomen shows dilatation of the inferior vena cava; compare with the aorta. This is one sign of impending cardiovascular compromise. If hydrops develops, the prognosis is dismal.*

Vascular Malformations



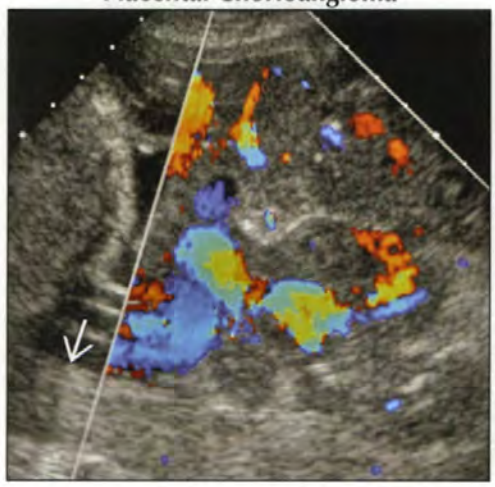
Vascular Malformations



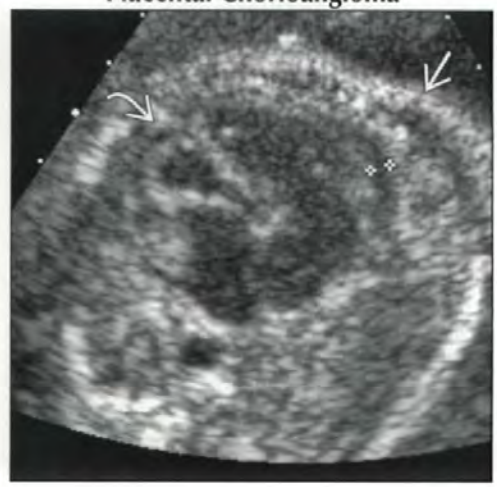
(Left) *Axial oblique ultrasound of the fetal head shows a midline, elongated, cystic structure, ventriculomegaly, and scalp edema.* ***(Right)*** *Axial oblique pulsed Doppler ultrasound demonstrates dramatic blood flow in this "cyst", confirming an arteriovenous fistula (vein of Galen malformation). Significant shunting leads to high output failure and hydrops. The ventriculomegaly was secondary to ischemic changes.*

HYDROPS

Placental Chorioangioma



Placental Chorioangioma



(Left) *Color Doppler ultrasound of the placenta shows a large, lobular, hypoechoic mass, which demonstrates marked vascularity. A portion of normal placenta is also seen.* ***(Right)*** *Axial ultrasound of the fetal chest in the same case shows cardiomegaly, skin edema, and a small pericardial effusion (calipers). The large volume of vascular shunting caused hydrops in the fetus.*

Turner Syndrome (XO)



Turner Syndrome (XO)



(Left) *Gross pathology of mid-trimester fetus with Turner syndrome shows a large hygroma and diffuse hydrops. The abdomen is distended from ascites. There is dramatic subcutaneous edema with characteristic dorsal pedal edema.* ***(Right)*** *Sagittal ultrasound of the foot in another case of Turner syndrome shows marked dorsal pedal edema.*

Turner Syndrome (XO)



Trisomy 21



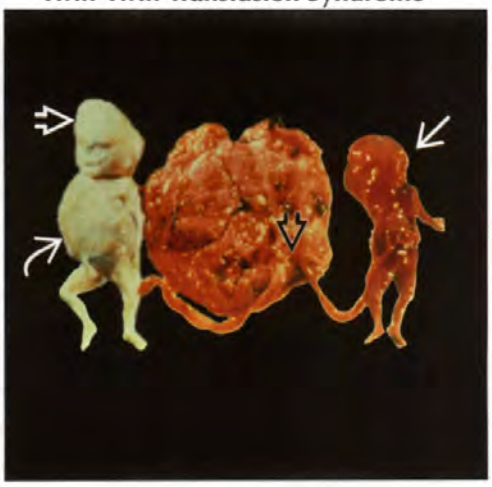
(Left) *Axial ultrasound of a late 1st trimester fetus shows diffuse edema and a large posterior hygroma with two large cysts. Turner syndrome typically has larger, more complex hygromas and more severe hydrops than does trisomy 21.* ***(Right)*** *Coronal ultrasound shows a 1st trimester fetus with diffuse skin edema. Chromosomes showed trisomy 21.*

HYDROPS

Twin-Twin Transfusion Syndrome

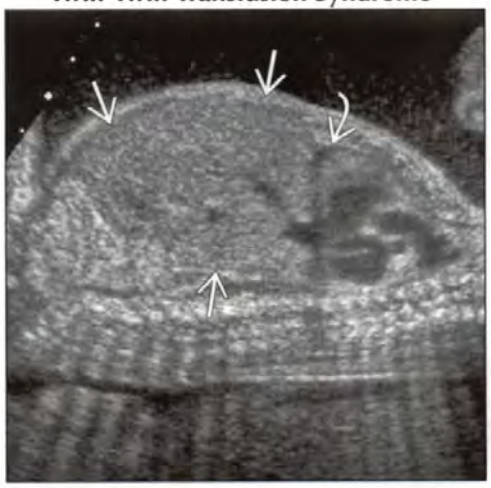


Twin-Twin Transfusion Syndrome

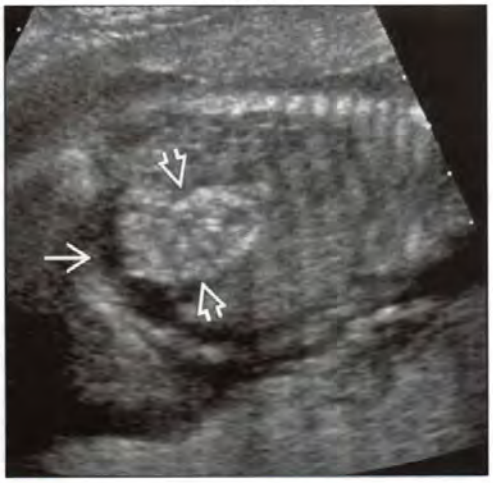


(Left) *Axial ultrasound through the abdomens of monochorionic diamniotic twins shows no apparent membrane, because it is tightly wrapped around the donor twin. The recipient is severely hydropic with skin edema and massive ascites.* ***(Right)*** *Gross pathology from a different case shows a hydropic recipient twin with edema and ascites, and a small donor twin. Note the close proximity of the cord insertions.*

Twin-Twin Transfusion Syndrome

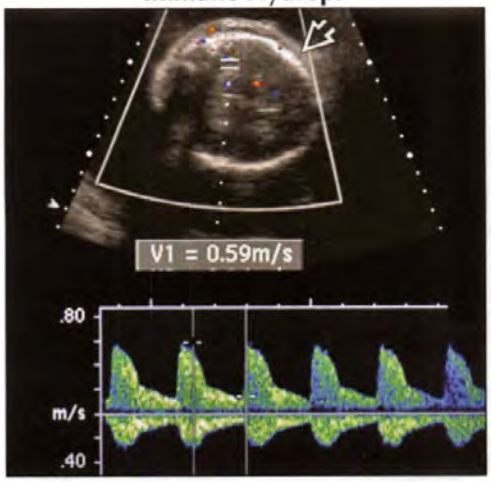


Infection

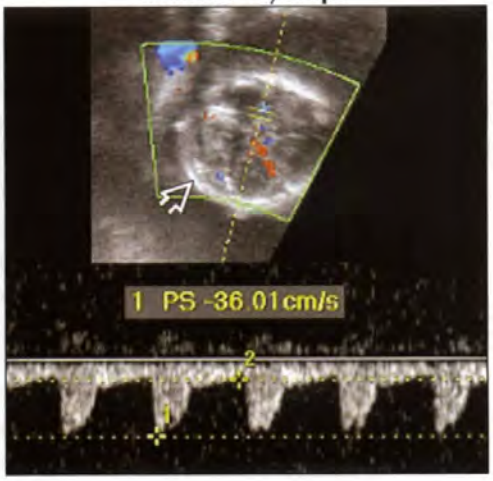


(Left) *Sagittal ultrasound of the recipient twin in another TTTS case shows hepatomegaly and cardiomegaly with a small pericardial effusion.* ***(Right)*** *Sagittal oblique ultrasound shows ascites. This was part of generalized hydrops, which also included skin edema and pleural fluid. The clue to the diagnosis is the echogenic bowel, which suggests infection. This fetus had a parvovirus infection that caused fetal anemia and subsequent hydrops.*

Immune Hydrops



Immune Hydrops



(Left) *Axial pulsed Doppler ultrasound shows a typical example of hydrops from anemia. MCA Doppler interrogation showed a high peak systolic velocity, consistent with fetal anemia. The fetus had scalp edema, body wall edema, and polyhydramnios.* ***(Right)*** *Pulsed Doppler ultrasound ten days after the transfusion shows the scalp edema has resolved and the MCA peak systolic value has decreased from 59 cm/sec to 36 cm/sec (normal for gestational age).*

FETAL ANEMIA

DIFFERENTIAL DIAGNOSIS

Common

- Alloimmunization
  - Rh Incompatibility
  - Other Antibodies
- Infection
  - Parvovirus B19

Rare but Important

- Hemangioendothelioma
- Leukemia
- Fanconi Anemia

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Clinical history is often key to diagnosis
  - Watch for fetal anemia with maternal history of isoimmunization
    - Usually with Rh negative maternal blood types
    - Can have other minor antibody sensitizations
  - Obtain history of potential viral exposures
- Look for associated fetal abnormalities which can cause fetal anemia
  - Tumors
  - Vascular malformations
- Always consider anemia when a fetus presents with hydrops
- Middle cerebral artery (MCA) Doppler peak systolic velocity (PSV) measurements elevated in anemia
  - Very accurate noninvasive method to screen for fetal anemia

Helpful Clues for Common Diagnoses

- **Rh Incompatibility**
  - Occurs in women who are Rh (-)
    - Indicates absence of D antigen in erythrocyte membrane
  - Results from maternal immune response against RhD antigen on fetal red blood cells
    - Maternal anti-D antibodies cross placenta
    - Leads to lysis of fetal erythrocytes
  - Prophylaxis should be given routinely if woman is Rh (-) and when fetal and maternal blood could be in contact
    - Ectopic pregnancy
    - Post chorionic villus sampling or amniocentesis
    - Following spontaneous or elective abortion
    - Maternal trauma
    - At time of delivery of an Rh + infant
    - Prophylactic dose given at approximately 28 weeks gestation
  - If no prior history of antibody sensitization → correlate with antibody titers to assess for risk of fetal anemia
  - If history of prior sensitization → plan for increased surveillance and monthly antibody titer until critical titer reached (generally 1:8-1:16)
    - Once critical titer reached, then follow serial MCA-PSV
  - Fetal hemolytic disease similar to or more severe in subsequent pregnancies
  - Serial ultrasound evaluation
    - Monitor for hydrops
    - Check PSV in MCA
    - Increased PSV indicates worsening fetal anemia
    - Follow-up interval and intervention based on PSV measurements compared to normative scale (Mari zones A-D)
  - Cordocentesis and transfusion if anemia severe
- **Other Antibodies**
  - Most sensitizations caused by incompatible blood transfusions
    - Includes Kell, Duffy, Kidd, E, C, c and multiple other antigens
  - Similar management of pregnancy as with Rh alloimmunization
- **Parvovirus B19**
  - 20-30% of women who become infected during pregnancy transmit to fetus
    - 4% risk of fetal hydrops
    - Highest risk of fetal death if infected < 20 wks gestation
  - Fetal anemia causes sonographic findings
    - Parvovirus attacks red blood cell precursors
    - Involvement of cardiac myocytes may contribute to hydrops
  - Ascites most common presenting finding
    - Progression to hydrops in severe cases
  - Maternal infection should be managed by high risk specialist

- Weekly ultrasound for 10-12 weeks after seroconversion
  - Check for developing hydrops
  - Monitor MCA Doppler PSV to assess for fetal anemia
  - Intrauterine transfusion warranted for fetal anemia

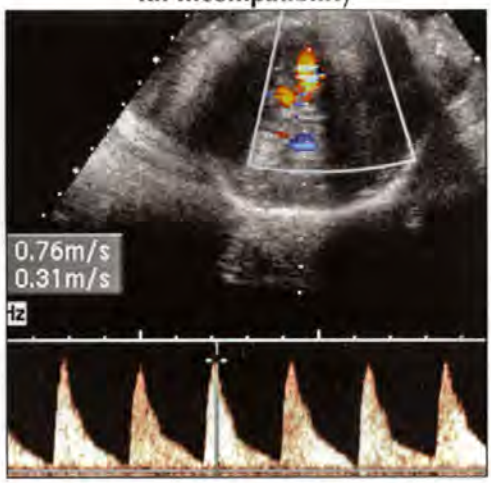
Helpful Clues for Rare Diagnoses

- **Hemangioendothelioma**
  - Typically solid, well-defined large mass
  - Variable echogenicity: Can be hypoechoic, hyperechoic or mixed echogenicity
  - Very vascular mass with arteriovenous shunting on color Doppler
  - Arteriovenous shunting may result in hemolytic anemia (Kasabach-Merritt sequence)
  - Both vascular shunting and anemia contribute to development of hydrops
- **Leukemia**
  - Usually presents with hepatosplenomegaly
    - Unlike other liver tumors, most commonly diffuse enlargement rather than focal mass
  - Associated with Down syndrome
  - Hydrops can develop due to multiple factors
    - Fetal anemia
    - Leukemic infiltration of myocardium
    - Visceral fibrosis with increased vascular resistance
- **Fanconi Anemia**
  - Inherited bone marrow failure syndrome
  - Autosomal recessive
    - Multiple known gene mutations
    - Can be tested prenatally with amniocentesis and chromosome breakage studies
    - Usually Jewish ancestry or family history will spur mutation test for more common gene defects
    - If fetus is index case then likely will not be able to test for all known mutations until birth
  - Prenatal ultrasound shows mainly musculoskeletal abnormalities
    - Radial ray defect
    - Absent or hypoplastic thumb
    - Microcephaly, micrognathia
  - Can also have genitourinary or cardiac malformations
  - May have non-immune hydrops or intrauterine growth restriction

Other Essential Information

- Kasabach-Merritt sequence
  - Rare condition associated with vascular lesion such as hemangioendothelioma or high-flow arteriovenous fistula
  - Results in consumptive thrombocytopenia, coagulopathy, hemolytic anemia
    - Fetal disseminated intravascular coagulation can result
  - May present with cardiomegaly and hydrops due to anemia and high output heart failure

Rh Incompatibility



Axial color Doppler ultrasound of the middle cerebral artery shows an elevated peak systolic velocity, measuring up to 76 cm/sec. This plots in zone A for 31 weeks, indicating severe fetal anemia.

Rh Incompatibility



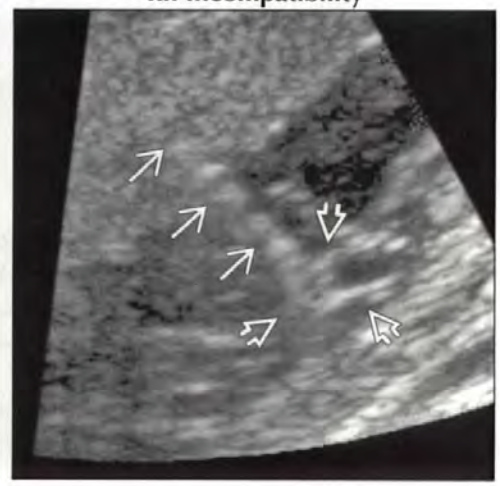
Ultrasound during the transfusion shows the echogenic needle tip in the umbilical vein at the cord insertion. This is a typical location for umbilical vein access. Subsequent MCA peak velocity measured 52 cm/sec.

FETAL ANEMIA

Rh Incompatibility



Rh Incompatibility



(Left) *Axial ultrasound shows an echogenic needle tip in the intra-abdominal portion of the umbilical vein during fetal transfusion. This approach can be used if the cord base is difficult to access.* ***(Right)*** *Ultrasound during fetal transfusion shows the echogenic needle entering a free loop of umbilical cord. This approach can also be used when access to the placental cord insertion site is limited.*

Parvovirus B19



Parvovirus B19



(Left) *Axial ultrasound shows hydrops with skin edema and ascites.* ***(Right)*** *Axial ultrasound through the chest in the same case shows cardiomegaly and pleural fluid. The MCA peak systolic velocity was elevated for a 23 week gestation. Fetal transfusion was attempted; however, the procedure was not tolerated, and the fetus expired. Fetal blood sampling showed a hematocrit of 6 and low platelets of 9000.*

Parvovirus B19



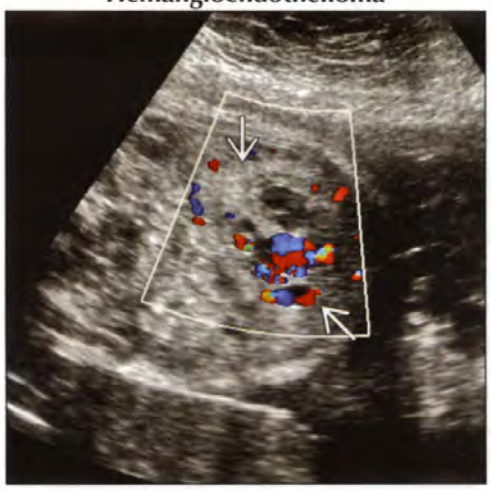
Parvovirus B19



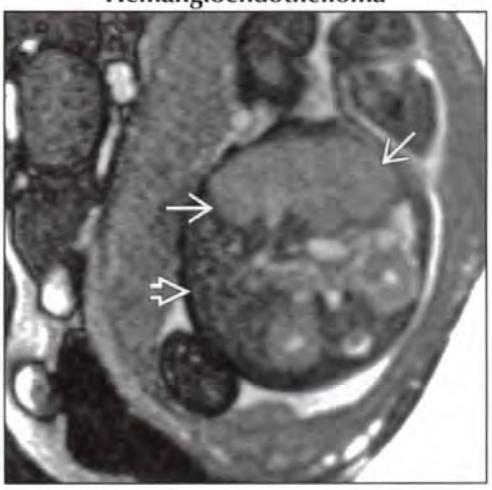
(Left) *Coronal ultrasound shows skin edema even involving the head and face. Realtime imaging demonstrated very little fetal movement. At the initial 17 week scan, the fetus was already 1.5 weeks behind in growth.* ***(Right)*** *Coronal ultrasound of the same fetus shows diffuse skin edema with the head hyperextended in an unusual position. Ascites was also present. Amniocentesis confirmed parvovirus infection.*

FETAL ANEMIA

Hemangioendothelioma

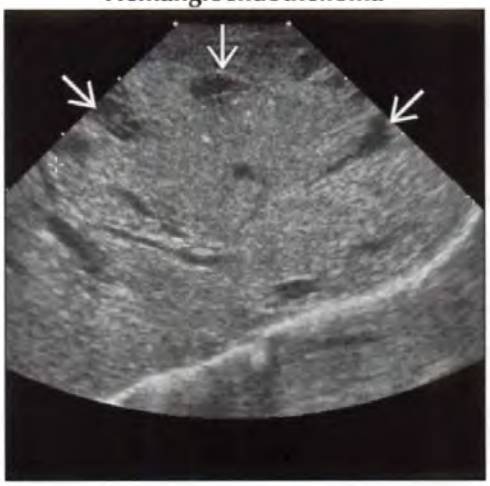


Hemangioendothelioma

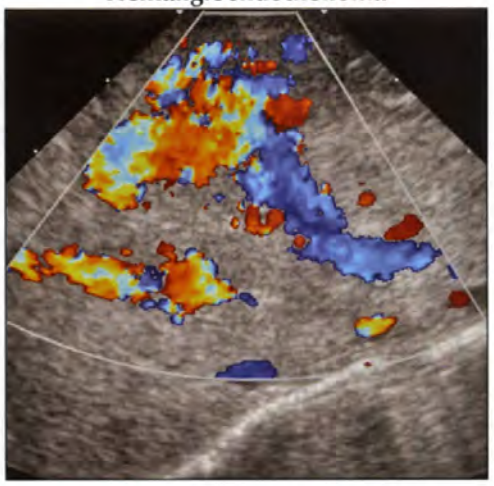


(Left) *Axial color Doppler ultrasound through the fetal abdomen shows a heterogeneous vascular mass ➡. No normal liver could be identified.* ***(Right)*** *Axial T2WI MR in the same case shows a lobular, intermediate signal mass ➡, within the left lobe of the liver. A normal right lobe ➡ is identified. MR is often better able to define the extent of the mass.*

Hemangioendothelioma

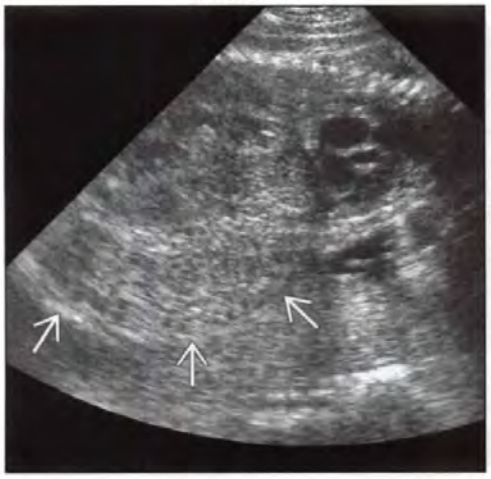


Hemangioendothelioma



(Left) *Axial ultrasound in the same case immediately after delivery shows the entire left lobe of the liver replaced by a heterogeneous, mildly hyperechoic mass (cursors). Note the multiple large vessels ➡ within the mass.* ***(Right)*** *Color Doppler ultrasound confirms marked flow within this lesion. Vascular tumors such as this can cause platelet trapping and a consumptive coagulopathy resulting in fetal anemia (Kasabach-Merritt sequence).*

Leukemia



Leukemia



(Left) *Sagittal ultrasound shows massive hepatomegaly ➡ in a fetus with congenital leukemia. The spleen was also grossly enlarged.* ***(Right)*** *Gross pathology of the same case shows gross hepatomegaly with the liver essentially filling the entire abdomen. Fetal leukemia has a strong association with trisomy 21. If fetal leukemia is suspected, cordocentesis for both blood cell analysis and karyotyping should be performed.*

ABNORMAL FETAL PRESENTATION

DIFFERENTIAL DIAGNOSIS

Common
- Occiput-Posterior
- Complete Breech
- Frank Breech
- Footling Breech

Less Common
- Transverse Lie
- Incomplete Breech

Rare but Important
- Funic Presentation

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Normal presentation at term
  - Vertex and occiput-anterior
    - Back of head faces pubis
  - 43% vertex at 15-22 weeks
  - 90% vertex at 31-35 weeks
- 3-4% of term fetuses are breech
  - 87% deliver by cesarean section
  - External cephalic version
    - 40% success in nulliparous women
    - 60% success in multiparous women
- Etiology of malpresentation
  - Idiopathic
  - Prematurity
  - Placenta previa
  - Uterine anomaly
  - Abnormal fetal movement

Helpful Clues for Common Diagnoses
- **Occiput-Posterior**
  - Vertex + face to pubis
  - Vaginal delivery attempted
    - Labor typically longer
- **Complete Breech**
  - Buttocks presenting
  - Flexed legs (feet down)
- **Frank Breech**
  - Buttocks presenting
  - Extended legs (feet up)
- **Footling Breech**
  - Foot or feet presenting

Helpful Clues for Less Common Diagnoses
- **Transverse Lie**
  - Fetus is sideways
    - Head in one flank, bottom in other
  - Obligatory cesarean delivery
- **Incomplete Breech**
  - Hybrid of complete and frank breech
  - One leg extended and one leg flexed

Helpful Clues for Rare Diagnoses
- **Funic Presentation**
  - Umbilical cord slips in front of fetus
    - More common with nonvertex presentation
  - Cord may prolapse into vagina
  - Emergency cesarean delivery

Other Essential Information
- Twins
  - Presenting twin vertex for vaginal delivery
  - Second twin position less important

Occiput-Posterior



Sagittal ultrasound shows a late term fetus in a cephalic position; however, the face ➡, not the occiput, is facing the anterior uterine wall. The finding may be relevant if the patient is in active labor.

Complete Breech



Sagittal ultrasound of a fetus at term shows a flexed knee ➡ that lies close to the fetal trunk ➡. The tibia and fibula extend inferiorly ➡. The fetal bottom ➡ is largely obscured by the leg but is near the cervix.

ABNORMAL FETAL PRESENTATION

Complete Breech



Frank Breech

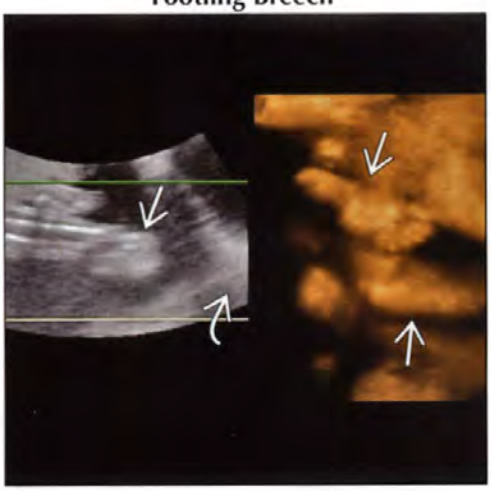


(Left) *3D ultrasound in the second trimester shows a fetus in complete breech position. The lower extremities are flexed at the knees ➔. **(Right)** 3D ultrasound in the second trimester shows a fetus in frank breech position with an extended leg (knee ➔ and foot ➔). Breech positioning is common in the second trimester, but the majority of fetuses convert to a cephalic presentation by term.*

Footling Breech

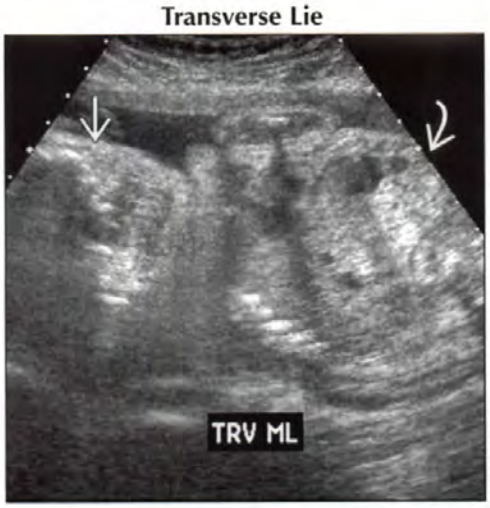


Footling Breech

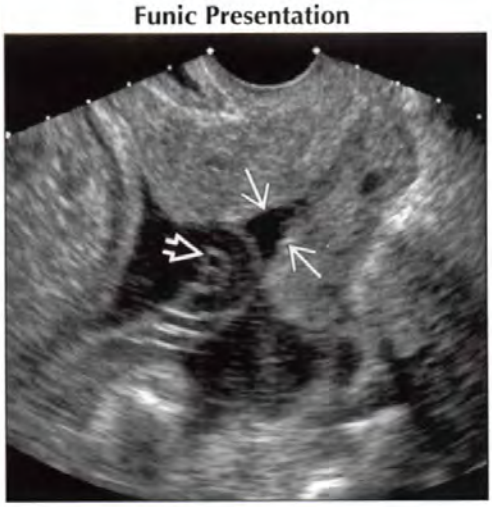


(Left) *Sagittal ultrasound in a near term fetus shows that the feet ➔ and lower legs ➔ are lower than the fetal pelvis and are the presenting parts. **(Right)** Sagittal 3D ultrasound in a 30 week pregnancy shows the feet ➔ are the presenting parts adjacent to the cervix ➔. This position may change with advancing pregnancy.*

Transverse Lie



Funic Presentation



(Left) *Transverse ultrasound in the third trimester shows the fetal head on the right ➔ and the fetal trunk ➔ on the left. The pregnancy was complicated by a complete placenta previa. **(Right)** Sagittal transvaginal ultrasound of another case shows dilatation of the internal os of the cervix ➔ and a loop of cord at the cervical os ➔. The cord is the presenting structure and is at risk for prolapse through the cervix.*

SECTION 12
Uterus-Cervix

Short Cervix	12-2
Uterine/Cervical Mass	12-6

SHORT CERVIX

DIFFERENTIAL DIAGNOSIS

Common

- Mimics
  - Poor Scan Technique
  - Nabothian Cyst
  - Prominent Mucus Plug
  - Curved Cervix
- Incompetent Cervix
- Multiple Gestation
- Failed Cerclage
- Uterine Duplication

Less Common

- Loop Electrosurgical Excision Procedure (LEEP)
- Trachelectomy

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- Short cervix defined as length < 26 mm at 24 weeks
  - Increases relative risk for preterm delivery by a factor of 6.19
  - Sensitivity of 37% for delivery before 35 weeks, positive predictive value of 18%
- Rates of preterm birth continue to rise despite extensive research
  - Measurement of cervical length is one method used in attempt to identify at-risk group who might benefit from intervention
  - However, effective interventions to prevent preterm birth remain elusive
- Correct scan technique vital for accurate evaluation of cervical length
- Options include transabdominal (TA), translabial (TL), & transvaginal (TV)
  - TA: Use amniotic fluid as acoustic window to cervix in second trimester
    - Transducer inferior to maternal umbilicus, angled inferiorly
    - Scanning in suprapubic area → shadowing by pubic symphysis
    - Full bladder or lower segment contraction may artificially elongate cervix → missed diagnosis of short cervix
    - Contraction may mimic appearance of fluid within endocervical canal
    - In 3rd trimester, presenting part may obscure detail: Perform TL or TV scans
  - TL: Used in 3rd trimester
    - Transducer placed at introitus
    - Coapted vaginal walls act as acoustic window to cervix
    - Safe in patients with ruptured membranes
  - TV: Best technique for evaluation of cervical length, dynamic changes, relationship to placenta, & to exclude vasa previa
    - Bladder empty
    - Transducer placed in vagina under direct visual monitoring
    - Advance until cervix in focus, retract until cervix just within focal zone
    - Anterior lip & posterior lip should be same thickness
    - Measure length along endocervical canal: Bright echogenic line
    - Observe for several minutes for dynamic changes ± apply fundal or suprapubic pressure
    - Bladder neck mucosal echo is anatomic landmark for internal os
    - Ruptured membranes is a relative contraindication for TV sonography
- Obtain measurements
  - Shortest length of closed cervix on good image
  - Degree of dilatation of internal os
  - Length of funneling of membranes
  - Length of cervix above cerclage ± length cervix below cerclage
    - Recent review suggests absent cervix above cerclage is associated with preterm delivery, chorioamnionitis, & preterm premature rupture of membranes

Helpful Clues for Common Diagnoses

- **Mimics**
  - **Poor Scan Technique**
    - Spurious short measurement may alter management & have patients needlessly placed on bedrest, etc.
    - Spurious long measurement may lead to missed opportunity to intervene in at-risk patients
  - **Nabothian Cyst**
    - May cause confusion with fluid in cervix
    - Check for normal echogenic endocervical canal "stripe"

- Scan from side to side: Nabothian cysts are thin walled, should be seen separate from endocervical canal
- Large nabothian cysts may distort cervix, alter contour of canal

◦ **Prominent Mucus Plug**
- Prominent hypoechoic material within endocervical canal
- No dynamic changes
- No internal os dilatation

◦ **Curved Cervix**
- Measure in series of straight lines & sum for total length

• **Incompetent Cervix**
◦ Cervix dilates without pain: Most likely to present with mid-trimester loss

• **Multiple Gestation**
◦ Cervical length shorter in multiples than singletons
◦ Higher order multiples → decreasing cervical length
◦ Recent reviews indicate that cerclage does not prevent preterm birth in multiples & may, in fact, be detrimental

• **Failed Cerclage**
◦ Membranes prolapse to, or through, level of cerclage

• **Uterine Duplication**
◦ Müllerian duct anomalies overrepresented in infertility/recurrent abortion population
◦ In addition to variant uterine morphology, cervix may be shortened

Helpful Clues for Less Common Diagnoses

• **Loop Electrosurgical Excision Procedure (LEEP)**
◦ Performed for cervical intraepithelial neoplasia (CIN)
◦ Women with a history of LEEP & cold knife conization have shorter cervical lengths than low-risk controls
◦ Cervical lengths are similar to women who have had a previous spontaneous preterm birth

• **Trachelectomy**
◦ Cervix absent, rather than short, due to fertility sparing surgery for early cervical cancer
- Cervix removed
- Inferior aspect of remaining uterus closed with purse string suture

◦ Delivery requires C-section

Other Essential Information

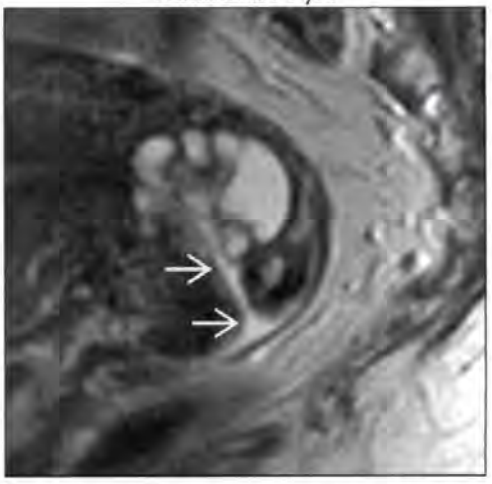
• Management of short cervix remains controversial
• Additional risk factors for preterm birth
◦ Prior spontaneous preterm birth
◦ History of 1 or more midtrimester losses
◦ History of multiple cervical procedures (cone biopsy, LEEP, multiple dilation, & curettage procedures)
◦ Uterine anomaly
◦ Excessive uterine distension (e.g., multiple gestations, polyhydramnios)

Nabothian Cyst



Sagittal transvaginal ultrasound shows several nabothian cysts ➡ separate from the endocervical canal ➡. The endocervical canal must be shown in its full length before measurements are made.

Nabothian Cyst



Sagittal T2WI MR in a nonpregnant patient shows how nabothian cysts can be confusing. The canal ➡ is normal in length, but could be measured short (between the arrows) if the cysts are not appreciated.

SHORT CERVIX

Prominent Mucus Plug



*(Left) Sagittal transvaginal ultrasound shows a 2.9 cm cervix in the third trimester. The endocervical canal ➡ appears mildly dilated, but this is merely a prominent mucous plug. **(Right)** Sagittal transvaginal ultrasound shows the correct way to measure the length of the cervix when it is curved. A straight line measurement from internal ➡ to external os ➡ would underestimate the length.*

Curved Cervix

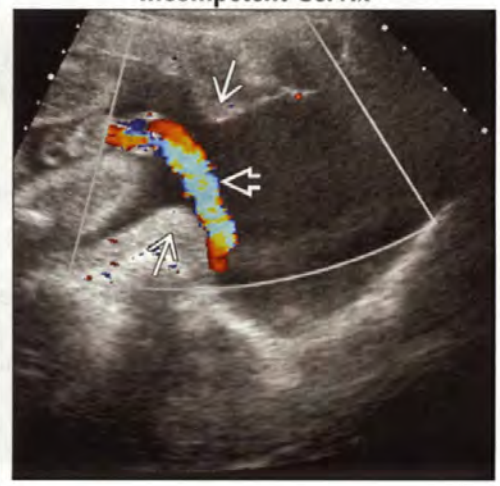


Incompetent Cervix



*(Left) Sagittal ultrasound shows cervical dilatation ➡ with the fetal feet in the cervix and a large balloon of membranes ➡ in the vaginal vault. **(Right)** Sagittal color Doppler ultrasound in the same patient shows funic presentation with a loop of cord ➡ extending into the ballooned membranes at the vaginal vault in a patient with an incompetent cervix ➡.*

Incompetent Cervix

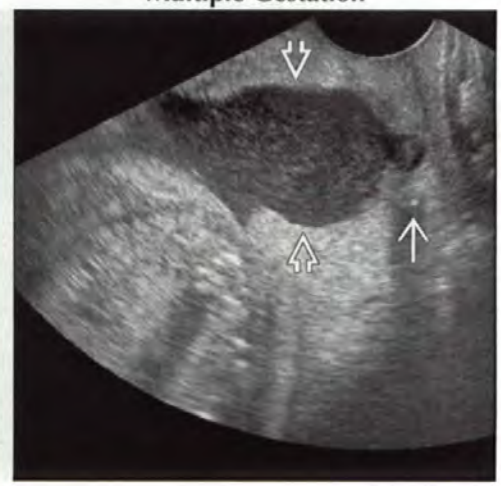


Multiple Gestation



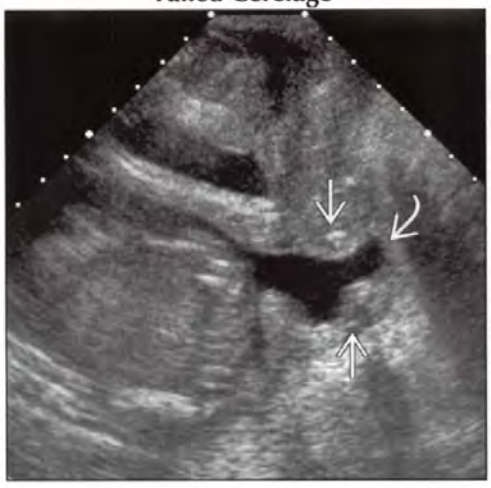
*(Left) Ultrasound shows a quadruplet ➡ pregnancy. At 24 weeks gestation the cervical length was 1.4 cm, a common finding with higher order multiples. **(Right)** Transvaginal ultrasound shows marked funneling of membranes into a dilated cervix ➡. The patient had a triplet pregnancy with emergency cerclage ➡ placed at 18 weeks. The membranes prolapsed through the cerclage, with premature rupture of membranes at 22 weeks.*

Multiple Gestation

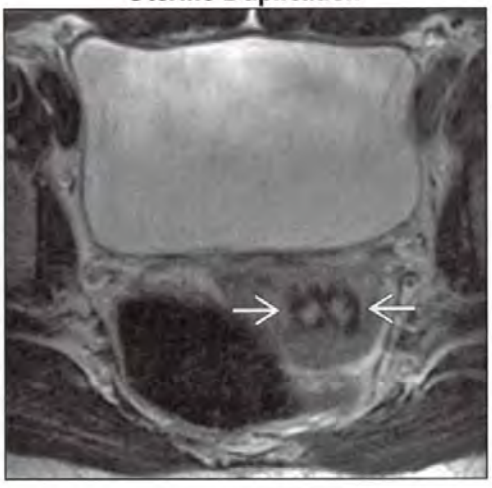


SHORT CERVIX

Failed Cerclage

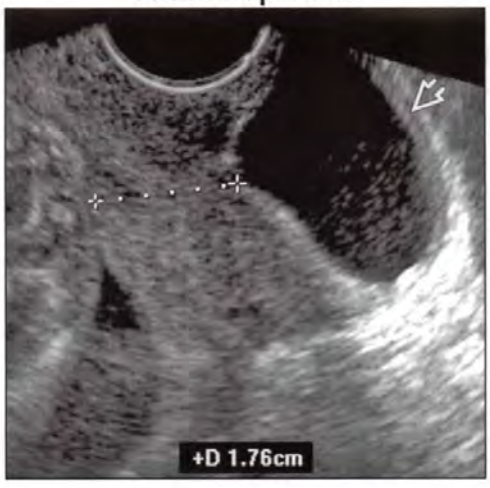


Uterine Duplication

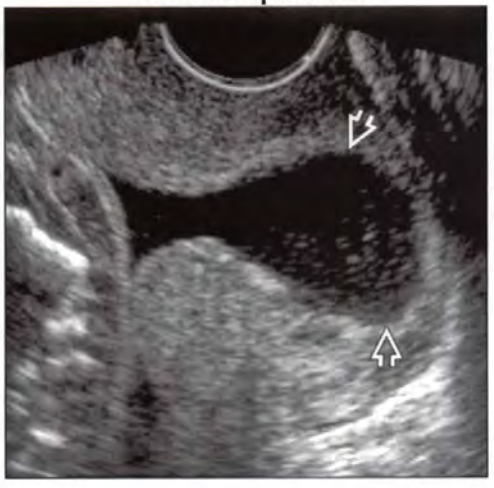


(Left) *Sagittal ultrasound shows a different patient with a failed cerclage. The membranes have prolapsed through the cerclage. This patient had a live delivery at 32 weeks.* ***(Right)*** *Axial T2WI MR shows cervical duplication. Patients with Müllerian duct anomalies are at risk for preterm labor. Number, appearance, and length of cervices should be documented in these patients.*

Uterine Duplication



Uterine Duplication



(Left) *Sagittal oblique ultrasound shows a confusing appearance. There is an apparently short, but closed cervix (calipers) measuring 1.76 cm, yet there is a balloon of membranes seen in the vaginal vault.* ***(Right)*** *Sagittal oblique transvaginal ultrasound in the same patient shows that the membranes are ballooning through the other cervix. This patient had a known uterine duplication.*

Loop Electrosurgical Excision Procedure (LEEP)



Trachelectomy



(Left) *Sagittal transvaginal ultrasound shows mild cervical shortening at 18 weeks in a patient with a history of LEEP for cervical intra-epithelial neoplasia.* ***(Right)*** *Sagittal transvaginal ultrasound shows the appearance of a post-trachelectomy lower uterus. The cervix was removed and the inferior uterus was closed with a cerclage type suture buried beneath the vaginal mucosa.*

UTERINE/CERVICAL MASS

DIFFERENTIAL DIAGNOSIS

Common
- Focal Myometrial Contraction
- Leiomyoma
- Placental Abruption, Mimic
- Uterine Duplication
- Adenomyosis

Less Common
- Chorioangioma, Mimic

Rare but Important
- Gestational Trophoblastic Neoplasia
  - Invasive Mole
  - Choriocarcinoma
- Cervical Cancer
- Uterine Sarcoma

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Is the finding constant?
  - Myometrial contractions are transient and change over course of the exam
  - Hematomas evolve over days to weeks
  - Fibroids may grow or degenerate
- Is it truly within the myometrial wall?
  - Retroplacental hemorrhage between wall and placenta
  - Chorioangioma originates from the placenta

Helpful Clues for Common Diagnoses
- **Focal Myometrial Contraction**
  - Transient myometrial thickening, which changes during course of examination
  - May appear mass-like with elliptical shape and no defined borders
  - Inner myometrium affected more than outer
  - Iso- to hyperechoic compared to myometrium
- **Leiomyoma**
  - Involves myometrium or cervix
    - Submucosal, intramural, subserosal, pedunculated
  - Generally round, well-defined, hypoechoic mass
  - May grow or degenerate during pregnancy
  - Degenerated fibroids more heterogeneous and variable in appearance
    - Hyperechoic with hemorrhage
    - Cystic often with thick, irregular septations
    - Calcified with dense shadowing
  - Color Doppler
    - Hypovascular compared to surrounding myometrium
    - May see uterine vessels splayed around mass
  - Increased complications if placental implantation is on fibroid
    - Abruption
    - Spontaneous abortion
    - Preterm labor
    - Intrauterine growth restriction
    - Postpartum hemorrhage
  - Cervical or lower uterine segment fibroids may obstruct delivery
- **Placental Abruption, Mimic**
  - Echogenicity varies according to age
    - Acutely more echogenic and may be similar to placenta
    - Becomes more hypoechoic and heterogeneous over time
  - Most are marginal abruptions
    - Begin at placental edge and dissect under chorionic membrane
    - Have a crescentic or lentiform configuration and usually not confused with a mass
  - Retroplacental abruption appears more "mass-like"
    - Hematoma contained between placenta and uterus
    - May give erroneous appearance of either an enlarged placenta or a retroplacental fibroid
- **Uterine Duplication**
  - Second horn in a duplicated uterine anomaly may give appearance of uterine mass
  - Always look for echogenic endometrium within center of the "mass"
  - 3 types of Müllerian duct anomalies (didelphys, bicornuate, septate) have two endometrial cavities, which may be confused with a uterine mass
  - Key to diagnosis is visualization of external uterine contour
    - Didelphys: Two separate uteri, each with its own cervix

- Bicornuate: Concave or heart-shaped external fundal contour
- Septate: Fundus mildly convex to mildly concave

- **Adenomyosis**
  - Uterine enlargement without well-defined mass
  - Usually seen in multiparous women
  - Very heterogeneous appearing with fine, linear areas of attenuation throughout the thickened wall ("rain shower" appearance)
    - Endometrium may be obscured
  - Myometrial cysts (2-6 mm) in 50%, highly specific for diagnosis
  - May be focal (adenomyoma)
    - May potentially be confused with fibroid
    - Adenomyoma has an elliptical shape rather than round
    - Poor definition of borders
    - Color Doppler shows speckled pattern of increased vascularity, without peripheral draping vessels seen in leiomyomas

Helpful Clues for Less Common Diagnoses

- **Chorioangioma, Mimic**
  - Benign, vascular placental tumor
  - Large ones may abut adjacent uterine wall causing confusion with a uterine mass
  - Most common on fetal side of placenta, near cord insertion

Helpful Clues for Rare Diagnoses

- **Invasive Mole**
  - Echogenic cystic mass invading myometrium
  - Markedly vascular on Doppler images
  - ↑ Human chorionic gonadotropin (hCG) levels
  - 12-15% of complete hydatidiform moles progress to invasive mole
- **Choriocarcinoma**
  - Suspect choriocarcinoma if ↑ hCG after any type gestational trophoblastic neoplasia (hydatidiform or invasive mole) or any pregnancy (ectopic, abortion or normal)
  - Uterine findings quite variable ranging from normal to infiltrating heterogeneous mass
  - Enlarged cystic ovaries (theca lutein cysts)
  - Lung, brain, liver metastases common
- **Cervical Cancer**
  - Arises from squamocolumnar junction with 80-90% being squamous cell
    - Adenocarcinoma and small cell are majority of remainder (both have worse prognosis)
  - Most tumors exophytic in younger woman
  - Cervical cancer appears as a hypoechoic mass on ultrasound but can be easily missed, especially in the early stages
  - MR best imaging for staging
- **Uterine Sarcoma**
  - Uncommon in women < 40 years old
  - Multiple histologic types including leiomyosarcoma, adenosarcoma, malignant mixed mesodermal modality tumor

Focal Myometrial Contraction



Axial ultrasound shows a "mass" ➡ protruding into the amniotic cavity. Note how it blends imperceptibly into the myometrial wall and that the outer surface of the myometrium ➡ is only minimally distorted.

Focal Myometrial Contraction



Axial ultrasound 15 minutes later shows the contraction ➡ has largely resolved.

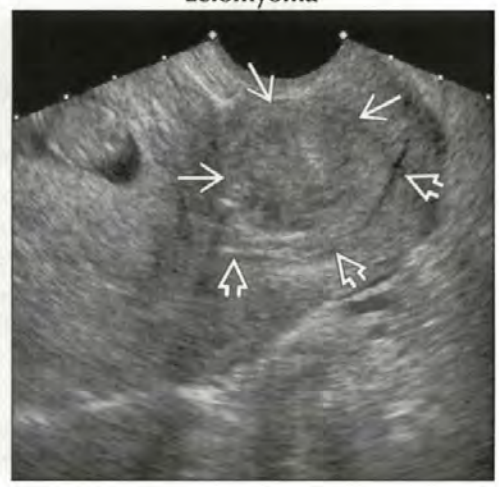
UTERINE/CERVICAL MASS

Focal Myometrial Contraction

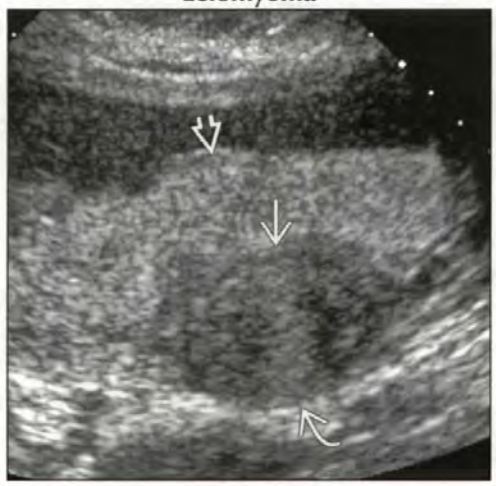


(Left) *Sagittal ultrasound shows a contraction ➡ in the lower uterine segment, which is distorting the inner wall of the myometrium. It is slightly hyperechoic compared to the adjacent myometrium. It resolved by the end of the study.* ***(Right)*** *Sagittal transvaginal ultrasound shows a leiomyoma ➡ in the anterior wall of the cervix. The endocervical canal ➡ bows posteriorly. Fibroids can grow during pregnancy, and if large enough, may potentially obstruct labor.*

Leiomyoma



Leiomyoma

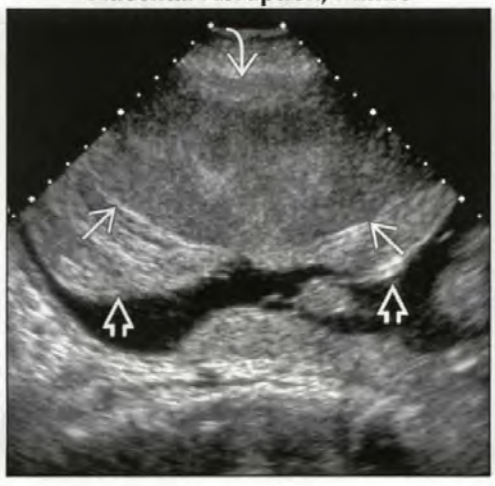


(Left) *Sagittal ultrasound shows placental implantation ➡ on a leiomyoma ➡. The external contour of the uterus ➡ is distorted, helping to differentiate this from a retroplacental abruption, which does not distort the external uterine contour.* ***(Right)*** *Axial color Doppler ultrasound shows a large, complex, cystic leiomyoma ➡ adjacent to an IUP ➡. No internal flow is seen on Doppler. A cystic appearance is often secondary to chronic changes from necrosis.*

Leiomyoma



Placental Abruption, Mimic



(Left) *Axial ultrasound shows a hypoechoic acute blood clot ➡ lifting the placenta ➡ off the myometrium. A distinct separate myometrial wall is seen ➡, which has a normal contour. This rules out a fibroid. Follow-up ultrasound showed evolution of the blood products.* ***(Right)*** *Transverse transabdominal ultrasound shows 2 separate cornua ➡ with a gestational sac on the right ➡. Note the classic heart-shaped external contour typical of a bicornuate uterus.*

Uterine Duplication



UTERINE/CERVICAL MASS

Uterine Duplication



Uterine Duplication

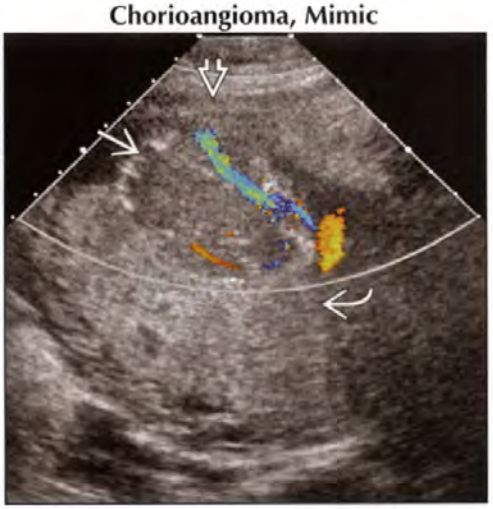


(Left) *Axial transabdominal ultrasound shows a soft tissue "mass" on the left side of the uterus, next to the left ovary. The fetus is on the right.* ***(Right)*** *Coronal oblique ultrasound focused on the left side shows a triangular echogenicity, which is a second endometrial cavity. There is a hypoechoic uterine septum and a normal external contour, typical of a septate uterus.*

Adenomyosis



Chorioangioma, Mimic

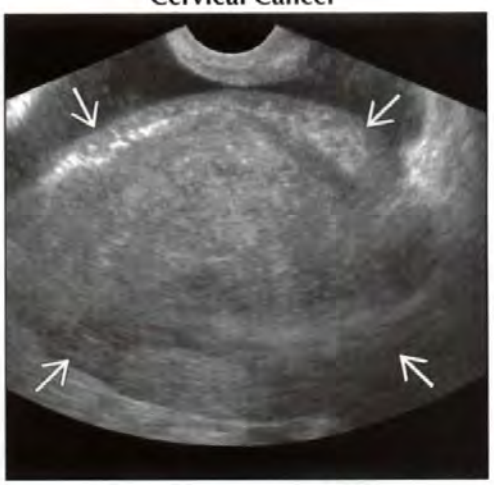


(Left) *Sagittal ultrasound shows a diffusely enlarged, heterogeneous uterus with linear reverberation artifacts. There is tiny fluid collection, but the endometrium is obscured so it is difficult to tell if this is an early gestational sac versus a myometrial cyst, which is common with adenomyosis.* ***(Right)*** *Color Doppler ultrasound shows a vascular mass that is juxtaposed to the uterine wall. The mass is actually emanating from the placenta, which is posterior.*

Invasive Mole



Cervical Cancer



(Left) *Sagittal transvaginal ultrasound of the uterus shows heterogeneous material with small cystic spaces, which is invading into the myometrium. The hCG was markedly elevated. Medical treatment was successful.* ***(Right)*** *Transverse ultrasound shows a bulky cervical mass. Biopsy showed poorly differentiated squamous cell carcinoma. The majority of cervical cancers in reproductive age women are exophytic and often difficult to see with ultrasound.*

SECTION 13
Maternal Conditions in Pregnancy

Uterine Size < Dates	13-2
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Antepartum Hemorrhage	13-6
Acute Abdomen in Pregnancy	13-10
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UTERINE SIZE < DATES

DIFFERENTIAL DIAGNOSIS

Common
- Intrauterine Growth Restriction (IUGR)
- Oligohydramnios

Less Common
- Fetal Demise

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Fundal height (FH) vs. menstrual age (MA)
  - FH = clinical measurement from pubic symphysis to top of uterus
  - FH in cm = MA in wks
  - FH is most accurate at 18-32 wks
  - Ultrasound more accurate than FH

Helpful Clues for Common Diagnoses
- **Intrauterine Growth Restriction (IUGR)**
  - Estimated fetal weight < 10th percentile
  - Causes
    - Placental insufficiency
    - Anomalous fetus
    - Intrauterine infection
  - Doppler assessment
    - ↑ Umbilical artery resistance
    - ↑ Uterine artery resistance
    - ↑ Ductus venosus resistance
    - ↓ Middle cerebral artery resistance
  - Associated with oligohydramnios
- **Oligohydramnios**
  - Fetal causes
    - Bilateral renal agenesis
    - Bilateral renal anomaly
    - Bladder outlet obstruction
  - Non-fetal causes
    - Placental insufficiency
    - Post-term pregnancy (> 42 wks)
    - Premature rupture of membranes
    - Maternal dehydration
  - Idiopathic not common

Helpful Clues for Less Common Diagnoses
- **Fetal Demise**
  - Fetal death after 10 wks
  - 6.9/1000 births in U.S.
  - Maternal causes
    - Prolonged pregnancy (> 42 wks)
    - Diabetes (poorly controlled)
    - Preeclampsia
    - Antiphospholipid syndrome
  - Fetal causes
    - Multiple gestation
    - Intrauterine growth restriction
    - Anomalous fetus
    - Infection
  - Other causes
    - Cord accident
    - Abruption
    - Vasa previa
    - Premature rupture of membranes

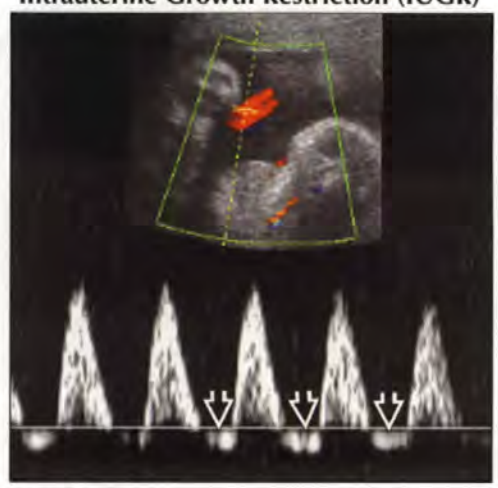
Other Essential Information
- Role of biophysical profile assessment (BPP)
  - Helps identify fetal acidosis
  - Consider formal BPP in 3rd trimester with oligohydramnios &/or IUGR

Intrauterine Growth Restriction (IUGR)



Coronal oblique ultrasound shows oligohydramnios and asymmetric IUGR. All measurements were small, but the body ⇒ was much smaller than the head ⇒. Amniocentesis results showed triploidy.

Intrauterine Growth Restriction (IUGR)



Pulsed Doppler ultrasound of the umbilical artery in a normal fetus with late onset IUGR shows reversal of diastolic flow ⇒. The finding prompted early delivery.

UTERINE SIZE < DATES

Oligohydramnios



Oligohydramnios



(Left) *Sagittal ultrasound shows severe oligohydramnios in the 3rd trimester. Amnioinfusion with blue-tinted warm saline was performed.* ***(Right)*** *Axial transvaginal ultrasound of the same fetus, after amnioinfusion, shows a fetal bladder ➡, ruling out severe bilateral renal anomalies as a cause for the low fluid. In this case, the cause for the oligohydramnios was premature rupture of membranes with slow leak (diagnosed with the amnioinfusion).*

Oligohydramnios

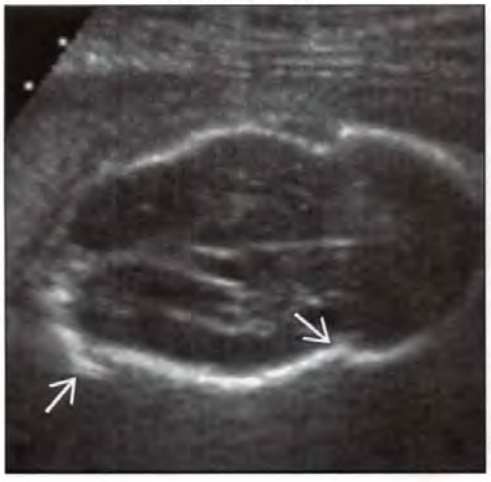


Oligohydramnios



(Left) *Sagittal ultrasound shows the absence of amniotic fluid surrounding a 2nd trimester fetus in cephalic lie ➡. The uterine size is less than expected because of anhydramnios.* ***(Right)*** *Coronal color Doppler ultrasound of the same fetus, after amnioinfusion, shows bilateral renal agenesis. The aorta ➡ is seen well, but no renal arteries arise from it. The final diagnosis in this case was VACTERL association.*

Fetal Demise



Fetal Demise



(Left) *Axial ultrasound of the calvarium of a 30 wk fetus with in utero demise shows oligohydramnios and overriding calvarial sutures ➡, also known as the Spalding sign. The head is misshapen secondary to compressive forces from the uterus. The cause for demise was not determined in this case.* ***(Right)*** *Axial ultrasound shows skin edema (calipers) in a fetus who had immune hydrops and died in utero. Anasarca is a postmortem finding regardless of cause of death.*

UTERINE SIZE > DATES

DIFFERENTIAL DIAGNOSIS

Common
- Macrosomia
- Polyhydramnios
- Multiple Gestations

Less Common
- Leiomyoma

Rare but Important
- Ovarian/Paraovarian Mass

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Fundal height (FH) vs. menstrual age (MA)
  - FH = clinical measurement from pubic symphysis to top of uterus
  - FH in cm = MA in wks
  - FH most accurate at 18-32 wks
  - Ultrasound more accurate than FH

Helpful Clues for Common Diagnoses
- **Macrosomia**
  - Estimated fetal weight > 90th percentile
  - ↑ Abdominal circumference (AC) is hallmark feature
  - Associations
    - Polyhydramnios
    - Diabetes
    - Beckwith-Wiedemann syndrome
- **Polyhydramnios**
  - 2/3 idiopathic
  - 1/3 with maternal or fetal cause
    - Maternal diabetes
    - Fetal gastrointestinal obstruction
    - Abnormal fetal swallowing
    - Abnormal fetal movement
    - Hydrops
- **Multiple Gestations**
  - Determine chorionicity and amnionicity
    - Best in 1st trimester

Helpful Clues for Less Common Diagnoses
- **Leiomyoma**
  - Fibroids grow in first 20 wks of pregnancy
  - Variable appearance
    - Diffusely hypoechoic (most common)
    - Complex, cystic from degeneration
    - Calcified in older patients
  - Complications with retroplacental myoma
    - Abruption
    - Preterm labor
    - Intrauterine growth restriction

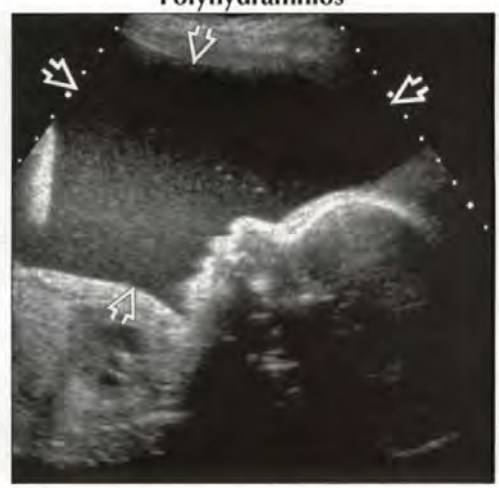
Helpful Clues for Rare Diagnoses
- **Ovarian/Paraovarian Mass**
  - Common
    - Hemorrhagic corpus luteum
    - Teratoma
    - Ovarian hyperstimulation
  - Less common
    - Paraovarian cyst
    - Theca lutein cysts (associated with gestational trophoblastic neoplasia)
    - Ovarian neoplasm
  - Management
    - Surgery if mass is suspicious
    - Optimal time is 14-18 wks
    - Emergent surgery for torsion

Macrosomia



Axial ultrasound shows a 41 week 6 day AC measurement. The uterine fundal height was greater than expected for menstrual age of 38 weeks. Increased AC is a hallmark feature of macrosomia.

Polyhydramnios



Sagittal ultrasound shows excessive amniotic fluid leading to increased uterine size. In this case, the polyhydramnios was from fetal duodenal atresia. The fetal chromosomes were normal.

Multiple Gestations



Multiple Gestations



(Left) *Coronal T2WI MR shows a large uterus ➡ secondary to twins. The twinning is monochorionic and diamniotic; in this case, the MR was performed for another reason.* ***(Right)*** *Sagittal ultrasound during the 1st trimester, in another case of twins, shows dichorionic twinning. Two separate gestational sacs with a thick separating membrane ➡ is seen. The uterine size ➡ was greater than expected for menstrual age, and twins were suspected at clinical exam.*

Leiomyoma

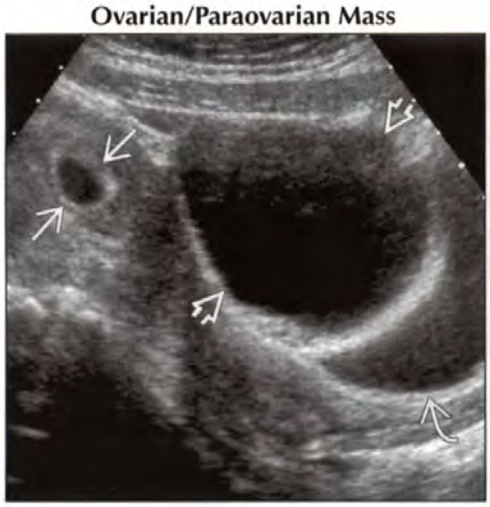


Leiomyoma



(Left) *Sagittal ultrasound in the 1st trimester shows a 6 cm posterior myoma (calipers) that had elevated the uterus ➡ out of the pelvis. On clinical exam, the uterus felt large for menstrual dates.* ***(Right)*** *Sagittal ultrasound of the uterus, in a 2nd trimester case, shows a large myoma ➡ near the fundus of the uterus. The fundal height was greater than expected for menstrual dates. In addition, the fibroid is retroplacental ➡, which confers a greater risk to the pregnancy.*

Ovarian/Paraovarian Mass



Ovarian/Paraovarian Mass



(Left) *Sagittal transabdominal ultrasound shows a 9 cm cyst ➡ that displaces the bladder ➡ in a 1st trimester pregnancy ➡. Subsequently, the patient presented with acute pain, and paraovarian cyst torsion was diagnosed at surgery.* ***(Right)*** *Sagittal transvaginal ultrasound in a 5 week pregnancy shows a malignant pelvic mass extending into the cul-de-sac ➡ with thick septations ➡ and papillary wall nodules ➡.*

ANTEPARTUM HEMORRHAGE

DIFFERENTIAL DIAGNOSIS

Common
- Placental Abruption
- Placenta Previa
- Marginal Sinus Previa

Less Common
- Placenta Accreta Spectrum

Rare but Important
- Vasa Previa

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Defined as vaginal bleeding from 28 weeks gestation until term
  - Many cases are idiopathic, though most likely from placental bed
  - Non-placental causes (e.g., cervicitis, severe monilial infection) should be apparent on clinical evaluation
    - Typically lighter bleeding (e.g., smear of blood-stained mucus)
- Evaluation of placental location is important part of every obstetric scan
- Scan technique important for correct diagnosis
  - Must see placenta and internal os in same image
  - If fetal parts or maternal habitus preclude visualization abdominally, perform transvaginal (TV) scans
    - Most effective method to visualize cervix
  - In patients with ruptured membranes translabial (transperineal) scanning may be helpful
- Scan from side to side and anterior to posterior
  - Lateral wrap placentae may have low-lying/marginal insertion, which is not apparent on a true midline image
  - Use color Doppler to evaluate for vasa previa
- Beware of full bladder, focal myometrial contraction
  - Spurious elongation of "cervix" → incorrect diagnosis of placenta previa
- If low-lying placenta seen at mid-trimester scan, re-image at 32-34 weeks with TV technique
  - 5% of women have a low-lying placenta at 18 weeks
  - ~ 1% of them have placenta previa at term

Helpful Clues for Common Diagnoses
- **Placental Abruption**
  - Abdominal pain, dark red vaginal bleeding
  - Look for hypoechoic blood clot near or behind placenta
  - Marginal
    - Hemorrhage from edge of placenta
    - Can see raised edge in 50%
    - Curvilinear clot near placenta
    - Hemorrhage dissects under chorionic membrane → clot at a distance from placenta, look in front of cervical os
  - Retroplacental
    - Central hematoma between placenta and uterus
    - Large detachment more likely than with marginal abruption → ↑ risk of fetal morbidity
    - Appears acutely as "placentomegaly"
    - Power Doppler delineates clot from placenta
  - Preplacental
    - Hematoma on fetal surface of placenta
    - Clot may compress cord
- **Placenta Previa**
  - Painless, bright red vaginal bleeding
  - Symmetric complete placenta previa
    - Placenta centrally implanted on cervix
    - Will not resolve with advancing pregnancy
  - Asymmetric complete placenta previa
    - Small part of placenta crosses internal os
    - May resolve with advancing pregnancy
    - If > 15 mm crosses internal os then less likely to resolve
  - Marginal: Placental edge within 2 cm of internal os
    - Gestational age > 20 weeks, TVUS done for maximal resolution
  - Low-lying placenta
    - Preferred term if < 20 wks and TVUS not done
    - Asymptomatic, majority resolve by 34 weeks
- **Marginal Sinus Previa**
  - Maternal placental veins at edge of solid placenta, near or cross internal os

- Do not confuse with vasa previa (fetal vessels in membranes anterior to internal os)

Helpful Clues for Less Common Diagnoses

- **Placenta Accreta Spectrum**
  - Abnormal penetration of placental tissue beyond endometrial lining of uterus
  - Look for
    - Loss of subplacental hypoechoic zone
    - Placental sonolucencies ("swiss cheese appearance")
    - Abnormal vascularity of placenta ("tornado vessels")
    - Breach of bladder mucosa seen as defect in echogenic line
    - Use Doppler to look for large vessels in bladder wall
  - Some series claim better resolution with MR
    - May be helpful to look for spread into broad ligament
    - Also helpful if there's history of other surgery that increases risk (e.g., posterior myomectomy)
  - MR technique
    - Fast T2 weighted sequences
    - Full bladder
    - Surgilube in vagina
    - Gadolinium not necessary (contraindicated in pregnancy)
  - Risk factors
    - Reported 5% risk with placenta previa, no prior C-section
    - 10% of patients with > 4 cesarean sections and no previa
    - 67% if previa and 4 prior C-sections
    - Prior uterine instrumentation

Helpful Clues for Rare Diagnoses

- **Vasa Previa**
  - Fetal vessels running in membranes crossing the internal cervical os
  - Risk factors include
    - Placenta previa or low-lying placenta earlier in pregnancy
    - Multiple gestation
    - Succenturiate lobe
    - Low-lying placenta
  - Use transvaginal sonography and Doppler
  - Fetal arterial pulse confirms diagnosis
  - Indication for C-section prior to onset of labor
    - If spontaneous rupture of membranes tears fetal vessels, fetal exsanguination ensues

Other Essential Information

- Placenta Accreta Spectrum
  - Major cause of maternal and neonatal morbidity
  - Maintain high index of suspicion in at-risk patients
- Abruption can be a difficult diagnosis to make in acute stages
  - Negative imaging should not alter clinical management in suspicious cases

Placental Abruption



Axial color Doppler ultrasound shows a subacute marginal abruption with a hypoechoic clot ➡ dissecting beneath the membranes at the placental margin.

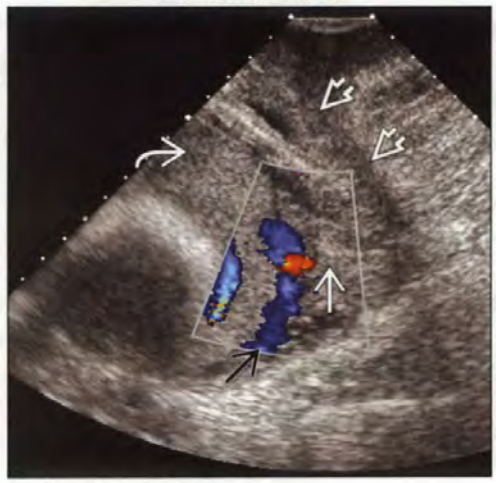
Placental Abruption



Sagittal color Doppler ultrasound shows dissection of subacute clot ➡ to the cervix ➡. Abruption often presents with dark red bleeding rather than bright red, fresh blood seen with placenta previa.

ANTEPARTUM HEMORRHAGE

Placenta Previa



Placenta Previa

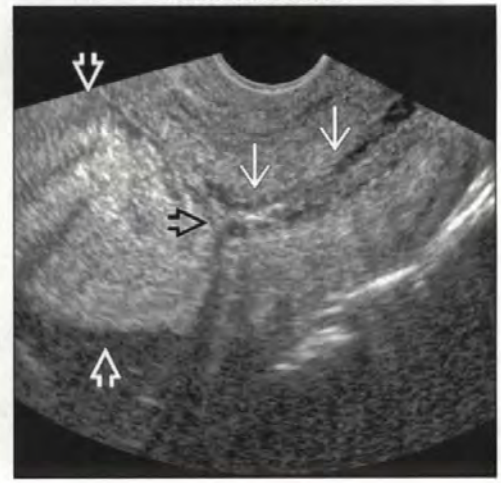


(Left) *Sagittal translabial ultrasound shows placenta and subplacental vessels completely covering the internal cervical os. The coapted vaginal walls provide an acoustic window to the cervix.* ***(Right)*** *Sagittal ultrasound shows an apparent placenta previa covering the cervical canal. A prominent contraction in the lower uterus caused spurious elongation of the cervix. This is an important pitfall, and other sources of bleeding should be sought.*

Placenta Previa



Placenta Previa

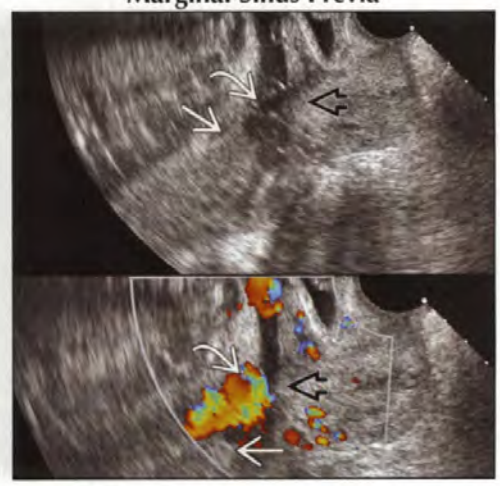


(Left) *Sagittal ultrasound shows an anterior placenta. The endocervical os is not clearly seen, and the apparent cervix seems abnormally long. It is not clear if this is a placenta previa vs. contraction that creates the impression of one.* ***(Right)*** *Sagittal transvaginal ultrasound in the same case clearly shows that the placenta completely covers the internal os, confirming complete placenta previa. The endocervical canal is also seen.*

Placenta Previa



Marginal Sinus Previa

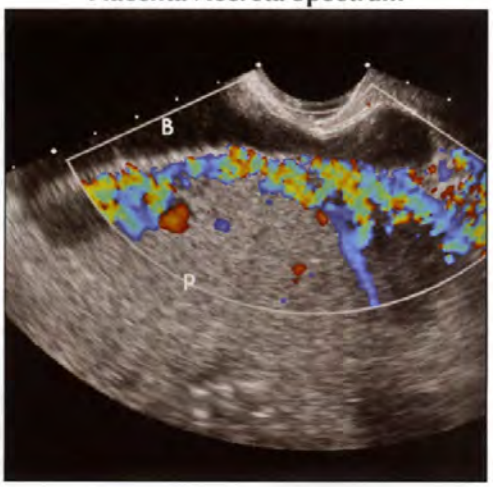


(Left) *Gross pathology shows the typical location of a complete, central placenta previa that completely covers the endocervical os.* ***(Right)*** *Sagittal transvaginal ultrasound images of the cervix show the typical appearance of a marginal sinus previa. The marginal sinus vessels encroach upon the internal os, even though the solid placental edge does not. Color Doppler is very helpful to confirm this diagnosis.*

Placenta Accreta Spectrum



Placenta Accreta Spectrum



(Left) *Transvaginal ultrasound shows an intact echogenic bladder mucosa ➡; however, there is only a tiny rim of hypoechoic tissue ➡ between it and the placenta ➡.* ***(Right)*** *Axial color Doppler ultrasound in the same case shows that the hypoechoic rim is composed of subplacental vessels. The patient had two prior C-sections. At surgery there was placenta increta without bladder invasion.*

Placenta Accreta Spectrum

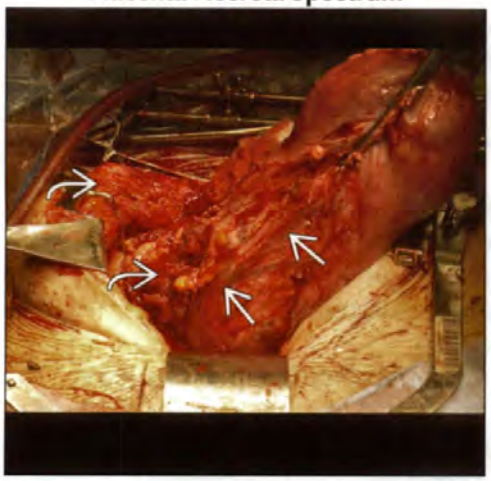


Placenta Accreta Spectrum

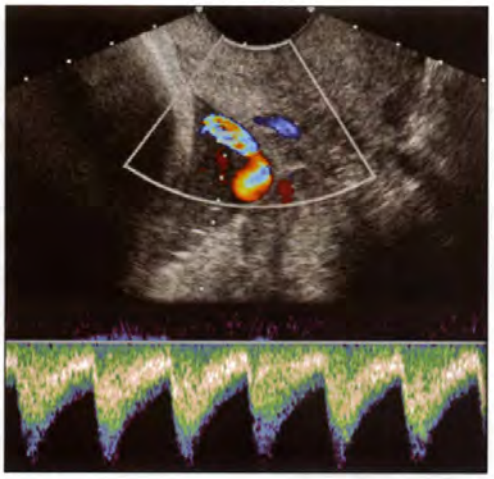


(Left) *Axial ultrasound shows an anterior to right lateral placenta with concern for loss of myometrial rim ➡ on the right. Normal subplacental hypoechoic zone ➡ is seen anteriorly.* ***(Right)*** *Axial T2WI MR in the same case confirms suspicion for placenta percreta. The myometrial signal is absent ➡ along the right lateral uterus, with only a flow void ➡ from a parametrial vessel seen where there should be myometrium.*

Placenta Accreta Spectrum



Vasa Previa



(Left) *Intra-operative photograph, in the same case of placenta percreta, shows placental invasion of the broad ligament ➡ with dilated right-sided uterine vessels ➡. There was also bladder invasion (not shown).* ***(Right)*** *Pulsed Doppler ultrasound shows fetal arterial flow in vessels crossing the internal os, confirming vasa previa. This was associated with a velamentous cord in this case.*

ACUTE ABDOMEN IN PREGNANCY

DIFFERENTIAL DIAGNOSIS

Common
- Placental Abruption
- Appendicitis
- Renal Stone Disease
- Pyelonephritis
- Cholecystitis

Less Common
- Torsion
- Fibroid Degeneration
- Trauma

Rare but Important
- Uterine Rupture
- HELLP Syndrome

ESSENTIAL INFORMATION

Helpful Clues for Common Diagnoses

- **Placental Abruption**
  - Difficult diagnosis to make sonographically: Occult in up to 50% of cases
    - Acute clot is isoechoic to placenta, no flow on Doppler evaluation
    - Clot may be marginal, preplacental, retroplacental
- **Appendicitis**
  - Appendix is displaced out of pelvis by enlarging gravid uterus
  - US
    - Blind ending, non-compressible tube
    - Diameter > 6 mm
    - Look for appendicolith: Echogenic focus with distal acoustic shadowing
    - May see echogenic periappendiceal fat from inflammation
    - May see focal fluid collection if ruptured
    - EV sonography very helpful when appendix drops into pelvis, behind pregnant uterus
  - CT
    - Same anatomic features
    - More sensitive for focal perforation, presence of appendicolith, inflammation of fat
  - MR
    - Increasingly used if US non-diagnostic as no ionizing radiation
    - T1, T2, T2 FS sequences (Gadolinium contraindicated in pregnancy)
    - Same anatomic features as seen with US or CT
- **Renal Stone Disease**
  - Collecting system dilatation
  - Ureteric dilatation, particularly suspicious for stone if dilatation stops abruptly
    - Physiologic dilatation tapers at pelvic brim
  - Look for ureteric jets with color Doppler
    - Have patient in decubitus position with side of concern elevated
  - Measure intrarenal resistive indices (RI)
    - Physiologic caliectasis does not cause elevated RI
    - Look for difference of > 0.1 side-to-side
    - Not specific for renal stone disease, as can also be seen with other other acute renal conditions
  - Coronal MR, with heavily T2 weighted sequences, shows stones as low signal filling defects within column of high signal urine
- **Pyelonephritis**
  - US
    - Enlarged kidney ± parenchymal edema
    - Look for complicating conditions such as abscess or pyonephrosis (an obstructed, infected system), which require drainage
  - CT
    - Delayed ± striated nephrogram
    - Focal areas of diminished enhancement on delayed images
- **Cholecystitis**
  - Gallstones
  - Gallbladder wall thickening
  - Pericholecystic fluid
  - Positive sonographic Murphy sign
  - Remember that right upper quadrant pain and abnormal liver function tests may be seen in preeclampsia

Helpful Clues for Less Common Diagnoses

- **Torsion**
  - Adnexal mass can undergo torsion in pregnancy
  - Maximum risk at 12-14 weeks and immediately postpartum
  - Look for
    - Ovarian or paraovarian mass as lead point
    - Echogenic stroma, ovarian enlargement, peripheral follicles from edema

- Hemorrhage/necrosis/infarction
- Use Doppler to assess flow
  - Lack of venous flow most suspicious finding
  - Documentation of flow does not exclude the diagnosis in presence of strong clinical suspicion or other imaging findings of concern

- **Fibroid Degeneration**
  - Larger fibroids at greater risk for acute red (hemorrhagic) degeneration
    - Severe abdominal pain may mimic abruption
    - Often requires narcotic analgesia for control
  - Inhomogeneous echoes in center of fibroid
  - No flow in inhomogeneous area on Doppler interrogation
  - Placental implantation over large fibroid carries increased risk for abruption
    - Look for changes of abruption, as well as fibroid degeneration, in setting of acute pain
- **Trauma**
  - Imaging evaluation should not be compromised because the patient is pregnant
  - When feasible, limit radiation exposure, or use US/MR if possible
  - Fetus at significant risk even if maternal injuries seem relatively minor
    - Placental shear injury → abruption, infarction
    - Maternal hypotension → decreased placental perfusion
    - Most fetal ischemic injury takes time to be visible on imaging
    - Wait 10-14 days post injury and consider performance of fetal MR to look for intracranial hemorrhage, ischemic encephalopathy

Helpful Clues for Rare Diagnoses

- **Uterine Rupture**
  - Most common in labor, uterus ruptures at site of old hysterotomy site
  - May be a complication of abdominal trauma
  - Look for disrupted myometrium, continuity of extrauterine fluid with endometrial cavity
- **HELLP Syndrome**
  - Patients present with preeclampsia and progress to more severe condition with the following
    - **H**emolysis
    - **E**levated **L**iver function tests
    - **L**ow **P**latelets
  - Low platelets → ↑ risk of spontaneous bleed → subcapsular liver hematoma

Other Essential Information

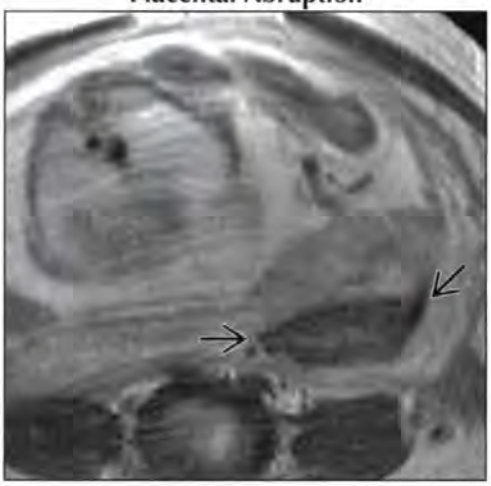
- Fetal well being depends on maternal well being
- Do not compromise evaluation because of pregnancy

Placental Abruption



Sagittal ultrasound at 22 weeks shows a wide open cervix ➡ and extensive intrauterine clot ➡ from an abruption. Preterm labor ensued, complicated by disseminated intravascular coagulopathy.

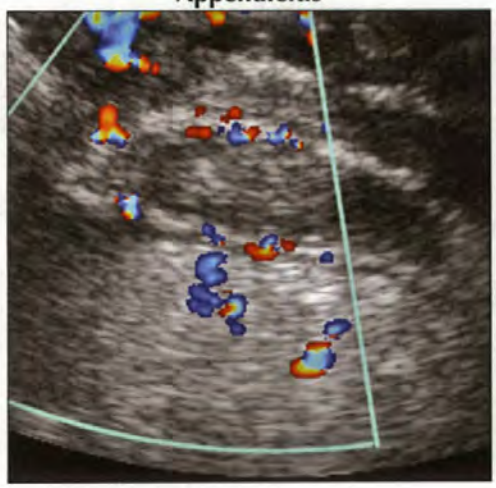
Placental Abruption

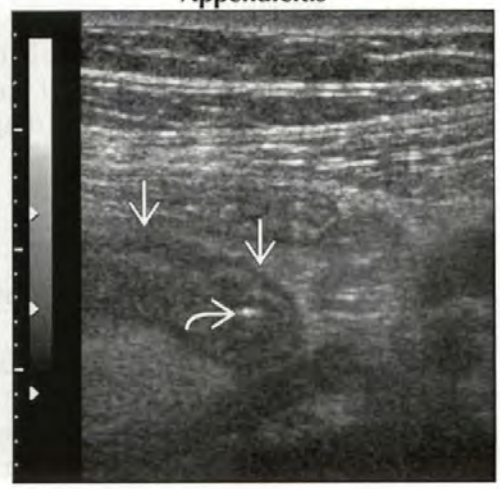


Axial T2WI MR shows a retroplacental abruption ➡ in a patient with pelvic fractures due to a car crash. The MR was performed to confirm sonographic findings of fetal ischemic brain injury.

ACUTE ABDOMEN IN PREGNANCY

Appendicitis





(Left) *Transvaginal (TV) color Doppler image shows a hyperemic, blind-ending, tubular structure surgically confirmed to be an inflamed appendix. TV sonography is helpful if the pain localizes deep in the pelvis.* ***(Right)*** *Ultrasound shows the typical appearance of appendicitis using a high frequency linear transducer at the site of maximum tenderness. This was a non-compressible, blind-ending, tubular structure ➡ that contained an appendicolith ➡.*

Renal Stone Disease



Pyelonephritis

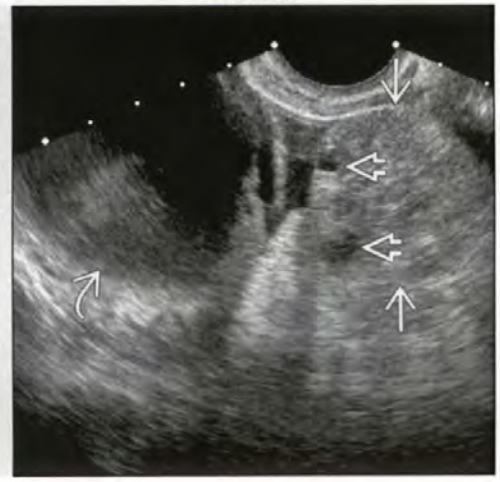


(Left) *Coronal oblique ultrasound shows an echogenic focus ➡ in the lower pole of the right kidney in a patient with pain suggestive of renal colic. Subsequent images (not shown) confirmed a stone obstructing the dilated ureter ➡.* ***(Right)*** *Ultrasound shows edema and loss of corticomedullary differentiation in the transplant kidney ➡. This patient was admitted with systemic CMV infection, as well as bacterial pyelonephritis.*

Cholecystitis



Torsion



(Left) *Ultrasound shows gallstones ➡, gallbladder wall thickening ➡, and pericholecystic fluid ➡ in a patient with a positive sonographic Murphy sign, indicating acute cholecystitis.* ***(Right)*** *Transvaginal ultrasound shows a paraovarian cyst ➡ beside an enlarged left ovary ➡ with no detectable flow on Doppler. Multiple peripherally located follicles ➡ are a sign of torsion. Pathology showed adnexal torsion with a paramesonephric duct cyst.*

Fibroid Degeneration

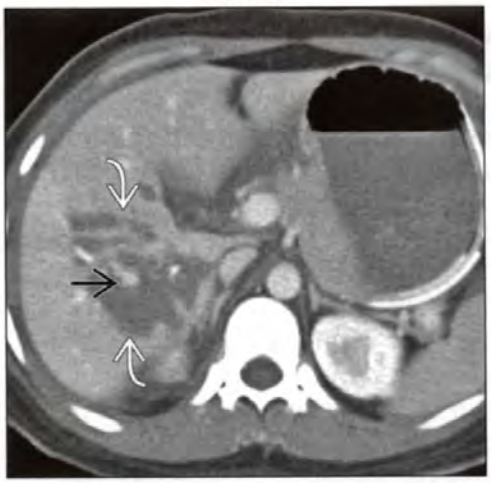


Fibroid Degeneration

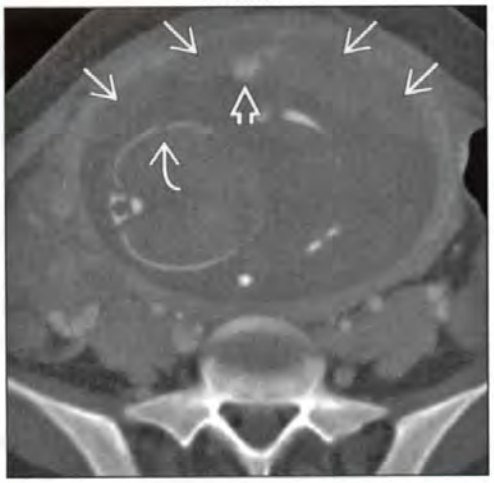


(Left) *Ultrasound shows mixed echogenicity in a transmural fibroid (calipers) in the second trimester. The patient was focally tender over the fibroid and required narcotics for pain control.* ***(Right)*** *Ultrasound shows the fetus helpfully pointing to the cause of its mother's pain! This patient had multiple admissions to the hospital with severe abdominal pain that localized to this fibroid* ➔.

Trauma

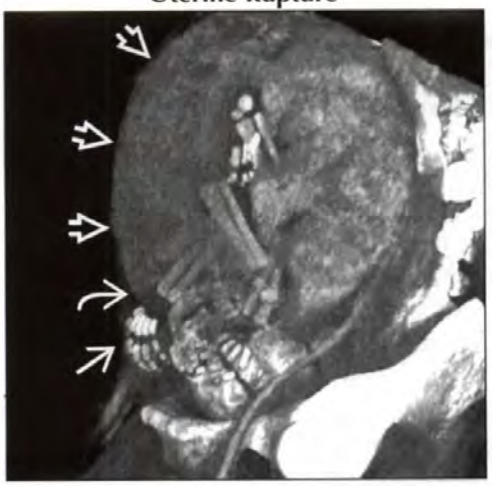


Trauma

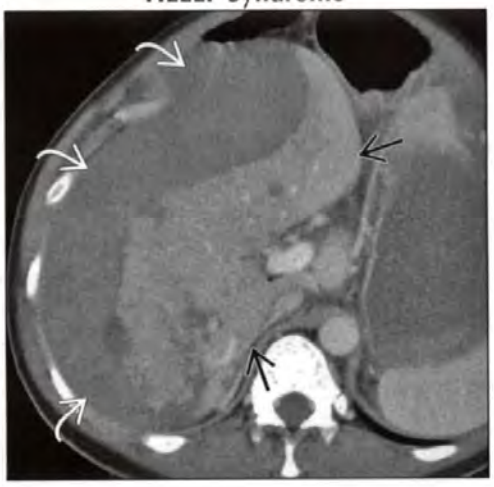


(Left) *Axial CECT shows a large liver laceration* ➔ *with active extravasation of contrast* ➔ *in a pregnant patient involved in a high speed motor vehicle crash.* ***(Right)*** *Axial CECT shows a fetal rib fracture* ➔ *and large areas of nonenhancing placenta* ➔, *with an area of active extravasation* ➔ *in the placental bed. The fetus was dead on arrival. The mother survived severe solid organ injuries.*

Uterine Rupture



HELLP Syndrome



(Left) *Sagittal oblique CECT shows the fetal hand* ➔ *extending through a defect* ➔ *in the anterior myometrium* ➔ *at the site of prior hysterotomies. The patient had had eight prior C-sections.* ***(Right)*** *Axial CECT in a woman with HELLP syndrome shows a dramatic subcapsular hematoma* ➔ *exerting mass effect on the liver* ➔. *Spontaneous subcapsular bleeding is attributed to the low platelet count in this condition.*

ELEVATED MSAFP

DIFFERENTIAL DIAGNOSIS

Common
- Open Neural Tube Defect
  - Spina Bifida
  - Exencephaly, Anencephaly
  - Encephalocele
- Abdominal Wall Defect
  - Gastroschisis
  - Omphalocele
  - Body Stalk Anomaly
- Multiple Gestations

Less Common
- Perigestational Hemorrhage
- Fetal Demise
- Placental Insufficiency

Rare but Important
- Maternal Tumors

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Alpha fetoprotein (AFP)
  - Normally produced from several sources in developing fetus
    - Yolk sac
    - Fetal gastrointestinal tract
    - Fetal liver
  - Circulation of AFP
    - Fetus ⇒ amniotic fluid ⇒ maternal serum
- Maternal serum alpha fetoprotein (MSAFP) test
  - Drawn at 15-20 weeks menstrual age
    - Part of genetic screening
    - Screening for anomalies (spina bifida)
  - Adjusted for maternal factors
    - Weight
    - ± Diabetes
    - Ethnicity
  - MSAFP results
    - Reported as multiples of the median (MOM)
    - > 2-2.5 MOM considered positive screen
- Reasons for positive MSAFP screen results
  - Incorrect menstrual dates (20%)
    - Recalculate result after ultrasound
  - Unsuspected multiple gestations
    - Twin MSAFP = approximately twice singleton values
    - Use of MSAFP screening in known multiple gestation is controversial
  - Fetal anomalies
    - Any anomaly with ↑ fetal internal organ exposure to amniotic fluid
  - Placental abnormalities
    - Primarily from fetal-placental hemorrhage
    - Fetal levels 100,000 times > maternal
    - Small hemorrhage easily causes ↑ MSAFP

Helpful Clues for Common Diagnoses
- **Open Neural Tube Defect**
  - **Spina Bifida**
    - Bony vertebral defect + neural content exposure to amniotic fluid
    - 73% lumbar
    - 17% sacral
    - 9% thoracic
    - 1% cervical
    - 80% with overlying sac
    - Almost all with Chiari II malformation of brain
    - 14% with aneuploidy
  - **Exencephaly, Anencephaly**
    - Absent calvarial vault
    - Variable amount of supratentorial brain
    - Brain exposed to amniotic fluid, which erodes neural tissue and leaves angiomatous stroma
    - Face: Proptotic eyes, cleft lip/palate
  - **Encephalocele**
    - Herniation of intracranial structures through skull defect
    - Occipital most common
    - Parietal, vertex, frontal more rare
    - Associations: Trisomy 13, trisomy 18, Meckel-Gruber syndrome
    - Look carefully for other anomalies
- **Abdominal Wall Defect**
  - **Gastroschisis**
    - Bowel herniation through right paramedian defect
    - Normal cord insertion
    - No covering membrane
    - Progressive bowel dilatation often seen
    - Intrauterine growth restriction (IUGR) common in 3rd trimester
    - Almost all with ↑ MSAFP
  - **Omphalocele**
    - Herniation of abdominal contents into base of umbilical cord
    - Midline abdominal wall defect
    - Most often involves liver

- Covering membrane present (difficult to see early, without ascites)
- 70% with ↑ MSAFP (30% normal MSAFP)
- 30% with aneuploidy
- Omphaloceles that contain only bowel more associated with aneuploidy

- **Body Stalk Anomaly**
  - Fetal evisceration
  - Visceral organs attached to placenta
  - Short or absent umbilical cord
  - No covering membrane
  - Scoliosis is prominent feature
  - Amniotic bands seen in 40%
- Other abdominal wall defect diagnoses
  - Pentalogy of Cantrell
  - OEIS syndrome
  - Bladder/cloacal exstrophy

- **Multiple Gestations**
  - Determine chorionicity/amnionicity
  - Dichorionic diamniotic twins
    - Most common type of twins
    - 2 placentae
    - "Twin peak" if adjacent placentae
    - Thick separating membrane
  - Monochorionic diamniotic twins
    - Single placenta
    - No "twin peak" sign
    - Thin separating membrane
    - Twins are same gender
  - Monochorionic monoamniotic twins
    - Single placenta
    - No separating membrane
    - Rarely conjoined

Helpful Clues for Less Common Diagnoses

- **Perigestational Hemorrhage**
  - Placental detachment
    - Blood primarily accumulates in subchorionic space
    - Bleed into placenta or amniotic fluid also occurs
    - Mix with maternal blood ⇒ ↑ MSAFP
  - Hematoma appearance is variable
    - Initially isoechoic to placenta
    - Becomes more hypoechoic with time
    - Eventually anechoic and resolves
  - Often no hematoma seen at time of ↑ MSAFP or screening ultrasound
- **Fetal Demise**
  - Tissue break down causes ↑ MSAFP
  - Look for anomalies to explain demise
- **Placental Insufficiency**
  - Unexplained ↑ MSAFP may present with IUGR, likely from subclinical placental hemorrhage
  - Follow-up for growth, fluid, and Doppler
    - IUGR often late onset
    - Associated with oligohydramnios

Helpful Clues for Rare Diagnoses

- **Maternal Tumors**
  - Yolk sac ovarian tumor
  - Hepatocellular carcinoma

Other Essential Information

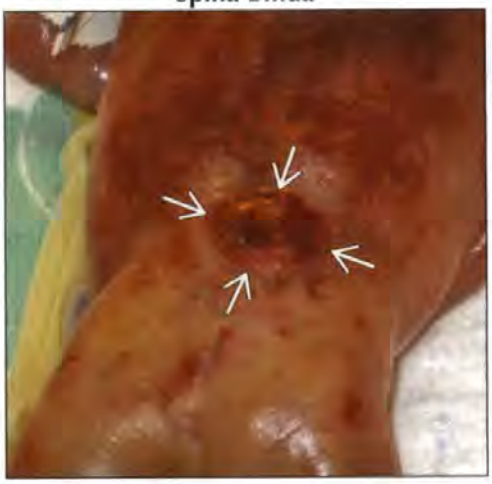
- ↑ MSAFP and amniocentesis
  - Not routinely performed
  - Anomalies associated with ↑ MSAFP are seen well with ultrasound

Spina Bifida



Sagittal T2WI MR shows a meningomyelocele ➡ involving the lumbo-sacral spine. Exposure of fetal neural tissue to amniotic fluid results in elevated levels of AFP in the amniotic fluid and maternal serum.

Spina Bifida



Gross pathology of a second trimester fetus with myeloschisis shows the open neural tube defect ➡. In this case, there is no meningomyelocele sac.

ELEVATED MSAFP

Exencephaly, Anencephaly



(Left) *3D ultrasound shows anencephaly in a 17 week fetus. The orbits are proptotic, and there is no calvarium superior to the face. The 3D reconstruction image shows brain tissue in direct contact with amniotic fluid.* ***(Right)*** *Axial transvaginal ultrasound of another second trimester fetus shows an occipital calvarial defect and herniation of a large amount of brain and meninges through the defect.*

Encephalocele

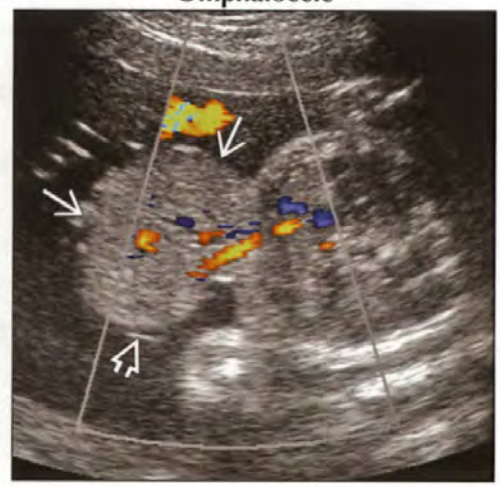


Gastroschisis



(Left) *Coronal T2WI MR performed to evaluate a uterine septum shows free floating bowel in a fetus with known gastroschisis. MR nicely demonstrates the direct contact between fetal bowel and amniotic fluid, leading to ↑ MSAFP levels.* ***(Right)*** *Axial color Doppler ultrasound in another case shows a large median abdominal wall defect and extra-corporeal liver. A small portion of the covering membrane is seen. This fetus had trisomy 18.*

Omphalocele



Body Stalk Anomaly



(Left) *Coronal ultrasound shows fetal evisceration. The bowel is outside of the body, and an amniotic band is seen. The fetal anatomy was difficult to assess as most of the intra-abdominal and chest contents were extracorporeal.* ***(Right)*** *Axial ultrasound of the same case shows the fetus intimately associated with the placenta. In addition, the umbilical cord is short and "clumped". The fetus also had scoliosis and extremity defects.*

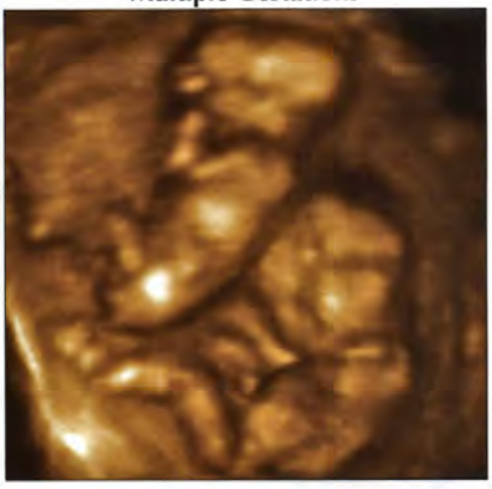
Body Stalk Anomaly



Multiple Gestations

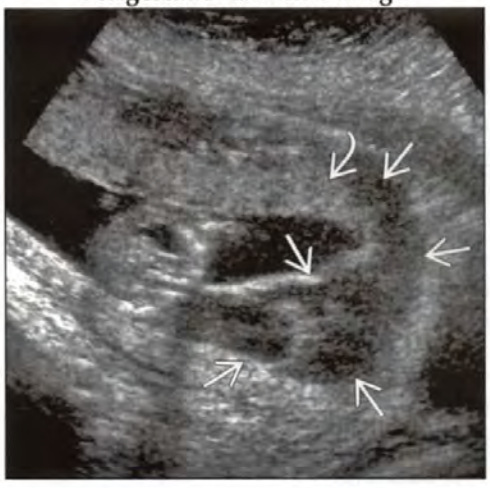


Multiple Gestations



(Left) *3D ultrasound shows dichorionic diamniotic twins with a thick separating membrane* ➡. ***(Right)*** *3D ultrasound shows monochorionic diamniotic twins. A thin separating membrane was seen with 2D ultrasound but was not resolved with 3D ultrasound. Unsuspected twins account for 10% of ↑ MSAFP cases.*

Perigestational Hemorrhage



Perigestational Hemorrhage

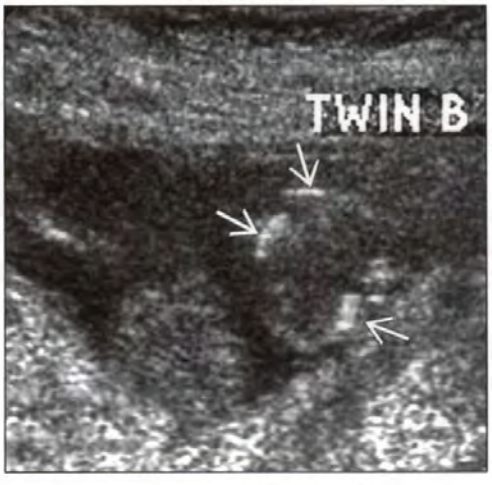


(Left) *Sagittal ultrasound shows detachment of the inferior placenta* ➡ *and a large perigestational bleed* ➡. *Subchorionic blood can mix with maternal blood and lead to ↑ MSAFP levels.* ***(Right)*** *Sagittal ultrasound shows a large perigestational hemorrhage* ➡ *with a fluid-fluid level* ➡. *A second smaller hemorrhage* ➡ *is also seen. Despite such impressive placental hemorrhage, the pregnancy advanced normally.*

Fetal Demise



Fetal Demise



(Left) *Sagittal transvaginal ultrasound shows a liver-containing omphalocele* ➡ *in a second trimester fetus presenting with ↑ MSAFP and fetal demise. A search for fetal anomalies is recommended, if possible, when demise is diagnosed. Chromosome analysis in this case was unsuccessful.* ***(Right)*** *Axial ultrasound shows overlapping calvarial sutures* ➡, *the Spalding sign, in a twin case complicated by demise of twin B.*

ABNORMAL BETA HCG

DIFFERENTIAL DIAGNOSIS

Common
- Imprecise Menstrual Dates
- Multiple Gestations
- Ectopic Pregnancy
- Retained Products of Conception (RPOC)

Less Common
- Complete Hydatidiform Mole
- Trisomy 21
- Trisomy 18

Rare but Important
- Invasive Mole
- Choriocarcinoma
- Embryonic/Fetal Demise

ESSENTIAL INFORMATION

Helpful Clues for Common Diagnoses

- **Imprecise Menstrual Dates**
  - Maternal serum screening provides numerical risk for certain conditions based on expected levels of certain hormones
    - Levels reported as multiples of the median (MOM) for gestational age
    - Incorrect menstrual data → incorrect gestational age assessment at time of blood draw → spurious ↑ or ↓ of levels
  - All patients with abnormal serum screen should have ultrasound as next step
    - Verify gestational age
    - Look for sonographic signs of aneuploidy
  - If dates are incorrect, risk often recalculated or test redrawn
- **Multiple Gestations**
  - Maternal serum hormone levels are higher in multiple than singletons
    - Greater amount of fetal tissue per gestational age week
- **Ectopic Pregnancy**
  - Ectopic pregnancies are not normal pregnancies
    - Beta hCG often lower than for corresponding gestational age (GA) in a normal early pregnancy
    - Normal "doubling" of beta hCG every 48 hours in early pregnancy may not occur
  - Tubal ectopic is commonest type
    - No intrauterine pregnancy + tubal mass + echogenic cul-de-sac fluid
    - Echogenic fluid in a pregnant patient has a 90% positive predictive value for ectopic pregnancy
  - Look for ring-like mass separate from ovary
    - May have yolk sac or embryonic pole with or without cardiac activity
  - "Ring of fire": Increased flow seen in trophoblastic tissue around ectopic
    - Do not confuse with increased flow around corpus luteum
  - Amorphous adnexal mass may be due to hematosalpinx or clot adherent to ruptured tube
  - Pseudogestational sac sign
    - Decidual cast surrounds fluid collection central in endometrial cavity
    - Lacks double decidual sac sign of normal intrauterine pregnancy
- **Retained Products of Conception (RPOC)**
  - Residual trophoblastic tissue continues to produce beta hCG
    - Levels do not fall to 0 as expected
    - If levels remain elevated > 4 months post delivery, consider persistent gestational trophoblastic neoplasia
  - Solid, heterogeneous, echogenic mass in endometrial cavity
  - Persistent, thickened endometrium
  - Irregular interface between endometrium and myometrium
  - May see high velocity, low resistance flow

Helpful Clues for Less Common Diagnoses

- **Complete Hydatidiform Mole**
  - No embryo or fetus
  - Beta HCG markedly elevated
  - Enlarged uterus with "swiss cheese" endometrium
    - Heterogeneous, echogenic intrauterine mass
    - Completely fills uterine cavity
    - Individual cysts that vary in size can be seen
    - Mass is vascular with high velocity, low impedance flow (mean resistive index of 0.55)
  - Bilateral, complex theca lutein ovarian cysts in 25-50%
- **Trisomy 21**
  - High beta hCG associated with low unconjugated estriol, alpha fetoprotein
  - First trimester ultrasound findings

- Increased nuchal translucency, absent nasal bone, abnormal ductus venosus flow
- Second trimester ultrasound findings
  - Increased nuchal fold, short femur/humerus, duodenal atresia, atrioventricular septal defect
  - More minor finding include echogenic bowel, mild pyelectasis, mild ventriculomegaly, echogenic intracardiac focus

- **Trisomy 18**
  - Low beta hCG associated with low unconjugated estriol, alpha fetoprotein
  - Multiple anomalies, intrauterine growth restriction

Helpful Clues for Rare Diagnoses

- **Invasive Mole**
  - Echogenic, cystic mass filling uterus and invading into myometrium
  - Foci of invasive tumor "light up" with color Doppler
  - MR
    - Heterogeneous, intermediate signal mass
    - Disruption of uterine zonal anatomy on T2WI
    - Tumor enhances with gadolinium
- **Choriocarcinoma**
  - Malignant gestational trophoblastic neoplasia
    - Distinguished from invasive mole by its tendency to metastasize
  - Uterine findings quite variable
    - May be no detectable uterine mass
  - Bilateral ovarian theca lutein cysts
  - Metastatic disease patterns
    - Lung, brain, liver metastases common, often large and heterogeneous
    - Brain metastases often hemorrhagic
    - Irregular pulmonary nodules with surrounding halo of ground glass secondary to hemorrhage
  - Absence of villous pattern on histopathology, in contrast to complete hydatidiform or invasive mole
- **Embryonic/Fetal Demise**
  - Failed first trimester pregnancy associated with low beta hCG levels for GA or inadequate increase in levels
  - Look for failure to meet normal early pregnancy milestones on transvaginal ultrasound
    - Yolk sac must be seen when mean sac diameter (MSD) > 10 mm
    - Embryo must be seen when MSD > 18 mm
    - Cardiac activity must be seen when embryo > 5 mm
  - Look for other signs of failed/failing pregnancy
    - Empty amnion
    - Poor decidual reaction
    - Abnormal sac shape or size
    - Size smaller than expected
    - Less than expected interval growth
    - Embryonic bradycardia

Imprecise Menstrual Dates



Axial transvaginal ultrasound shows a small gestational sac with a yolk sac ➡ and embryo ➡. The patient had irregular cycles, and the hCG was less than expected for LMP, but concordant with sac size.

Multiple Gestations



Axial oblique ultrasound shows dichorionic twins with one gestational sac ➡ in each horn ➡ in a patient with uterus didelphys.

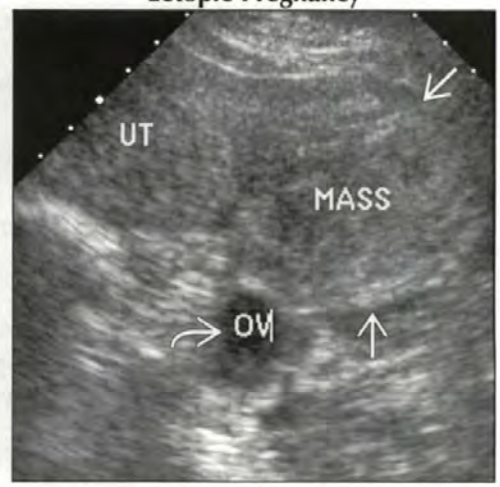
ABNORMAL BETA HCG

Multiple Gestations

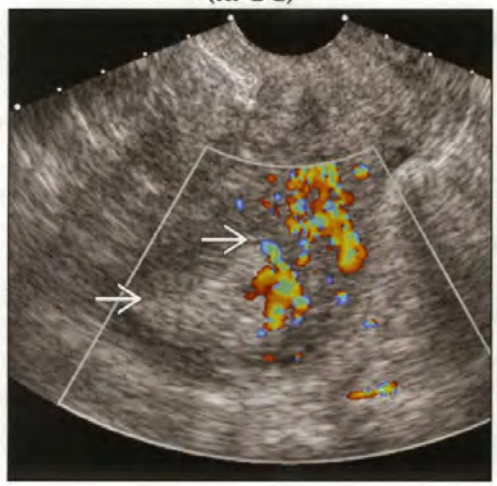


(Left) *Sagittal ultrasound shows trichorionic triplets, with demise of the embryo in the smallest sac. Beta hCG is greater than expected in multiple gestations.* ***(Right)*** *Transvaginal ultrasound shows an amorphous adnexal mass adjacent to the uterus (UT) and ovary (OV). The ovary contains a simple cyst, likely a corpus luteum cyst. In ectopic pregnancy, the HCG is usually lower than expected and does not double appropriately.*

Ectopic Pregnancy

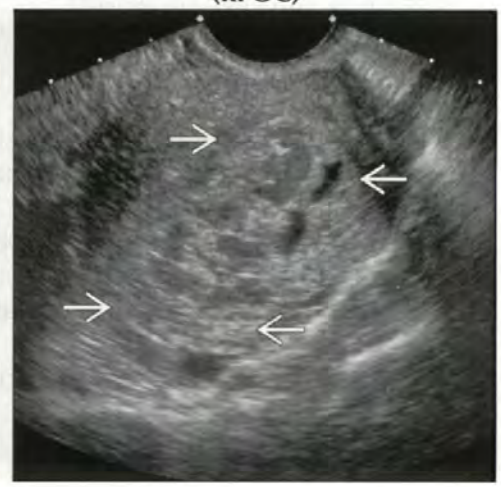


Retained Products of Conception (RPOC)



(Left) *Sagittal oblique color Doppler ultrasound shows hypervascular, echogenic material in the uterus, typical for RPOC. Lack of flow, however, does not exclude the diagnosis.* ***(Right)*** *Transvaginal ultrasound in a postpartum woman with bleeding shows a large amount of mixed echogenicity material in the uterus. Dilatation and curettage confirmed RPOC. Trophoblastic tissue will continue to produce HCG even after delivery of the infant.*

Retained Products of Conception (RPOC)

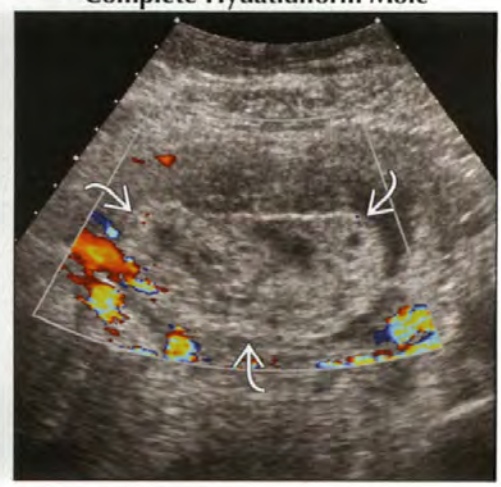


Complete Hydatidiform Mole



(Left) *Axial ultrasound shows a multicystic mass completely filling the uterus (calipers). The patient presented with vaginal bleeding and had markedly elevated levels of beta hCG, typical for complete hydatiform mole.* ***(Right)*** *Axial color Doppler ultrasound shows increased flow in the surrounding myometrium. The mass is well circumscribed, with no features to suggest myometrial invasion.*

Complete Hydatidiform Mole



ABNORMAL BETA HCG

Trisomy 21



Trisomy 18

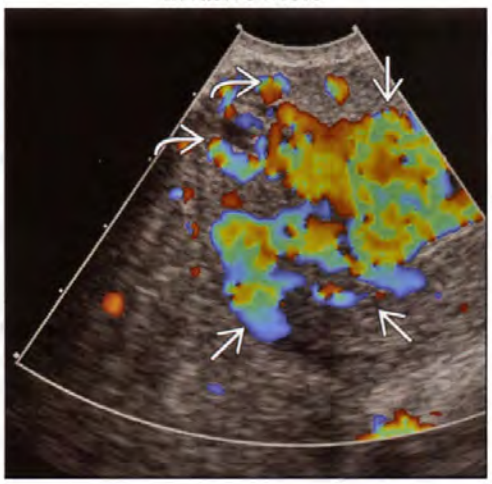


(Left) *Sagittal ultrasound shows an increased nuchal translucency measuring 3.2 mm in a patient with high beta hCG, associated with low unconjugated estriol and alpha fetoprotein. Trisomy 21 was confirmed by chorionic villous sampling.* ***(Right)*** *Axial ultrasound shows a "strawberry-shaped" head in a fetus with low beta HCG, associated with low unconjugated estriol and alpha fetoprotein. Amniocentesis confirmed trisomy 18.*

Trisomy 18

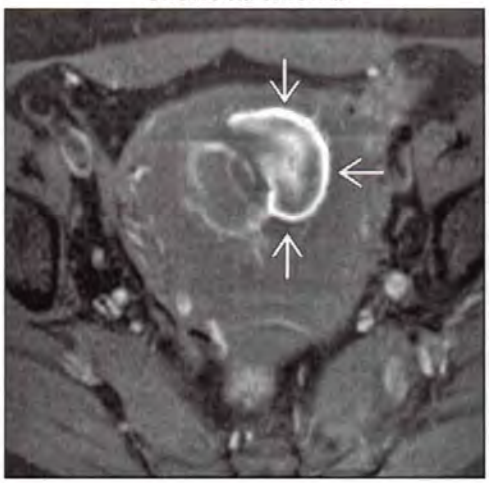


Invasive Mole

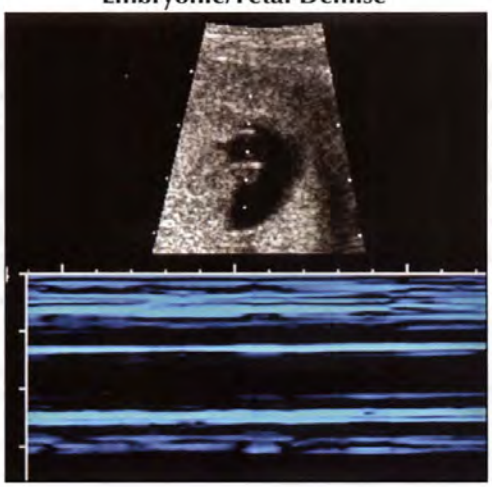


(Left) *Ultrasound with the corresponding clinical photograph shows a clenched hand with overriding second finger ⇨, a characteristic finding in trisomy 18.* ***(Right)*** *Transvaginal ultrasound with color Doppler shows a hypervascular mass ⇨ filling the uterus and invading the myometrium ⇨. The patient had persistently elevated beta hCG following a spontaneous abortion.*

Choriocarcinoma



Embryonic/Fetal Demise



(Left) *Axial T1WI MR with fat-saturation post-gadolinium administration shows intense enhancement around an intrauterine soft tissue mass ⇨. The patient had an elevated beta hCG and lung metastases.* ***(Right)*** *M-mode ultrasound shows demise with loss of cardiac activity in an embryo that had been seen alive on an earlier scan. Failure of beta hCG levels to double every 48 hours in early pregnancy is associated with poor outcomes, including demise.*

CESAREAN SECTION COMPLICATIONS

DIFFERENTIAL DIAGNOSIS

Common

- During Pregnancy
  - Placenta Accreta Spectrum
  - Dehiscence
  - Uterine Rupture
- Postpartum
  - Bladder Flap Hematoma
  - Endometritis
  - Normal Cesarean Section Scar (Mimic)

Less Common

- Uterine Rupture, Delayed
- Cesarean Section Scar Ectopic
- Endometrioma in Cesarean Section Scar

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues

- During pregnancy
  - Determine history of prior cesarean section (C-section)
  - Visualize lower uterine segment
    - Confirm hypoechoic myometrial layer present
    - Risk of rupture/dehiscence increases as lower uterine thickness decreases
    - Myometrium absent if uterine rupture/dehiscence has occurred
  - Document placental location
    - Placenta previa associated with accreta spectrum in 5% of cases
    - Take care to show preservation of subplacental hypoechoic zone if implantation on the C-section scar
    - Use Doppler liberally to assess for abnormal vascularity
- Postpartum
  - Immediate postoperative complications often difficult to differentiate due to gas and edema in soft tissues from delivery
  - Healed C-section scar normally associated with focal mild thinning of myometrium

Helpful Clues for Common Diagnoses

- **Placenta Accreta Spectrum**
  - Abnormal penetration of placental tissue beyond endometrial lining of uterus
    - Accreta: Attached to myometrium without muscular invasion
    - Increta: Chorionic villi invading myometrium
    - Percreta: Penetration of chorionic villi through uterine wall
  - Loss of subplacental hypoechoic zone
    - Should normally be present over entire placental surface
    - Hypoechoic zone may be difficult to see when placenta is anterior
    - Switch to high resolution transducer
  - Placenta previa present in almost all cases
  - Thinning of underlying myometrium ≤ 2 mm can be a sign of placental invasion
  - "Swiss cheese" placenta
    - Multiple hypoechoic placental vascular lacunae ("tornado-shaped" vessels)
    - Better positive predictive value than loss of subplacental hypoechoic zone
  - Interruption of bladder wall-uterine interface
    - Normal bladder mucosa is echogenic
    - Large vessels, or nodularity, extending through bladder wall can be seen with percreta
- **Dehiscence**
  - Incomplete rupture: Disrupted myometrium but intact serosa
    - Loss of hypoechoic myometrial layer
    - Usually at site of prior C-section
  - In pregnancy, patient may be followed carefully to allow fetus to mature
    - Delivery indicated for any sudden pain
    - Reports of successful repair and continuation of pregnancy
  - MR may be helpful to assess for location and size of dehiscence
- **Uterine Rupture**
  - Full thickness tear of uterine wall
    - May occur during pregnancy, labor, or puerperium
    - Highest risk in patients with history of C-section
  - Focal discontinuity of the myometrium at rupture site
    - Usually anterior lower uterine segment or site of prior myomectomy
    - Fetal parts and fluid in peritoneal cavity
  - During labor, imaging rarely performed as patient is emergently delivered operatively
    - Simultaneous maternal-fetal distress
    - Risk of rupture during spontaneous labor with prior C-section ≈ 5/1,000

- In setting of trauma, CT useful → look for maternal solid organ injury also

- **Bladder Flap Hematoma**
  - Hypoechoic clot at site of C-section on serosal surface
    - Between lower uterine segment and bladder wall
- **Endometritis**
  - Most commonly occurs after delivery or termination
    - Associated with postpartum fever and pelvic pain
  - Endometrium may appear normal or have nonspecific findings
    - Thickened, heterogeneous endometrium
    - Echogenic foci of gas in endometrial cavity can be seen but also commonly related to delivery
    - Large amount of echogenic fluid concerning for pyometra
  - Retained products of conception (RPOC) is a risk factor
    - Sonographic appearance of RPOC overlaps with endometritis
    - Use Doppler to check for vascularity within endometrial contents
- **Normal Cesarean Section Scar (Mimic)**
  - Focal triangular or wedge-shaped defect in myometrium
    - Anterior lower uterine segment
  - Small amount of fluid in scar is normal
    - Cystic-appearing C-section scar
    - May be confused with ectopic gestational sac in early pregnancy
    - Doppler ultrasound shows an avascular collection of fluid

Helpful Clues for Less Common Diagnoses

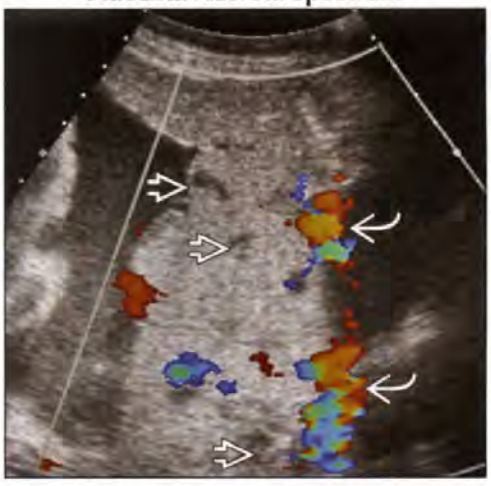
- **Uterine Rupture, Delayed**
  - Postpartum full thickness tear of uterine wall
    - Following delivery, especially if patient had vaginal birth after prior C-section
    - Due to incompletely treated endometritis
    - After uterine instrumentation with a chronic healed scar
  - Myometrial defect can be seen with ultrasound, CT, or MR
    - If the patient presents with an acute abdomen, CT may be initial imaging modality to exclude other etiologies
  - Look for free intraperitoneal fluid or hemorrhage
- **Cesarean Section Scar Ectopic**
  - Eccentric sac within the anterior myometrium at site of C-section scar
    - Minimal or no myometrium between sac and bladder
    - Empty uterine cavity
  - Color Doppler shows marked peritrophoblastic flow around ectopic sac
  - High risk for uterine rupture and massive bleeding if untreated
- **Endometrioma in Cesarean Section Scar**
  - Homogeneous, hypoechoic mass in C-section scar
  - History of endometriosis

Placenta Accreta Spectrum



Gross pathology shows a cesarean hysterectomy specimen with placenta ➡ extending through the myometrium ➡ in the area of prior C-section.

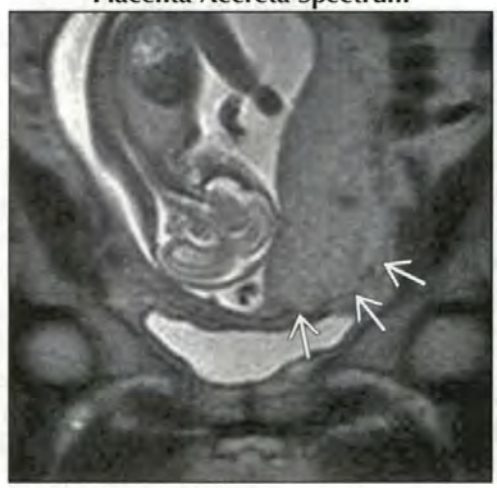
Placenta Accreta Spectrum



Sagittal color Doppler ultrasound shows no myometrium deep to the placenta in a patient with a prior C-section. Note lucent areas ➡ and large vessels ➡ extending into the bladder wall.

CESAREAN SECTION COMPLICATIONS

Placenta Accreta Spectrum

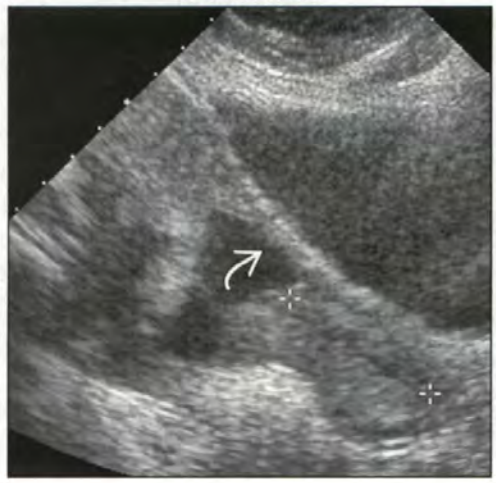


(Left) *Coronal T2WI MR shows a partial placental implantation on the C-section scar toward the left. No visible myometrium is seen in this area. Placenta increta was seen on histology at time of cesarean hysterectomy.* ***(Right)*** *Coronal T2WI MR shows a 34 week fetus with scar tissue near the site of prior C-section. Toward the left, there is loss of the already thinned myometrium, suspicious for a uterine window. The bladder is deviated leftward.*

Dehiscence

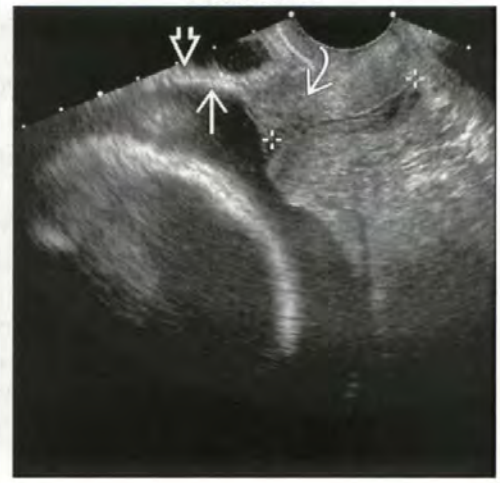


Dehiscence

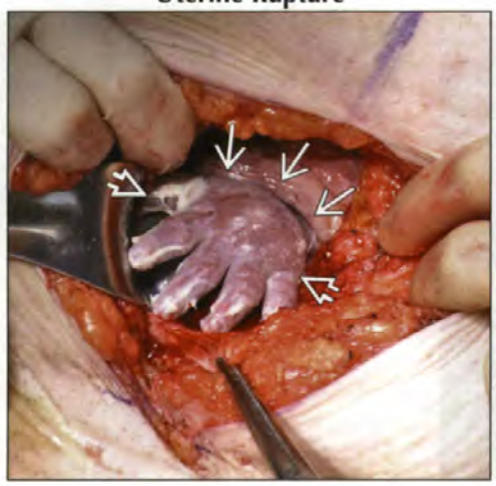


(Left) *Sagittal transabdominal ultrasound shows thin or absent anterior myometrium in the lower uterine segment. The cervix is marked by calipers.* ***(Right)*** *Sagittal transvaginal ultrasound of the same patient shows that the uterine serosa and bladder mucosa form 2 echogenic lines. There is discontinuity of the anterior lip of the cervix with the lower uterine segment myometrium. Calipers denote the endocervical canal.*

Dehiscence



Uterine Rupture



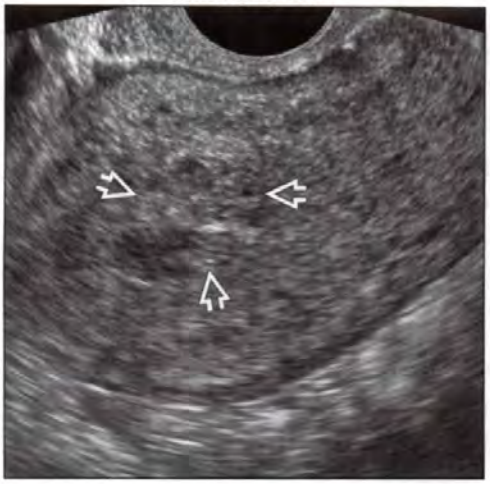
(Left) *Intra-operative photograph shows the fetal hand extending out of a ruptured C-section scar. No hysterotomy had been made; the hand had spontaneously poked through the rupture site.* ***(Right)*** *Sagittal transvaginal ultrasound shows clot in the uterine cavity following C-section delivery. Adjacent to the incision site, there is an external hypoechoic hematoma (calipers). The bladder is partially seen.*

Bladder Flap Hematoma



CESAREAN SECTION COMPLICATIONS

Endometritis



Normal Cesarean Section Scar (Mimic)

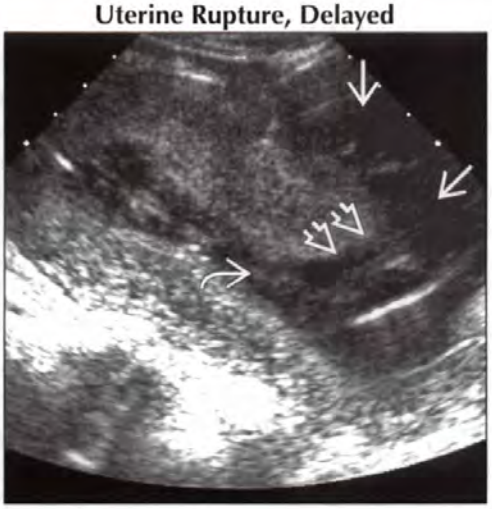


(Left) *Sagittal ultrasound of a patient with clinical endometritis shows a distended endometrial cavity filled with heterogeneously echogenic debris. Endometritis can be a complication after either vaginal or C-section delivery.* ***(Right)*** *Sagittal ultrasound shows the typical appearance of a healed C-section scar in the lower uterine segment. There is tenting of the endometrial cavity at the scar, with a focal narrowing of the myometrium.*

Normal Cesarean Section Scar (Mimic)



Uterine Rupture, Delayed



(Left) *Sagittal T2WI MR shows a healed C-section scar with a contained fluid collection. This is often seen incidentally when imaging the uterus postpartum & does not indicate dehiscence. It should not be confused with a nabothian cyst, which occurs in the cervix.* ***(Right)*** *Sagittal ultrasound shows a myometrial defect in the anterior lower uterine segment. Clot extends anterior to the uterus, communicating freely with the endometrial cavity.*

Cesarean Section Scar Ectopic



Endometrioma in Cesarean Section Scar



(Left) *Sagittal ultrasound shows a gestational sac implanted within a C-section scar. The decidualized endometrium is seen centrally within the uterus.* ***(Right)*** *Axial transvaginal ultrasound shows a homogeneous, hypoechoic mass (calipers) at the site of a C-section scar. Although the appearance is similar to a fibroid, the patient had a history of endometriosis, and this proved to be an endometriotic implant within the C-section scar.*

ADNEXAL MASS IN PREGNANCY

DIFFERENTIAL DIAGNOSIS

Common
- Corpus Luteum Cyst
- Teratoma
- Theca Lutein Cysts

Less Common
- Other Ovarian Masses
  - Endometrioma
  - Cystadenoma
  - Epithelial Ovarian Carcinoma
  - Sex-Cord Stromal Tumor
- Non-Ovarian Adnexal Masses
  - Paraovarian Cyst
  - Pedunculated Fibroid
  - Ectopic Pregnancy
  - Hydrosalpinx
  - Abscess

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Adnexa should be routinely evaluated during pregnancy
  - Usually with transvaginal ultrasound in the first trimester
  - Identify and characterize ovaries
    - If not visible in pelvis, use transabdominal approach
    - Ovary can be displaced by large associated mass or torsion
  - If mass present, characterize as ovarian versus paraovarian in location
- Ovarian mass
  - Cystic ovarian mass usually related to corpus luteum
    - Most often simple cyst of varying sizes
    - Can be complicated or even solid-appearing
    - If large, consider close follow-up and postpartum ultrasound to exclude benign ovarian tumor
  - Solid ovarian mass most often a dermoid
    - Should have classic appearance
    - May be bilateral
  - If ovarian mass is suspicious with cystic and solid components, consider removal during pregnancy
- Paraovarian cysts will not change over time and may have been seen on prior pelvic ultrasounds
- If in the right lower quadrant, assess for appendicitis/appendiceal abscess
- Adnexal mass in 1st trimester
  - Could this be an ectopic or heterotopic pregnancy?
  - Correlate with any history of in vitro fertilization or reproductive assistance

Helpful Clues for Common Diagnoses
- **Corpus Luteum Cyst**
  - May be anechoic or hemorrhagic
    - Commonly complicated by hemorrhage
  - Can have thick, vascular, hyperechoic cyst wall
    - May be mistaken for ectopic gestational sac with decidual reaction
    - Ovarian ectopic pregnancy is exceedingly rare
  - Should decrease in size over pregnancy
    - Some functional cysts may persist
  - Can follow expectantly if no malignant features
    - Postpartum pelvic ultrasound to exclude benign ovarian neoplasm
    - Check for any prior pelvic ultrasounds to see if present before pregnancy
- **Teratoma**
  - Most common incidental ovarian mass seen in pregnancy
    - 10% bilateral
  - May see hair, teeth, osseous structures which gives characteristic complex sonographic appearance
  - Dermoid plug often present
    - Echogenic keratin "plug"
    - Posterior acoustic shadowing
  - If large, risk of ovarian torsion
- **Theca Lutein Cysts**
  - Multiple cysts within enlarged ovaries bilaterally
    - May see typical "spoke-wheel" appearance
    - Occasionally unilateral
  - Reaction of ovaries to elevated hormone levels
    - Multiple gestation pregnancies
    - Assisted reproduction patients
    - Rarely singleton pregnancy with underlying high level of beta hCG
  - Associated pregnancy may be abnormal
    - Molar pregnancy - partial or complete mole

- Hydrops - skin edema, ascites, pleural effusions
- Triploidy - multiple fetal anomalies, IUGR

◦ Look for signs of hyperstimulation syndrome
- Maternal effusions, ascites, oliguria
- Seen in the setting of in vitro fertilization due to hormonal stimulation
- May occur before intrauterine pregnancy identified

Helpful Clues for Less Common Diagnoses

• **Endometrioma**
◦ Homogeneous, low-level echoes

• **Cystadenoma**
◦ Serous - unilocular anechoic cyst, thin septations
◦ Mucinous - hypoechoic with internal mucin, more often multilocular with thicker septations

• **Epithelial Ovarian Carcinoma**
◦ Complex cystic ovarian mass
◦ Vascularized, thick, irregular septations with soft tissue masses
◦ Look for ascites or other indications of peritoneal spread

• **Sex-Cord Stromal Tumor**
◦ Solid, homogeneous ovarian mass
◦ May be hormonally active

• **Non-Ovarian Adnexal Masses**
◦ **Paraovarian Cyst**
- Located in broad ligament
- Does not change in size with hormonal fluctuations
- Use gentle transducer pressure to show it separate from ovary

◦ **Pedunculated Fibroid**
- Attached somewhere to uterine wall even if exophytic or pedunculated
- Can be very heterogeneous if degeneration present

◦ **Ectopic Pregnancy**
- Extrauterine gestational sac ± yolk sac, embryo
- Echogenic free fluid
- May only see adnexal hematoma from rupture

◦ **Hydrosalpinx**
- Dilated tubular "mass"
- Can be mistaken for ovarian mass - use transvaginal ultrasound to clarify

◦ **Abscess**
- Secondary to tubo-ovarian infection
- Possibly ruptured appendix if in the right lower quadrant

Other Essential Information

• Be sure to characterize adnexal mass as ovarian versus paraovarian to narrow differential diagnosis
• If patient has had hormonal manipulation and/or IVF, always consider possible heterotopic pregnancy

Corpus Luteum Cyst



Transvaginal ultrasound shows a 4.5 cm persistent, anechoic, right ovarian corpus luteum cyst ➡ at 28 weeks gestation. Given the benign appearance, removal is not required during pregnancy for confirmation.

Corpus Luteum Cyst



Color Doppler ultrasound of a corpus luteum cyst shows the relative hypervascularity around the cyst ➡, which should not be mistaken for an ectopic pregnancy.

ADNEXAL MASS IN PREGNANCY

Corpus Luteum Cyst

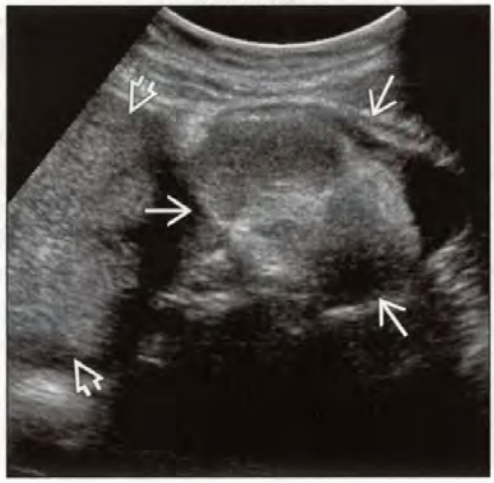


Corpus Luteum Cyst

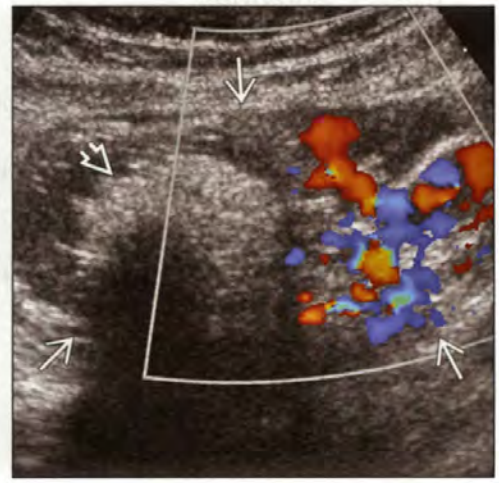


(Left) *Coronal transvaginal ultrasound shows a hemorrhagic corpus luteum cyst with retracting blood clot ➡. After two weeks, the cyst became anechoic. The sequence of findings is typical for a resolving hemorrhagic cyst.* ***(Right)*** *Sagittal transvaginal ultrasound of the right ovary (calipers) shows a solid-appearing homogeneous mass ➡. Normal adjacent ovarian tissue is also present ➡. This is a hemorrhagic corpus luteum, which resolved over time.*

Teratoma



Teratoma



(Left) *Transverse ultrasound of the left adnexa shows a cystic and solid, heterogeneous, incidental mass ➡ adjacent to the uterus (placenta ➡).* ***(Right)*** *Color Doppler ultrasound shows a benign mature teratoma ➡ noted incidentally in the second trimester. This mass measured 8 cm with an echogenic "plug" ➡. Dermoid cysts are the most common ovarian neoplasms discovered during pregnancy.*

Theca Lutein Cysts



Theca Lutein Cysts



(Left) *Axial ultrasound shows a thick placenta containing multiple small and large cysts ➡. In addition, the fetus has intrauterine growth restriction and anomalies (not shown).* ***(Right)*** *Axial ultrasound of the same patient shows the ovaries are enlarged and contain theca lutein cysts (right ovary, ➡). These findings are associated with triploidy.*

ADNEXAL MASS IN PREGNANCY

Endometrioma



Cystadenoma



(Left) *Sagittal US shows a cystic mass with diffuse low-level echoes, typical of an endometrioma. The mass was managed expectantly and remained unchanged during the pregnancy.* ***(Right)*** *Axial oblique T2WI MR shows a simple cystic left ovarian mass posterior to the uterus, without septations or papillary projections. At C-section delivery, a mucinous cystadenoma was removed.*

Epithelial Ovarian Carcinoma

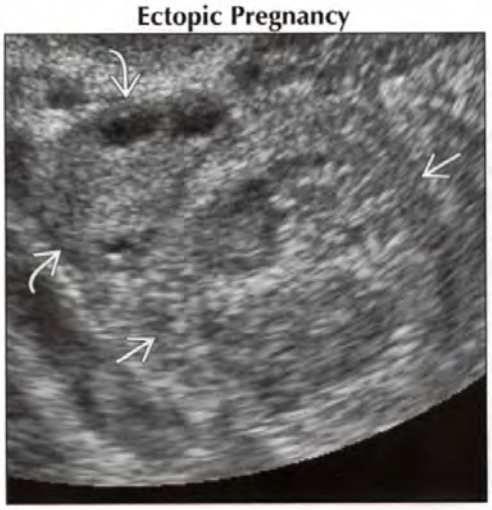


Paraovarian Cyst



(Left) *Color Doppler ultrasound shows an incidental ovarian mass found during a first trimester ultrasound. There is a cystic right ovarian mass with vascular papillary excrescences. Images of the ovaries should be obtained in every obstetrical exam. Pathology showed a serous cystadenocarcinoma, stage 1.* ***(Right)*** *Transvaginal ultrasound shows a normal right ovary with a corpus luteum cyst, and an adjacent large paraovarian cyst.*

Ectopic Pregnancy



Hydrosalpinx



(Left) *Sagittal oblique ultrasound of the right adnexa shows a normal ovary with follicles. Adjacent to the ovary is a heterogeneous mass. A ruptured ectopic pregnancy was removed at surgery.* ***(Right)*** *Sagittal ultrasound shows a cystic, folded tubular structure in the right adnexa, consistent with a chronic hydrosalpinx. This could be confused with a complicated cystic ovarian mass with thin septations.*

BREAST MASS IN PREGNANCY

DIFFERENTIAL DIAGNOSIS

Common
- Normal Fibroglandular Tissue
- Fibroadenoma
- Simple Cyst
- Complicated Cyst

Less Common
- Breast Cancer
- Lactating Adenoma
- Abscess
- Phyllodes Tumor

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Start with breast ultrasound for palpable lumps in pregnancy
- Differentiate whether cystic or solid
  - Simple vs. complicated cyst (both benign)
    - Complicated cyst could be galactocele
  - Solid mass requires more careful evaluation
    - If benign-appearing, may follow in a young patient, though even benign lesions may grow in pregnancy
    - If suspicious do not hesitate to perform mammogram and ultrasound-guided core biopsy
- If ultrasound negative, clinical follow-up warranted to assess for any changes

Helpful Clues for Common Diagnoses
- **Normal Fibroglandular Tissue**
  - Slightly more echogenic parenchyma
  - Usually very dense on mammographic imaging due to hormonal stimulation of tissue and milk production
- **Fibroadenoma**
  - Most common breast mass across all age groups
    - Peak age of occurrence → 20-30 years old
  - Can significantly enlarge during pregnancy
    - Many involute postpartum or after menopause
  - Circumscribed homogeneous mass growing parallel to breast tissues
    - "Wider than tall"
    - Isoechoic or slightly hypoechoic to fat
  - Variable effect on acoustic transmission
    - May have posterior acoustic shadowing, no effect, or increased posterior enhancement
  - If involuted fibroadenoma, may see echogenic, internal coarse "popcorn" calcifications
  - Atypical fibroadenomas may also have irregular or microlobulated margins
    - Should be sampled at any age if appears atypical
- **Simple Cyst**
  - Presents as painful palpable mass, often with acute onset
  - Anechoic circumscribed mass
    - Smooth wall without irregularities or solid components
    - Increased through transmission
    - May have thin nonvascular septa
- **Complicated Cyst**
  - Low-level internal echoes
    - Proteinaceous debris, cellular debris, or cholesterol crystals
    - Movement often visible with realtime imaging
    - Consider galactocele (cyst filled with milk) if during lactation
  - Can have fluid-debris level
  - Color or power Doppler ultrasound can show active swirling of debris
  - Benign finding; not an indication for removal or biopsy unless patient symptomatic and desires aspiration
    - Aspiration may require 18-gauge needle as fluid can be tenacious

Helpful Clues for Less Common Diagnoses
- **Breast Cancer**
  - ~ 3% of breast cancers occur during pregnancy or lactation
    - Cancers are considered pregnancy-associated if occurs during pregnancy or within 1 year of delivery
  - Palpable hypoechoic, irregular mass; no specific appearance in pregnancy
    - May have posterior acoustic shadowing and echogenic halo
  - Do not hesitate to biopsy if lesion suspicious
    - Breast cancer can occur at any age
  - Late diagnosis common when found during pregnancy

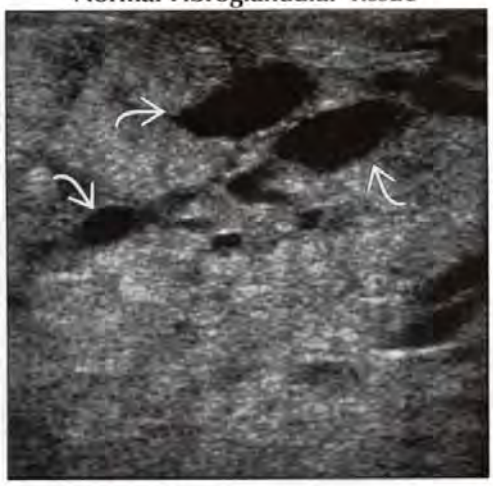
- Could be due to difficult breast exam, age of patient, rapid growth in setting of many physiologic changes
- Treatment should be initiated despite pregnancy
  - Including surgery and chemotherapy
  - No significant known risk to the fetus in the 2nd-3rd trimester
  - Radiation therapy usually deferred
- Most report similar outcome to patients with breast cancer diagnosed outside of pregnancy
  - If compare patients with similar age and stage at diagnosis
  - Some studies suggest prognosis worse during pregnancy if advanced stage at diagnosis
- If chemotherapy given, can cause premature menopause

- **Lactating Adenoma**
  - Not sonographically distinguishable from fibroadenoma
    - Abundant proliferative changes on histology, without atypia
    - Pregnancy-associated changes with secretory activity
  - Occurs during pregnancy and lactation
    - More commonly occurs during pregnancy than lactation
- **Abscess**
  - Correlate with clinical history of infection
  - Usually erythematous/edematous overlying skin
  - Focal, complicated fluid collection on ultrasound
    - Can have irregular margins and echogenic halo of surrounding inflammation
    - May be difficult to distinguish from neoplasm unless correlated with clinical history
  - Ultrasound-guided drainage indicated to aid in resolution of infection
    - May require multiple aspirations during course of treatment until resolution
- **Phyllodes Tumor**
  - Solid, inhomogeneous, lobular circumscribed mass
    - May contain small cystic spaces
    - Posterior acoustic enhancement
  - Not definitively distinguishable from fibroadenoma or carcinoma
  - May be benign, malignant, or borderline

Other Essential Information

- FNA should always be performed for a clinically suspicious lump even if normal ultrasound and mammogram
- If any biopsy performed, notify pathologist that patient is pregnant or lactating to avoid confusion in interpretation
- Core biopsy can be safely performed in pregnancy and lactation
  - Rare reports of milk fistula occurring
  - Can be managed expectantly even if milk fistula develops

Normal Fibroglandular Tissue



Ultrasound of the periareolar region in the late 3rd trimester of pregnancy shows mildly distended ducts and diffusely echogenic parenchyma, consistent with pregnancy/lactational changes.

Normal Fibroglandular Tissue



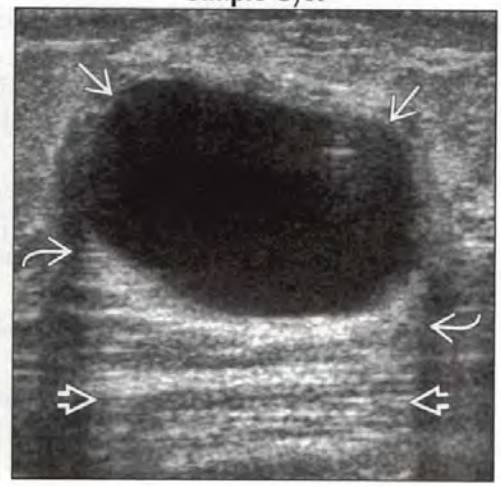
Ultrasound of lactating patient after delivery shows echogenic parenchyma with focal ductal dilation in the axillary tail related to symptoms of a blocked duct. There was a tender palpable lump on examination.

BREAST MASS IN PREGNANCY

Fibroadenoma



Simple Cyst

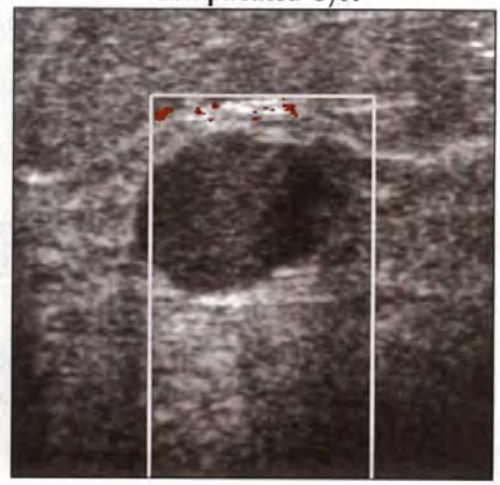


(Left) *Ultrasound shows a solid, oval, gently lobulated, circumscribed, hypoechoic mass at the site of a palpable lump. It is growing parallel to normal tissue planes without posterior acoustic shadowing. Biopsy at the patient's request showed a fibroadenoma.* ***(Right)*** *Ultrasound shows an anechoic, circumscribed, oval, simple cyst at the site of a palpable lump. There is edge shadowing and posterior acoustic enhancement, signs of a benign lesion.*

Complicated Cyst

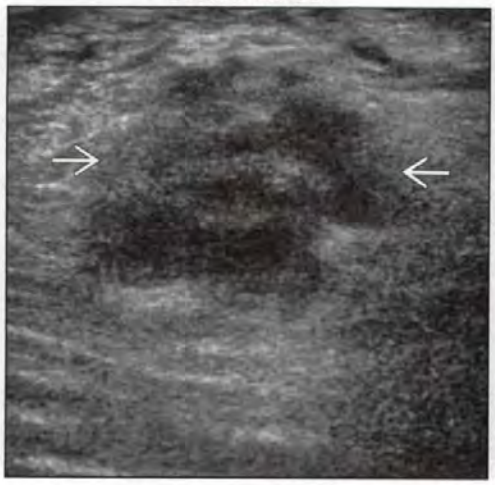


Complicated Cyst

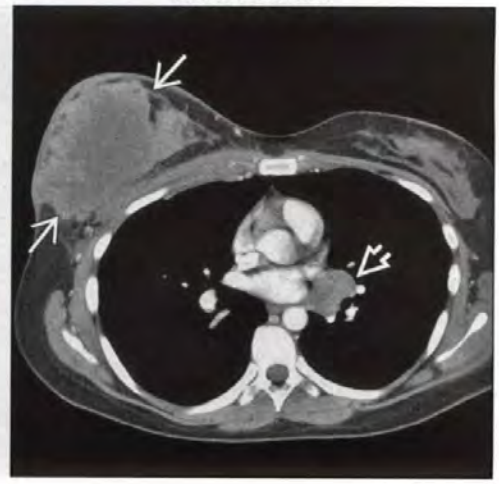


(Left) *Ultrasound shows a fluid-debris level in a complicated cyst, with a punctate echogenic calcification as well. Note the hypoechoic debris portion is sharply demarcated without suggestion of underlying mass.* ***(Right)*** *Color Doppler ultrasound shows no Doppler flow in the layering portion of the complicated cyst. Despite the complicated appearance, this is a benign lesion.*

Breast Cancer



Breast Cancer



(Left) *Ultrasound of a palpable lump in a 27 yo pregnant woman at 35 wks gestation shows a 3 cm, ill-defined, hypoechoic mass (biopsy showed high grade, invasive ductal cancer). A healthy baby was delivered at 37 weeks.* ***(Right)*** *Axial CECT of the same patient 4 months later shows rapid enlargement of the tumor despite chemotherapy. Hilar adenopathy is also visible. The patient died of diffuse metastases 7 months after diagnosis.*

BREAST MASS IN PREGNANCY

Lactating Adenoma



Abscess

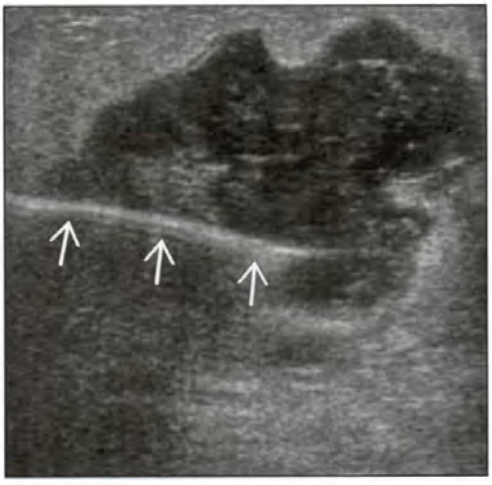


(Left) *Ultrasound of a new lump in a lactating patient shows a lobulated mass ➡ with an adjacent artery ➡. Core biopsy confirmed lactating adenoma.* ***(Right)*** *Ultrasound of a nontender lump shows an irregular mass ➡ with an echogenic halo ➡ and an incidental simple cyst ➡. The patient had partial response to antibiotics for presumed mastitis. The abscess was drained with complete resolution of the imaging findings.*

Abscess



Abscess



(Left) *Ultrasound in a lactating patient 3 wks postpartum shows a complicated fluid collection at the site of skin erythema & lump ➡. Note surrounding echogenic background parenchyma, consistent with lactation.* ***(Right)*** *Ultrasound guided drainage of the abscess was performed; the needle is visible ➡. Despite IV antibiotics, the abscess was drained twice more before full resolution. Repeat drainage is preferred over surgical evacuation to minimize breast trauma.*

Phyllodes Tumor



Phyllodes Tumor



(Left) *Sagittal T2WI MR shows a heterogeneous cystic mass ➡ in a patient just after delivery, nearly replacing all of the normal parenchyma. This presented during pregnancy and grew rapidly throughout the pregnancy.* ***(Right)*** *Axial T1WI MR shows the relative size of left breast (with mass) ➡ compared to the normal right breast ➡. On exam the discrepancy was initially thought to be asymmetric breast hypertrophy. Core biopsy & excision showed phyllodes tumor.*

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Postpartum Complications

Postpartum Hemorrhage	14-2
Postpartum Pain/Fever	14-6

POSTPARTUM HEMORRHAGE

DIFFERENTIAL DIAGNOSIS

Common
- Uterine Atony
- Intrauterine Blood Clot
- Retained Products of Conception

Less Common
- Placenta Accreta Spectrum
- Uterine Dehiscence/Rupture
- Endometritis

Rare but Important
- Gestational Trophoblastic Disease

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Primary postpartum hemorrhage (PPH) is defined as loss of > 500 mL of blood within 24 hours of delivery
  - Vulval/vaginal lacerations are managed clinically & do not require imaging
  - Retained placenta diagnosed by inspection of delivered placenta
    - Managed with manual evacuation or immediate curettage; imaging does not play a major role in diagnosis
- Secondary PPH is defined as abnormal bleeding from the genital tract, from 24 hours after delivery until 6 weeks postpartum
- Risk factors
  - Antepartum hemorrhage in current pregnancy
  - Placenta previa (15x risk)
  - Multiple pregnancy (5x risk)
  - Pre-eclampsia
  - Cesarean section (9x risk for emergency, 3x risk for elective)
- Important to differentiate conditions requiring surgical intervention from others
  - Most cases of retained products of conception (RPOC) are managed with dilation & curettage (D&C)
  - Intrauterine clot, without RPOC, managed conservatively
  - Endometritis may require hospitalization & intravenous antibiotics
  - Gestational trophoblastic disease (GTD) requires D&C ± chemotherapy, radiation therapy
- Remember that it is normal to have some blood & air within the uterus in the puerperium
  - In one study, 21% of healthy women had gas visible up to 3 wks after uncomplicated spontaneous vaginal delivery
  - In another study, 64% of healthy women requesting sterilization had intrauterine blood on CT at 24 hours postpartum
  - Uterine involution takes 6-8 weeks for uterus to return to normal size

Helpful Clues for Common Diagnoses
- **Uterine Atony**
  - Uterus does not contract after delivery of placenta
  - Immediate postpartum event treated with massage, Pitocin, other uterotonic medications
  - May necessitate emergency hysterectomy for control of bleeding
  - Imaging generally not performed
  - Increased risk with multiparity, excessive uterine distension (multiple gestation, polyhydramnios)
- **Intrauterine Blood Clot**
  - Hypoechoic material in endometrial cavity
  - No perfusion to endometrial contents
- **Retained Products of Conception**
  - Mass in endometrial cavity
    - Echogenic material much more suspicious for RPOC than hypoechoic
  - Use color Doppler to look for perfusing vessels from myometrium
    - Echogenic material with feeding vessels highly suggestive of RPOC, but lack of flow does not exclude the diagnosis

Helpful Clues for Less Common Diagnoses
- **Placenta Accreta Spectrum**
  - Abnormal penetration of placental tissue beyond endometrial lining of uterus
  - Placenta fails to separate after delivery with potentially catastrophic bleeding
    - If diagnosis made prior to onset of labor, delivery is planned in tertiary center with neonatal intensive care, appropriate subspecialty surgeons alerted, blood banked
  - Strong association with placenta previa & prior C-section
  - For preemptive diagnosis, maintain high level of suspicion and look for

- Loss of normal subplacental hypoechoic zone
- Abnormal placental lacunae: "Swiss cheese" placenta or "tornado" vessels
- Interruption in bright reflector of bladder mucosa
- Large vessels or nodularity in bladder wall

- **Uterine Dehiscence/Rupture**
  - Dehiscence or rupture most common at site of cesarean hysterotomy
  - Most commonly occurs in labor
    - Surgical emergency with high maternal morbidity & potential fetal demise
    - Imaging rarely performed
  - Delayed rupture presents with pain, bleeding
    - Look for defect in myometrium
    - Often clot in the defect in continuity with intrauterine fluid
- **Endometritis**
  - Generally a clinical diagnosis based on combination of fever, pelvic pain, elevated white cell count, & tender "boggy uterus" in a patient with PPH
  - Imaging findings of endometritis are nonspecific and overlap with normal postpartum state & RPOC
    - Uterus often large
    - Increase flow on Doppler typical, but not always present (lack of flow does not rule out endometritis)
    - Mixed echogenicity material in cavity (may have coexistent RPOC)
    - Bright echoes in cavity from gas
    - May appear normal & still have clinical endometritis
  - More common after C-section than after vaginal birth
  - May be complicated by ovarian vein thrombosis

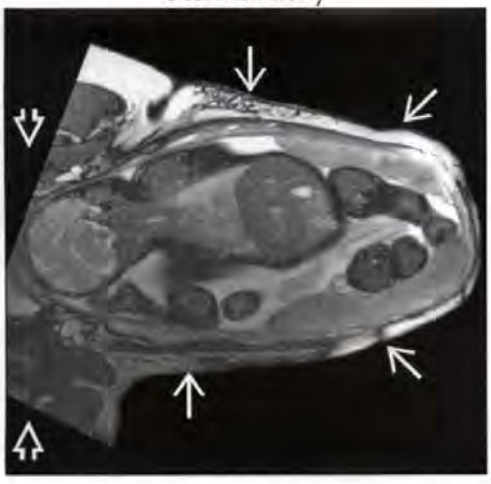
Helpful Clues for Rare Diagnoses

- **Gestational Trophoblastic Disease**
  - Can occur after spontaneous abortion, ectopic pregnancy, or rarely, after normal pregnancy
  - Choriocarcinoma
    - Most commonly follows a molar pregnancy
    - Uterine findings variable from no detectable abnormality to multicystic, hypervascular mass
    - Size of uterine mass does not relate to presence of metastases
    - Theca lutein ovarian cysts from elevated human chorionic gonadotropin (beta hCG)
    - May present as acute respiratory or neurological symptoms after pregnancy
    - Chest X-ray is appropriate to diagnose lung metastases
    - CT scanning is preferable for intra-abdominal metastases
    - MR or CT scanning for brain metastases

Other Essential Information

- Undiagnosed blood dyscrasia may present as unexplained PPH

Uterine Atony



Axial oblique T2WI MR shows marked uterine distension ➡ (maternal hips ➡) in a patient with twins, one of whom had an anomaly (not shown). Excess uterine distension is a risk factor for atony.

Intrauterine Blood Clot

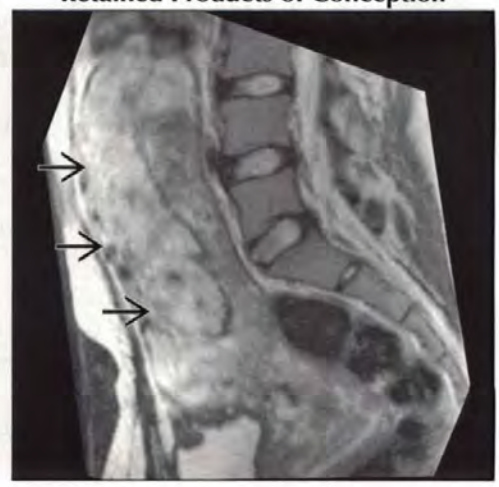


Sagittal transvaginal ultrasound shows the cervix ➡ distended by hypoechoic clot and some active bleeding ➡. The clot was removed manually and hemostasis secured by inflating a catheter within the uterus.

POSTPARTUM HEMORRHAGE

Retained Products of Conception

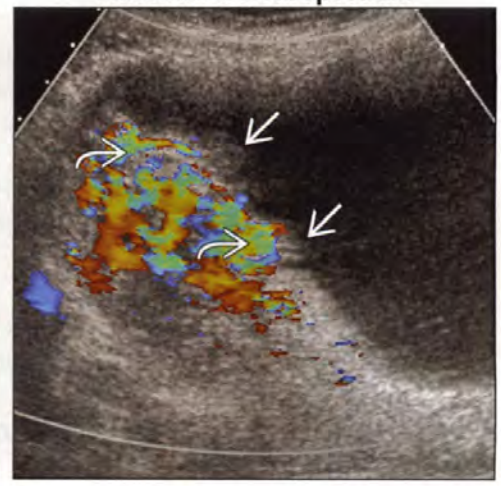




(Left) *Sagittal ultrasound shows apparently intact placental cotyledons ➡ in the uterus in a patient with PPH.* ***(Right)*** *Sagittal T2WI MR in the same case confirms a large amount of placental material ➡ in the enlarged uterus, which reaches the umbilicus (i.e., the size of a 20 week gestation). Contrast enhanced images showed no evidence of myometrial invasion. Final diagnosis was RPOC, with a large portion of the placenta remaining in the uterine cavity.*

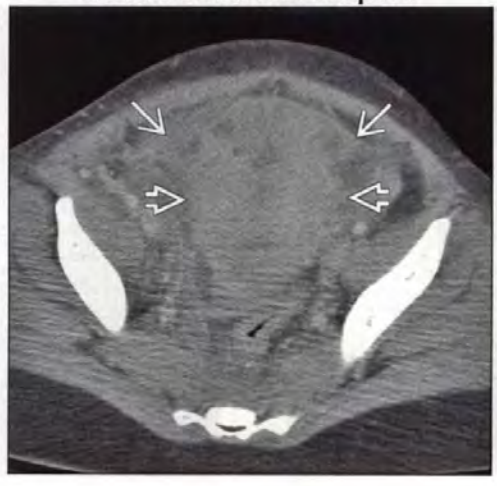
Placenta Accreta Spectrum

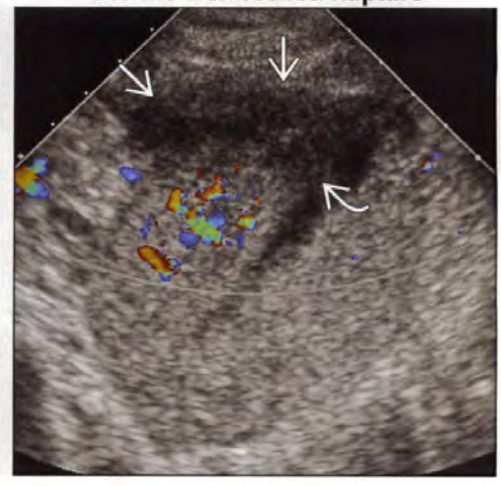




(Left) *Axial ultrasound shows "tornado-shaped" vessels ➡ and multiple areas of abnormal placental extension ➡ into the myometrium. Look carefully in all patients at risk for accreta spectrum in order to avoid its potentially lethal presentation with uncontrolled acute PPH.* ***(Right)*** *Sagittal color Doppler ultrasound shows placental vessels ➡ invading the nodular bladder wall ➡. The patient needed a partial cystectomy at the time of cesarean hysterectomy.*

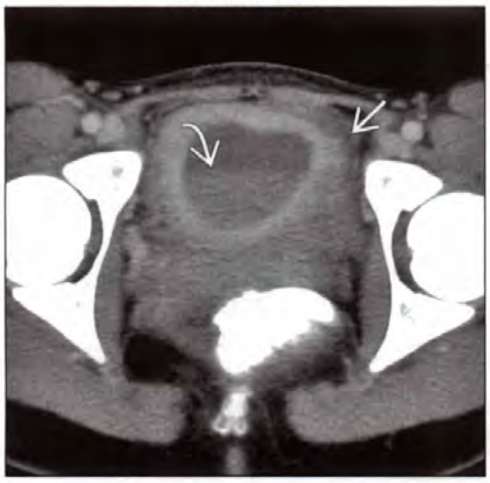
Uterine Dehiscence/Rupture



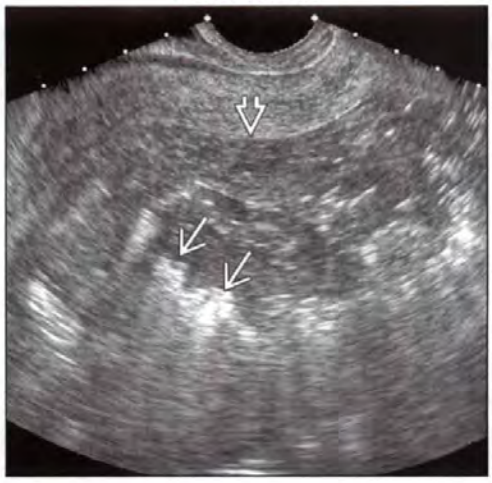


(Left) *Axial CECT shows loculated fluid ➡ around the irregularly marginated uterus ➡. The uterus was necrotic, ruptured, and surrounded by pus. The patient presented with pain, fever, & vaginal bleeding and quickly developed septic shock.* ***(Right)*** *Sagittal color Doppler ultrasound shows delayed rupture with a defect ➡ at the hysterotomy site. Note that clot ➡ extends anterior to the uterine defect. This woman presented with pain and delayed PPH.*

Endometritis



Endometritis

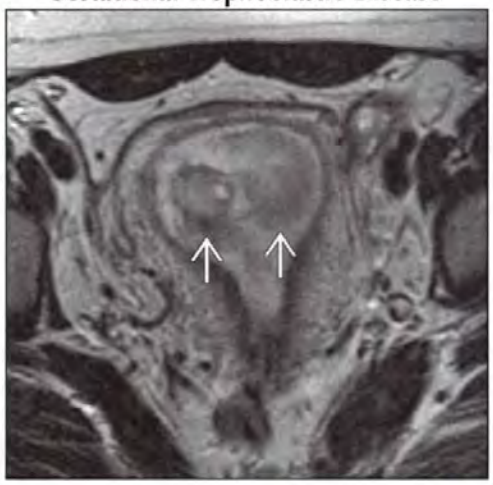


(Left) *Axial CECT shows a pyometrium with a distended endometrial cavity, which contains a fluid-fluid level. Surrounding inflammatory changes are seen in the parametrial fat.* ***(Right)*** *Transvaginal ultrasound in another case shows a distended endometrial cavity filled with echogenic debris and bright, echogenic gas bubbles in a patient with pyometrium. Both patients presented with fever, pain, and elevated white cell count, as well as PPH.*

Gestational Trophoblastic Disease



Gestational Trophoblastic Disease



(Left) *Axial CECT shows an enhancing intrauterine mass. There was no myometrial breach or distant metastases. Other images showed theca lutein cysts in the ovaries.* ***(Right)*** *Axial T2WI MR in a different patient with choriocarcinoma shows small intrauterine masses. All women with persistent bleeding after delivery or abortion should have their beta hCG checked to rule out GTD.*

Gestational Trophoblastic Disease



Gestational Trophoblastic Disease



(Left) *Sagittal T2WI MR in a patient with PPH and elevated beta hCG shows a mixed signal intensity mass invading the myometrium. The fundal serosa remains intact. Theca lutein ovarian cysts were present, and a chest X-ray showed multiple lung metastases.* ***(Right)*** *Gross pathology from the same patient shows the intrauterine choriocarcinoma and an enlarged ovary, with multiple theca lutein cysts.*

POSTPARTUM PAIN/FEVER

DIFFERENTIAL DIAGNOSIS

Common
- Endometritis
- Bladder Flap Hematoma

Less Common
- Adnexal Torsion
- Ovarian Vein Thrombosis
- Uterine Rupture

ESSENTIAL INFORMATION

Key Differential Diagnosis Issues
- Puerperal fever is defined as fever ≥ 38° C within 14 days of giving birth
- Aim to differentiate pregnancy-related from other sources of fever
  - Check for vaginal bleeding, discharge
  - History of vaginal vs. cesarean delivery
  - History of prior C-section
- Post-operative population at risk for pneumonia, venous thromboembolic disease, urinary tract infection

Helpful Clues for Common Diagnoses
- **Endometritis**
  - Mixed echogenicity debris in endometrial cavity
  - Punctate bright echoes with distal shadowing suggest gas in cavity
  - Increased risk with retained products of conception, C-section
- **Bladder Flap Hematoma**
  - Occurs after C-section
  - Typical location between lower uterine segment and bladder wall

Helpful Clues for Less Common Diagnoses
- **Adnexal Torsion**
  - Increased risk in puerperium as ovaries descend back into pelvis
  - Ovary enlarged, edematous with peripheral follicles
  - Use Doppler to evaluate flow; venous flow obliterated before arterial
- **Ovarian Vein Thrombosis**
  - Difficult diagnosis on ultrasound but contrast-enhanced CT shows typical findings
    - Distended ovarian vein with surrounding inflammation
    - Typical course extending from pelvis to inferior vena cava (IVC) on right, pelvis to renal vein on left
    - Inflammation can extend to surrounding bowel; do not confuse with appendicitis or other bowel pathology
- **Uterine Rupture**
  - More common in labor but may occur as delayed event
  - History of prior C-section or uterine surgery
  - Anterior uterine segment hematoma at site of dehiscence ± intraperitoneal fluid

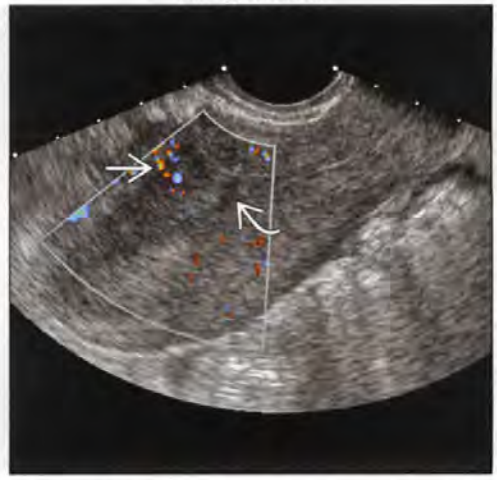
Other Essential Information
- Mother should pump and discard breast milk for 24 hours after contrast-enhanced CT due to iodine excretion in breast milk

Endometritis



Sagittal ultrasound shows punctate echoes ➡ in the anterior myometrium and an area of inhomogeneous echoes between the uterus ➡ and the bladder ➡ in a patient with fever and pain following C-section.

Endometritis



Sagittal color Doppler ultrasound shows hyperemia ➡ in the anterior myometrium and fluid tracking into the hysterotomy site ➡ in a patient with endometritis post C-section.

Bladder Flap Hematoma



Bladder Flap Hematoma

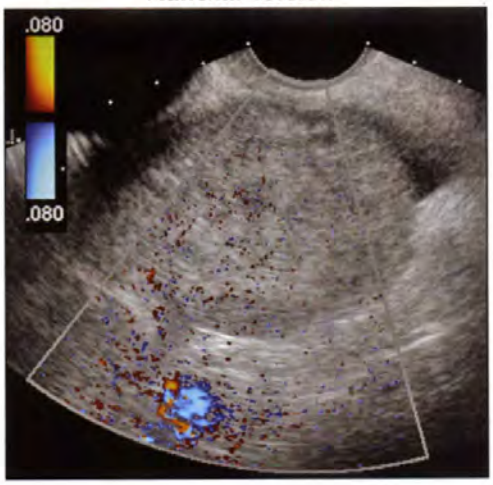


(Left) *Axial CECT shows soft tissue edema and a loculated fluid collection posterior to the bladder.* ***(Right)*** *Sagittal CECT reconstruction in the same patient shows that the loculated fluid collection is in the typical location for a bladder flap hematoma. The patient was morbidly obese, which made the surgery more technically challenging. The complication rate is increased in obese patients.*

Adnexal Torsion



Adnexal Torsion



(Left) *Transvaginal ultrasound shows an enlarged ovary (calipers) with a fluid-fluid level in a cyst within it. The patient developed acute pain within hours of delivery.* ***(Right)*** *Color Doppler ultrasound in the same patient shows no flow in the edematous parenchyma, consistent with torsion. The diagnosis was surgically confirmed.*

Ovarian Vein Thrombosis



Uterine Rupture



(Left) *CECT shows a thick, enhancing vessel wall with low density intraluminal clot. It is important to follow the course of this tubular structure to confirm it is the ovarian vein and to evaluate for propagation of clot into the IVC.* ***(Right)*** *CECT shows disruption of the anterior myometrium and multiple rim-enhancing fluid collections in a patient transferred in septic shock after a vaginal delivery. The uterus was necrotic and ruptured, and there was a liter of pus in the abdomen.*

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