

A case of Fetal intestinal volvulus and Cystic fibrosis.

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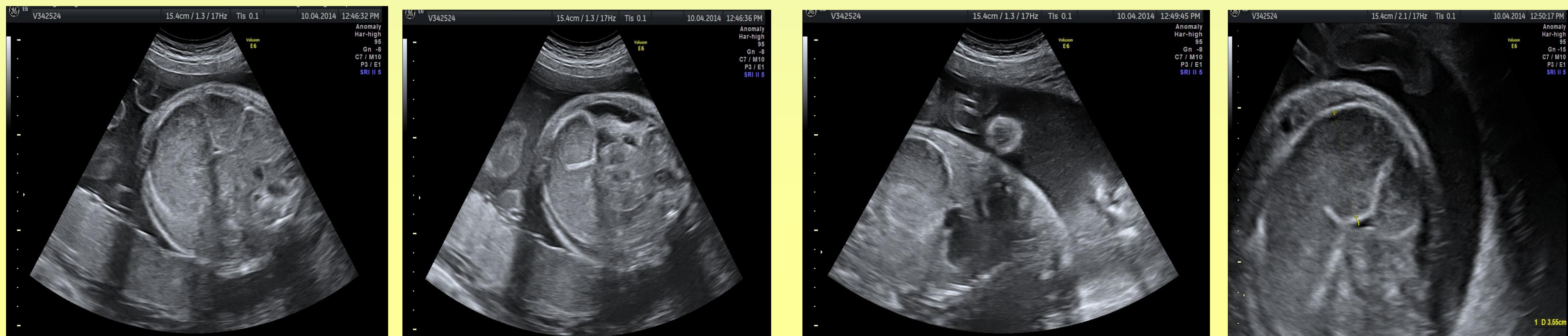
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Introduction:

Fetal intestinal volvulus can be diagnosed in utero in presence of dilated fetal bowel. It can be associated with Cystic fibrosis (CF) (meconium ileus), intestinal obstruction/atresia, and cloacal malformations. We present a case of incidental finding of fetal volvulus that lead to diagnosis of CF, that needed neonatal intestinal resection but with a satisfactory outcome

Case:

A 30-year-old, Caucasian, primigravida attended labour ward at 32 weeks gestation with reduced fetal movements. On ultrasound normal fetal growth was noted. However, there were dilated bowel loops, ascites and polyhydramnios. Bloods were taken to exclude TORCH infection, and an amniocentesis was carried out to check for Cystic Fibrosis. An elective c-section was performed at 32 +2 weeks because of reduced fetal movements and suspected volvulus. A female baby was born and underwent bowel resection because of confirmed necrotic volvulus. Amniocentesis results confirmed the diagnoses of compound heterozygote Cystic Fibrosis. Fetus is a carrier for p.Phe508 mutation



Discussion:

- CF is the most common inherited disease in white populations. The prevalence is 1 in 2,500 newborn, with calculated carrier frequency of 1 in 25.
- This case is an unusual presentation of Cystic Fibrosis. The most frequent ultrasound finding in fetus with CF is a hyperechoic bowel. However, the presence of fetal volvulus can be associated with CF.
- Unlike postnatal volvulus, most prenatal volvulus occurs without intestinal malrotation and is a life-threatening condition due at risk of intestinal necrosis, perforation, peritonitis and preterm delivery.
- In addition to dilated loops of bowel, the “whirlpool sign” and the “coffee bean” or “kinked loop sign” are signs of intestinal volvulus.
- In the prenatal diagnosis of CF the only risk factor is a family history of the condition.
- Hyperechoic bowel is a non specific finding of the second trimester that can occur as a normal variant or in association with intrauterine infection, aneuploidy, CF and other entities.
- About 15% of infants with CF are born with meconium ileus, and nearly 50% are classified as complicated.
- Meconium ileus is the impaction of abnormally thick and sticky meconium in the distal ileum as a result of pancreatic enzyme deficiency; the distal ileum is plugged by thick meconium, leading to a functional obstruction. Around 95% of these cases have CF.
- This case emphasizes the role of examination of the fetal bowel during growth ultrasounds in the third trimester and the importance of quick decision to delivery in cases of suspected fetal volvulus may prevent bowel necrosis.
- Prenatal screening for CF should be indicated in all pregnancies with ultrasound patterns of specific intestinal disorders, specially bowel obstruction or volvulus.

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