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Antenatally Diagnosed Omphalocele in Mid Trimester: A Case Report

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Abstract Omphalocele is a defect in anterior abdominal wall with extrusion of abdominal organs that is covered by a membrane consisting of an outer layer of amnion and an inner layer of peritoneum with cord inserting through these covering. Widespread use of antenatal ultrasound has made the detection of omphalocele with increasing incidence in second trimester of pregnancy. Prevalence of omphalocele ranges between 1.5 and 3 per 10,000 births. A case of omphalocele diagnosed antenatally in mid trimester is reported with Ultrasound imaging features and brief review of literature.
Keywords: Omphalocele, Ultrasound, Antenatal.

INRODUCTION

Anterior abdominal wall defects comprise a group of congenital malformations that includes Gastroschisis, omphalocele, which are relatively common, and ectopia cordis, bladder exstrophy, and cloacal exstrophy, which are extremely rare. The prevalence of gastroschisis and omphalocele are increasing with increased use of ultrasound.

The incidence of omphalocele reported in the literature ranges from0.8 to 3.9 cases per 10,000 births [1]. Omphalocele is known to be a part of various syndromes such as Beckwith-Wiedemann syndrome, pentalogy of Cantrell, omphalocele exstrophy imperforate anusand spinal defects syndrome. Structural anomalies are associated in with omphalocele in 27% to 91% [2] of fetuses and chromosomal anomalies ietrisomy 13 and trisomy 18 in 20%–50%.

CASE REPORT

A 25 year old woman was refereed for routine mid trimester anomaly scan. Ultrasound was done with state of the art equipment and diagnosis of omphalocele was based upon herniation of the intestinal loops and/or abdominal organs into the umbilical cord with abdominal wall defect. Foetal parameters for assessment of gestational age were taken. BPD, HC, HL and FL correlated well with her gestational age by LMP. However the abdominal circumference was less due to herniation of abdominal contents and did not correlated with other parameters.

In our case, there was defect in anterior abdominal wall with herniation of bowel and liver in to umbilical cord. Liver was identified by presence of distended Gall bladder in to herniated sac and bowel by small echogenic structures. Umbilical vessels were anechoic with two arteries and one vein showing colour flow. There were no associated anomalies. Liquor was adequate for gestational age and placenta was normal in anterior segment. Following birth the ultrasound diagnosis of omphalocele was confirmed.

DISCCUSSION

Up to 10weeks, intestines are present outside abdomen and by 10 weeks they should be within the fetal abdomen. Their persistence beyond 10 weeks should suspect anterior abdominal wall defect. The exact mechanism leading to omphalocele is controversial, which may be either due to failure of reduction of physiologic embryonic umbilical hernia or failure of the embryonic lateral folds to fuse in the midline [3].

Omphalocele (also known as exomphalos) is an uncommon type of anterior abdominal wall defect through which abdominal organs herniate in to sac outside the abdomen [4]. Most common organs to herniate are liver and intestines, as in the present case. Severity of the omphalocele depends on the extent of the hernia and other associated anomalies. Incidence rate is 2.5/10,000 births (small omphalocele 1: 5000 and large one1; 10000 with associated cardiac (50%) and neural tube defects (40%). High mortality (25%) is seen with 15 % of live-born having chromosomal abnormalities.

Gastroschisis represents congenital defect in the anterior abdominal wall through which the abdominal contents freely protrude with reported incidence of 1 in 6000 births. The abdominal wall defect is located at the junction of the umbilicus and normal skin, and is almost always to the right of the umbilicus with no overlying sac. Most common

Syndromic association of omphalocele is with pentalogy of Cantrellseen in- 1/100,000 live births with suspected familial association of X chromosome at Xq25-26.1 region. Pentalogy of Cantrell comprises Omphalocele, cardiac ectopia, absence of distal portion of the sternum, absence of anterior diaphragm and absence of the parietal pericardium.

Omphalocele has to be differentiated from Gastroschisis as the prenatal and post natal outcomes are different. Sonological differentiation has to be made antenatally so that appropriate management can be undertaken. Small and medium sized omphalocelescan be repaired after delivery which show good prognosis and larger ones require delivery by cesarean, followed by repair.

Table 1: Important differentiating features of omphalocele from Gastroschisis

	Omphalocele	**Gastroschisis**
Incidence	1:5000 to 1:10,000	1:20,000-30,000
Covering Sac	Present (may be ruptured)	Absent
Wall Defect	Small to large	Small (vascular compromise)
Cord Attachment	Umbilical the herniated sac	Abdominal wall
Herniated Bowel	Protected	Edematous and matted
Other organs	Liver is often in the sac	Remain in abdomen.
IUGR	Less common	Common
NEC	If sac is ruptured	18%
Assoc. Anomalies		
- Overall	55% to 80%	10% to 15%
- GI	37 % (Midgut volvulus Meckel's Diverticulum, atresia, duplications)	18 % (stenosis and atresias)
-Cardiac	20 %	2%
- Trisomy	30 %	Not associated

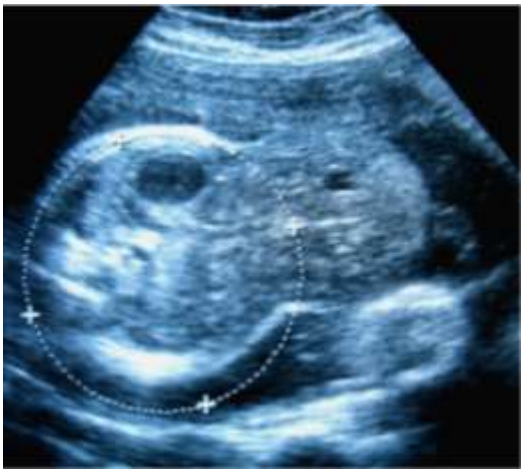

Fig. 1

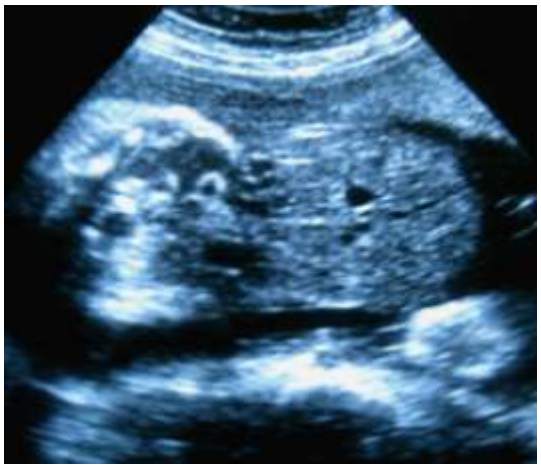

Fig. 2

Fig 1and Fig 2: Axial ultrasound image of fetal abdomen at the level of stomach showing protrusion of liver and small bowel outside the abdominal wall in to Umbilicus

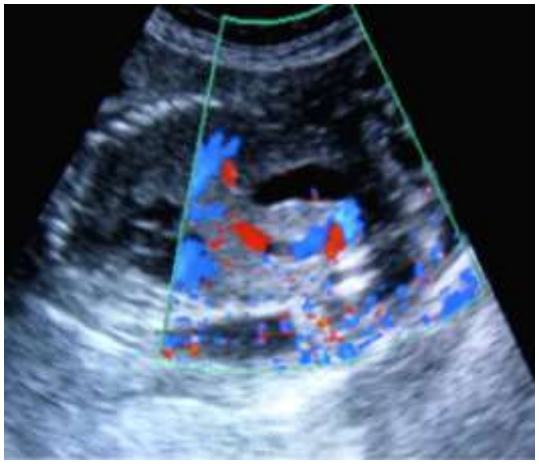

Fig. 3: Longitudinal image of fetal abdomen showing herniation of fetal liver and small bowel

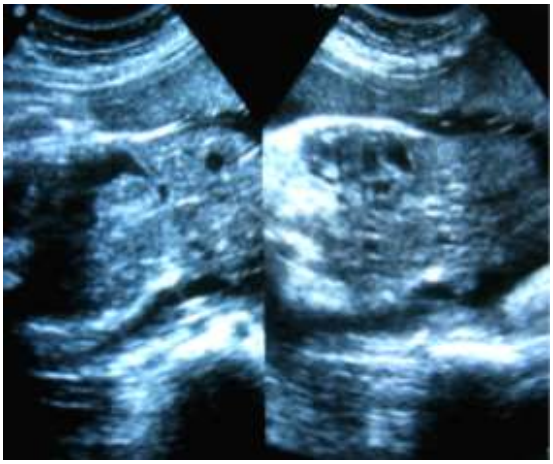

Fig 4: Liver herniation with portal vein and fetal Gall bladder on color Doppler

CONCLUSION

Antenatal ultrasound can diagnose majority of anterior abdominal wall defects with accurate distinction of omphalocele and gastroschisis which would permit an opportunity to counsel the family members and prepare for optimal postnatal care. Omphalocele with chromosomal aberrations are usually associated with other structural anomalies with high mortality compared to simple omphalocele not associated with chromosomal aberrations.

A thorough, well-documented fetal sonographic examination, combined with Doppler examination in equivocal cases, can increase the accuracy of in utero diagnoses of omphalocele and gastroschisis. The diagnosis of such anomalies is important in assuring appropriate obstetric and perinatal management. If the pregnancy is maintained with a known diagnosis of omphalocele, the obstetrician can minimize injury to the bowel at the time of delivery. Fluid loss and incidence of peritonitis can be reduced if surgical closure is performed within 6-8 hr of birth.

Favorable prognosis is seen in Fetuses with a solitary finding of intestinal herniation into the umbilical cord and a normal karyotype .Parents should be counseled about the possibilities of omphalocele which can be surgically treated after delivery or omphalocele can be transient which may disappear during gestation. However the fetus may have a rare syndrome that cannot be definitively diagnosed inutero .Follow-up ultrasound examinations may be needed throughout the gestation and even after delivery.

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