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# Diagnosis of Omphalocele by Ultrasound Scan

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**Abstract:** Omphalocele is an anterior abdominal wall defect at the base of the umbilical cord, with herniation of the abdominal contents. We report a case of a 39-year old gravida 7 para 6 +0 of Sudan origin who presented complain of amenorrhea for 6mnoths and LMP = 27/11/2011, decreased fetal movement and subrapubic pain.
**Keywords:** polyhydramnios, Omphalocele, US.

## INTRODUCTION

Omphalocele is an anterior abdominal wall defect at the base of the umbilical cord, with herniation of the abdominal contents. The herniated organs are covered by the parietal peritoneum. After 10 weeks' gestation, the amnion and Wharton jelly also cover the herniated mass.[1, 2, 3, 4]. The etiology of omphalocele is not known. Various theories have been postulated; these include failure of the bowel to return into the abdomen by 10-12 weeks, failure of lateral mesodermal body folds to migrate centrally, and persistence of the body stalk beyond 12 weeks' gestation [4].

Omphaloceles are associated with other anomalies in more than 70% of the cases; the severity of the associated anomalies determines the prognosis.[5] The mortality rate is 80% when associated anomalies are present, and it increases to 100% when chromosomal and cardiovascular abnormalities are present.[6, 7, 8, 9] Most associated anomalies are chromosomal.

## CASE PRESENTATION

We report a case of a 39-year old gravida 7 para 6 +0 of Sudan origin who was seen in the ultrasound department of SHIFA AL-ELALEEL with amenorrhea for 6mnoths and LMP = 27/11/2011. Presented complain of decreased fetal movement and subrapubic pain. A careful ultrasound assessment led to the diagnosis of a fetal ventral solid mass is seen at the cord insertion site containing bowel and liver (Fig 1-A). The herniated contents are covered by a membrane, increased amniotic fluid volume (polyhydramnios Fig 1-B), the largest vertical pocket measured < 9cm. Markedly short fetal lower limb was seen (Fig 2).GA: BPD = 23wks+ 6days, FL = 19 weeks + 6days.

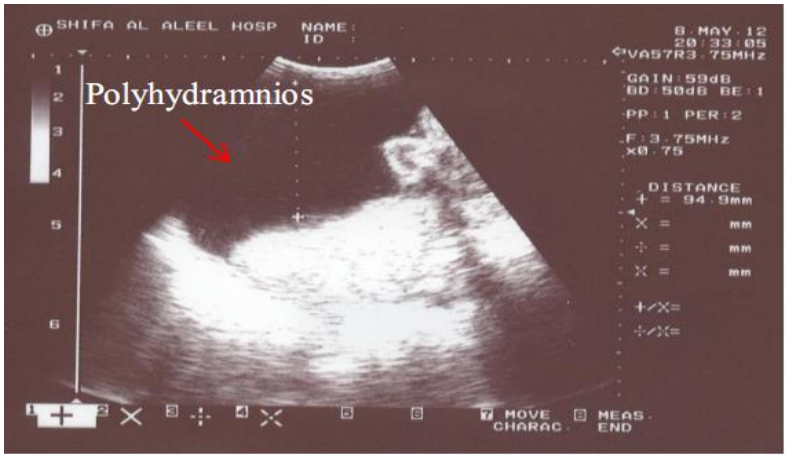


**Fig (1- A): TAS showing Poly hydramnios (arrow)**


**Fig (1 -B): TAS showing Cross section of fetal abdomen (arrow)**

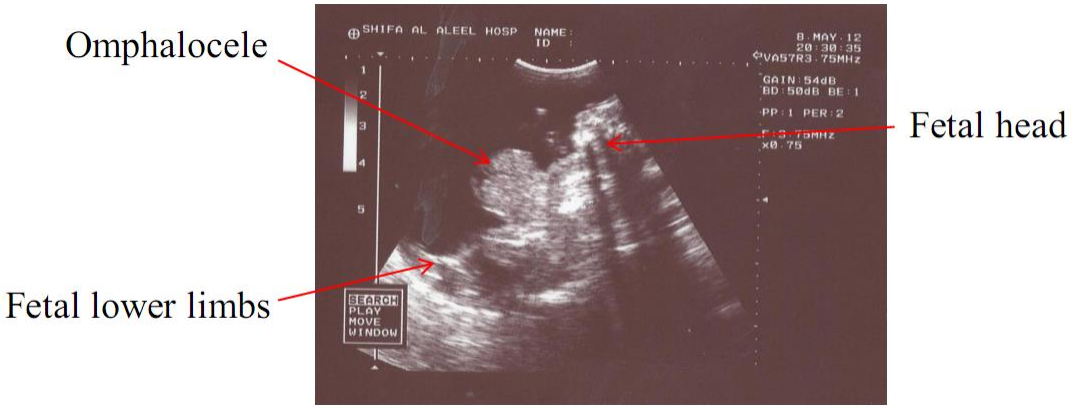


**Fig 2: TAS showing fetal head, Omphalocele and Fetal lower limbs (arrows)**

**DISCUSSION**

The diagnosis is usually made after the 12th week of gestation once the normal physiological hernia has resolved (bowel containing omphaloceles). Liver herniation is not a feature of normal physiological bowel herniation, and therefore eviscerated liver permits diagnosis of omphalocele at any age. Liver containing omphaloceles are more homogeneous and less echogenic than normal.

First trimester diagnosis of liver containing omphaloceles have been made [10, 11]: Omphalocele at 13 weeks as an echogenic tumor at the umbilicus; the fetus was subsequently found to have trisomy [12]. Omphalocele containing liver at 10 weeks, but retrospective examinations of the sonograms obtained at 6–9 weeks did not reveal any abnormality; the diagnosis was confirmed after delivery [12]. Pagliano *et al.;* [6] reported the diagnosis of omphalocele containing liver and bowel in a 10-week fetus. Heydanus *et al.*; [13] reported the diagnosis of omphalocele in three fetuses at 12–14 weeks; in one there was an associated ectopiacordis and hydrops and the pregnancy was terminated, in the second there was an associated two-vessel cord and intrauterine death occurred and, in the third with isolated exomphalos, there was an infant death. Van Zalen-Sprock *et al.*; [14] reported the findings of 14 cases with omphalocele diagnosed at 11–14 weeks of gestation. In eight cases, there was increased nuchal translucency thickness (3.5–10 mm) and seven of these had chromosomal abnormalities, mainly trisomy 18. The contents of the omphalocele were bowel only in the chromosomally abnormal group and liver as well as bowel in those with a normal karyotype. In the chromosomally normal group, there were four with other defects, such as tetra logy of Fallot and Meckel–Gruber syndrome; only three infants were live born. An ultrasound screening study of 622 high-risk pregnancies at 10–13 weeks correctly diagnosed the two cases of omphalocele [15].

In two other screening studies of low-risk patients, involving 1632 pregnancies at 12–14 weeks [16] and 1473 pregnancies at 10–14 weeks [17] respectively, there were four cases of Omphalocele (two in each) and they were all diagnosed in the first-trimester scan. In a screening study for chromosomal abnormalities by assessment of fetal nuchal translucency thickness at 10–14 weeks of gestation, there were 15 726 pregnancies with a minimum

gestation of 11 weeks and 4 days and, in this group, there were 18 cases of omphalocele. In seven cases, the karyotype was normal, in nine there was trisomy 18, in one trisomy 13 and in one triploidy. The prevalence of omphalocele in fetuses with trisomy 18 was 23%, in those with trisomy 13 it was 9%, in those with triploidy it was 13% and in those with no evidence of these chromosomal defects it was 0.045%. This study demonstrated that both the prevalence of omphalocele and the associated risk for chromosomal defects increase with maternal age and decrease with gestational age [18].

Gastroschisis is a defect in the abdominal wall, usually to the right of the umbilicus that allows bowel to protrude into the amniotic cavity. On ultrasound, multiple, round, freely floating structures along the abdominal wall suggests the diagnosis. The loops of bowel may be distinguished from loops of umbilical cord by the presence of blood flow in the latter on Doppler. The defect is thought to arise from disruption of blood flow (the right omphalomesenteric artery) to the affected area [19].The prognosis is generally favorable with survival rates between 77% and 100% [20, 21]. Gastroschisis occurs with an incidence of 1:10,000 births, and for unclear reasons is seen more frequently in younger mothers. The defect is also seen more frequently in mothers who use vasoactive substances such as nicotine and cocaine [22, 23].

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