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BRIEF COMMUNICATION

Sonographic Diagnosis of Omphalocele in the Second Trimester of Pregnancy

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Abstract Omphaloceles or exomphalos are congenital midline abdominal wall defects at the base of the umbilical cord insertion with herniation of gut and/or liver or occasionally other content, out of the fetal abdomen. Prenatal detection has significantly increased with a wide use of ultrasound and alpha-fetoprotein screening. Omphaloceles detected early in pregnancy and containing only bowel, have increased risk of associated anomalies and chromosomal disorders.

Keywords Developmental anomaly · Examphalos · Fetal anomaly ultrasound scan · Gastroschisis · Omphalocele · Obstetric ultrasound

Introduction

The word omphalocele is derived from the Greek word Ομφαλός meaning navel and cele—meaning pouch [1]. The estimated occurrence can be up to 1:4000 live births [2]. It appears to be more common in pregnant women of extreme reproductive age (<20 and more than 40 years). The chromosomal abnormality is more likely if omphalocele contains only small intestine [3]. Approximately 15 % of live-born infants with omphalocele have chromosomal abnormalities [1, 3]. Omphalocele is associated with severe malformations such as cardiac anomalies (50 %) and neural tube defect (40 %) and has a high mortality rate (25 %) [4].

Report of Case

A 23-year-old gravida 2 para 1 with history of lower segment cesarian section (LSCS) in the past and the death of a female child soon after birth (cause not known), had her first visit to the antenatal clinic. Routine obstetric sonography revealed a single live fetus in breech position with average gestational age of 21 weeks. An echogenic mass in the midline of the anterior abdominal wall (Fig. 1) with membrane covering the mass was detected (Fig. 2). Color Doppler revealed insertion of the cord in the apex of the mass (Fig. 3). After counseling, the patient opted for medical termination of pregnancy and a male child of 500 g weight with omphalocele was delivered (Fig. 4).

Hemoglobin was 9.8 gm/dL, leucocytes 7.6 × 10^3/mm^3, differential white cell count: polymorphs 74 % lymphocytes 22 %, monocytes 2 %, basophils 2 %, red blood cell count 4.17 × 10^6/mm, packed cell volume (PCV) 32 %, blood group was A+ve platelets 257,000/mm^3, bleeding time 2 min 15 s, clotting time 4 min 15 s, random blood sugar 70 mg/dL, blood urea 15 mg/dL, serum creatinine 0.70 mg/dL, serum glutamic-oxaloacetic transaminase (SGOT) 34 U/L, serum glutamic pyruvic transaminase (SGPT) 24 U/L, bilirubin (total) 0.28 mg/dL, conjugated 0.15 mg/dL, unconjugated 0.13 mg/dL, total serum proteins 6.3 g/dL, albumin 3.11 g/dL, globulin 3.2 g/dL, albumin to globulin (A/G ratio) 0.97.

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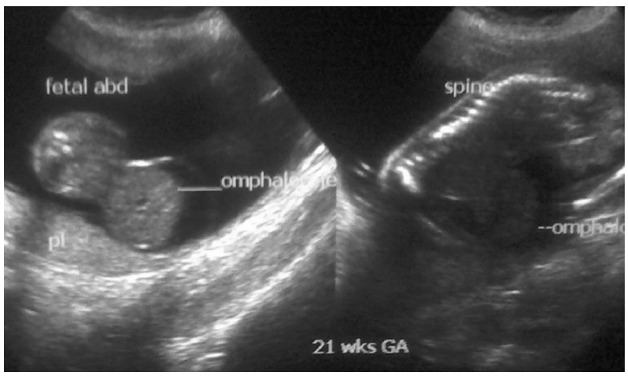

Fig. 1 Circular central mass with a covering membrane and ascites protruding from the anterior abdominal wall forming an acute angle

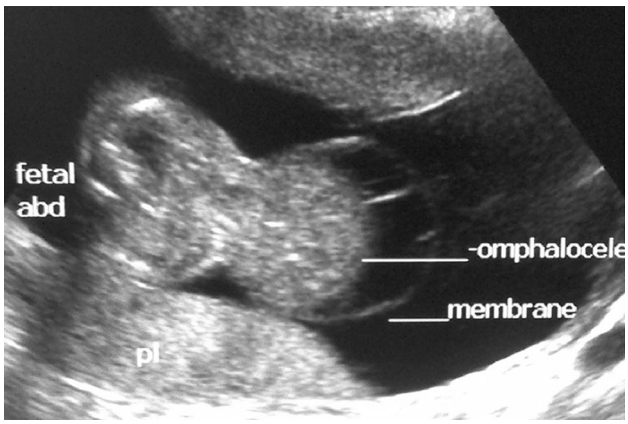

Fig. 2 Omphalocele with covering membrane and herniated liver

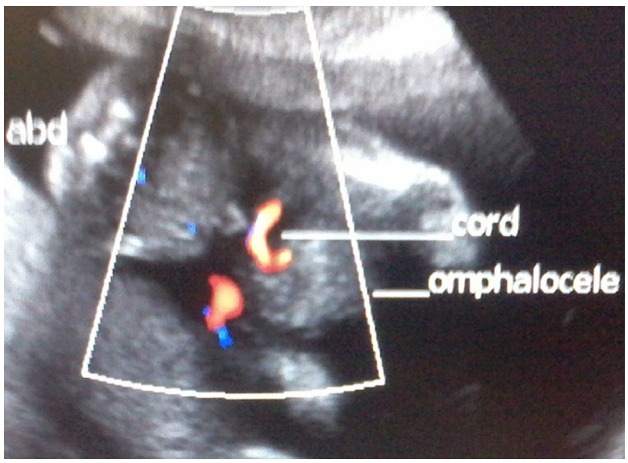

Fig. 3 Central insertion of umbilical cord into the herniated mass of omphalocele

Sickling for RBC negative
HIV (spot) + HBs Ag (spot) + VDRL/RPR:Nonreactive
Urinalysis was normal.

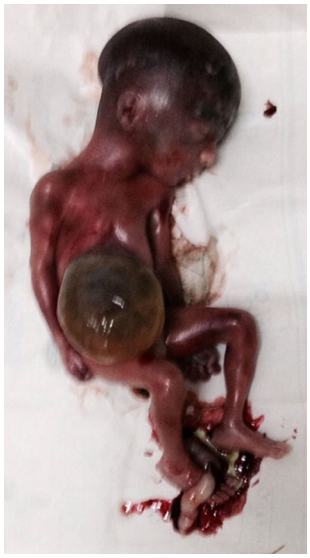

Fig. 4 Male baby with omphalocele after MTP

Discussion

Omphalocele is a midline abdominal defect with herniation of the abdominal content such as liver and/or bowel into the base of the umbilical cord covered by a peritoneal membrane as well as amnion. The umbilical cord typically inserts at or near the apex. There can be other organs protruding into the omphalocele such as large intestine, bladder, stomach, and spleen.

Omphalocele has to be differentiated from physiologic bowel herniation which starts around 8th week of gestation and disappears before 12 weeks of gestation. It never contains herniated liver. One should note that physiologic midgut herniation should not exceed 7 mm in diameter and should not be apparent in fetuses with a crown rump length >44 mm.

There are three main theories for the etiology of omphalocele formation [5].

1. Persistence of the primitive body stalk
2. Failure of the bowel to return to the abdominal cavity during the 10th week. This process is called the reduction of the midgut hernia which is completed at 12 weeks gestation [2].
3. Another possibility is that omphalocele results from the failure of the embryonic lateral folds to fuse in the midline at approximately 3–4 weeks postconception, which allows the abdominal contents to herniate into the sac [2].

There are few case reports of transient fetal omphalocele or delayed reduction of the physiologic midgut hernia. Therefore, the importance of this finding: Does it represent

Table 1 Differential diagnosis of gastroschisis and omphalocele

Feature	Gastroschisis	Omphalocele
Time at which can be visualized	After 12 weeks gestation when physiologic gut herniation resolves	Persists beyond 12 weeks gestation
US features	Covering membrane not present, ragged appearance in sonography	Covered by membrane comprising of amnion and peritoneum with Wharton's jelly between the two layers; well-defined rounded, smooth appearance
US characteristics of sac	Edge of hernia sac forms obtuse angle with abdominal wall	Edge of hernia sac forms an acute angle with abdominal wall
Size of defect in US	Diameter not more than 7 mm; physiologic gut herniation not seen in CRL of 44 mm	Size varies from small to large, 2.5–5 cm diameter
Insertion of umbilical cord	Umbilical cord entering the abdomen on left side of gastroschisis	Cord entering in central portion/peak of the sac
Contents of hernia sac	Containing only bowel, not liver	Containing bowel and/or liver or other organs
Associated anomalies	Associated anomalies uncommon	Associated anomalies (15–20 %) are present. Cardiac defects 50 %, neural tube defects 40 %
Prognosis	Good	Good (80–90 %), if small in size and does not contain bowel; mortality rate 80 % if associated with chromosomal disorder
False diagnosis	Less common	More common
Amniocentesis	Usually not indicated	Definite indication

CRL crown rump length, *US* ultrasonography

a malformation or a delayed physiologic process?, still raises a concern [6].

The most common associated findings are cardiac defects (50 %) such as atrial and ventricular septal defects and tetrology of Fallot and gastrointestinal defects (40 %) [7, 8].

Ultrasonography is a safe, noninvasive, real-time technique that is widely available. It remains the imaging modality of choice for the prenatal assessment of the fetus [9].

The frequency of associated abnormalities is high 27–91 % [4] and are thought to be even commoner with smaller omphalocele containing bowel only [10]. When an omphalocele is suspected on antenatal sonograms, diagnostic amniocentesis, a targeted ultrasonographic examination to search for associated anomalies, fetal echocardiography, and karyotyping should also be performed [11].

The diagnosis is usually made after the 12th week of gestation once the normal physiological hernia has resolved. Eviscerated liver permits diagnosis of omphalocele at any age [10, 11]. Chromosomal anomalies can occur in 20–50 % of cases; the risk of an associated anomaly gets higher when the omphalocele is detected earlier in gestation. Trisomy 18 is the most common associated chromosomal anomaly while the others could be trisomy 13, trisomy 21, Turner syndrome, Kleinfelter syndrome, and Pallister–Killian syndrome [7].

Sonographic findings in omphalocele [4] include a midline ventral abdominal wall defect (2.5–5 cm) covered by a membrane composed of two layers (the inner peritoneal membrane and outer amnion) containing liver and/or bowel with insertion of umbilical cord into the apex of the defect with widening of the cord where it joins the abdominal skin. Ascites within the sac is commonly present. Differentiating features between gastroschisis and omphalocele have been depicted in Table 1.

Color Doppler is a useful tool to demonstrate the umbilical cord insertion, which is usually in the membranes in the midline of omphalocele. If membrane ruptures, it may be difficult to differentiate it from gastroschisis.

Prenatal MRI enhances the fetal anatomic evaluation; it can be a valuable adjunct to ultrasonography before surgical intervention for selected life-threatening fetal defects. MRI helps in corroborating and refining the ultrasonographic diagnosis of complex fetal defects [12]. MRI is not operator dependent and maternal obesity or oligohdramnios is not a limiting factor in making a diagnosis [12]. Fetal movements can make MRI difficult. The large field of view, high resolution of the soft tissues, and its accuracy has been reported to be superior to that of sonography [12].

MRI is considered safe for the developing fetus. At present, no clinical or experimental evidence suggests that MRI causes teratogenic or other adverse affects during pregnancy, although a few studies in laboratory animals have shown that prolonged, high-level exposure to electromagnetic radiation might result in teratogenicity [14]. A recommendation from the National Institute of Health Consensus Development Conference states, "MRI should be used during the first trimester pregnancy only when

there are clear medical indications and when it offers clear advantage over other modalities" [12].

The prognosis and mortality rate in omphalocele is determined more by the presence of associated anomalies, such as cardiovascular and chromosomal defects, than by the omphalocele itself. Prenatal MRI can be used to screen for anomalies such as complex cardiac defects and nervous system anomalies [12].

In the assessment of giant omphalocele, MRI-based observed/expected total lung volume of <50 % was predictive of increased postnatal morbidity [13].

An omphalocele has a higher morbidity and mortality than in gastroschisis. Fetuses with a small omphalocele and no associated anomalies have a very good prognosis with a survival around 80–90 % [6]. Mortality rates can approach 80 % when associated anomalies are present and increase to nearly 100 % when chromosomal or cardiovascular anomalies exist. However, if omphalocele is found in isolation, then the mortality rate decreases to 10 % [14].

Compliance with Ethical Standards

Conflict of interest None.

Informed Consent Consent from the patient has been obtained. Permission from institutional review committee has been obtained.

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