Prenatal Diagnosis of an Inguinoscrotal Hernia in a Fetus With Cystic Fibrosis

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A detailed anatomic survey of the fetus by an experienced sonographer can detect a multitude of structural anomalies. Although relatively common in the neonate, an inguinoscrotal hernia is a condition that is rarely seen prenatally. In a male fetus, this abnormality typically appears in the third trimester as a complex scrotal mass. Active intestinal peristalsis within an enlarged scrotum is diagnostic of this condition. We report a case that involves the prenatal association between cystic fibrosis and a congenital inguinal scrotal hernia. Serial sonograms from 30 to 37 weeks’ gestation provided the opportunity to follow both the abnormal bowel appearance and the progression of the hernia. Postnatal follow-up confirmed both diagnoses.

Case Report

A 26-year-old married woman was referred to the Regional Perinatal Center for counseling concerning the couple’s confirmed cystic fibrosis carrier status. The patient’s cystic fibrosis genetic mutation is written as 1898+1, G>A. The father of the fetus was identified as carrying the most common cystic fibrosis gene change, ∆F508. Prenatal genetic testing was offered and declined. Because sonography is nondiagnostic for cystic fibrosis, the couple was provided with a cord blood kit and instructions for testing at the time of delivery.

At 17 weeks’ gestation, a detailed obstetric sonogram obtained with a 3.5-MHz curvilinear transducer (Acuson 128XP/10OB; Siemens Medical Solutions, Mountain View, CA) was unremarkable. On follow-up at 30 weeks’ gestation, echogenic loops of bowel were noted in the lower abdomen. At 34 weeks, the prominent fetal bowel pattern was highly suggestive of cystic fibrosis (Figure 1). In an attempt to confirm the sex, an incidental finding of...
the external genitalia was noted. The fetus was identified as male, and a left-sided inguinal scrotal hernia was noted. It appeared as a complex mass in the left hemiscrotum with active peristalsis observed. The left scrotal mass measured $36 \times 30$ mm, and the right side measured $24 \times 18$ mm. The right testis was identified within its scrotal sac. The left testis could not be isolated from the small bowel within the scrotum. The question was raised of whether the unusual bowel pattern within the fetal abdomen could be secondary to the hernia or a result of a fetus affected with cystic fibrosis. A follow-up scan at 37 weeks’ gestation showed that the left hemiscrotum had increased to $48 \times 32$ mm, and the right side measured $30 \times 19$ mm. Sequential sonographic images documented the active peristalsis within the left scrotal sac (Figure 2). The amniotic fluid volume was normal throughout gestation. The bowel in the lower abdomen appeared dilated and filled with echogenic meconium.

The patient went into labor and had a spontaneous vaginal delivery of a 2676-g male neonate with Apgar scores of 8 at 1 minute and 9 at 5 minutes. On physical examination, there was a large but very easily reducible left inguinal hernia (Figure 3). Intermittent spitting and feeding intolerance were also observed in the neonate. The cord blood sample was obtained, and genetic testing resulted a compound heterozygote for $\Delta F508$ and the 1898+1, G>A mutation, which is consistent with a diagnosis of cystic fibrosis.

A single supine radiograph of the abdomen showed findings of a dilated bowel loop, suspected to be a sigmoid colon, seen beneath the inguinal ligament and coursing into the left scrotal sac (Figure 4). Fairly dramatic colonic gaseous

**Figure 1.** In the third trimester, a prominent abnormal fetal bowel pattern is shown on this cross-sectional sonogram. This sonographic appearance is similar to that observed in documented cases of cystic fibrosis.

**Figure 2.** At 37 weeks’ gestation, sequential sonographic images of the fetal scrotum help show the peristalsis of the bowel within the left hemiscrotum (arrowheads).
distention, particularly involving the transverse colon, was observed. The small bowel was outlined by gas but was not distended. At 3 weeks of age, the neonate underwent surgical repair of the hernia. At the time of surgery, in addition to the presence of the large left inguinal hernia, a small right hernia sac was also repaired, and a routine circumcision was performed. The neonate tolerated the procedure and was sent home in satisfactory condition. The cystic fibrosis was to be treated medically by the cystic fibrosis clinic.

Discussion

Inguinal hernias are among the most common surgical conditions of infancy and childhood.1 Several case reports have introduced the ability to prenatally diagnose this condition with the use of high-resolution obstetric sonography. A review of the journal literature establishes a consistent sonographic appearance of this abnormality involving observable peristalsis of the fetal bowel within an enlarged scrotum in the third trimester.2–6 In neonates, the prevalence is 1% to 5%, with a male-female ratio of 9:1. Approximately 60% are on the right side; 25% are on the left side; and 15% are bilateral.1 An increased prevalence of indirect inguinal hernias is seen in children with positive family histories of hernias, cystic fibrosis, congenital dislocation of the hip, connective tissue disorders, undescended testes, ambiguous genitalia, hypospadias, epispadias, ascites, and congenital abdominal wall defects. In addition, up to 50% of female neonates with testicular feminization have an inguinal hernia. The premature neonate has a higher prevalence of inguinal hernias and incarceration. Up to 7% of boys born earlier than 30 weeks' gestation have inguinal hernias compared with only 0.6% of boys born later than 36 weeks' gestation. In addition, the prevalence of hernias in premature neonates weighing less than 1500 g is 20 times greater than in larger neonates, with a prevalence of incarceration approaching 30% in this patient population.6

During the third month of gestation, the processus vaginalis develops as an outpouching of the peritoneum in the region of the internal ring, and it descends along the inguinal canal to the scrotum. The testes, which are initially located within the urogenital ridge in the retroperitoneum, descend to the area of the internal ring by approximately 28 weeks' gestation and descend into the scrotum through the inguinal canal external to the processus vaginalis by approximately 29 weeks' gestation. The ovaries
also descend into the pelvis from the urogenital ridge but do not exit the abdominal cavity. The processus vaginalis in girls extends into the labia majoris through the inguinal canal. During the last few weeks of gestation or shortly after birth, the layers of the processus vaginalis normally fuse together and obliterate the entrance to the inguinal canal in the vicinity of the internal ring. Failure of obliteration results in a variety of inguinal anomalies, including hernias.6

An inguinal hernia usually appears as a bulge in the inguinal region and extends toward or into the scrotum. The parent is usually the first person to notice this bulge, which may appear only during crying or straining. Physical examination reveals an inguinal bulge at the level of the internal or external ring or scrotal swelling that is easily reducible or fluctuates in size. The treatment of choice is surgical repair. An inguinoscrotal hernia does not resolve spontaneously. The operation should be carried out electively shortly after diagnosis because of the risk of incarceration. The results of inguinal hernia repair in infants and children are excellent, with a 2% complication rate.6

As the imaging capabilities of diagnostic sonography equipment improve, they increase the observer’s ability to identify a multitude of abnormalities that have not been recognized with consistency in the past. Although many of these findings may not be life threatening, there is an opportunity for the parents and the care provider to be prepared. Concerns can be discussed before delivery, which may alleviate stress and confusion in the immediate postnatal period. One purpose of this report is to acknowledge the association between cystic fibrosis and a congenital inguinoscrotal hernia. More importantly, a review of several ultrasound texts and atlases failed to discover any mention of congenital inguinal hernias or to find any sonographic images as references.8–11 This case may serve as an educational tool by providing visual correlation between the prenatal sonographic appearance in conjunction with radiography and a photograph of the actual infant.

References