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CASE REPORTS

Altering Appearance of Fetal Enteric Duplication Cysts: The Gut Signature Sign and Other Indications for Prenatal Diagnosis

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Abstract Gatrointestinal duplication cysts are congenital anomalies that can be associated with severe adverse outcomes, such as hemorrhage, intestinal obstruction, or intestinal torsion. Despite their substantial postnatal impact, antenatal scans have been reported to identify only 20–30% of such cases. Although the gut signature sign is considered to be relatively specific in the diagnosis of enteric duplication cysts, the classic five-layered appearance is not always easy to demonstrate in the prenatal setting. We present a case of a fetal enteric duplication cysts that presented as a migrating, peristaltic cyst prenatally with the positive 'bilayer sign.' Given the challenging circumstances for diagnosis, we also provide a few insights on the altering appearance of the gut signature sign and describe other practical ultrasonic features used to diagnose enteric duplication cysts.

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Keywords Enteric duplication cyst · Bowel duplication cyst · Prenatal diagnosis · Gut signature sign · Congenital anomalies · Bilayer sign

Background

Duplication cysts are rare congenital gastrointestinal anomalies (1:4500 live births) that can be associated with severe adverse outcomes, such as hemorrhage, intestinal obstruction, or intestinal torsion. Despite their substantial postnatal impact, antenatal scans have been reported to identify only 20–30% of such cases.[1] Although the gut signature sign is considered to be relatively specific in the diagnosis of enteric duplication cysts, the classic five-layered appearance is not always easy to demonstrate in the prenatal setting. Owing to the diminished permeability of the ultrasonic beam through duplication cysts, their walls are detected as simple, linear linings. Herein, we present a case of a fetal enteric duplication cyst that presented as a migrating, peristaltic cyst prenatally with the positive 'bi-layer sign.' Given the challenging circumstances for diagnosis, we also provide a few insights on the altering appearance of the gut signature sign and describe other practical ultrasonic features used to diagnose enteric duplication cysts.

This is the first case to report serial ultrasonic findings of duplication cysts throughout pregnancy, and indicate four features for the identification of duplication cysts: the gut signature sign, peristalsis, altering position (depending on the site of the duplication), and altering size.

Case Report

A 34-year-old pregnant woman, gravida 2 para 1, was referred for a fetal abdominal cyst detected at 27 + 0 gestational weeks. Fetal ultrasound was performed with 2–6 MHz RM6C convex probes (Voluson E10, GE, Tokyo Japan), and showed a low echogenecity cyst (22 × 19 mm) with peristalsis, located right above the urinary bladder. The cyst changed its location during pregnancy. A migrating cyst with peristalsis indicated mesenteric or intestinal origin and we, therefore, evaluated the cyst wall to detect the characteristic sign of intestinal wall layers, also known as the "gut signature sign." At 31 + 6 weeks, the cyst was confirmed to have two partial linings, an appearance suggestive of the gastrointestinal wall (Fig. 1), known as the 'bi-layer sign'. The cyst enlarged at 32 + 6 weeks (41.9 × 35.9 mm), compressing the duodenum at the time and diminished in size at 35 + 6 weeks (19.3 × 16.5 mm). The bi-layer sign was more evident at 36 + 6 weeks (Fig. 2). A fetal enteric duplication cyst was considered to be the most likely diagnosis. The classic five layered lining of the gut signature sign was not evident at the time. As the cyst shrunk and acquired ridges, the gastrointestinal wall was more detectable, making the inner hyperechoic mucosa and outer hypoechoic muscular layer visible.

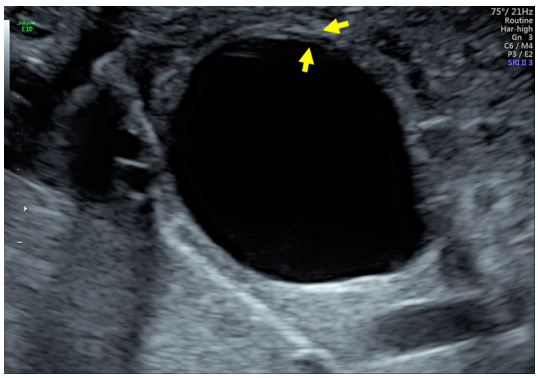

Fig. 1 At 31 + 6 gestational weeks; BMI(body mass index) of the pregnant woman: 21.2, AFI(amniotic fluid index): 13.9 cm. The arrows show two partial linings of the cyst wall (bi-layer sign), an appearance suggestive of gastrointestinal wall

The mother gave birth to a 2760 g female baby at 39 + 0 weeks. Postnatal transabdominal ultrasound clearly detected the bi-layer sign of the duplication cyst, that possessed a common muscular layer with the bowel. The shared muscular layer had not been detected prenatally. The bi-layer sign appeared to be more obvious in the postnatal setting, presumably due to the increased permeability of the ultrasound beam (Fig. 3a).

The surgery was performed at 19 days post-partum. Intraoperatively, a duplication cyst was found 5 cm above Treitz' ligament, confirming the diagnosis. The pathological findings of the typical gastrointestinal layers are shown in Fig. 3b, as contrasted with the postnatal ultrasound scan. There was a tiny fistula between the jejunum and the mass,

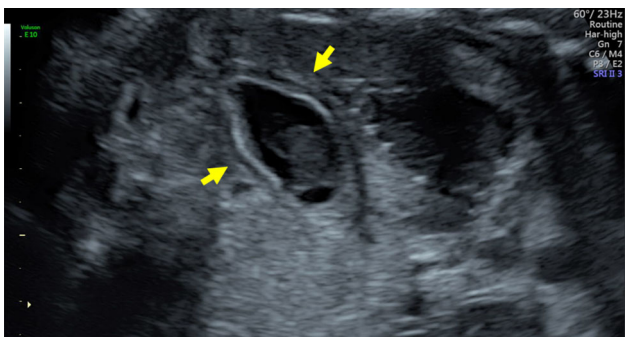

Fig. 2 At 36 + 6 gestational weeks; BMI of the pregnant woman: 21.9, AFI: 16.3 cm. The cyst has diminished in size and the cystic wall is wrinkled, enabling us to detect the bi-layer sign more evidently

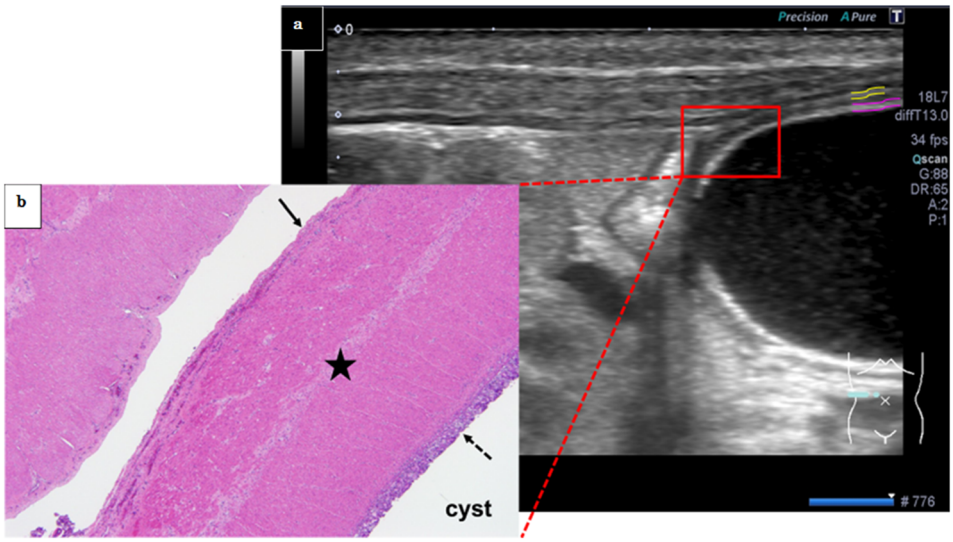

Fig. 3 A high-resolution linear probe shows two layers that correspond to the hyperechoic inner layer representing mucosa, surrounded by a hypoechoic external layer, representing the muscular layer (**a**). Hyperechoic serosa can be detected when the ultrasonic beam penetrates perpendicularly to the cyst wall (yellow line as serosa, pink line as mucosa). Pathological findings show three layers of the cystic wall (serosa (solid arrow), muscularis propria (★) and mucosa (dotted arrow)) (**b**). Submucosa of the duplication cyst is not formed, and the cystic serosa is rather thin compared to the normal bowel. This explains why all classic five layers of the enteric duplication cysts are not detectable

explaining the repetitive change in the size of the cyst. The postoperative course was uneventful and the baby was discharged on postoperative day 14.

Opt-out consent was obtained from the patient for the publication of this manuscript and its accompanying images.

Discussion

The authors indicate four features for the identification of duplication cysts observed in this case: the gut signature sign, peristalsis, altering position (depending on the site of the duplication), and altering size.

The gut signature sign is a term used to describe the appearance of the gastrointestinal wall. It represents five layers, corresponding to the innermost hyperechoic mucosa, hypoechoic muscularis mucosa, hyperechoic submucosa, hypoechoic muscularis propria, and the outermost hyperechoic serosa [1–3]. However, not all five layers are detectable consistently; the term then also refers to a hyperechoic inner layer, representing the mucosa, surrounded by a hypoechoic external layer, representing the muscular layer [2, 4, 5]. Therefore, it rather presents as a bi-layer in the prenatal setting [1, 4, 6]. When the direction of the high frequency ultrasound beam is perpendicular to (90 degrees) the cyst wall, the gut signature sign tends to be more evident as a result of receiving more beam reflections from the cyst wall. This explains why the characteristic layered appearance was observed only partially in some sections of the examination (Fig. 1). For the same reason, this sign can be detected clearly when the cyst wall is thickened or wrinkled (Fig. 2).

Peristalsis is another distinct feature that indicates that the tumors are intestinal in origin. Cysts of other origins such as ovarian or from Meckel's diverticulum do not have peristaltic walls and can easily be differentiated from duplication cysts. However, these cysts can also present a layered wall that could simulate the gut signature sign [3, 7]. Confirming peristalsis when a bi-layered cystic wall is present is highly useful to differentiate intestinal duplications from other abdominal cysts. Change in the location of the cysts is also a sign suggesting intestinal origin since bowel is only partially fixed to the retroperitoneum.

Change in the size of duplication cysts has been reported in several reports [4, 8]. Although altering size is not a unique finding suggestive of duplication cysts, it could be explained by the fact that some duplication cysts have communication with the bowel. There are two general types of gastrointestinal duplication cysts: cystic duplications, which account for approximately 80% of the cases and have no communication with the bowel lumen, and tubular duplications, which account for approximately 20% of the cases and communicate directly with the bowel lumen [5]. When the size of the cyst changes, it is suggestive of a communication between the intestinal duplication cysts and the bowel lumen.

Duplication cysts are typically attached to the gastrointestinal tract, have smooth muscle in their walls, and are lined with gastrointestinal epithelium [9]. Since they

usually share a muscular wall with the normal gastrointestinal tract, this shared muscular layer could be a specific sign for the diagnosis of duplication cysts. Due to limited spatial resolution, we could not detect the common muscular wall between the cyst and the jejunum prenatally. Nonetheless, on closer postnatal evaluation, the shared muscular wall was distinctly visible in the 1-day old neonate. Assuming that under certain circumstances this "shared muscular sign" is detectable, prenatal diagnosis of duplication cysts can be more accurate.

Implications for Clinical Practice

Given the pitfalls of relying on the bi-layer sign alone in the diagnosis of enteric duplication cysts, we believe that the gut signature sign remains highly practical when combined with other ultrasonic findings such as peristalsis, location, and size. Careful and serial evaluation of the wall structure, movement, and cyst location is essential to diagnose duplication cysts.

Even though enteric duplication cysts are rare congenital anomalies, prenatal diagnosis or suspicion of this rare condition allows planning delivery and postnatal critical care to achieve the most optimal outcomes.

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