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**CASE REPORT****Prenatal Diagnosis of Omphalocele on Antenatal Sonography: A Case Report***Rajasbala P. Dhande<sup>1</sup>, Varun Singh<sup>1\*</sup>, Amruta Verma<sup>1</sup>, Shreya Tapadia<sup>1</sup>**<sup>1</sup>Department of Radio-diagnosis, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences, Sawangi, Meghe -442004 (Maharashtra) India*

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**Abstract:**

A wide spectrum of anterior abdominal wall defects exist having varying outcomes, the most common being omphalocele and gastroschisis followed by bladder, cloacal exstrophy, Pentalogy of Cantrell and body stalk deformity. The final outcome of these defects is significantly affected by the presence of additional structural and / or chromosomal abnormalities, Hence, it is very important to accurately diagnose and classify these defects as they have different prognosis and management. The key features to differentiate these lesions are based on the-1) site of abdominal defect—midline, para midline, infraumbilical, 2) umbilical cord insertion-normal, abnormal 3) size of the defect and organs eviscerated, 4) presence or absence of covering membrane and 5) any associated congenital/ chromosomal anomaly. Here we are reporting a case of omphalocele, which was diagnosed on routine antenatal ultrasonography and will be discussing the diagnostic algorithm to differentiate other entities of anterior wall defects

**Keywords:** Omphalocele, Ultrasonography, Anterior abdominal defect

**Introduction:**

Anterior abdominal wall develops in multiple steps by the 12 menstrual week, any abnormality in these steps leads to a spectrum of anterior abdominal wall defects like omphalocele, gastroschisis, pentalogy of Cantrell, limb body wall complex, cloacal and bladder exstrophy being infraumbilical in location [1]. The two most important and frequently encountered congenital

anterior wall defects are the omphalocele and gastroschisis. They are often associated with other congenital anomalies like trisomy 12, 18, 21, Pentalogy of Cantrell (Ectopia cordis, midline supraumbilical abdominal defect, sternal cleft and intracardiac defect), charge syndrome and cloacal exstrophy which in turn determine the final outcome. Omphalocele is known to have a high mortality rate, it is stated that only about 60% of children with such type of malformations survive until the end of first year of age [2-3]. Accurate and early detection of these defects, by a skilled radiologist is of utmost importance as it affects the patient's management and prognosis.

**Case Report:**

A 36 year old pregnant woman came to Outpatient Department (OPD) of Radiology Department for her chromosomal scan at a gestational age of 17 weeks 3 days from Last Menstrual Period (LMP). She had an obstetric score of G4P3L1, her previous two children were diagnosed with omphalocele, out of which fetus of second pregnancy (G2) was aborted as it was diagnosed with omphalocele at 18 weeks of gestation. The fetus of third pregnancy (G3) died after surgical correction of omphalocele.

**Ultrasonography Findings:**

The present scan revealed a single intra uterine live fetus, with calculated gestational age by Ultrasonography (USG) being 17 weeks 3 days, a

detailed examination of the fetus revealed- a midline abdominal wall defect of 2 cm, through which there was herniation of intra-abdominal contents mainly consisting of part of liver and few bowel loops, which were seen as echogenic round mass on USG. The herniated mass was well defined and was covered with a thin echogenic membrane. The umbilical cord showed abnormal insertion into the herniating mass forming the apex of the mass. (Figs. 1 and 2). Amniotic Fluid Index (AFI) was 8.5 which was suggestive of oligohydramnios. An additional finding was noted during the evaluation of fetal brain, there was presence of a small cystic space of approximate size  $8 \times 6$  mm in the posterior fossa, which was connecting to the fourth ventricle with the hypoplastic vermis, highly suggestive of Dandy Walker spectrum (Fig. 2).



**Fig. 1: Axial Grey Scale USG image showed an Anterior Wall Defect through which Liver has Herniated into a Sac (1) Umbilical Cord was Seen Forming the Apex of Omphalocele, (2) Amniotic Memberane was Covering the Herniated Organs, (3) Echogenic Liver Seen as A Content**



**Fig. 2: (1) Wharton's Jelly seen as Content between the Amniotic Membrane and Herniated Bowel and Liver. (2) Midline Anterior Wall Defect of 2 cm**



**Fig. 3: (1) Small Cystic Space of Approximate Size  $8 \times 6$  mm in the Posterior Fossa, Which was Connecting to the Fourth Ventricle with the Hypoplastic Vermis, Highly Suggestive of Dandy Walker Spectrum**

Following the USG diagnosis of omphalocele, patient underwent counseling by the Obstetric Department and was managed by terminating the pregnancy.



**Fig. 4: Post Cesarean Gross Image of Fetus showing Anterior Abdominal Wall Defect Having Herniation of Liver and Bowel Loops were Contained in Sac Lined by the Amniotic Membrane. The Cord can be Seen Getting Inserted into the Sac.**

#### **Discussion:**

Omphalocele is derived from Greek term Exomphalos meaning “Outside the navel” [2]. Incidence of omphalocele is recorded as 1 in 4000 live births, most commonly seen in multiparous mothers of advancing age(>35 years) as seen in our case [4]. Broadly, there are two accepted theories known for the underlying pathology of omphalocele:

- 1) Failure of the lateral body wall to migrate and close in the center, this cause has been proposed when the underlying content of the omphalocele is predominantly liver.
- 2) Failure of the reduction of physiologic embryonic umbilical hernia after 12 weeks of gestational age results in omphalocele, predominantly containing bowel loops [5-6].

Antenatal ultrasound is the modality of choice to diagnose anterior body wall defects, the common imaging findings seen in omphalocele are -a centrally placed abdominal wall defect located at the base of the umbilical cord insertion thus, the cord insertion forms the apex of the defect as seen in our case. Prenatal USG also demonstrates herniated organs which are separated from the amniotic cavity by membranes characteristic for this abnormality. The inner membrane is formed by the peritoneum, and the outer layer represents the amnion. Sometimes loose mesenchymal tissue (Wharton jelly) can also be visualized between the membranes. The size of abdominal opening in an omphalocele may range from a simple hernia containing only bowel loops to giant omphaloceles in which large part of liver protrudes. The liver is the most frequently eviscerated organ followed by bow loops.

The prognosis of the fetus totally depends upon additional findings like associated pulmonary hypoplasia, restrictive lung disease, oligohydramnios, polyhydramnios and other congenital anomalies which can complicate the outcome. The most common chromosomal anomalies include trisomy 18 or 13, Beckwith-Wiedemann Syndrome, Pentalogy of Cantrell, Meckel-Gruber syndrome and cardiac defects [4, 7].

According to a study conducted by Mayer *et al.* intrauterine death occurs in 10% of cases if no other anomalies are present, whereas, mortality increases to 80% for infants with one or more concurrent malformations and to approximately 100% for those with chromosomal or cardiovascular abnormalities [8]. The most common differential diagnosis of omphalocele is gastroschisis and umbilical hernia. Gastroschisis is a right para midline anterior wall defect in which on USG free-floating extra-abdominal bowel loops are seen herniating from the defect, with no associated membrane covering. Unlike omphalocele fetus with gastroschisis have a survival rate of 90-94% and generally occur as isolated defects without any other chromosomal anomalies [7]. An umbilical hernia is a relatively common entity

that results from a fascial defect at the umbilicus and is always covered by an intact skin and subcutaneous tissue. The USG findings include the intestine protruding to only the base of the hernia and a normal umbilical cord insertion site. Umbilical hernia are generally an isolated finding and have a better prognosis than omphalocele [9]. There are other few complex anterior wall defects in the fetus which are much rarer to omphalocele and gastroschisis. 1) These consist of Pentalogy of Cantrell (consists of congenital heart disease and defects in the lower sternum, anterior diaphragm, diaphragmatic pericardium, and supraumbilical abdominal wall), 2) limb body wall complex, 3) cloacal and bladder exostrophy. A simple diagnostic algorithm can be followed to differentiate these defects on basis of USG (Table 1).

**Table 1: Diagnostic Algorithm to Differentiate Anterior Abdominal Wall Defects [4, 7]**

Position of the defect	Diagnosis to consider	Cord insertion	Other imaging features
Midline	Omphalocele	Abnormal – Defect involves the cord insertion site.	<ul style="list-style-type: none"> <li>• Limiting membrane present</li> <li>• Ascitis common</li> <li>• Liver and bowel as contents (liver being more common)</li> <li>• Associated anomalies common</li> </ul>
	Pentalogy of Cantrell		<ul style="list-style-type: none"> <li>• Findings of omphalocele plus-</li> <li>• Congenital heart disease</li> <li>• Defect in- lower sternum, anterior diaphragm, diaphragmatic pericardium</li> </ul>
Right Paraumbilical	Gastroschisis	Normal	<ul style="list-style-type: none"> <li>• Small defect- with bowel as its content</li> <li>• No associated membrane covering</li> <li>• Associated bowel complication like- atresia, malrotation, obstruction</li> <li>• Other anomalies absent</li> </ul>

*Continued...*

Position of the defect	Diagnosis to consider	Cord insertion	Other imaging features
Other	<b>Limb body wall complex</b>	Abnormal- <ul style="list-style-type: none"> <li>• Cord is very short or absent</li> <li>• Cord insertion site not identified</li> </ul>	<ul style="list-style-type: none"> <li>• Rare and very lethal</li> <li>• Abdominal wall does not develop.</li> <li>• Peritoneal cavity is open to the extra embryonic coelom</li> <li>• Complete evisceration of the abdominal contents, which are adherent to the placental surface</li> <li>• Wide spectrum of limb anomalies such as – clubfoot and absent limbs are noted</li> </ul>
<b>Infraumbilical</b>	<b>Bladder exstrophy</b>	Normal-cord insertion	<ul style="list-style-type: none"> <li>• The hallmark of bladder exstrophy is a persistently “absent” urinary bladder.</li> <li>• The bladder halves are exposed and everted which is visible at ultrasonography as an irregular contour of the lower anterior abdominal wall.</li> </ul>
	<b>Cloacal exstrophy</b>	Lower defect – may involve cord insertion site	<ul style="list-style-type: none"> <li>• Absence of a visible bladder, and exstrophy of all the structures that form the cloaca (ie, rectum, bladder, and lower genitourinary tract).</li> </ul>

**Conclusion:**

Prognosis of fetus with omphalocele is generally more fatal than gastroschisis due to its associations with other chromosomal defects, USG between 12-20 weeks can accurately diagnose anterior wall defects making it very

importance to conduct a fetal anomaly scan by a skilled radiologist. Early diagnosis permits an opportunity to counsel the family and to prepare for optimal postnatal care.

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