Gastroschisis—antenatal diagnosis

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MJAFI 2011;67:169–170

INTRODUCTION

Gastroschisis also called paraomphalocoele, laparoschisis, abdominoschisis or abdominal hernia is a type of inherited congenital abdominal wall defect in which the intestines and sometimes other organs develop outside the fetal abdomen through an opening in the abdominal wall. It is believed that this defect is the result of obstruction of the omphalomesenteric vessels during development. The rapid development of antenatal ultrasonography has prompted an early diagnosis of this entity and hence can guide the gynaecologist as well as paediatric surgeon for appropriate management.

CASE REPORT

Twenty-six-years-old primigravida unbooked lady presented to our institute for antenatal ultrasonography at 26 weeks of gestation. There were no associated comorbidities. On examination the height of uterus corresponded to 26 weeks. Foetal movements were felt and foetal heart sound was heard. Rest of the general and systemic examination was insignificant. The alfa fetoprotein (AFP) was markedly elevated. The biochemical parameters were within normal range. The ultrasonography scan showed reduced abdominal circumference for period of gestation (POG) with free floating bowel loops in the amniotic fluid (Figure 1). The biparietal diameter, head circumference and femoral length were corresponding to the period of gestation (POG). The insertion of cord was normal while the herniation of bowel loop was seen through a right parasagittal defect located in the anterior abdominal wall. Rest of the abdominal viscera was normal in appearance. The amniotic fluid index (AFI) was within normal limits for POG. A diagnosis of gastroschisis was offered. Post abortus specimen confirmed the above findings (Figure 2).

DISCUSSION

Gastroschisis, derived from the Greek word meaning “belly cleft”, is a relatively small defect (2–4 cm) in the abdominal wall involving all the layers of abdominal wall laterally; usually right side to the intact umbilical cord. Gastroschisis is generally sporadic with incidence of 1 in 10,000 live births but its occurrence with other anomalies is rare.¹ Various theories regarding this defect have been proposed. Failure of mesoderm

Figure 1 Axial ultrasound image of the foetus showing herniated bowel loops (white arrow) through the anterior abdominal wall and to the right of umbilical cord.

Figure 2 Post abortus specimen confirming the herniated bowel loop through the anterior abdominal wall.

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Received: 07.12.2009; Accepted: 06.06.2010

to form in the body wall (Duhamel, 1963); Rupture of the amnion around the umbilical ring with subsequent herniation of bowel (Shaw, 1975); abnormal involution of the right umbilical vein leading to weakening of the body wall and gut herniation (deVries, 1980); disruption of the right vitelline (yolk sac) artery with subsequent body wall damage and gut herniation (Hoyme et al, 1981) and abnormal folding of the body wall results in a ventral body wall defect through which the gut herniates (Marcia et al, 2007) are five main contenders to explain this entity.

It is proposed embryologically during the fourth week of development the lateral body folds move ventrally and fuse in the midline to form the anterior body wall. Incomplete fusion results in a defect that allows abdominal viscera to protrude through the abdominal wall. The bowel typically herniates through the rectus muscle, lying to the right of the umbilicus.

The abdominal contents herniate through this small defect in utero and are free floating within the amnion without any covering membrane unlike omphalocele. Most commonly it is the small bowel which eviscerates often accompanied by the large bowel and sometimes stomach but liver and other solid organs are rarely involved. One of the reported hallmarks in diagnosis of gastroschisis is the normal insertion of umbilical vessels into the foetal abdomen. However, identification of the position of these vessels can be difficult, particularly when the cord is absent or rudimentary as in body-stalk anomaly, which is the most severe form of gastroschisis.² Few authors have advocated for use of colour Doppler as an aid in the diagnosis of the umbilical vessels in such tricky situation. Almost all the patients with gastroschisis demonstrate either bowel malrotation or non-rotation. Intestinal ischaemia or stenosis may be present in 1/3rd of cases. This ischaemia may lead to bowel gangrene, perforation or meconium peritonitis.³ The umbilical cord inserts in anterior abdominal wall, which distinguishes it from omphalocele where cord inserts into the sac.⁴ Foetus with gastroschisis is more often associated with prematurity and intra-uterine growth retardation.⁵

Gastroschisis and omphalocele also differ markedly in prevalence and nature of additional malformations. Omphalocele is associated with a high rate of additional malformations and frequently is associated with syndromes and chromosomal anomalies.⁶ Neural tube defects, diaphragmatic hernia and foetal heart defects are the most commonly associated malformations.² Although gastroschisis rarely is associated with chromosomal anomalies or other system malformations more frequently intestinal complications as jejunoileal disruption and intestinal atresia may be present.⁵

Prenatal detection of abdominal wall defects by obstetric ultrasonography has greatly modified their postnatal management. Sonographic examination is of aid in most cases in classifying the abnormality, detecting associated malformations, establishing the prognosis and then optimising the management through a multidisciplinary approach.

CONFLICTS OF INTEREST

None.

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