Abstract
Diagnostic ultrasound in the field of obstetrics is used to evaluate fetal anomalies and well-being. These images are used to develop a differential diagnosis that can later be correlated with postnatal outcomes. The following cases are a small sample of recent challenging fetal anomalies at our institution that were detected on prenatal ultrasound.

Objectives
• Interpret ultrasound images of fetal anomalies
• Assimilate the interpretation of images into a differential diagnosis
• Correlate prenatal imaging with postnatal outcomes

Keywords: Fetal anomaly, ultrasound, megacystis microcolon, neuroblastoma, facial hemangioma, goiter, myotonic dystrophy, abdominal wall defect, hydrancephaly.

INTRODUCTION
Diagnostic ultrasound in the field of obstetrics is used to evaluate fetal anomalies and well-being. Fetal ultrasound imaging is also an indispensable skill that has become an integral part of the training of obstetrician gynecologists, sonologists, and sonographers.

The following cases illustrate a series of challenging fetal anomalies at our institution that were detected with prenatal ultrasound. These images are used to develop differential diagnoses and are later correlated with postnatal outcomes.

CASE 1
A 34-year-old gravida 6 para 5004 female had an ultrasound performed for uncertain dates at 15 gestational weeks. During this ultrasound a large cystic mass measuring 24 × 18 × 14 mm, was noted to be occupying the fetal pelvis and abdomen. Her past obstetrical history was significant for gestational diabetes in and a history of a neonatal demise of uncertain etiology (Figures 1 to 3).

The right and left renal pelvices measured approximately 3 mm. The bladder did not empty during the exam and the amniotic fluid volume was normal (Figure 4).

The fetal stomach appeared normal (Figure 5).

An amniocentesis was performed and fetal karyotype was 46 XX. A repeat ultrasound was performed at 18 gestational weeks. At the this time, the right and left renal pelvices measured approximately 6 mm and the cystic pelvic mass had enlarged to 35 × 23 mm (Figures 6 and 7).

An intrauterine bladder tap was performed and urine electrolytes were normal. A second tap was performed two days later and urine electrolytes were again normal, but the beta 2 microglobulin was elevated at 3, which indicated possible poor renal function (Figure 8).

Repeat ultrasound at 24 gestational weeks showed continued dilation of the renal pelvices (14 mm) and of the cystic mass (61 × 79 mm) (Figures 9 and 10).

At 32 gestational weeks polyhydraminos was noted with a maximum vertical pocket of fluid of 14 cm. Two vesicoamniotic shunts were placed to assist in expediting a vaginal delivery (Figures 11 to 13).

The patient presented to labor and delivery at 33 gestational weeks with shortness of breath. A therapeutic amnioreduction was preformed, followed by an induction of labor given the poor fetal prognosis. Spontaneous vaginal delivery of the infant followed with Apgar scores of 3, 4, and 4. Comfort care was given and the infant expired at 2 hours of life.
Figure 1: Sagittal sonogram of fetus at 15 gestational weeks with a cystic mass occupying the pelvis and abdomen

Figure 2: Sagittal sonogram of the cystic mass, measuring 24 mm

Figure 3: Transverse sonogram of the cystic mass, measuring 14 x 18 mm

Figure 4: Right and left renal pelvises at 15 gestational weeks measured 3 mm

Figure 5: Transverse sonogram of the fetal abdomen, showing a normal fetal stomach

Figure 6: Right and left renal pelvises at 18 gestational weeks measured 6 mm
Figure 7: At 18 gestational weeks, the cystic mass measured 35 x 23 mm

Figure 8: Transverse sonogram of the fetal bladder post-tap

Figure 9: Right and left renal pelvises at 24 gestational weeks measured 14 mm

Figure 10: Transverse sonogram of the cystic mass at 24 gestational weeks, measuring 61 x 79 mm

Figure 11: At 32 gestational weeks, maximum vertical amniotic fluid pocket was 14 cm

Figure 12: Vescioamniotic shunt in the fetal bladder
What is your diagnosis?

a. Duplicated collecting system
b. Posterior urethral valves
c. Megacystic microcolon
d. Urethral atresia
e. Urethral reflux

Answer:
c. Megacystic microcolon¹²

An autopsy was performed. On gross external examination, the infant had female genitalia and a distended abdomen (Figure 14).

Plain films showed the vesicoamniotic shunts within the fetal bladder (Figure 15).

Hepatosplenomegaly was noted as well as a markedly dilated bladder and renal pelvises. No posterior urethral valves were noted. A microcolon also was not seen, but pathology examination was felt to be most consistent with megacystic microcolon given the female gender of the infant and the transient nature of microcolon (Figures 16 and 17).
CASE 2

A 29-year-old gravida 1 female was noted on second trimester ultrasound to have a low lying placenta. Fetal anatomy appeared normal (Figures 18 and 19).

A repeat ultrasound was performed at 34 weeks gestation to re-evaluate placentation and a facial mass was noted covering the right eye and nose of the fetus (Figures 20 to 24).

Significant vasculature was noted within the mass on power Doppler (Figure 25).

Intracranial anatomy was normal (Figure 26).

What is Your Diagnosis?

a. Nasopharyngeal teratoma
b. Facial hemangioma
c. Frontal encephalocele
d. Dacrocystocele
e. Proboscis

Figure 18: Coronal sonogram of the fetal face at 18 gestational weeks

Figure 19: Sagittal sonogram of the facial profile at 18 gestational weeks

Figure 20: 34 gestational week fetus with a right-sided facial mass

Figure 21: Sagittal sonogram of the fetal face with a mass involving the fetal nose

Figure 22: Coronal sonogram of the fetal face showed a mass covering the right eye

Figure 23: Coronal sonogram of the facial mass
An elective cesarean section was performed at 39 gestational weeks. The facial hemangioma rapidly involuted post delivery (Figures 27A and B).

**CASE 3**

A 15-year-old gravida 1 female presented to labor and delivery at 36 gestational weeks with elevated blood pressures and new onset proteinuria. Ultrasound examination showed an estimated fetal weight of 2138 grams, which was less than the tenth percentile and the amniotic fluid index was 16 centimeters. A fetal bilobular neck mass was noted measuring 8 cm in largest diameter (Figures 28 to 33).

Significant vasculature was noted within the mass on color Doppler (Figure 34).

Cardiomegaly and a pericardial effusion were also seen (Figure 35).

What is your diagnosis?

a. Fetal goiter
b. Cervical teratoma
Figure 28: Oblique view of the fetal face and neck

Figure 29: Oblique coronal sonogram of the fetal face and neck

Figure 30: Transverse sonogram at the level of the cervical spine showing a bilobed mass

Figure 31: Transverse sonogram of the neck mass, measuring 8.69 x 5.84 cm

Figure 32: Sagittal sonogram of the neck mass, measuring 5.29 cm

Figure 33: Sagittal sonogram depicting the position of the mass in relation to the fetal head and heart
c. Cervical neuroblastoma
d. Cystic hygroma
e. Thyroglossal duct cyst

A fetal MRI confirmed the mass and a cesarean section was performed secondary to head extension and severe pre-eclampsia. An ear, nose, and throat specialist was present at the delivery. The infant had Apgar scores of 4 and 9 and poor respiratory effort, which required intubation. On physical exam a soft, mobile anterior neck mass with a central isthmus was palpable. The diagnosis was fetal goiter. Initial thyroid function tests showed a free T4 of 0.4 ng/dL (0.6 to 2 ng/dL) and a TSH greater than 100 mIU/mL (2.5 to 17.4 mIU/mL), consistent with hypothyroidism. The infant was started on thyroxine 20 mcg orally daily (Figure 36).

On day of life seven, thyroid function tests were within normal limits and direct bronchoscopy confirmed the thyroid goiter. Trachomalacia and left bronchomalacia was also seen. Echocardiogram showed normal left ventricular function. The goiter resolved rapidly.

**CASE 4**

A 21-year-old gravida 1 female had an elevated alpha fetoprotein on maternal serum quadruple screen. An ultrasound was performed at 19 gestational weeks and an abdominal defect was noted. The mass was anterior to the fetal abdomen and measured 6 cm in length (Figures 37 to 40).

The following images show the fetal umbilical cord insertion in relation to the mass (Figures 41A to C).

The fetal sternum was intact (Figures 42 and 43).

An amniocentesis was performed and fetal karyotype was 46 XX. Fetal echocardiogram was normal. A repeat

**Figure 34:** Color Doppler showed significant vasculature within the mass

**Figure 35:** Four chamber view of the heart with a pericardial effusion and cardiomegaly

**Figure 36:** Gross image of the fetal neck with a 7 cm soft, mobile neck mass

**Figure 37:** Sagittal sonogram at 19 gestational weeks with a mass anterior to the abdomen, 6 cm in length
ultrasound at 23 gestational weeks showed a solid and cystic mass anterior to the fetal abdomen (Figures 44 and 45).

The fetal stomach was not visualized (Figure 46).

Figure 38: Transverse sonogram of the abdominal wall defect

Figure 39: The mass measured 3.98 x 2.54 cm on transverse imaging

Figure 40: Transverse sonogram of the mass adjacent to the fetal thorax

Figures 41A to C: Umbilical cord insertion in relation to the mass

What is your diagnosis?

a. Omphalocele
b. Ruptured omphalocele
c. Body stalk anomaly
d. Gastrochisis
e. Bladder extrophy

The infant was delivered by cesarean section at 36 gestational weeks after the patient presented with premature rupture of membranes. Light meconium was noted at the
time of membrane rupture. Apgars scores were 6 and 8 and reduction of the gastroschisis in the delivery room was unsuccessful. A silo was placed and on day of life one the infant required increased respiratory support and developed compartment syndrome with multiple organ failure. The bowel and liver became necrotic because reduction was not possible and support was withdrawn on day of life 3.

**CASE 5**

A 22-year-old gravida 2 para 1 female had an ultrasound performed at 11 gestational weeks that was significant for a fetal midgut herniation. A repeat ultrasound at 15 gestational weeks continued to show an abdominal wall defect. The
fetal stomach was not visualized. Amniocentesis was performed and fetal karyotype was 46 XX (Figures 47 and 48).

The following figures show the cord insertion in relation to the mass (Figures 49A and B).

The upper and lower extremities were moving normally. (Figures 50 and 51).

A repeat ultrasound at 18 gestational weeks showed limited movement in the upper and lower extremities and posturing that was suggestive of contractures (Figures 52A to 53B).

At 20 gestational weeks, continued contractures were noted at the elbows, wrists, ankles, and knees. Flexion or extension was absent at these joints. The abdominal defect was still present (Figures 54 to 56).

The abdominal defect was re-examined by ultrasound at 24 gestational weeks. The fetal stomach was still absent (Figures 57A and B).
Figures 52A and B: Upper extremities at 18 gestational weeks with contractures

Figures 53A and B: Lower extremities at 18 gestational weeks with contractures

Figure 54: Upper extremities at 20 gestational weeks with contractures

Figure 55: Lower extremities at 20 gestational weeks with contractures

Figure 56: Transverse sonogram at 20 gestational weeks of the fetal abdomen with the cord insertion and the abdominal wall defect

What is your diagnosis?

a. Restrictive dermopathy
b. Distal arthrogryposis
c. Spinal muscular atrophy
d. Multiple pterygium syndrome
Answer:

c. Spinal muscular atrophy.

The infant delivered at 37 gestational weeks by cesarean section. Apgar scores were 4, 6 and 9. An abdominal defect superior to the umbilicus with herniation of the total abdominal viscera including the liver was present. This defect was surgically reduced. Arthrogryposis was noted in the infant’s extremities. A muscle biopsy of the latissimus dorsi showed absent anterior horn cells, consistent with spinal muscular atrophy. Poor muscle integrity may have contributed to the unusual location of the abdominal wall defect. The infant required numerous abdominal surgeries including a small bowel resection for necrotizing enterocolitis. The infant subsequently developed multiple organ failure and support was withdrawn at 6 weeks of life.

CASE 6

A 26-year-old gravida 1 female underwent fetal ultrasound evaluation at 21 gestational weeks. The fetal face and profile was normal (Figures 58 and 59).

However, the fetal intracranial anatomy was abnormal. (Figures 60A to C).

The cerebellum measured appropriately for the fetal gestational age (Figure 61).

The lateral ventricle measured 5 mm (Figure 62).

What is your diagnosis?

a. Porencephaly
b. Subdural hemorrhage
c. Holoprosencephaly
d. Hydrocephalus
e. Hydranencephaly

The pregnancy was terminated by misoprostol induction. An autopsy was performed and massive cerebral necrosis consistent with hydranencephaly was present. Periventricular infarctions and intraventricular grade 2 hemorrhage was seen in the third ventricle. The placental was small and underperfused with a superficial implantation site.
CASE 7

A 29-year-old gravida 1 female presented to labor and delivery at 30 gestational weeks with complaints of vaginal bleeding. Her pregnancy was complicated by a posterior complete placenta previa. On prior ultrasound examinations, fetal anatomy was normal. However, admission ultrasound showed that the fetal abdominal circumference measured greater than the ninetieth percentile and an intraabdominal mass was seen (Figures 63 and 64).

Color flow was noted around the mass (Figure 65).

Arterial blood flow was seen within the mass (Figures 66A and B).

Ascities was seen around the fetal liver and echodensities with the liver (Figure 67).

At 32 gestational weeks, an additional heterogeneous echolucent structure was seen on the left side of the fetal abdomen (Figures 68 and 69).

Color flow was noted within the mass (Figure 70).

What is your diagnosis?

a. Adrenal hemorrhage
b. Nephroblastoma (Wilms’ tumor)
c. Pheochromocytoma
d. Mesoblastic nephroma
e. Neuroblastoma

The patient had heavy vaginal bleeding at 32 gestational weeks and an emergent cesarean section was performed for the indication of bleeding placenta previa. The infant had Apgar scores of 2, 7 and 7 and was intubated for respiratory depression. The placenta was examined and had significantly infiltrated by neuroblastoma. The infant developed multiple organ failure and compartment syndrome. An exploratory laparotomy with mesh placement was performed.
performed. The infant’s condition deteriorated and support was withdrawn on day of life 3. Due to the impressive tumor burden in the placenta, the infant’s mother is underwent a metastatic neuroblastoma evaluation which was negative.

**CONCLUSION**

Obstetrical ultrasound imaging is the most important tool for diagnosing fetal anomalies. Ultrasound is the essential element from which a multidisciplinary plan for the pregnant patient and newborn is created.
Figure 68: Sagittal sonogram of the fetal abdomen at 32 gestational weeks with an additional intraabdominal mass measuring 6.79 x 1.47 cm

Figure 69: Transverse sonogram of the additional intra-abdominal mass measuring 4.18 cm

Figure 70: Power Doppler of the vasculature surrounding and in the mass

REFERENCES


