Obstetric Sonography Registry Review

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Purpose of the Lecture

- Recount rational facts about the fetal head and brain, fetal gastrointestinal system, fetal genitourinary system, chromosomal abnormalities, so that the information may be easily recalled in an assessment situation.
- Utilize the information from this lecture to appropriately respond to clinical situations.
- Recognize important sonographic terminology and definitions.

Fetal Brain Review

EMBRYOLOGIC DEVELOPMENT OF THE FETAL BRAIN

- Initially, the brain is separated into three primary vesicles termed the prosencephalon (forebrain), mesencephalon (midbrain), and rhombencephalon (hindbrain).

- The cerebrum can be divided into a right and left hemisphere by the interhemispheric fissure.

- The falx cerebri, a double fold of dura mater, is located within the interhemispheric fissure and can readily be noted on a fetal sonogram as an echogenic linear formation coursing through the midline of the fetal brain.

- These vesicles will continue to develop and form critical brain structures.

- Sonographically, the rhombencephalon may be noted within the fetal cranium during the first trimester.

THE CEREBRUM

- The cerebral hemispheres are linked in the midline by the corpus callosum, a thick band of tissue that provides communication between right and left halves of the brain.
THE CORPUS CALLOSUM

• The corpus callosum forms late in gestation, but should be completely intact between 18 and 20 weeks.

• The corpus callosum connects the two lobes of the cerebrum.

• The sonographic appearance of the corpus callosum is that of an echogenic band of tissue within the midline of the brain connecting the two cerebral hemispheres.

THE CAVUM SEPTUM PELLUCIDUM

• The cavum septum pellucidum (CSP) is a midline brain structure that can be identified in the anterior portion of the brain between the frontal horns of the lateral ventricles.

• It will appear as an anechoic “box-shaped” structure in the axial scan plane.

• Although the CSP should always be seen between 18 and 37 weeks, the closure of this structure is normal in later gestation and often occurs before birth or shortly thereafter.

• The cavum septum pellucidum does not communicate with the ventricular system and its absence is associated with multiple cerebral malformations.

THE THALAMUS

• The thalamus, a vital brain structure that has numerous functions, is a significant landmark for sonographers to locate within the fetal brain.

• The two lobes of the thalamus are located on both sides of the third ventricle.

• The massa intermedia passes through the third ventricle to connect the two lobes of the thalamus.

What is another name for the massa intermedia?

1. Cerebral aqueduct
2. Interthalamic adhesion
3. Zona irregularis
4. Sylvian union

What structures would be most likely be confused for the thalamus?

1. Cerebellar hemispheres
2. Cisterna magna
3. Corpus callosum
4. Cerebral peduncles

The Cerebellum

• The cerebellum is located in the posterior fossa of the cranium. The cerebellum consists of two hemispheres, right and left, that are coupled at the midline by the cerebellar vermis.

• The normal cerebellum is a dumbbell- or figure eight-shaped structure noted in the posterior cranium of the fetus.

• The two hemispheres of the cerebellum should be symmetric, although hypoplasia of one cerebellar hemisphere can occur, resulting in the hypoplastic hemisphere appearing smaller than normal.
What is another term for dolichocephaly?

1. brachycephaly
2. scaphocephaly
3. mesocephaly
4. hydrocephaly

THE CISTERNA MAGNA

• The cisterna magna, located in the posterior fossa of the cranium, is the largest cistern in the head.
• On sonography, the cisterna magna appears as an anechoic, fluid-filled space, posterior to the cerebellum, between the cerebellar vermis and the interior surface of the occipital bone.
• The cerebellum is located in the posterior fossa of the cranium.
• The cerebellum consists of two hemispheres, right and left, that are coupled at the midline by the cerebellar vermis.

THE CISTERNA MAGNA MEASUREMENT

• Sonographic measurement of the depth of the cisterna magna can be performed as well.
• The depth of the cisterna magna should not measure over 10 mm or less than 2 mm in the transcerebellar plane.
• Measurement over 10 mm is consistent with mega cisterna magna and Dandy-Walker complex, while a measurement of less than 2 mm is worrisome for Arnold-Chiari II malformation.

LATERAL VENTRICLE MEASUREMENT

• The diameter of the lateral ventricle can be easily measured with sonography.
• The lateral ventricle is measured in the transaxial plane at the level of the atrium.
• The atrium of the lateral ventricle is the optimal site for measuring the lateral ventricle because this is the first region where ventricular enlargement occurs.
• The calipers are placed at the level of the glomus of the choroid plexus.
• The normal lateral ventricle does not typically measure over 10 mm at the level of the atrium.

VENTRICULOMEGALY AND HYDROCEPHALUS

• The abnormal enlargement of the ventricles within the brain is referred to as ventriculomegaly.
• Hydrocephalus is typically reserved for cases of ventriculomegaly that are more severe and are caused by some type of obstruction to the flow of CSF.
• Therefore, obstructive hydrocephalus is the buildup of CSF within the ventricular system secondary to some type of obstruction.

VENTRICULOMEGALY AND HYDROCEPHALUS

• Ventriculomegaly has been cited as the most common cranial abnormality.
• Suspicion of ventricular dilatation occurs when the atrial diameter measures greater than 10 mm.
• The lateral ventricle that will be readily seen on sonography is most often the ventricle farthest away from the transducer.
• The sonographic finding of the "dangling choroid" sign describes the echogenic choroid plexus, hanging limp and surrounded by CSF, within the dilated lateral ventricle.
What is another name for the atrium of the lateral ventricle?

1. Trigone
2. Splenium
3. Rostrum
4. Genu

AQUEDUCTAL STENOSIS

- Aqueductal stenosis is the most common cause of hydrocephalus in utero.
- The cerebral aqueduct (aqueduct of Sylvius), located between the third and fourth ventricle of the brain, may be narrowed, thus preventing the flow of CSF from the third to the fourth ventricle.
- This obstruction level will cause the third ventricle and both lateral ventricle to expand, while the fourth ventricle remains normal.

Cerebral Malformations

HYDRANENCEPHALY

- Hydranencephaly is a fatal condition in which the entire cerebrum is replaced by a large sac containing cerebrospinal fluid.
- With hydranencephaly, the falx cerebri may be partially or completely absent, while the brainstem and basal ganglia are maintained and surrounded by cerebrospinal fluid.
- There will be no cerebral cortex identified.

HYDRANENCEPHALY

- Hydranencephaly can be difficult to differentiate with the sonographic findings of severe hydrocephalus and alobar holoprosencephaly.
- It is important to note that with both hydrocephalus and holoprosencephaly there will be a rim of cerebral tissue maintained, while with hydranencephaly there is no cerebral mantle present.
- Hydranencephaly is typically a fatal condition, with death occurring in the first year of life.

HOLOPROSENCEPHALY

- Holoprosencephaly is a midline brain anomaly that is associated with not only brain aberrations but also atypical facial structures.
- It may be detected with endovaginal imaging as early as the first trimester.
- There are three main types of holoprosencephaly: alobar, semilobar, and lobar.
- While the lobar form can be consistent with life, alobar holoprosencephaly is the most severe form, often resulting in neonatal death.
HOLOPROSENCEPHALY

- Alobar holoprosencephaly is diagnosed when there is absence of the corpus callosum, cavum septum pellucidum, third ventricle, interhemispheric fissure, and falx cerebri.
- There will also be evidence of a horseshoe-shaped monoventricle and the lobes of the thalamus will be fused and echogenic in appearance.
- Conversely, the cerebellum and brainstem remain intact.

- Cyclopia, a condition in which the orbits are fused and contain a single eye, and proboscis, a false nose situated above the orbits, are two of the most disturbing external findings associated with holoprosencephaly.
- Other facial anomalies such as anopthalmia, hypotelorism, median cleft lip, and cebocephaly may be detected during a fetal sonogram as well.
- With the less devastating forms of holoprosencephaly, such as lobar, there are varying degrees of fusion of the midline structures. Infants with lobar holoprosencephaly may experience severe mental retardation.

What chromosomal abnormality is more likely linked with holoprosencephaly?

1. Trisomy 21
2. Trisomy 13
3. Trisomy 18
4. Trisomy 8

HOLOPROSENCEPHALY and Trisomy 13

- Trisomy 13, or Patau syndrome, is present in 50 to 70% of fetuses diagnosed with holoprosencephaly.

DANDY-WALKER MALFORMATION

- Dandy-Walker malformation (DWM) is actually a classification within a larger group of anomalies referred to as the Dandy-Walker complex.
- Dandy-Walker complex is a spectrum of posterior fossa abnormalities that involve the cystic dilatation of the cisterna magna and fourth ventricle.
- DWM is thought to be caused by a developmental abnormality in the roof of the fourth ventricle.
- The sonographic findings of DWM include the presence of an enlarged cisterna magna that communicates with a distended fourth ventricle through a defect in the cerebellum.

- The cerebellar vermis is either completely absent or hypoplastic.
- As a result, the tentorium, the structure that separates the cerebrum from the cerebellum, is elevated.
- There are often other midline brain abnormalities present as well.
- For instance, agenesis of the corpus callosum, ventriculomegaly, holoprosencephaly, and cephaloceles are all associated anomalies that can co-exist with DWM.
MEGA CISTERNA MAGNA

- Mega cisterna magna, which is the enlargement of the cisterna magna without the involvement of the fourth ventricle, may be confused with Dandy-Walker malformation.
- Mega cisterna magna is present when only the cisterna magna is enlarged, measuring more than 10 mm in depth.
- Consequently, the fourth ventricle is normal with mega cisterna magna and enlarged with DWM.
- It is important to note that in the early second trimester, the inferior portion of the cerebellar vermis may not be formed, thus making it appear as if the fetus has partial agenesis of the vermis.

AGENESIS OF THE CORPUS CALLOSUM AND CAVUM SEPTUM PELLUCIDUM

- There can be partial or complete absence of the corpus callosum.
- Most often, if the corpus callosum is absent, the cavum septum pellucidum will be absent as well.
- Their nonexistence has been linked to as many as 50 to 200 different syndromes and anomalies, such as holoprosencephaly, Dandy-Walker malformation, aqueductal stenosis, trisomy 18, trisomy 8, and trisomy 13.
- There are several distinct sonographic findings consistent with agenesis of the corpus callosum, excluding the obvious absence of this structure.

AGENESIS OF THE CORPUS CALLOSUM AND CAVUM SEPTUM PELLUCIDUM

- The “sunburst” manifestation of the sulci is a straightforward and discernible sonographic finding. In the normal brain, the sulci within the cerebrum typically travel parallel to the corpus callosum, but with agenesis of the corpus callosum they tend to have a more perpendicular or radial arrangement and often appear to have a “spoke wheel” pattern.
- Colpocephaly, small frontal horns and enlarged occipital horns, is often present as well and offers a distinct teardrop shape to the lateral ventricles.
- With absence of the cavum septum pellucidum and corpus callosum, the third ventricle tends to migrate more superiorly and appear dilated.

SCHIZENCEPHALY

- Schizencephaly is associated with the development of fluid-filled clefts within the cerebrum.
- The sonographic appearance of schizencephaly is that of a cerebrum containing clefts filled with anechoic, cerebrospinal fluid.
- There are several associated anomalies, such as agenesis of the corpus callosum, agenesis of the cavum septum pellucidum, and ventriculomegaly.

LISSENCEPHALY

- Lissencephaly literally means “smooth brain.”
- It is a condition in which there is no gyrí within the cerebral cortex.
- Agyria, and the absence of sulci within the brain, is not typically diagnosed until the third trimester or postnatally and almost always carries a poor prognosis.

CHORIOID PLEXUS CYSTS

- Choroid plexus cysts are cysts located within the choroid plexus of the lateral ventricles.
- These small cysts are frequently encountered during a routine sonographic examination and typically regress by the end of the third trimester, although they are associated with an increased risk of Trisomy 18.
- A choroid plexus cyst will be located within the choroid plexus of the lateral ventricle, measure greater than 2 mm, appear round and anechoic, and have smooth walls.
Neural Tube Defects

NEURAL TUBE DEFECTS AND THE BRAIN

- Neural tube defects occur when the embryonic neural tube fails to close.
- Anencephaly and spina bifida are the most common neural defects, occurring in 1 per 1,000 pregnancies.

NEURAL TUBE DEFECTS AND THE BRAIN

- Fortunately, studies have shown that a supplement of 4 mg of folate (folic acid) in a woman's diet significantly reduces the likelihood of her fetus developing a neural tube defect.
- Maternal serum screening, also referred to as the triple screen, combines the laboratory values of human chorionic gonadotropin, estriol, and maternal serum alpha-fetoprotein (MSAFP).
- Particularly helpful for detecting neural tube defects is the MSAFP component of this test.

ACRANIA (ANENCEPHALY & EXENCEPHALY)

- Acrania remains one of the most common neural tube defects.
- Acrania is defined as the absence of the cranial vault above the bony orbits.
- It can be further divided into two main subtypes that are related to the amount of cerebral tissue present, anencephaly and exencephaly.

ACRANIA (ANENCEPHALY & EXENCEPHALY)

- Anencephaly is considered when there are no cerebral hemispheres present, while exencephaly denotes the presence of a normal amount of cerebral tissue.
- Nonetheless, the cranium is absent, making this condition fatal.
- The sonographic appearance of anencephaly has been described as having "froglike" faces, or bulging eyes, and absence of the cranial vault.
ARNOLD-CHIARI II MALFORMATION AND SPINA BIFIDA

- Arnold-Chiari II or Chiari II malformation is a group of cranial abnormalities associated with the neural tube defect spina bifida.
- Spina bifida may result in a mass that protrudes from the spine.
- This mass can be referred to as a meningocele or myelomeningocele, depending on its contents.
- The most common location of spina bifida is within the distal lumbosacral region.

ARNOLD-CHIARI II MALFORMATION AND SPINA BIFIDA

- In the presence of spina bifida, there are several notable changes that occur within the brain and skull.
- The frontal bones become flattened and will yield a lemon shape to the cranium, which is referred to as the "lemon" sign, often referred to as scalloping of the frontal bones.
- The cerebellum will become displaced inferiorly and posteriorly and appear curved in the presence of spina bifida, which is referred to as the "banana" sign.

ARNOLD-CHIARI II MALFORMATION AND SPINA BIFIDA

- As a result of the cerebellum being displaced inferiorly, the cisterna magna is completely obliterated.
- A posterior fossa abnormality such as Chiari II malformation should be suspected if the cisterna magna is not visualized.
- The lateral ventricles will also be distorted in shape.
- The frontal horns will be small and slit like, while the occipital horns will be enlarged, a condition known as colpocephaly.

CEPHALOCELES

- Cephaloceles are protrusions of intracranial contents through a defect in the skull.
- The most common location for a cephalocele is in the occipital region.
- However, cephaloceles may also have frontal and parietal positions.
- Cephaloceles are common findings in Meckel-Gruber syndrome and have varying sonographic appearances based on their content.

FETAL INTRACRANIAL VASCULAR ANOMALIES

- The vein of Galen aneurysm is an arteriovenous malformation that occurs within the fetal brain.
- The sonographic findings of a vein of Galen aneurysm is that of a large, anechoic mass within the midline of the cranium that when interrogated with color and pulsed Doppler fills with turbulent venous and arterial flow.
- The fetus will also have signs hydrops and cardiomegaly.
- Newborns with this condition are prone to suffer from congestive heart failure in the postnatal period.

The Fetal Gastrointestinal System
Polyhydramnios, or excessive amniotic fluid, can be noted in the presence of multiple anomalies. During fetal development, there are several structures that are thought to produce amniotic fluid. Initially, in early embryologic development, the origin of amniotic fluid is thought to result from an osmotic process, as water crosses the amniotic space freely. In later gestation, somewhere around 12 weeks, the fetal kidneys begin to produce urine, a liquid that eventually comprises most of the amniotic fluid.

The Fetal Gastrointestinal System

For instance, the fetus who suffers from esophageal atresia or duodenal atresia cannot transport amniotic fluid into the intestines. The fluid exits back out of the esophagus and absorption cannot take place. Consequently, there is a buildup of amniotic fluid resulting from the continual production of urine by the fetal kidneys.

Abdominal Wall Defects
Abdominal Wall Defects and AFP

- Gastroschisis and omphalocoele are two of the most common abdominal wall defects.
- AFP can exit the fetus through an abdominal wall defect, thereby increasing the level of maternal serum alpha-fetoprotein (MSAFP).
- If an opening is present, a greater amount of AFP is allowed to pass into the maternal circulation.
- Although MSAFP screening is not specific for abdominal wall defects, elevated levels of MSAFP are found in the presence of omphalocoele and gastroschisis and thus can be used as a reliable screening test for the early detection of these and other abnormalities.

Physiologic Bowel Herniation

- As part of normal fetal development, the midgut during the sixth menstrual week herniates into the base of the umbilical cord; this is termed physiologic bowel herniation.
  - The intestines return to the abdomen by the twelfth gestational week.
  - Therefore, a diagnosis of an abdominal wall defect, such as omphalocoele or gastroschisis, is difficult before twelve weeks, and follow-up examinations are often needed if the diagnosis is suspected in the first trimester.

Gastroschisis

- Gastroschisis is the herniation of abdominal contents through a right-sided, periumbilical abdominal wall defect.
- Gastroschisis is thought to be caused by a vascular incident occurring to either the right umbilical vein or omphalomesenteric artery.
  - Most often, there is herniation of the small intestine, but with larger defects the stomach and other organs may be found outside of the abdomen.
  - The bowel that is exposed to amniotic fluid may become dilated, thick-walled, and have decreased peristaltic activity.

Gastroschisis

- Gastroschisis, unlike omphalocoele, does not have a strong association with chromosomal abnormalities.
- Sonographically, normal cord insertion into the abdomen is noted, and most often the right-sided periumbilical mass will be easily identified.
  - Recognizable loops of bowel are often noted outside of the abdomen floating in the amniotic fluid.

Omphalocoele

- The evidence of persistent herniation of the bowel, and potentially other abdominal organs, into the base of the umbilical cord leads to the diagnosis of an omphalocoele.
  - An omphalocoele is located within the midline of the abdomen.
  - The umbilical cord will insert into this mass. The entire contents is contained and covered by peritoneum and amnion.
  - Ascites is often noted within an omphalocoele, as well as within the abdomen of the fetus.

Omphalocoele

- Ascites may be helpful in demarcating the contents of the mass.
- It is important to note whether the mass contains liver, as a poorer prognosis corresponds with this type of omphalocoele.
- The sonographic appearance of an omphalocoele is that of a midline abdominal mass that contains bowel, liver, or other abdominal organs.
- Omphalocoele has a more significant risk for heart defects and chromosomal anomalies than gastroschisis.
The Fetal Urinary Tract

The Fetal Urinary Tract

VACTERL Association

- VACTERL stands for vertebral anomalies, anal atresia, cardiac anomalies, tracheoesophageal fistula or esophageal atresia, renal anomalies, and limb anomalies.
- Patients are considered to have this association if three of the organ systems listed have abnormalities.
- Therefore, if an irregularity is noted within one structure, this should prompt the sonographer to further investigate the other systems for associated anomalies.
- VACTERL association may also be referred to as VATER sequence, VATER, or VACTEL syndrome.

Renal Abnormalities and Oligohydramnios

- Renal abnormalities are the most frequent cause of oligohydramnios.
- Urine comprises the greater part of amniotic fluid after 16 weeks.
- In circumstances in which the fetus has a renal abnormality, specifically those that are linked with bilateral renal agenesis, inadequately functioning kidneys, or obstruction of the urinary tract, oligohydramnios will be present, and in some cases anhydramnios may occur.

Renal Agenesis

- Failure of a kidney to form is referred to as renal agenesis.
- Renal agenesis can be unilateral or bilateral.
- There are two sonographic findings that are helpful in making the sonographic diagnosis of renal agenesis.
  - First, when the kidney is absent in the abdomen, the adrenal gland can be noted in a parallel, flattened position, a sonographic finding known as the "lying down" adrenal sign.
  - Secondly, color Doppler can be employed over the renal artery branches of the abdominal aorta.
- When there is absence of the kidney, there will be no identifiable renal artery branches.

Renal Agenesis

- Bilateral renal agenesis, also known as Potter's syndrome, is typically a fatal condition.
  - Absence of both of the fetal kidneys can be difficult to detect sonographically, secondary to the lack of amniotic fluid surrounding the fetus.
  - Therefore, it is extremely beneficial to utilize color Doppler to investigate the renal area.
  - Non-visualization of the urinary bladder and kidneys, with associated severe oligohydramnios, are considered to be trustworthy findings consistent with bilateral renal agenesis.
  - Bilateral renal agenesis may be seen in conjunction with sirenomelia and various cardiovascular malformations.

Congresswoman’s ‘miracle baby’ may be first to survive Potter’s Syndrome

Abigail Rose Beutler
Renal Agenesis

- Fortunately, unilateral renal agenesis is much more common than bilateral renal agenesis.
  - Most often, with unilateral renal agenesis there is an average amount of amniotic fluid and the prognosis is good.
  - Before making the conclusion of unilateral renal agenesis, the sonographer should always analyze the fetal pelvis for a pelvic kidney, as this is the most common location of an ectopic kidney.
  - In the presence of unilateral renal agenesis, the contralateral kidney will enlarge, a condition known as compensatory hypertrophy.

ARPKD (Infantile Polycystic Disease)

- Autosomal recessive polycystic kidney disease (ARPKD) may also be referred to as autosomal recessive polycystic renal disease and infantile polycystic kidney disease.
  - The typical sonographic findings of a fetus affected by ARPKD are bilateral enlarged, echogenic kidneys, non-detectable urinary bladder, and oligohydramnios.
  - The kidneys may be as large as 3 to 10 times the normal renal size for the gestation.
  - One condition associated with ARPKD is Meckel-Gruber syndrome, which is a fatal disorder that is associated with renal cystic disease, occipital cephalocele, and polydactyly.

ARPKD (Infantile Polycystic Disease)

- Referring to this condition as a renal cystic disease can be puzzling to a sonographer because cysts are not always perceptible with sonography.
  - This is secondary to the size of the cysts, as the cysts with ARPKD are microscopic and not macroscopic.
  - It is significant to appreciate the differences in the sonographic appearance of ARPKD and multicystic dysplastic kidney (MCDK) disease.
  - Cysts are not identifiable in ARPKD but are evident in the multicystic dysplastic kidney.

MCDK (Multicystic Dysplastic Kidney Disease)

- Multicystic dysplastic renal disease may also be referred to as multicystic dysplastic kidney (MCDK) disease and multicystic renal dysplasia.
  - The sonographic findings of MCDK disease is the identification of unilateral or bilateral multiple, smooth-walled, noncommunicating cyst of varying sizes in the area of the renal fossa.
  - There is typically no normal-functioning renal tissue present on the side affected by MCDK disease.

MCDK (Multicystic Dysplastic Kidney Disease)

- Therefore, MCDK disease is fatal if bilateral, with the consistent associated findings of oligohydramnios and absent bladder.
  - Fortunately, most cases of MCDK disease are unilateral and consequently have a normal amniotic fluid volume.
  - Fetuses with MCDK disease can also have additional related anomalies, such as abnormalities of the gastrointestinal tract and central nervous system, limb anomalies, and further renal abnormalities.

ADPKD (Autosomal Dominant Polycystic Kidney Disease)

- Autosomal dominant polycystic kidney disease (ADPKD) may also be referred to as autosomal dominant polycystic renal disease.
  - The sonographic appearance of fetal kidneys with ADPKD is similar to that of ARPKD in that both kidneys will appear enlarged and echogenic.
  - A distinguishing difference between the two diseases is that in the fetus with ADPKD, the urinary bladder is often present and there is a normal amniotic fluid volume, while with ARPKD the bladder is absent and there is oligohydramnios.
ADPKD
(Autosomal Dominant Polycystic Kidney Disease)

- ADPKD does not typically manifest until approximately the fifth decade of life, at which time the adult will develop renal cysts and may die from end-stage renal failure.
- Adult renal cystic disease also is associated with the development of cysts within the liver, pancreas, and spleen.

Fetal Urinary Tract Obstructions

- An obstruction of the fetal urinary tract can lead to distension of the bladder, ureters, and renal collecting system.
- Although physiologically fundamental, it is quite imperative for the sonographer to understand the creation and flow of urine through the urinary tract in order to determine the origin of a urinary tract obstruction.
- Urine is produced by the kidney, exits the kidney by means of the renal pelvis, travels down the ureter, into the bladder, and exits the body via the urethra.
- Any obstruction to this normal succession will result in a back-up of urine.

Ureteropelvic Junction Obstruction

- Ureteropelvic junction (UPJ) obstruction is the most common cause of hydronephrosis in the neonate and the most common form of fetal renal obstruction.
  - The UPJ is located at the junction of the renal pelvis and ureter.
  - It is typically unilateral and more common in males.
- The sonographic appearance of a UPJ obstruction is dilation of the renal pelvis and renal calices.

Common Chromosomal Abnormalities
The Triple Screen

- The triple screen is a maternal blood test that can be helpful in the second trimester at detecting unusual levels of certain proteins or hormones in the presence of chromosomal abnormalities.
  - The three laboratory values that typically comprise the triple screen are alpha-fetoprotein (AFP), estriol and human chorionic gonadotropin (hCG).
  - AFP is produced in the yolk sac and fetal liver. Estriol and hCG are produced by the placenta.
  - The triple screen has a 60% detection rate for Down syndrome.

- Two supplementary proteins that can also be monitored are the PAPP-A, or pregnancy-associated plasma protein A, and the dimeric inhibin A.
  - Both of these proteins are produced by the placenta as well.

Down Syndrome (Trisomy 21)

- Down syndrome, or trisomy 21, is the most common chromosomal abnormality.
  - It occurs in 1 in 500-800 pregnancies.
  - Fetuses with trisomy 21 have an extra chromosome 21.

- Various sonographic features of Down syndrome include:
  - Duodenal atresia
  - Thickened nuchal translucency in the first trimester or increased nuchal fold thickness in the second trimester
  - Pyelectasis (hydronephrosis)
  - Echogenic bowel
  - Absent nasal bones
  - Maternal serum screening outcomes yield evidence of an elevated hCG level, while all other laboratory values are reduced.

Edward Syndrome (Trisomy 18)

- The majority of fetuses diagnosed with Edward syndrome, or trisomy 18, die either before birth or shortly after birth.
  - Fetuses with trisomy 18 have an extra chromosome 18.

- Various sonographic features of Edward syndrome include:
  - Strawberry-shaped skull
  - Choroid plexus cysts
  - Micrognathia
  - Club feet
  - Omphalocele
  - Single umb. artery
  - All laboratory values are decreased with Edward Syndrome.

Patau Syndrome (Trisomy 13)

- Holoprosencephaly and abnormal faces are common findings with Patau syndrome, or trisomy 13.
  - The fetus with Patau syndrome has an extra chromosome 13.
  - Unfortunately, this is almost a uniformly fatal condition, as the fetus typically dies in the neonatal period.
Patau Syndrome (Trisomy 13)

- Various sonographic features of Patau syndrome include:
  - Cyclopia
  - Facial clefting
  - Heart defects
  - Omphalocele
  - Polydactyly
- Maternal serum screening is not always beneficial in the diagnosis of this condition, though a possible decrease in MSAFP and inhibin A may be identified.

Triploidy

- Triploidy is a chromosomal abnormality in which the fetus has 69 chromosomes instead of the normal 46.
  - Specifically, the fetus has three sets of chromosomes, instead of the normal two.
  - Because there are multiple major structural anomalies associated with triploidy, most of the fetuses with triploidy die in the first trimester or early second trimester.

Triploidy

- Often, a partial molar pregnancy is found with a triploid fetus, thus resulting in a markedly elevated human chorionic gonadotropin level and bilateral ovarian theca lutein cysts.
- Sonographic features of triploidy include small, low-set ears, cardiac defects, syndactyly, and intrauterine growth restriction.

Turner Syndrome

- Turner syndrome is a disorder typically found in females.
  - It may also referred to as monosomy X, as most often the paternal sex chromosome is missing.
  - A fetus with this chromosomal anomaly classically presents with a nuchal cystic hygroma and non-immune hydrops.
  - Non-immune hydrops is the buildup of fluid within at least two fetal body cavities.
  - Therefore, ascites, pleural effusions, pericardial effusion, and subcutaneous edema are all common findings with Turner syndrome.

Turner Syndrome

- The sonographic diagnosis is initially suspected when there is visualization of a large, septated cystic hygroma located in the neck.
- Maternal serum screening reveals decreased levels of all laboratory findings when hydrops is present.
- Ovarian dysgenesis, webbed neck, short stature, motor deficits, hearing loss, and renal anomalies are common in those individuals who do survive birth and progress into adulthood.

Which of the following would be least likely associate with polyhydramnios?

1. Potter syndrome
2. Duodenal atresia
3. Tracheoesophageal fistula
4. Esophageal atresia
Which of the following would appear as several cystic spaces within the right renal fossa in an otherwise normal fetus?

1. ARPKD
2. Renal agenesis
3. MCDK
4. ADPKD

What chromosomal abnormality is typically linked with micrognathia?

1. Trisomy 18
2. Trisomy 13
3. Trisomy 21
4. Turner syndrome

Which of the following is most likely associated with oligohydramnios?

1. Potter syndrome
2. Bilateral renal agenesis
3. Unilateral renal agenesis
4. Sirenomelia

Which of the following would be most likely associated with a median facial cleft?

1. Trisomy 18
2. Triploidy
3. Trisomy 13
4. Trisomy 21

Thanks for listening!
I hope you enjoy the rest of the symposium.