Diagnostic Dilemmas

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U/S Detected Fetal Malformation
What Parents Want To Know

- Prognosis (as close as possible)
  - Physical
  - Mental
- Likelihood of other anomalies
- Need for further studies
- Risk of recurrence
Sonographer’s Tools

- Textbooks, (Smith’s Recognizable Patterns of Human Malformations, Mendelian Inheritance in Man)
- Internet, (Pubmed, Ovid)
- Journals, (Ultrasound in Obstetrics & Gynecology, Journal of Ultrasound in Medicine, and Prenatal Diagnosis)
Case #1
11+3/7 Weeks
Case #1
11+3/7 Weeks
Case #1
15+3/7 Weeks
Case #1
15+3/7 Weeks
Ultrasound Description

- Cystic mass in the chest
- Cardiac displacement / abnormal axis
Case # 1

Differential diagnosis

Thoracic Cyst

- Congenital diaphragmatic hernia
- Congenital cystic adenoid malformation
- Bronchogenic cyst
- Esophageal duplication cyst
- Neurenteric cyst
- Teratoma
- Neuroblastoma
Abnormal Cardiac Axis
Differential Diagnosis

- Absent lung
- Intrathoracic mass
- Cardiosplenic syndrome
- Congenital diaphragmatic hernia
- Pleural effusion
- Scimitar Syndrome
Congenital Diaphragmatic Hernia

- Herniation of the abdominal contents into the chest
- 80-90% left sided posterior through foramen of Bochdalek
- 10% right sided anterior through foramen of Morgagni
- <5% bilateral
Congenital Diaphragmatic Hernia
U/S Clues

- Cystic structure in the chest
- Absent stomach
- Peristalsis in the chest
- Hydramnios
- Use color doppler to identify hepatic vessels
- Abdominal circumference small
Congenital Diaphragmatic Hernia
50% have Associated Anomalies

- 30% CNS
- 20% cardiac
- Renal
- Spinal
- Aneuploidy in 16-37% (Trisomy 9, 13, 18, 21, Deletion 4p-, tetrasomy 12p)
Congenital Diaphragmatic Hernia

- Up to 85% contain herniated liver, usually left lobe next to the heart with the stomach displaced posteriorly.
- Can use color Doppler to follow portal vein.
- Right sided may be confused for chest mass and stomach is below the diaphragm, though gallbladder may be in chest.
Congenital Diaphragmatic Hernia

Genetics

- Sporadic 1:2-5K
- Aneuploidy
- Autosomal dominant
- Fryns Syndrome: autosomal recessive
  - Facial and digital abnormalities
  - Cleft lip and palate
  - CNS malformations
  - Micrognathia
Case #1
13+3/7 Weeks

Right Side

Left Side
Case #1
13+3/7 Weeks

Left

Right
Case #1
13+3/7 Weeks
First Trimester
Diaphragmatic Hernia
Case #1
11+3/7 Weeks
Increased NT
Diaphragmatic Hernia

- 1/4000
- Aneuploidy and other anomalies 50%
- Venous congestion probable cause of nuchal edema
- 37% of all cases of CDH
- 83% of those with NND secondary to pulmonary hypoplasia
- 22% of survivors
Case #2
Ascites
Isolated Ascites

- Anechoic fluid in the abdomen
- Rule out pseudoascites
- Rule out hydrops
  - Pleural and pericardial effusion
  - Edema
  - Hydramnios
  - Placentomegaly (>4 cm)
Isolated Ascites Differential

- Perforated bowel secondary to anomaly such as atresias, malrotation, volvulus
- Urinary ascites secondary to PUV, UPJ, UVJ
- Ruptured ovarian cyst
- Cloacal malformation with perforation
- Immune hydrops
- Nonimmune hydrops
Isolated Ascites
Nonimmune Hydrops

- Infection: Parvo B19, CMV, varicella, toxoplasmosis, syphilis
- Aneuploidy: 45X0 and trisomy 21 most common
- Cardiac anomaly or arrythmia
- Fetal mass: CCAM, PS, teratoma
- Chylous ascites
- Placental chorioangioma
Ascites
MCA Doppler

PSV = 27 cm/s
Ascites
Right Kidney
Ascites
Bladder @ 29 weeks

0.27 cm
Right Kidney

R KIDNEY 29 WEEKS
Right Kidney
Posterior Urethral Valve
Uroascites
Case # 3
Right Sided Abdominal Mass
28 Weeks Left Kidney
Right Side Abdominal Mass
28 Weeks
Kidney at 30 weeks

Kidney 55.8 mm
Mass 30 Weeks
Amniotic Fluid At 30 Weeks
Unilateral Solid Pelvic Mass

- Renal Tumor-Mesoblastic Nephroma, Wilms tumor, Rhabdoid tumor
- Multicystic dysplastic kidney
- Autosomal recessive polycystic kidney disease
- Autosomal dominant polycystic kidney disease
- Beckwith-Wiedemann Syndrome
- Adrenal lesions
- Retroperitoneal teratoma
- Crossed fused ectopy
Congenital Mesoblastic Nephroma

- Most common renal fetal neoplasm
Congenital Mesoblastic Nephroma

- Benign mesenchymal renal tumor composed of predominantly of spindle cells
- Diagnosis: solid hyperechoic renal mass + hydramnios, may have ring sign
- May displace abdominal organs and cause bowel obstruction
- Hydrops may occur (AV shunting vs venous obstruction)
- Hypercalcemia
Mesoblastic Nephroma Neonate

- Preterm Delivery
- Hypercalcemia
- Hypertension
- Large abdominal circumference
- Rare recurrence
Mesoblastic Nephroma

Treatment

- Amnioreduction
- Nephrectomy with wide margins
Case # 4
Kidneys 19 Weeks
Kidneys 27 Weeks
Kidneys 27 Weeks

Length 3.7 cm
Ultrasound Findings

- Enlarged echogenic kidneys
- Oligohydramnios
Enlarged Kidneys
Differential Diagnosis

- Autosomal dominant polycystic kidney disease
- Autosomal recessive polycystic kidney disease
- Bilateral multicystic dysplastic kidney
- Beckwith-Wiedemann syndrome
- Meckel-Gruber syndrome
- Trisomy 13
- Normal variant
- Tuberus sclerosis (usually not seen in utero)
Autosomal Recessive Polycystic Kidney Disease

- Ectatic distal convoluted tubules and collecting ducts with increasing volume of the medulla
- Incidence 1/20-50 K
- Gene (PKHD1) on Chromosome 6p12
ARPKD Management

- Amniocentesis / CVS
- Offer termination
- Monitor abdominal circumference
- Predelivery consultation with pediatric nephrologist
- Encourage autopsy
Case #5
22 Weeks Renal Fossa

2.4X1.7 cm
22 Weeks Renal Fossa
22 Weeks Renal Fossa
27 Weeks Renal Fossa

Fetal Head

Fetal Feet
Ultrasound Findings

- Unilateral left sided suprarenal mass
- Homogenously echogenic
- Normal appearing kidneys
- Both adrenals seen
Differential Diagnosis

- Teratoma
- Neuroblastoma
- Bronchopulmonary sequestration
27 Weeks Renal Fossa

Aorta
Subdiaphragmatic BPS

- Bronchopulmonary tissue that does not connect to the tracheobronchial tree with the feeding vessel originating from the aorta
- 10-15% of BPS are subdiaphragmatic
- 90% left-sided
- Spontaneous regression common
- Genetics: sporadic with no recurrence risk
- Associated anomalies in up to 50%
BPS

- MRI or CT after delivery
- Embolization of feeding vessel
- Surgical ligation and resection
Final Case
32 Week IUP
Referred for Decreased Fetal Movement
32 Week IUP
Decreased Movement
Velamentous Cord
Vasa Previa
IVF Patient
Transabdominal View of Cervix
34 Week Twins
Conceived with IVF
34 Week IVF Twins
Transvaginal U/S

FHR 142 bpm
Previa
Transvaginal Ultrasound
Previa
Transvaginal Ultrasound
Vasa Previa
Risk Factors

- Placenta previa
- Low lying placenta
- IVF patients
- Velamentous cord insertion
- Patients with vaginal bleeding
- Always look
Conclusion

- Describe the ultrasound findings of the abnormality
- Confirm by looking at abnormality from at least two different angles
- Look for other anomalies
- Use color Doppler for all cystic lesions, (Aneurysms), and to evaluate feeding vessels
- Always ask “What else could it be”
Conclusion
What Do You Tell The Patient

- Avoid being evasive, they can read your face
- Describe the findings
- Find the silver lining
Abdominal Tubular Cyst
18 Weeks
Abdominal Cyst
18 Weeks
Abdominal Cyst
Abdominal Cyst
Abdominal Cyst
Abdominal Cyst Differential

- Bowel Atresia (duodenal, jejunal, ileal)
- Ovarian cyst
- Enteric duplication
- Mesenteric cyst
- Choleductal cyst
- Hydrocolpos
- Urachal Cyst
- Most cystic abdominal masses are related to the urinary tract
Intestinal Atresias

- Hyperperistalsis
- Bowel contents, (succus entericus) are often echogenic
- Risk is perforation, (always consider this in isolated ascites)
- Normal colon is often prominent in the 3rd trimester with normal caliber 18 mm
Intracerebral Cyst
Intracerebral Cyst
Cranial 3-D Sectional Planes
Intracerebral Cyst Differential

- Glioependymal cyst
- Schizencephaly
- Teratoma
- Dandy Walker continuum
- Porencephalic cyst - replaces damaged brain
- Intracranial hemorrhage (subacute)
- Vein of Galen
- Arachnoid cyst
Arachnoid Cyst

- Smooth marginated anechoic cyst
- Remaining brain normal in most cases
- ACC in 5% of supratentorial cysts
- May have ventriculomegaly
Arachnoid Cyst
Location

- Cerebral convexities
- 1/3 in the posterior fossa
Arachnoid Cyst Management

- Amnio
- Monitor for growth
- Head size may impact timing of delivery