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Case Report

"Prenatally diagnosed gastroschisis: A case report" ☆,☆☆

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ABSTRACT

Gastroschisis represents a congenital malformation characterized by the herniation of abdominal contents through a defect in the abdominal wall, predominantly situated to the right of the umbilical cord. The defect is characterized by the absence of a covering membrane, resulting in the free floating of extruded abdominal contents. Major complications associated with this condition include stillbirth, preterm delivery, and intrauterine growth restriction. This report presents a case involving a 24-year-old female who received a prenatal diagnosis of gastroschisis and subsequently opted for termination of pregnancy. This anomaly presents considerable challenges in neonatal management, thereby necessitating timely and effective surgical intervention.

Categories: Gynaecology, Radiology, Obstetric, Surgery.

Introduction

Gastroschisis is characterized as a fetal anterior extra-abdominal wall defect exhibiting a full-thickness cleft, primarily located to the right of the umbilical insertion (paraumbilical). This defect facilitates the herniation of abdominal viscera, specifically the evisceration of bowel loops, and occasionally involves portions of the stomach or abdomen into the amniotic fluid [1]. Antenatal diagnosis is achieved in over 90% of gastroschisis cases. Infants diagnosed with gastroschisis exhibit survival rates of approximately 90%. However, the incidence of intrauterine death (IUD) is 7.5 times higher in comparison [2]. The etiology of gastroschisis is attributed to a failure in the development and formation of the ventral body wall during the embryonic period, culminating in bowel herniation [3]. There exists a slight male predominance in the incidence of gastroschisis.

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Gastroschisis is identified as a congenital defect of the abdominal wall, with a prevalence rate of roughly 5 in 10,000 live births. The defect in the anterior abdominal wall associated with gastroschisis predominantly occurs to the right of the umbilical ring, accompanied by the extrusion of the midgut. Notably, there is a lack of a membranous covering over the herniated segment. Recognized risk factors correlating with gastroschisis encompass maternal young age, low body mass index, racial determinants, tobacco use, alcohol consumption during pregnancy, low socioeconomic status, and the utilization of recreational drugs [4].

Ultrasound serves as the primary screening tool for the diagnosis of gastroschisis. Through ultrasound imaging, complications such as necrosis, atresia, perforation, stenosis, and volvulus can be identified. Predictive markers for gastroschisis, such as intra-abdominal or extra-abdominal bowel dilation, can also be discerned via ultrasound examination [5]. Affected neonates may exhibit intrauterine growth restriction. The prevalence of associated anomalies is comparatively lower in cases of gastroschisis than in omphalocele. Vascular compromise may lead to intestinal stenosis, malrotation, or atresia.

Timely diagnosis through prenatal ultrasound is imperative for effective management and planning. Antenatally diagnosed instances of gastroschisis, coupled with surgical planning and parental counseling, generally yield favorable prognoses. Surgical intervention is typically performed on the first day of life, which correlates with improved survival rates. This case report delineates the diagnosis and management of a fetus diagnosed with gastroschisis at 18 weeks of gestation.

Case presentation

A 24-year-old gravida 1, para 0 woman presented for routine check-up at obstetrics department at 18 weeks of gestation. Doctors advised her for routine anomaly ultrasound scan which is generally done at 18 weeks of gestation. The patient had no significant medical or surgical history. She had history of previous caesarean section and tobacco use. Her current pregnancy had been uncomplicated until this point.

Previous ultrasound scans were within normal limits. The ultrasound done at 18 weeks of gestation revealed a fetal abdominal wall defect, on the right side of the umbilical cord. No membrane was seen covering the defect. The loops of the bowel were seen floating freely in the amniotic fluid, confirming the diagnosis of gastroschisis. The fetus showed normal growth parameters appropriate for gestational age. The volume of amniotic fluid was within normal limits.

Following the diagnosis, the patient was referred back to an specialist obstetrician for further evaluation and management. Serial ultrasounds were performed weekly to monitor fetal growth, amniotic fluid levels, and the condition of the herniated bowel. Following the ultrasound findings, the patient was counselled regarding the diagnosis, potential complications such as bowel damage, intestinal atresia, and the potential need for multiple surgeries after birth, and prognosis was also explained. She was also told that although surgical repair is possible, there is a significant risk of morbidity and prolonged hospital stays. She had been given 2 options by the obstetrician, either continuation of pregnancy with postnatal surgical intervention or termination of pregnancy.

After extensive discussions with her healthcare provider and family members, the patient elected to terminate the pregnancy. The termination was carried out at 20 weeks of gestation. The procedure was conducted in a hospital setting with appropriate preprocedural preparations and postprocedural care.

The patient tolerated the procedure well with no immediate complications. She received postprocedural counseling and support, including discussions on future pregnancy planning and the recurrence risk of gastroschisis, which is generally low

The fetus of a 29 year old G2P1 was diagnosed on routine prenatal ultrasound in the early second trimester with gastroschisis (Fig. 1) On prenatal ultrasound the fetus was also noted to have talipes equinovarus deformity of the left foot and persistence of the left SVC on echocardiography (not in the figure). The mother's past medical history was significant for chronic hypertension, preeclampsia in a prior pregnancy, previous cesarean section, and tobacco use throughout this pregnancy. She delivered a live 1910 gram girl at 34 weeks gestational age by scheduled cesarean section due to suspected placenta previa and accreta. Placental pathology demonstrated increta/percreta with implantation site reaction focally extending into the endocervical canal. There were maturing chorionic villi with focal chorangiosis and focal broad changes consistent with fetal vascular insufficiency. The fetus of a 29 year old G2P1 was diagnosed on routine prenatal ultrasound in the early second trimester with gastroschisis (Fig. 1) On prenatal ultrasound the fetus was also noted to have talipes equinovarus deformity of the left foot and persistence of the left SVC on echocardiography (not in the figure). The mother's past medical history was significant for chronic hypertension, pre-eclampsia in a prior pregnancy, previous cesarean section, and tobacco use throughout this pregnancy. She delivered a live 1910 gram girl at 34 weeks gestational age by scheduled cesarean section due to suspected placenta previa and accreta. Placental pathology demonstrated increta/percreta with implantation site reaction focally extending into the endocervical canal. There were maturing chorionic villi with focal chorangiosis and focal broad changes consistent with fetal vascular insufficiency. The fetus of a 29 year old G2P1 was diagnosed on routine prenatal ultrasound in the early second trimester with gastroschisis (Fig. 1) On prenatal ultrasound the fetus was also noted to have talipes equinovarus deformity of the left foot and persistence of the left SVC on echocardiography (not in the figure). The mother's past medical history was significant for chronic hypertension, pre-eclampsia in a prior pregnancy, previous cesarean section, and tobacco use throughout this pregnancy. She delivered a live 1910 gram girl at 34 weeks gestational age by scheduled cesarean section due to suspected placenta previa and accreta. Placental pathology demonstrated increta/percreta with implantation site reaction focally extending into the endocervical canal. There were maturing chorionic villi with focal chorangiosis and focal broad changes consistent with fetal vascular insufficiency (Fig. 2).

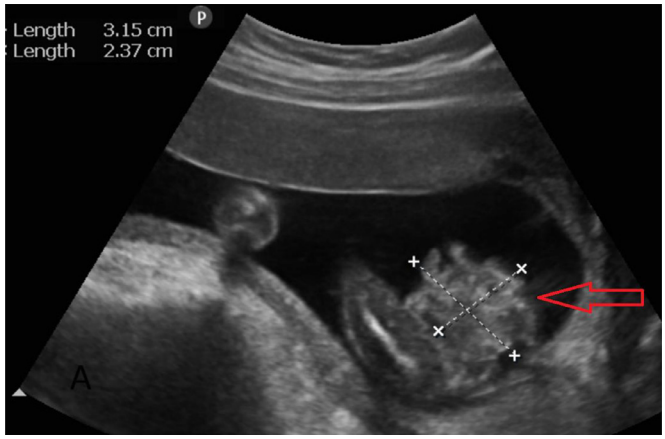

Fig. 1 – Greyscale B mode ultrasound image in sagittal oblique section of antenatal anomaly scan revealed herniated bowel loops (red arrow) measuring 3.1 x 2.3 cm through an abdominal defect.

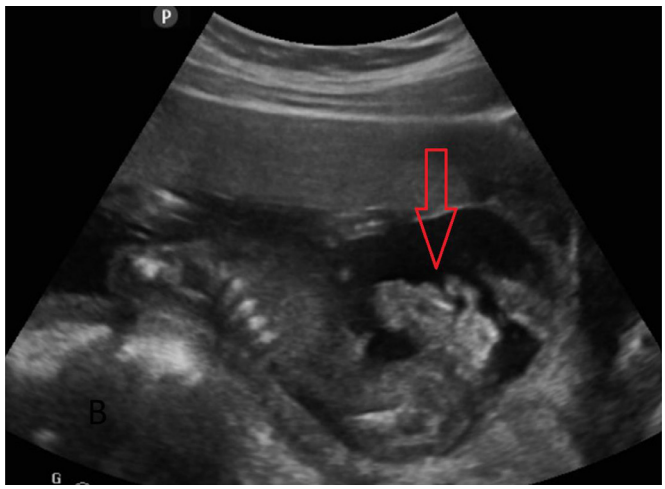

Fig. 2 – Greyscale B mode ultrasound image in sagittal oblique section of antenatal anomaly scan showed free floating bowel loops in the amniotic cavity (red arrow) through approximately 9 mm abdominal wall defect.

Discussion

Abdominal wall defects represent the most prevalent congenital anomalies observed in fetuses and neonates. An increase in the incidence of gastroschisis cases has been documented over recent decades. Abdominal wall defects encompass conditions such as omphalocele and gastroschisis [6]. In cases of omphalocele, the defect in the abdominal wall is situated at the base of the umbilical cord insertion, with the herniated visceral components enveloped by the peritoneal membrane. Conversely, in gastroschisis, the herniated portions are freely floating within the amniotic cavity, lacking any overlying peritoneal covering. Chromosomal abnormalities have been correlated with omphalocele but are not associated with gastroschisis [1].

Gastroschisis may manifest as either simple or complex, contingent upon the condition of the intestines at the time of birth. The presence of gastrointestinal complications categorizes it as complex gastroschisis, which may include congenital intestinal atresia, necrosis, stenosis, volvulus, or perforation. In instances of complex gastroschisis, multiple complications may coexist [5].

Intra-abdominal bowel dilatation, gastric enlargement, and polyhydramnios are potentially linked to poorer outcomes [7]. The condition of the bowel at birth serves as a critical prognostic indicator for neonatal outcomes [2]. The primary risk factor, notably young maternal age, is evident in our case study. The precise etiology underlying the occurrence of gastroschisis remains inadequately elucidated; however, contributory factors include tobacco use, administration of cyclooxygenase inhibitors such as aspirin and ibuprofen, utilization of decongestants like phenylpropanolamine and pseudoephedrine, as well as environmental exposures to nitrosamines such as atrazine [3].

Fetuses diagnosed with gastroschisis are generally smaller in size compared to their normal counterparts, albeit maintaining a normal karyotype. Associated intestinal complica-

tions are observed in 10%-20% of cases. Survival rates range from 90% to 95%. Instances of gastroschisis leading to fetal distress, intrauterine growth restriction, and intrauterine fetal demise have been documented [8]. The complications associated with gastroschisis primarily involve intrauterine bowel obstruction, perforation, and meconium peritonitis. Additional complications include motility dysfunction, short gut syndrome, necrotizing enterocolitis, fistula formation, and postrepair neonatal gastro-oesophageal reflux.

There exists ongoing debate regarding the appropriate timing and method of delivery; nevertheless, the typical delivery timeframe for fetuses with gastroschisis is noted to be around 37 weeks. The treatment approach is contingent upon the condition of the affected fetus. Immediate early delivery or elective cesarean section is not universally recommended for infants diagnosed with antenatal gastroschisis. Surgical interventions may involve primary closure, staged closure, and reduction to optimize surgical outcomes. Survival rates have shown continuous improvement, with current rates exceeding 90% now considered routine [9].

The advent of advanced sonographic technologies and biochemical screening via maternal serum alpha-fetoprotein during pregnancy has significantly contributed to the reduction of gastroschisis cases. Nonetheless, gastroschisis is associated with considerable perinatal mortality and morbidity, with stillbirth rates reported at 12.5% and neonatal mortality rates reaching up to 7.5% [1].

Surgical intervention for gastroschisis is typically conducted within a few hours postdelivery. Compromised bowel perfusion is regarded as an emergency requiring immediate attention. Various surgical modalities for the repair of gastroschisis have been established. The exposed bowel is repositioned into the abdominal cavity, followed by closure. Staged reduction of the herniated bowel utilizing a silastic pouch may be performed, with closure occurring at a subsequent date. A recent advancement involves the use of a spring-loaded, self-retaining silastic pouch, which obviates the necessity for general anesthesia. Definitive closure is executed at a later date.

Conclusions

Gastroschisis is a serious congenital condition that can be detected early through prenatal ultrasound. This case highlights the importance of thorough prenatal screening and counselling. The decision to terminate the pregnancy was made after careful consideration of the potential challenges and long-term outcomes associated with gastroschisis.

Early diagnosis of gastroschisis allows for timely and informed decision-making regarding the pregnancy. This case exemplifies the critical role of comprehensive prenatal care in managing complex fetal anomalies and providing patients with the necessary information to make informed choices about their pregnancies.

Patient consent

Informed and written consent was obtained from the patient.

Ethics approval and consent to participate

Written consent taken.

Availability of data and material

None.

Author contributions

SD and DN was involved in providing clinical details of the patient. PHP discussion on the pathology. GVM accumulated the results of the patient's radiological investigations. AK and RK was involved in collecting images and formatting data. All authors have read and approved the manuscript.

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