Case Report

Large Omphalocele with Limb Deficiency

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Abstract

Omphalocele is defined as herniation of abdominal contents into the base of umbilical cord and covered by a membrane. Large omphalocele contains liver. We report a case of twin pregnancy (monochorionic diamniotic) with one stillborn anomalous fetus, whereas the other was a live-born preterm low birth weight baby. The anomalous baby had a herniated sac at the insertion of umbilical cord covered by a membrane and absent left upper limb. Content of herniated sac included liver with part of it outside the sac. Microscopic examination of the membrane showed a layer of amnion.

Keywords: *Giant omphalocele, limb deficiency, liver*

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Introduction

Omphalocele or exomphalos is defined as herniation of abdominal contents into the base of umbilical cord. It is always covered by a membrane.[1] It consists of peritoneum and amnion.[1,2] It is the most common congenital midline abdominal wall defect. Incidence is 1 in 5000 live births. It is of two types, small and large (giant). Both the types have a different pathogenesis. However, large omphalocele always contains liver as one of the contents as in this case. In addition, the fetus also had absent left upper limb.

Case Report

A 22-year-old female presented with 8-month history of amenorrhea with complaints of pain abdomen, paravalvular leak, and decreased fetal movements. She has been married for 3 years (nonconsanguinous marriage).

Per abdomen examination showed the uterus of 34-week size. Multiple fetal parts were felt (monochorionic-diamniotic pregnancy). Twin 1 was in cephalic presentation while twin 2 was in breech presentation. Ultrasonography (USG) revealed one fetus in oblique lie with no anomaly and adequate liquor. The other fetus was in transverse lie with omphalocele and herniation of the liver and absent unilateral upper limb [Figure 1].

After delivery, twin 1 was sent to the neonatal intensive care unit due to preterm delivery and low birth weight. Twin 2 was a fresh stillborn anomalous baby and was received for autopsy. External examination revealed a male fetus with anal opening patent. The umbilical cord grossly showed two vessels. A herniated sac was seen at the insertion of umbilical cord measuring 6 cm × 4 cm and covered by a translucent membrane. The umbilical cord was attached to the apex of sac. The left upper limb was absent.

The fetus was cut opened using I-shaped incision from the chin to the pubic symphysis. *In situ* dissection through the Rokitansky method was done. Content of herniated sac included liver. A part of the liver was seen outside the sac also [Figure 2].

On microscopic examination of organs, the umbilical cord showed two vessels. Sections from membrane of sac showed a layer of amnion [Figure 3]. The liver present inside the sac showed areas of necrosis, dense lymphocytic infiltration, and acidophilic bodies [Figure 4].

On the basis of the above findings, the diagnosis of large omphalocele with limb deficiency was given.

Discussion

Omphalocele, also known as exomphalos, is defined as herniation of abdominal contents into the base of umbilical cord covered by a membrane.[1] It consists of peritoneum and amnion.[1,2]

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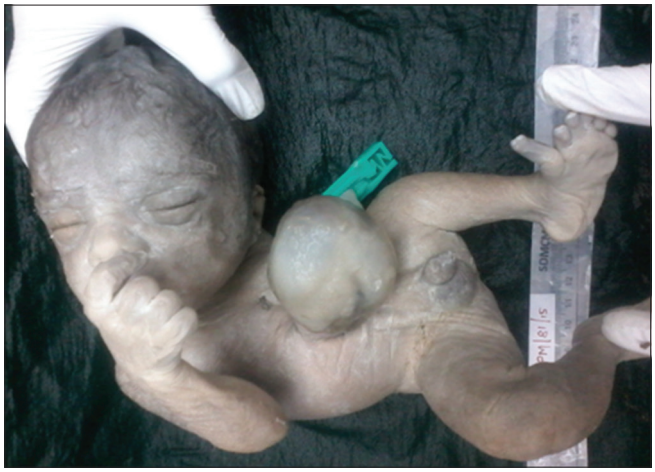

Figure 1: Fetus with herniated sac and absent left upper limb

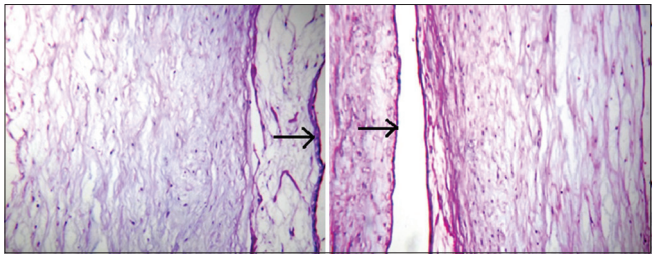

Figure 3: Microphotograph of membrane of sac showing a layer of amnion (arrows) (H and E, ×40)

The membranous sac is translucent and avascular. It has two layers: outer layer is amnion and inner layer is peritoneum. Wharton's jelly is present in between these two layers.[1] It protects against infection and loss of extracellular fluid. It may rupture *in utero* in 10%–18% of cases or from delivery process in 4% of cases. Approximate size is 2–10 cm.[2] It is the most common congenital midline abdominal wall defect. Incidence is 1 in 5000 live births.[2] The male: female ratio is 1:1[3]

Omphalocele can be associated with many defects such as diaphragmatic defects, pulmonary hypoplasia, neural tube defects, cardiac defects (atrial septal defect, ventricular septal defect, and tetralogy of Fallot), gastrointestinal defects (malrotation), renal agenesis, fetal valproate syndrome, Beckwith–Wiedemann syndrome, pentalogy of Cantrell, limb deficiencies, ossification defects of skull, cleft palate, polyhydramnios, OEIS syndrome (omphalocele, exstrophy of bladder, imperforate anus, and spinal defect), chromosomal abnormalities, i.e., trisomy 18, 13, 21, 14, and 15, Turner syndrome, and Klinefelter syndrome.[2,4]

Investigations needed for diagnosis are prenatal USG in the second trimester (14 weeks), polyhydramnios, maternal serum alpha-fetoprotein, and amniocentesis. Regarding prognosis, small omphalocele with no

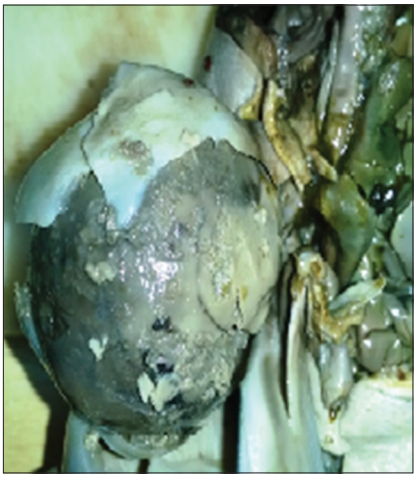

Figure 2: Herniated sac covered by translucent membrane and contains liver

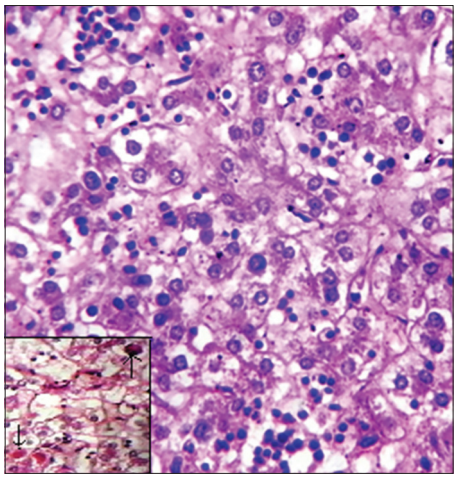

Figure 4: Microphotograph showing liver with dense lymphocytic infiltration (H and E, ×10). Left lower inset shows apoptotic bodies (H and E, ×40)

anomalies has a survival rate of 80%–90%. If omphalocele is associated with chromosomal and structural defects, then the mortality rate increases to 80%–100%. It is to be noted that omphalocele *per se* is not an indication for caesarean section. Usually, vaginal delivery is preferred in case of omphalocele. However, in case of intrauterine growth restriction baby may be born prematurely. Only giant omphaloceles containing a large portion of the liver are delivered by caesarean section.[5]

Differential diagnosis in our case includes physiological gut rotation, gastroschisis, cloacal or bladder exstrophy, and pentalogy of Cantrell (ectopia cordis). The most important differential diagnosis is gastroschisis where there is herniation of bowel usually through a small, off midline abdominal wall defect without any covering membrane. It can be associated with intestinal atresia and malrotation but never with chromosomal anomalies. It has a better prognosis.[5]

Maternal risk factors for fetus developing omphalocele include early pregnancy (less than 20 years), late pregnancy (more than 40 years), folic acid deficiency, obesity,medications such as saliscylates, acetaminophen, ibuprofen, pseudoephedrine, alcohol, smoking and drugs like marijuana and cocaine.[4-8]

The midgut extends from the second part of duodenum to the proximal two-third of transverse colon. It is supplied by the superior mesenteric artery. By the 6th week of development, the gut grows faster than abdominal cavity and herniates into extraembryonic coelom outside the abdominal cavity at the base of umbilical cord. This is known as physiological midgut herniation. The midgut loop has two segments.

Cranial segment includes distal duodenum, jejunum, and part of ileum, whereas caudal segment includes terminal ileum, cecum, appendix, ascending colon, and proximal transverse colon. The first rotation includes cranial segment in the extraembryonic coelom which rotates 90° anticlockwise which results in cranial segment being on the right and distal segment being on the left. The second rotation includes jejunum and ileum in the abdominal cavity which rotates 90° anticlockwise. The third rotation includes caudal segment in the abdominal cavity which rotates 90° anticlockwise such that cecum and appendix lie on the right side of liver. Hence, the whole rotation of the intestine amounts to 270° anticlockwise.[9]

If there is nonregression of physiological midgut herniation which usually regresses by the 12th week, then it results in simple small omphalocele which contains only bowel.[10] Other theory states that during the development of embryo which occurs in 4th-5th week, four areas of tissue form folds to close the abdominal wall. These four folds are cephalic, caudal, right lateral and left lateral. Lateral folds form lateral portions of abdominal wall, whereas cephalic and caudal folds make up the epigastrium and hypogastrium. Rapid growth of the liver and intestine also occurs at this time such that herniation of the liver and loops of bowel results in large/giant omphalocele due to failure of lateral folds to fuse. It is to be noted that the liver is never present in physiologic midgut rotation.[10,11]

Small omphalocele contains only small intestine, and it has more chances of chromosomal abnormalities and abnormal karyotype, whereas giant/large omphalocele contains liver (extracorporeal liver), colon, and stomach.[12]

Treatment depends on three factors, namely size of omphalocele, presence of other chromosomal abnormalities, and gestational age of baby.[5] Surgical treatment of small omphalocele should be done soon after birth. However, in case of large omphalocele, repair should be done in stages.[13]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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