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Case Report

Omphalocele: A Rare Case Report

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Abstract

Gastroschisis and omphalocele are commonly described anterior abdominal wall defects. Omphalocele is a midline anterior abdominal wall defect with extrusion of abdominal viscera, covered by a membranous sac, into the base of the umbilical cord. Omphaloceles occur in 1 in 3,000 to 10,000 live births. Contents of omphalocele are usually bowel, liver, or both of these. A male foetus of 21wks gestational age was delivered by MTP after being detected of having omphalocele on USG. The diagnosis was confirmed later by autopsy.

Keywords: Anterior abdominal wall defects, omphalocele, Exomphalos

Introduction

An omphalocele is a ventral defect of the umbilical ring resulting in herniation of the abdominal viscera. The incidence of omphalocele ranges between 1.5 and 3 per10,000 births [1-3]. Foetuses with omphalocele have increased incidence of anomalies up to 50-70% that includes chromosomal, cardiac genitourinary, central nervous system and skeletal system which is the cause of increased mortality in these cases [4-7]. Here we report a case of omphalocele delivered by MTP in a 21 weeks fetus containing liver and stomach as its content.

Case report

A 25-year-old primigravida presented to our hospital with complaints of decreased fetal movement along with lower abdominal pain since last 24 hours. She didn't had any previous ANC check-ups anytime in our hospital. However she was under Iron and folic acid supplementation and took 2 doses of Inj. TT, as guided by the ASHA worker of her village. Based on her complaints she was advised to go for USG. USG showed an intrauterine gestation of a single live fetus of 15 wks. There was a defect measuring 15 mm noted in the anterior abdominal wall with herniation of stomach and liver through it (omphalocele). No other gross congenital anomaly could be detected.

The placenta was posterior and low lying with liqor volume being adequate. The couple was counseled to go for MTP based on the dismal prognosis about the fetal condition and complications they would encounter, if they continued with the current pregnancy. With the consent of the couple, MTP was done and the fetus into to was send to our dept for histopathological analysis.

Autopsy was performed as per normal protocols with detailed anthropometry measurements. It was a male fetus having a large omphalocele containing intestines,

liver and spleen as its content. All organs were enclosed in single sac thus confirming the USG diagnosis. Sections were given from different organs for histopathology study, which on microscopy were compatible with age of the fetus.

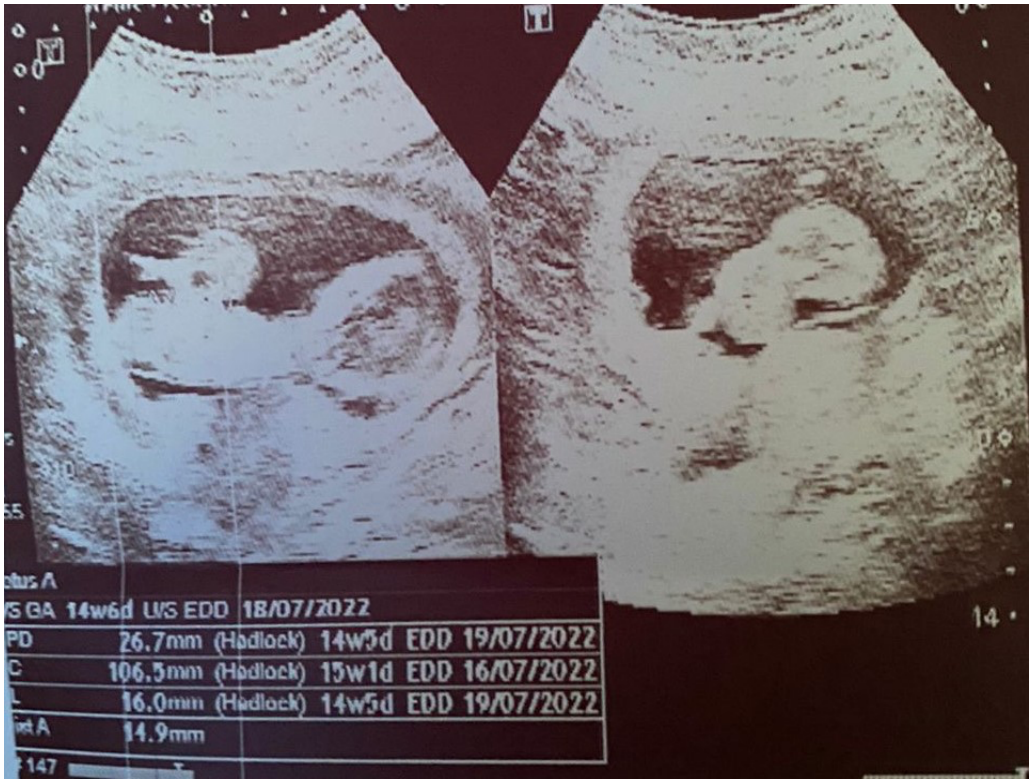

Figure 1: USG –There is a defect measuring 14.9mm noted in the anterior abdominal wall with herniation of stomach and liver through it (Omphalocele).

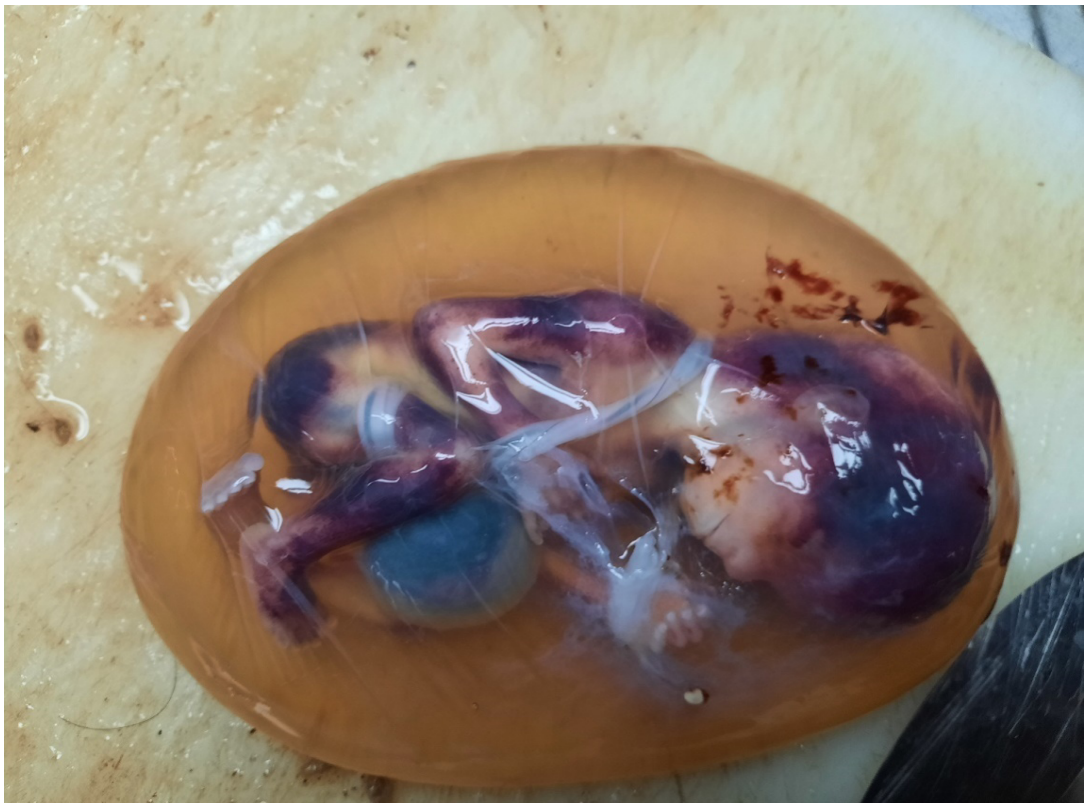

Figure 2: Gross pic showing Foetus with intact amniotic sac received Into to

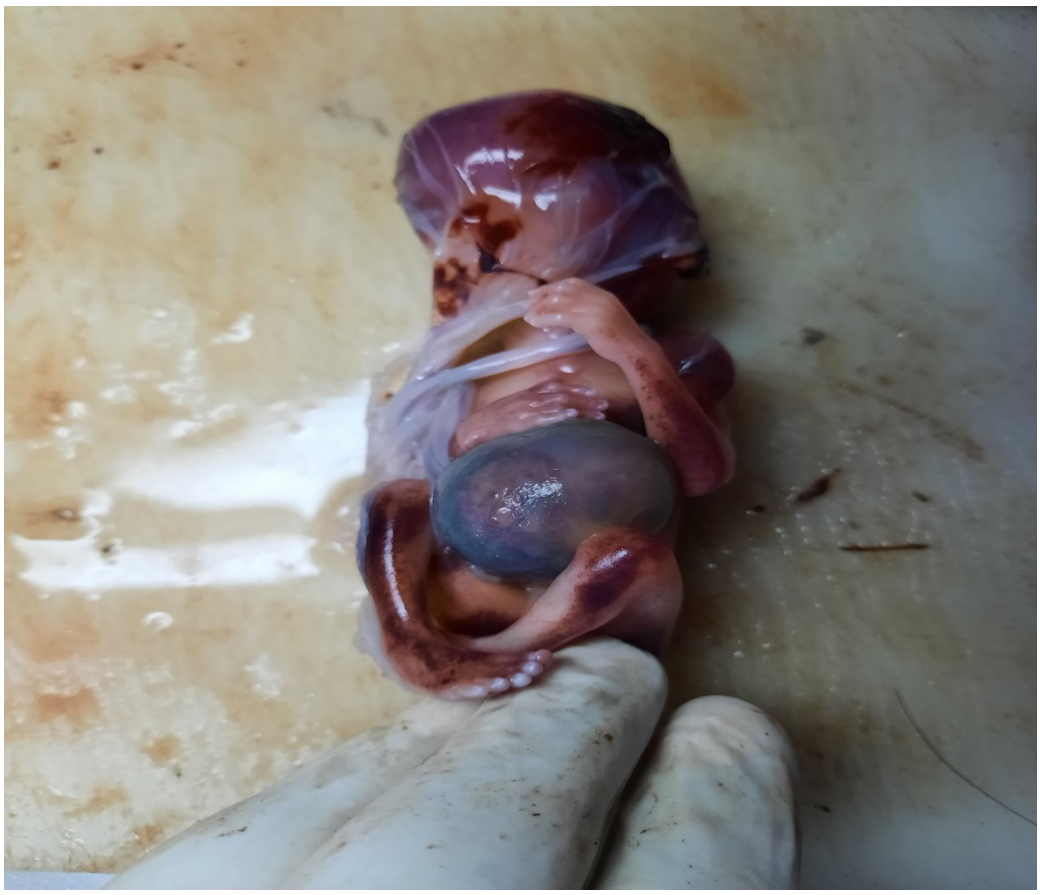

Figure 3

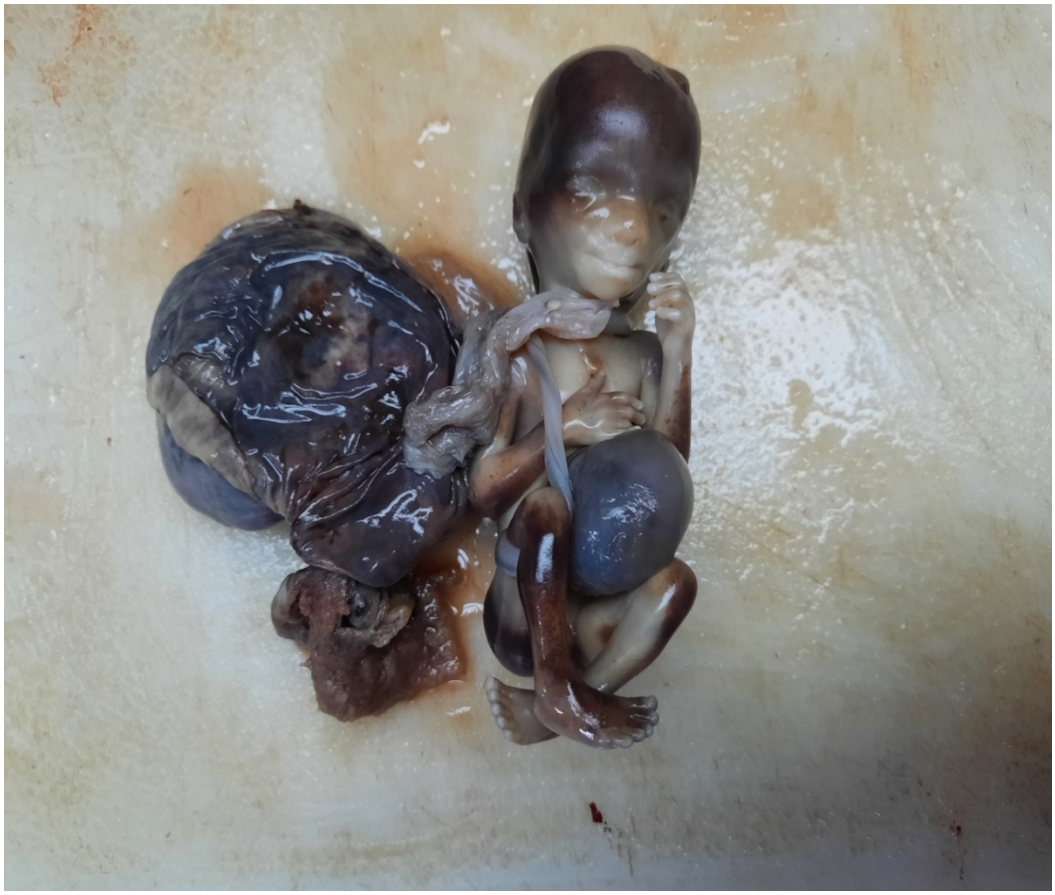

Figure 4: Gross Pic showing foetus with omphalocele sac and Placenta.

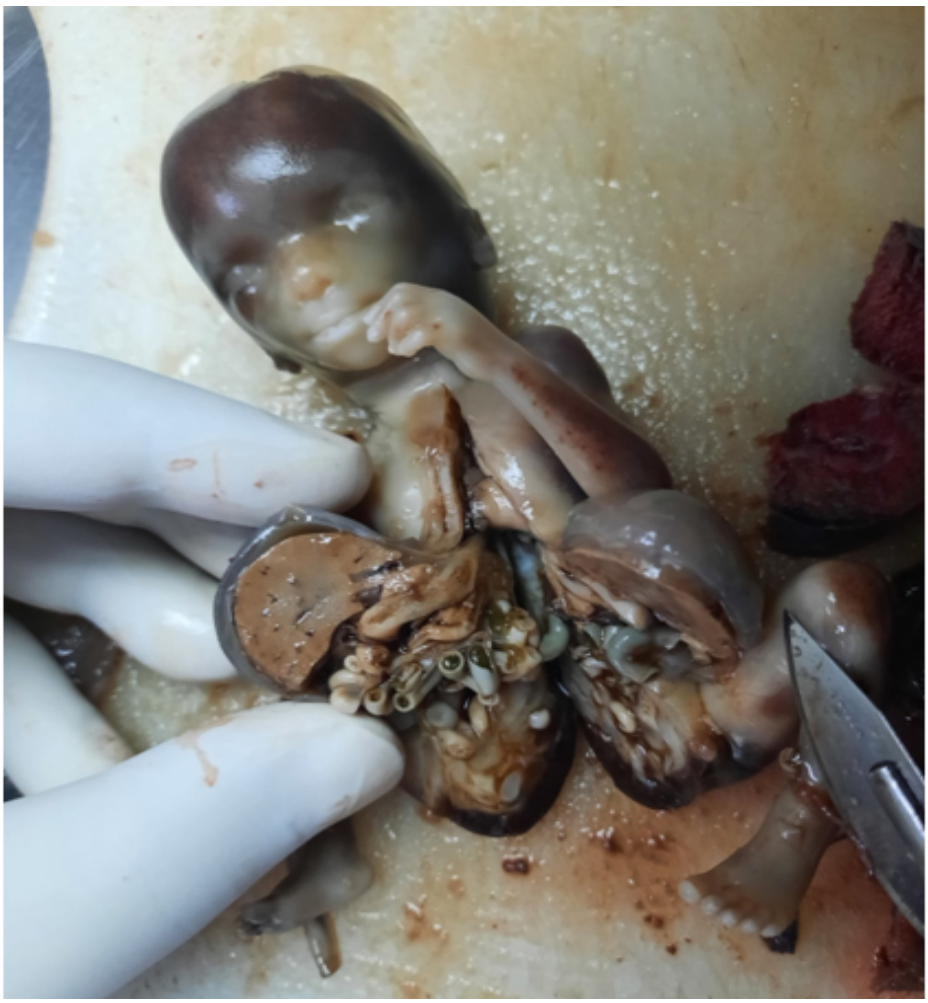

Figure 5: Gross Pic showing liver stomach and intestine as content of omphalocele

Figure 6: Showing sections given from different organs

Discussion

Two most common anterior abdominal wall defects are gastroschisis and omphalocoele or exomphalos. Overall incidence of anterior abdominal wall defects is 2.63 per 10000 live births, while that of omphalocele is 2.17 per 10000 live births which is lowest amongst Indian population. Exomphalos is literally translated from the Greek word which, means 'outside the navel'. Omphalocele is a congenital abnormality in which the contents of the abdomen herniate into the intact umbilical cord through the umbilical ring and the tissues being covered by a membrane. Gastroschisis on the other hand means 'stomach cleft' which is a congenital defect of the abdominal wall, usually to the right of the umbilical cord insertion and abdominal contents herniate into the amniotic sac with no membrane covering. These congenital malformations have a high mortality rate. Only about 60 % of children with such type of malformations survive until the end of first year of age. There is thin Line of demarcation between these two entities and some of the text books have grouped them together, but actually these are two different entities. The exact mechanism leading to these defects is not clear .Various opinions have been described in literatures regarding the pathogenesis that leads to these anterior abdominal wall defects. Anterior abdominal wall develops from fusion of four ectomesodermic folds that is cranial, caudal and two lateral folds. Failure of fusion of them results in omphalocele [8]. If omphalocele is associated with caudal fold failure; it results in extrophy of bladder or cloaca. If it is associated with cranial fold failure, it results in Pentalogy of Cantrell [9]. While other literatures have suggested during the 6th week of embryonic development, the rapid growth of bowel causes the intestinal loops to enter the extraembryonic celome. These bowel loops return to embryonic cavity by 3rd month of gestation. If due to some reason this returning process fails to occur, then this results in omphalocele formation [4].Gastroschisis is thought to result from an ischemic insult to the developing body wall [10]. It is probably caused by disruption of right omphalomesentric artery. As per the review of literature, Older mothers above age of 35 years carry 1.8 fold higher risk of having a baby with omphalocele [11,12] however in our case mother was only 18 yrs of age. cigarette smokers [13, 14] and use of recreational drug like cocaine, amphetamine, marijuana and alcohol [15, 16], in undernourished [17], who use over counter medication like ephedrine, pseudoephedrine etc [14, 18] are more prone to develop gastrochisis. No such specific risk factors are mentioned with regard to omphalocele. Both mainly affect male children of first para first gestational mothers. Both are associated with prematurity and low birth weight [19]. Omphalocele is associated with cardiac anomalies [20], gastrointestinal, genitourinary [21], neural tube [22] and musculoskeletal defects. Omphalocele is a part of Beckwith–Widemann syndrome [23], Pentalogy of Cantrell, [24] Meckel–Gruber syndrome [25], and lethal cleft palate Omphlocele syndrome [26]. Omphalocele is associated with higher incidence of other structural and chromosomal abnormalities like trisomy 13, 14, 15, 18 and 21 in 30 % of cases. In gastroschisis, the incidence of associated anomalies is between 10% and 20%, and most of the significant anomalies are in the gastrointestinal tract[2]. About 10% of babies with gastroschisis have intestinal stenosis or atresia that results from vascular insufficiency to the bowel. Prenatal ultrasound is an useful modality in detecting such abdominal defects. In normal embryogenesis, extra abdominal intestinal loops return to abdominal cavity by 11 weeks of gestation so in order to confirm diagnosis of such anterior abdominal wall defects the routine USG should be preferably delayed till 14 week of gestation [27]. Thus USG not only helps to identify and confirm findings of such defects but also gives an opportunity to gyneacologist to counsel the couple and

discuss the prognosis of the fetuses born with such defects and plan out further management for the betterment of the patient . Other serum markers that help us in reaching at diagnosis is serum markers like Maternal Serum Alfa Feto Protein (MSAFP). The median value of MSAFP in gastroschisis is 9.42 multiples of median (MOM). While in omphalocele, it is only 4.18 MOM making it little bit difficult to diagnose [2]. The mode of delivery in cases of such abdominal birth defects is controversisal. Review of literatures have suggested mixed opinion regarding this aspect . Studies by How *et al* have reported that these fetuses can be safely delivered by vaginal route[28], however other studies have shown improved outcome of fetus delivered by C section [29]. However mainstay of treatment of such anterior abdominal wall defects is reduction of the herniated viscera into the abdomen and to close with fascia and skin to create a solid abdominal wall with relatively normal umbilicus with minimal risk to the baby. Pregnancy termination options should be discussed with the couple depending on the gestational age of the fetus. If the couple still wishes to continue with the pregnancy ,then serial USG for fetal growth monitoring should be done and elective CS section must be planned in presence of neonatologist and a pediatric surgical management team. In our case patient decided to go for medical termination of pregnancy at 21 weeks. In comparison with gastroschisis, omphalocele doesn't require urgency in surgical management so long as the viscera are covered with membranes as in our case.

Conclusion

Prognosis of infant with omphalocele is fatal as it is commonly associated with chromosomal defects. It also depends largely on associated structural anomaly like cardiac defects and if it is a large omphalocele containg spleen, liver with intestine there is a higher chance of poor neonatal outcome .Such anterior abdominal wall defects can be prevented to a large extent by taking multivitamins during pregnancy . Review of literatures have suggested that use of multivitamins during pregnancy is associated with 60% reduction in risk of symptomatic Omphalocele [19].

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