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CASE REPORT

A case report of gastroschisis: Prenatal diagnosis in primary care settings matters

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Abstract

Gastroschisis may lead to serious congenital disability, as prolonged exposure to the amniotic fluid could compromise the mesenteric blood flow and irritate the foetal intestinal wall. The consequences of gastroschisis include spontaneous preterm birth, growth restriction and foetal demise. Awareness of this disease is essential, as a high mortality rate is linked to inadequate initial therapy. Early detection of the disease, mainly by primary care physicians as the initial point of contact, is crucial to improve the course of the condition through early monitoring and referral to tertiary care centres. With this case report, we hope to highlight the need of detecting gastroschisis before to the third trimester of pregnancy for better readiness, especially in primary care settings located far from tertiary centres.

Introduction

Gastroschisis is one of the most common congenital ventral wall body defects (VWBDs). However, diagnosing VWBDs is complex and requires extensive ultrasound experience.[1] An accurate evaluation of these congenital abdominal wall defects can help distinguish between isolated and multiple anomalies with chromosomal and genetic associations.[2] Early diagnosis of these anomalies at the primary care level can provide future opportunities for parental counselling and appropriate perinatal care. Herein, we present the case of a 23-year-old woman who delivered a child with gastroschisis. The baby was first diagnosed at 28 weeks of gestation during a regular antenatal visit. The prompt referral ensured foetal survival and yielded a favourable prognosis for the patient.

Case presentation

A 23-year-old Malay woman (gravida 2 para 1) visited the hospital for booking at 12 weeks of gestation. Her last menstrual period was on 27 September 2021, and the expected delivery date was 4 July 2022. The patient was a housewife and a passive smoker (her husband smoked) and came from a middle-income family. She was not taking any medication apart from haematinic during this pregnancy.

The booking visit was uneventful, except that it was noted that the pregnancy was poorly spaced since her last childbirth was in 2020. Infection screening revealed normal findings for the current pregnancy. She had a history of group B streptococcus infection in her previous pregnancy and a history of vacuum delivery owing to poor maternal effort.

The patient had an uneventful antenatal check-up until her visit at 28 weeks of gestation. During this visit, transabdominal ultrasound was performed as part of foetal growth monitoring. This examination showed a foetal abdominal circumference corresponding to 26 weeks of gestation and features of gastroschisis (herniated and thickened bowel [18.5 mm] loops floating freely in the amniotic fluid without membrane covering and side of defect at the paraumbilical area [**Figure 1**]). Nonetheless, the foetal heart was present. The umbilical cord had three vessels with normal Doppler arteries (resistance index=0.66). Other measures including head circumference, biparietal diameter and femur length corresponded to the gestational age. The placenta was at the anterior, upper segment.

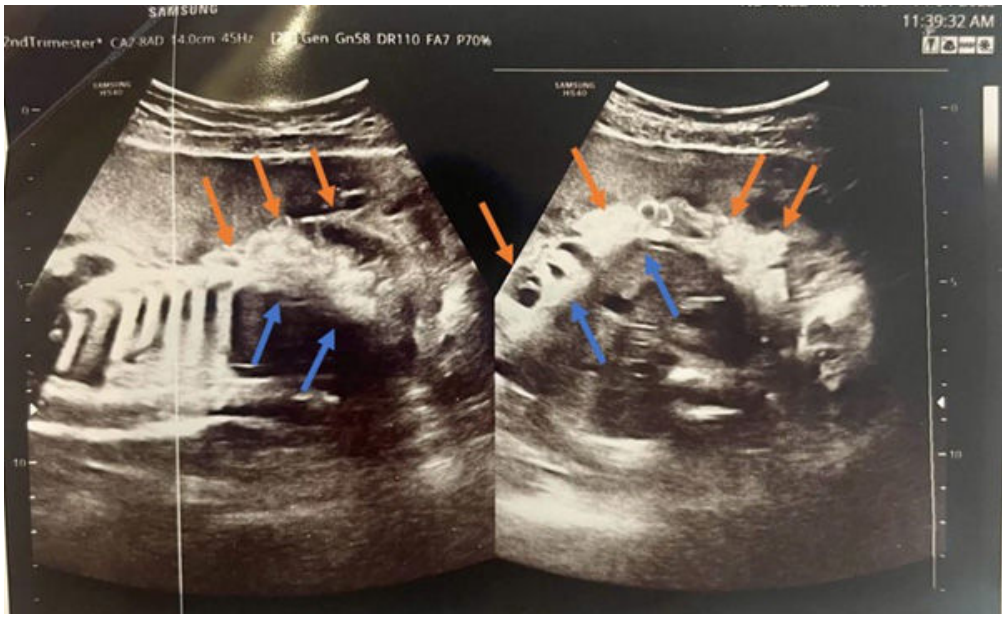

Figure 1. Ultrasound findings at 28 weeks of gestation: Orange arrows show numerous freely floating extra-abdominal cystic masses representing fluid-filled bowel loops; the bowel is thickened, dilated and eviscerated through the abdominal defect. Blue arrows show the abdominal wall.

The patient was referred to an obstetrics and gynaecology (O&G) specialist and was seen at 30 weeks of gestation. Similar ultrasound findings, together with the presence of oligohydramnios (amniotic fluid index=4.1) and intrauterine growth restriction, were observed by the O&G specialist. The patient was counselled on the risk of intrauterine demise and chromosomal abnormality, early neonatal death following delivery and prematurity in this current pregnancy. The non-invasive prenatal test showed a low risk for trisomy (T21 [down syndrome], T18 [Edwards syndrome] and T13 [Patau syndrome]).

The O&G team devised a plan to induce labour through vaginal delivery at 37 weeks of gestation. However, at 35 weeks and 6 days of gestation, the patient had preterm premature rupture of membrane and delivered a baby boy via spontaneous vertex delivery on the same day. The Apgar score of the baby was 8 at 1 min and 9 at 5 min. The birth weight of the baby was 2.037 kg.

After delivery, the baby was promptly attended to by the paediatric team. His abdomen was covered by an Alexis silo bag and a warm gauze (**Figure 2**). The baby was then admitted to the neonatal intensive care unit, and delayed primary closure and reduction of the viscera were conducted on day 7 of life by the paediatric surgical team (**Figure 3**). The baby was discharged well on day 40 of life and thrived well during the subsequent follow-up (**Figure 4**).

Figure 2. The baby was admitted to the neonatal intensive care unit, with his intestines covered by an Alexis silo bag and a gauze.

Figure 3. Post-delayed primary closure of the abdominal wall defect on day 7 of life.

Figure 4. The baby was thriving well at 3 months of age, with a healed scar over the abdomen.

Discussion

Gastroschisis is a congenital abdominal wall defect occurring in approximately 5 out of 10,000 live births.[3] It is a foetal anomaly of the front abdominal wall, characterised by the presence of a full-thickness cleft, typically to the right of the umbilical insertion. This defect results in herniation of the abdominal viscera into the amniotic fluid. The herniated viscera are not enclosed by a membrane sac, unlike in exomphalos. Differentiation between the two conditions is essential, as exomphalos (unlike gastroschisis) is associated with foetal chromosomal abnormalities.[4]

Although gastroschisis can be detected as early as 12 weeks of gestation, its prenatal imaging can be complex and challenging. This often requires attending physicians to have a high level of suspicion and knowledge of the imaging patterns of gastroschisis. The quality of ultrasound images also plays a valuable role in detecting the pathology.[1] Both factors mentioned could explain why the pathology was not detected before 28 weeks of gestation in the present case, despite an adequate number of controls and ultrasounds performed from 12 to 26 weeks of gestation. Our patient was referred to and seen by an O&G team at 30 weeks of gestation. Such early referral allowed for timely genetic counselling and non-invasive prenatal test. We did not perform karyotyping studies, as gastroschisis is rarely associated with either syndromic or non-syndromic anomalies.[5]

Early diagnosis facilitates better monitoring of pregnancy and helps determine whether gastroschisis is an isolated abnormality (simple) or associated with other gastrointestinal anomalies and complications such as intestinal atresia, perforation, necrosis or volvulus (complex).[6] Moreover, survival from gastroschisis highly depends on early prenatal diagnosis, delivery at a tertiary care facility with timely resuscitation, rapid intervention, complete parenteral feeding and availability of neonatal intensive care units. Our case was diagnosed as isolated gastroschisis and was referred promptly, yielding a good prognosis for the neonate.[7]

Conclusion

This case emphasises the significance of early detection of gastroschisis during antenatal examinations by primary care personnel, who should maintain a high index of suspicion for foetal ventral wall defects, especially in the presence of masses on the foetal anterior abdominal wall. Early detection can improve the course of the condition through early monitoring and referral to tertiary care centres for thorough screening for related abnormalities and a multidisciplinary approach. Primary care doctors should enhance their ultrasound skills, cultivate empathetic communication and acquire sufficient knowledge on genetic counselling to manage foetal ventral wall defects effectively.

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Conflicts of interest

All authors declare no conflicts of interest.

Author contributions

Abdul Hadi Said: Writing, Editing, Literature Review, Supervision
Mohd Zamir Md Nor: Writing, Editing, Literature Review
Mohd Daud Che Yusof: Writing, Editing, Literature Review, Supervision

Patient's consent for the use of images and content for publication

Informed consent was obtained from the patient before the preparation of this case report.

What is new in this case report compared to the previous literature?

- This case emphasises the importance of early detection of gastroschisis by primary care personnel.
- Most antenatal cases are seen at the primary care level; the present case underscores the need for primary care personnel to enhance their foetal ultrasound skills.
- The quality of ultrasound images plays a valuable role in detecting gastroschisis.

What is the implication to patients?

The detection of gastroschisis during prenatal monitoring may cause distress to the mother regarding the outcome of her pregnancy. A strong doctor–patient relationship and good communication are necessary to safeguard patients' physical and mental health in the current setting. In the present case, the potential distress was mitigated through early referral to a tertiary care centre for perinatal counselling and care. The mother was assured that the prognosis of gastroschisis was favourable with regular monitoring and a multidisciplinary approach. However, such favourable prognosis is only possible if the primary physician has sufficient knowledge of gastroschisis and is guided by high-quality ultrasound imaging, hence enabling early gastroschisis detection and referral.

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