Intraabdominal Cysts Turn Out to Be Distended Large Bowel – An Electively Terminated Fetus with Isolated Imperforate Anus
Yu Shi, MD¹, Maria A. Giraldo-Isaza, MD² and John L. Farber, MD¹
¹Departments of Pathology, ²Maternal Fetal Medicine, Thomas Jefferson University Hospital, Philadelphia, Pennsylvania

Patient
Clinical history: A 19-week male fetus of a 36-year old G7P4 mother was found by ultrasound to have intrabdominal “cysts” and oligohydramnios. At 15 weeks of pregnancy, by ultrasound examination, two intrabdominal cysts measuring 1.1x0.7x0.4 cm and 1.4x1.0x1.0 cm were noted. At 19 week gestation, the ultrasound was performed again showing anhydramnios, ascites, a normal appearing bladder, and multiple cystic structures in the abdomen most likely consistent with dilation of bowel (Figure1A, B). The mother was consulted and opted to proceed with KCL injection and induction of labor. The nonviable fetus was delivered at 19 5/7 week gestation. Rhogam was administered since the mother is Rh negative.

Past Medical/Surgical History
• Heterozygous for Factor V leiden with deep venous thrombus (DVT) diagnosed about 7 years ago
• Rh negative

Allergies
• No Known Drug Allergies

Medications
• On chronic Coumadin prior pregnancy, switched to lovenox and then heparin daily at the beginning of this pregnancy.

Social/Family History
• Smoking 1 pack per day prior to pregnancy, ½ pack per day thereafter. Denies alcohol and illicit drug uses. Family history is negative for birth defects or mental retardation.

Lab Data
Kleihauer-Betke: No Fetal Cells Seen
RPR Result: Nonreactive

Imaging and Autopsy Findings

Figure 1. Abdominal sonography showing multiple abdominal cysts and anhydramnios. A, Coronal image of the fetal abdomen and pelvis in the prone position. Asterisk indicates dilated fluid-filled distal colon; B, on the sagittal scan, the rectum is dilated (asterisk) with no visible amniotic pocket.

Figure 2. Gross examination of the macerated fetus. A. The fetus has abdominal distension with no facial or digital anomalies. B. The fetus shows ambiguous genitalia in the supine position. C. This photograph is taken from the inferior aspect of the fetus in the prone position and shows the perineal atresia. D. The distended large bowel is responsible for the echogenically cystic lesion (Asterisk). E. Bilateral ureters are dilated and tortuous (arrows).

Figure 3. Micrographs of the atretic rectum with presence of ganglion cells to rule out Hirschsprung’s disease. Arrows indicate a few immature ganglion cells in the submucosa. The mucosa is denuded due to maceration. Scale bars, 2mm.

Discussions
The fetus was found by ultrasound to have intrabdominal cysts and oligohydramnios. Gross examination identified that the large bowel ends blindly with anal imperforate. The distended bowel contains meconium and is responsible for the echonically cystic lesion. In addition, the ureters are dilated and tortuous suggesting urinary flow blockage, which can cause oligohydramnios.

The incidence of imperforate anus is 1 in 4000 to 5000 live births (1). Imperforate anus is often associated with the VACTERL (vertebral, anal, cardiac, tracheal, esophageal, renal, and limb) anomalies prompting further evaluation postnatally (1). However, imaging studies (echo-cardiogram and ultrasound) and gross examination were negative for fistula formation, cloacal anomalies, or VACTERL association. Clinic atresia may occur in combination with Hirschsprung disease. The presence of ganglion cells ruled out Hirschsprung megacolon. Cases of isolated imperforate anus are reported. The etiology of imperforate anus is unknown. There have been several cases of monozygotic twins concordant for imperforate anus, drawing attention to a genetic causation (2).

There is no report associating warfarin use with bowel atresia or imperforate anus. The teratogenicity of warfarin or warfarin embryopathy fetuses having the characteristic nasal hypoplasia (3), which are not seen in this case.

References