21-week Routine Antenatal Ultrasound Scan – Diagnosis of Gastroschisis

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Section 2 - Answer

CASE

A 24-year-old female presented to the obstetrics outpatient clinic for a routine antenatal checkup at 21 weeks of gestation. This was her first pregnancy, and there was no history of previous miscarriage. Earlier, the ultrasound was done at around 10 weeks of gestation, which was normal. Her physical examination was unremarkable. An ultrasound scan was performed for the patient, which is shown in Figure 1 and Video 1. What is your interpretation?

INTERPRETATION

On ultrasound examination, there was a large paraumbilical abdominal wall defect (approximately 3.5 cm) seen right to the midline. The umbilical cord insertion was normal. The bowel loops were seen eviscerated through the abdominal wall defect. Eviscerated bowel loops were mildly dilated. The liver and stomach were also seen herniating through the defect. The entire herniated contents were not seen surrounded by any membrane [Figure 1 and Video 1].

Next day, magnetic resonance imaging (MRI) was performed to evaluate the abnormality. An abdominal wall defect was seen adjacent to intact umbilical cord insertion on MRI images, while bowel loops were seen freely floating in amniotic fluid. The bowel loops were not surrounded by any membrane. No associated anomalies such as bowel atresia or stenosis were seen. The liver and stomach were also seen herniating through the defect [Figure 2]. Diagnosis of gastroschisis was made based on characteristic ultrasound and MRI findings. The pregnancy was terminated at the patient's request. The imaging findings were confirmed on the fetal autopsy.

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DISCUSSION

The prevalence and incidence of gastroschisis are 1/4000 and 0.4–2.3/10,000 live births, respectively.^[1,2] The differentiation of gastroschisis from other anterior abdominal wall defects such as omphalocele, body stalk anomaly, bladder, and cloacal exstrophy, and pentalogy of Cantrell is made by prenatal imaging. Evaluation of the defect is critical in the differentiation of these anomalies. The final outcome is affected by the presence of additional structural anomalies; hence, finding anterior abdominal wall defect requires further assessment by targeted ultrasonography, MRI, echocardiography, and karyotyping.^[3]

Abdominal wall defect includes a broad spectrum of disorders. Detection and categorization of various types of abdominal wall defects on ultrasonography are essential because of their impact on management, prenatal counseling, the appropriate mode of delivery, prenatal transfer to the appropriate health-care center, and surgical closure method as precise prenatal characterization can affect the outcome.^[3] The difference between gastroschisis and omphalocele is shown in Table 1.

Initially, vascular insult was considered the cause of gastroschisis, but now, the abnormality of lateral wall folding with deficient mesenchyme is considered the cause of gastroschisis. Maternal smoking, abuse of vasoactive substances, and younger maternal age are considered risk factors for gastro schisis. There is no sex predilection. The mortality rate for gastroschisis is 10%–15%, and the risk of death depends on the degree of bowel abnormality, herniation of other viscera such as liver and stomach, and low birth weight.^[4] Liver and stomach herniation through gastroschisis is extremely rare and is associated with high mortality.^[5]

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Radiological features	Gastroschisis	Omphalocele
Size	<5 cm	Variable (2-10 cm)
Content of externalized viscera	Small bowel, sometimes large bowel and stomach	Small bowel, sometimes liver, spleen, stomach, bladde
Location of defect	Full-thickness periumbilical defect (mostly right side)	Central, umbilical
Covering sac/membrane	Absent	Present
Umbilical cord and its insertion	Cord insertion in abdominal wall at paraumbilical location	Normal cord insert over the apex of the sac
Complications	Bowel complication greater than omphalocele	Ascites
Associated findings	Oligohydramnios > polyhydramnios	Polyhydramnios
	Intra-uterine growth restriction is common	
Chromosomal abnormality	Sporadic anomaly, not common	Common
Other structural abnormalities	Gastrointestinal most common	Cardiac, gastrointestinal, central nervous system, genitourinary and musculoskeletal

Table 1. Differentiating features between gastroschisis and omphalocele



Figure 1: (a-c)Antenatal ultrasound scan showing anterior abdominal wall defect with herniation of bowel loops (B), stomach (S), and liver (L) through the defect. The herniated contents are freely floating in the amniotic fluid and are not covered by any membrane. The umbilical cord is seen on color. Doppler scan normally entering the abdominal wall (arrow in d)

Our case presented at 21 weeks of gestation with gastroschisis associated with liver and gastric herniation. The learning objective behind presenting this case is the importance of prenatal ultrasound in diagnosing the abdominal wall defects and recognizing the importance of differentiating between the two most common abdominal wall defects for appropriate patient management. Our case also had liver and gastric herniation, which is extremely rare.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Nil.



Figure 2: (a-d) Antenatal magnetic resonance imaging showing anterior abdominal wall defect with herniation of bowel loops (B), stomach (S), and liver (L) through the defect. The herniated contents are freely floating in the amniotic fluid and are not covered by any membrane

Conflicts of interest

There are no conflicts of interest.

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