Fetal small bowel obstruction: report of two cases

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Summary

Fetal small bowel obstruction is usually diagnosed by sonography in the late second or early third trimester. We report two such cases of different etiology: a case of meconium ileus due to cystic fibrosis and a case of intestinal atresia. The only sonographic finding that allowed differentiation between them was the echogenic bowel in the case of cystic fibrosis.

Key words: Fetal small bowel obstruction; Meconium ileus; Cystic fibrosis; Intestinal atresia.

Introduction

Sonographic diagnosis of fetal small bowel obstruction relies on demonstration of multiple interconnecting, overdistended bowel loops with active peristalsis often seen during real-time observation. We report two such cases identified antenatally which had different etiology: a case of meconium ileus due to cystic fibrosis and a case of intestinal atresia.

Case Report 1

A 23-year-old primipara presented for the first time to our department in the 32nd week of gestation. Ultrasound examination revealed marked distention of the small bowel loops (length up to 25 mm and diameter 15 mm) with vigorous peristalsis. The fetal abdomen was distended – abdominal circumference corresponded to 35 weeks of gestation – while the biparietal diameter and the femur length corresponded to 32 and 31 weeks, respectively. Other findings were polyhydramnios and an abnormal area of increased abdominal echogenicity (Figure 1). There were no other abnormalities seen and the gallbladder was inconspicuous. In the follow-up ultrasounds the findings were stable and an amniocentecis revealed a normal 46 XY karyotype. There was no evidence of fetal distress in the biophysical profile score, computerized cardiotocography or Doppler studies. At the 39th week of gestation a 3,790 g neonate was delivered by selective caesarian section, with Apgar scores of 7 and 9 at 1 and 5 min, respectively. Physical examination of the neonate revealed marked abdominal distention with large veins seen coursing over the abdomen and splayed scalp sutures. During the first day of life a laparotomy was performed and rupture of the ileus with obstruction of the distal segment by thick meconium were found. Denaturing gradient gel electrophoresis (DGGE) established the diagnosis of cystic fibrosis as the neonate was a compound heterozygote (R1070Q / N1303K). On day 13 of life the neonate was re-operated due to adhesional ileus, and on the 48th day developed klembsiella septicemia and died.

Case Report 2

A 28-year old para 2 (previous caesarian section) also presented for the first time to our department at the 36th week of gestation, with ultrasonographically demonstrated marked distention of the small bowel loops (length up to 46 mm and diameter 26 mm) and hyperperistalsis. The fetal abdomen was distended – abdominal circumference corresponded to 39 weeks of gestation – while the biparietal diameter and the femur length corresponded to 35 and 33 weeks, respectively (Figure 2). The quantity of the amniotic fluid was increased and in the follow-up studies there was no evidence of fetal distress. In the 38th week of gestation a 2,500 g female neonate was delivered by selective caesarian section, with Apgar scores of 6 and 7 at 1 and 5 min, respectively. Physical examination of the neonate revealed a distended painless abdomen, bile-stained secretions and slight cyanosis. On day 2 of life a laparotomy was performed and type II intestinal atresia was found to be the cause of fetal small bowel obstruction (Figure 3). Urinary tract studies, which were performed later because of Candida albicans urinary infection, were normal. The neonate finally survived and thrives well.

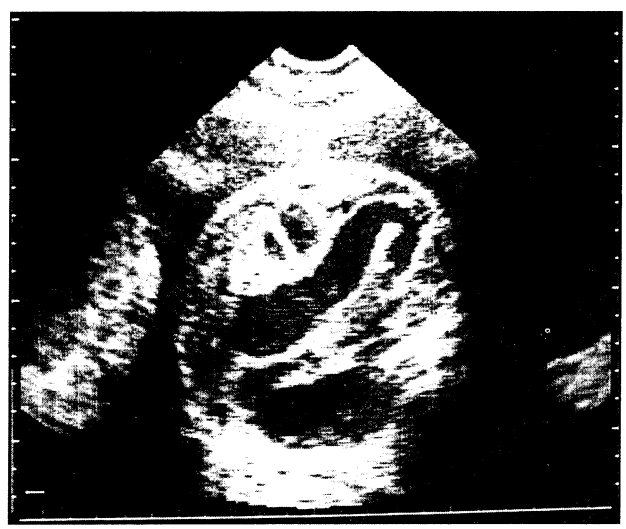

Figure 1. — Dilated loop of small bowel located centrally with echogenic abnormal area anteriorly in the fetus with cystic fibrosis.

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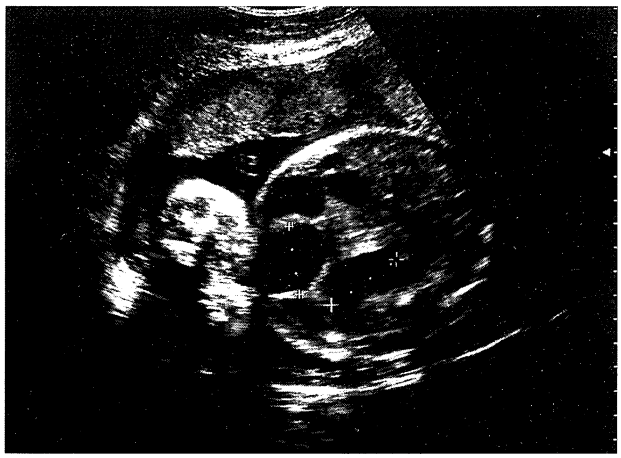

Figure 2. — Dilated loops of small bowel located laterally in the fetus with intestinal atresia.

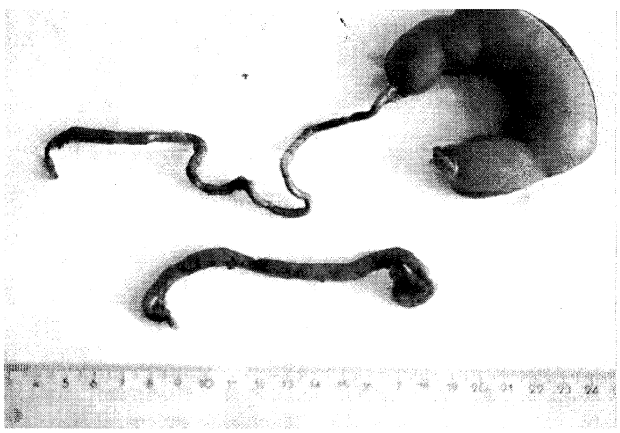

Figure 3. — Type II atresia of the ileum.

Discussion

Cystic fibrosis (CF) is an inherited multisystem disorder characterized chiefly by obstruction and infection of the airways and by maldigestion and its consequences, with an average lifetime of 25-30 years. In Greece, CF occurs in approximately 1/2,500 live births [1] and according to the Hardy-Weinberg law heterozygote frequency in the population is 4%. CF is inherited as an autosomal recessive trait and is caused by more than 800 mutations in CFTR, a gene that is located on chromosome 7 and codes for a 1480 amino acid membrane protein, the cystic fibrosis transmembrane conductance regulator (CFTR) [2]. In the Greek population, a panel of common mutations that will account for 85.6% of all CFTR mutations may be assayed, with the ΔF508 being the most frequent mutation [1]. Thus it is feasible that 73% of heterozygote couples can be detected.

On the other hand, small intestine atresia is a major cause of neonatal intestinal obstruction with a reported incidence of 1/330 to 1/1,500 live births, and is equally distributed between the jejunum (51%) and ileum (49%). There are four types of small intestine atresia: type I (32%) characterized by mucosal (septal) atresia with an intact bowel wall and mesentery; type II (25%) where the two atretic blind ends are connected by a band of fibrous tissue (cord) and the mesentery is intact, and type IIIa (15%) where the two atretic blind ends are separated by a gap (V-shaped defect) in the mesentery; type IIIb (11%) or "apple-peel atresia", and type IV (17%) of multiple atresias (string of sausages). The interruption of the blood supply to a segment of bowel during fetal development seems to be the main mechanism of atresia, though in some cases failure of recanalization has been proposed as the underlying cause. Associated abnormalities and chromosomal defects are rare [3].

Fetal small bowel dilation occurs in a variety of disorders, including both obstructive and non-obstructive causes (congenital chloride diarrhea and megacystis-microcolon-intestinal hypoperistalsis syndrome), the second being more rare [3]. The most common obstructive cause of fetal small bowel dilation is intestinal atresia, while the prevalence of CF is reported to be 33%. Five to 15% of neonates with CF have meconium ileus, while other manifestations of CF include ileal or jejunal atresia, meconium plug syndrome and meconium peritonitis secondary to bowel perforation. On the other hand, 80% of neonates with meconium ileus will have CF [4]. Bowel obstruction in fetuses with meconium ileus due to CF is thought to result from impaction of abnormally thickened, viscous meconium. From the comparison of the ultrasonographic findings of the present two cases, only the area of increased abdominal echogenicity differentiated the case of CF. Concerning the prognoses of these two cases, it is evident that CF carries the worst postoperative risk for the neonate. However, the efficient detection of the heterozygous parents and the prenatal diagnosis by CVS or amniocentesis of the affected fetus (25% risk of being affected) minimizes the risk of an unwanted birth. Furthermore, ultrasonograhically hyperechoic bowel of the second trimester should raise the possibility of CF (13.3%) [5] and amniocentesis would clarify the underlying cause of this ambiguous finding. It is important to report that, except from a transient normal variant, other unfavorable outcomes associated with hyperechoic bowel in the second trimester may be fetal infections (cytomegalovirus and toxoplasmosis), chromosomal abnormalities (triploidy, trisomies 13, 18 and 21) (20%), increased perinatal mortality (16.7%) and intrauterine growth retardation (23.3%) [3, 5, 6]. However the observer's subjectivity in detecting and interpreting this finding mandates an evidence-based consensus in managing the second trimester fetus with hyperechoic bowel.

In conclusion, it is difficult to distinguish sonographically meconium ileus from small bowel atresia, and these two entities may coexist. Possibly only the echogenic bowel would allow differentiation between these two cases.

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