Case Report

Omphalocoele - A Salvaged Baby

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Abstract

Gastroschisis and Omphalocoele (Exomphalos) are the two commonnest ventral abdominal wall defects. Failure of bowel loops to return to the foetal abdominal cavity following physiological umbilical hernia during sixth to tenth week of intrauterine development results in Omphalocoele. It is related with a greater short and long term risk of infant morbidity and mortality. Diagnosis and management of these ventral abdominal wall defects still pose a great challenge for clinicians. Early diagnosis (as early as 11weeks to 20 weeks) and prompt correction by surgery improves the prognosis and enhances the neonatal survival. We are reporting a rare case of Exomphalos which was diagnosed following routine anomaly scan and successfully corrected surgically in postnatal period.

Keywords: Ventral abdominal wall defects, Omphalocoele, Exomphalos, Gastroschisis

Introduction

Omphalocoele is protrusion of viscera which is covered by peritoneum and amnion into the base of umbilical cord.[1, 2] The worldwide prevalence of Omphalocoele is 2-3% per 10000 births and incidence of intestinal herniation into cord is 1 in 5000 births; liver and intestinal herniation into cord is 1 in 10,000 births.[3,4]

Rapid growth of intestines during the sixth week of early intrauterine development leads to herniation through the umbilical ring causing physiological umbilical hernia. During tenth week the herniated bowel loops undergo a 270° anti-clockwise rotation and return to the abdominal cavity fixing the gut in normal axis, the failure of which results in exomphalos.

Majority (upto 75%) of infants with omphalocoele have associated congenital anomalies like Beckwith Wiedemann syndrome, Cloacalexostrophy, Fibrochondrogenesis, Marshall-Smith syndrome, Meckel-Gruber syndrome, Pentalogy of Cantrell, Trisomy 13, Trisomy 18. Non chromosomal multiple and isolated congenital anomalies with involvement of musculoskeletal system is 24%, urogenital system is 20%, cardiovascular system is 15%, central nervous system is 9%.Survival rate can be upto80%.

In Exomphalos minor, the defect is small (<4cm) at umbilicus (central), sac is small (peritoneum), covered by whartons jelly and layer of amniotic membrane, contains one loop of small intestine or meckel's diverticulum, complications include umbilico enteric fistula.

In Exomphalos major, the defect is larger (>4cm) in the centre of abdominal wall (usually supraumbilical, eccentric cord), sac is large (peritoneum), covered by only a layer of amniotic membrane, contents include any abdominal viscera like liver (specifically, it is adherent to the sac), stomach, bowel, spleen etc, complications include rupture of sac and coverings may occur leading to infections, peritonitis (cause of death).

Prognosis and survival of children born with omphalocoele depends on the associated anomalies usually isolated cases with omphalocoele have good prognosis.[5,6,7] Inspite of improved antenatal care and availability of high quality radioimaging techniques

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(MRI, high resolution and 3D ultrasound), a large proportion of the associated anomalies will remain undiagnosed until birth which influences the outcome. [8]

Case Report

Reporting the case of a 21 year old woman who visited Department of Obstetrics & Gynecology at 22 weeks of gestation in her first pregnancy for antenatal checkup. Her pregnancy until then was uneventful with a normal early anomaly scan. During this visit, an Anomaly scan was done which showed a sac containing bowel loops and fluid herniating through umbilicus- likely omphalocoele. (Figure 1)

Opinion from the Paediatric surgeon was taken and the mother was counselled regarding the birth defect and was advised to continue pregnancy to term.

She was followed up and monitored closely until term when she was induced with Cerviprime gel and delivered a live male baby weighing 2.46kg vaginally. Postpartum period was uneventful.

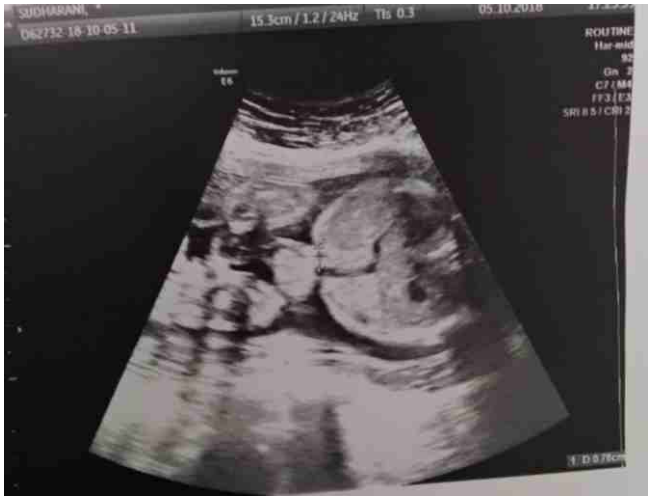

Figure 1. Anomaly scan showing Omphalocele

Figure 2. Neonate with Omphalocele

Baby did not cry immediately after birth, cried after AMBU for 2 minutes. Apgar at 1 min was 5 and 5 min was 7. On examination, Omphalocoele was present. (Figure 2) Under aseptic precautions umbilical cord was clamped and cut. 2 arteries and 1 vein was noted. Baby was admitted in NICU.

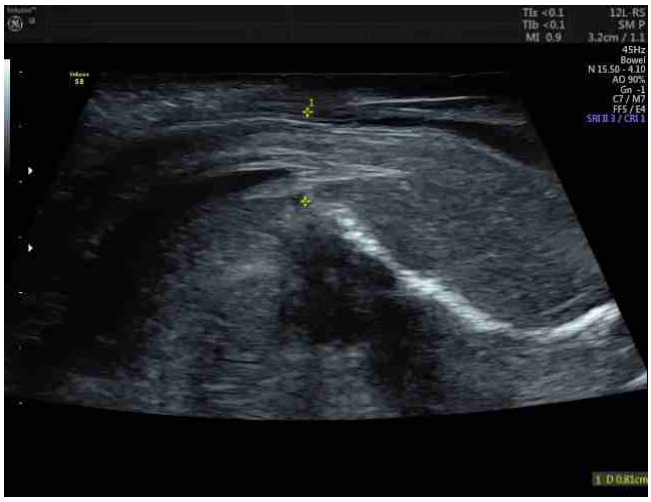

Figure 3. USG of neonate showing Omphalocele

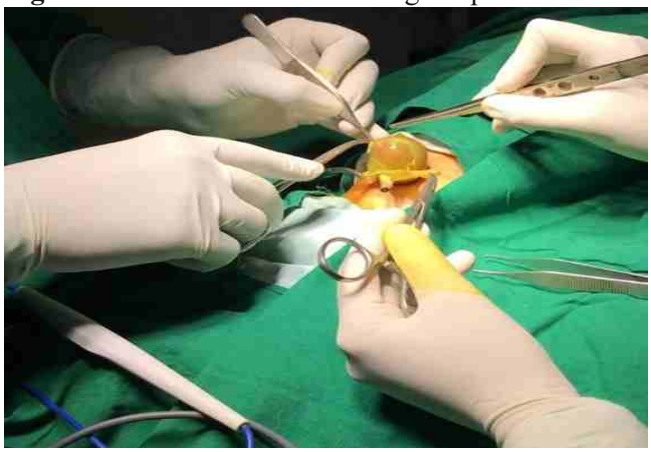

Figure 4. Identification of sac and contents

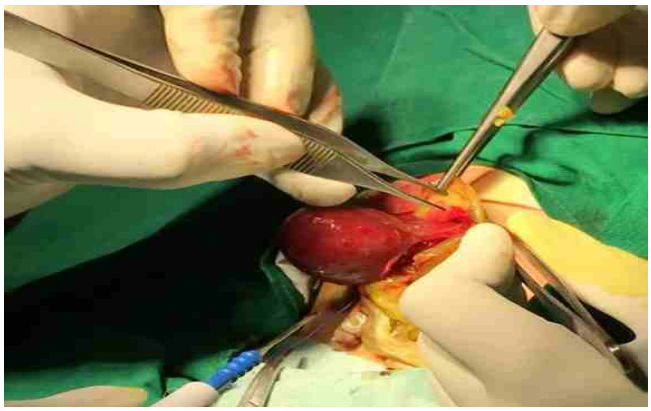

Figure 5. Repair in process

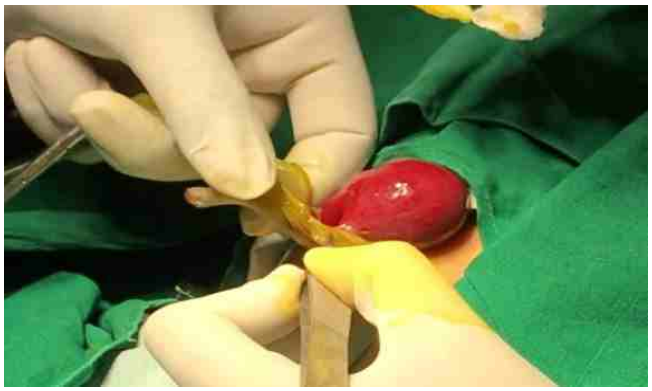

Figure 6. Release of adhesions

Figure 7. Umbilicoplasty

Ultrasound abdomen was done on the first day of life. A defect of 8 to 10 mm was noted with herniating contents left lobe of liver and gall bladder seen with in the sac. Normal vascularity was noted within. (Figure 3)

2D echo was taken on same day which showed Situs solitus, levocardia, AML prolapse with moderate mitral regurgitation, 5 mm ostium secundum ASD with left to right shunt, normal biventricular function.

Plan for elective repair of omphalocoele was done on day 3 of life. Procedure included circumferential incision separating sac and skin, delineating rectus sheath, content was liver and gall bladder, adhesions were released all around, rectus muscle incised laterally, Gall bladder and liver reverted back to abdomen and umbilicoplasty done. (Figure 4,5,6,7). Intra operative and post operative period was uncomplicated and the baby was discharged from Intensive Care Unit on tenth day and advised close follow up.

Discussion

In clinical practice, gastroschisis and omphalocoele are commonly encountered anterior abdominal wall defects. Omphalocoele is a weakness in the ventral abdominal wall with absent skin, fascia and abdominal muscles which is enclosed by peritoneum and amnion.[9] It varies from a few centimetres to almost full ventral abdominal wall.

Development of anterior abdominal wall is by fusion of cranial, two lateral and caudal ectomesodermic folds. Failed fusion results in omphalocoele.[10] Caudal fold failure results in low or hypogasticomphalocoele as seen in bladder or cloacalexostrophy. Cranial fold fusion failure results in high or epigastricomphalocoele as seen in Pentalogy of Cantrell. [11] Defect in lateral fold results in Classic omphalocoele with a mid abdominal defect.

Gastroschisis is a full thickness abdominal wall defect which is secondary to incomplete lateral fold closure during the 6th week of intrauterine growth.[12] There are various theories explaining the occurrence of gastroschisis including discrete teratogenic insult resulting in an isolated defect in differentiation of the somatopleural mesenchyme, due to rupture of an umbilical hernia in utero or due to disrupted right omphalo-mesenteric artery.

In India, the overall incidence of ventral abdominal wall defects, Gastroschisis and Omphalocoele is 2.6, 0.5 and 2.2 per ten thousand live births respectively. Women under 25 years of age have 2.8 fold greater risk of having a baby affected with gastroschisis. Whereas women above 35 years have 1.8 fold risk of having a baby affected with omphalocoele.[13] Both mainly affect male children of primiparous mothers.

Early diagnosis of anterior abdominal wall defects (as early as 10-12 weeks) is important and can be made by Transvaginal sonography, when an echogenic mass will be found anterior to the fetal abdomen. Use of 3D transvaginal examination may facilitate this diagnosis early in gestation.

Anterior wall defects can also be diagnosed prenatally by biochemical marker like Maternal Serum Alfa Feto Protein (MSAFP). Cut off value of MSAFP is 9.4 multiples of median (MOM) and 4.2 MOM in gastroschis is and omphalocoele respectively, posing a difficulty in early diagnosis of omphalocoele.[13] The

detection rate for omphalocoele is poorer because of the presence of an intact amnioperitoneal membrane covering the abdominal cavities, in contrast to direct exposure of intestines to amniotic fluid in gastroschisis.

Nowadays chromosomal analysis is recommended strongly as multiple studies have documented a high rate of karyotype abnormalitie.

Significant pulmonary hypoplasia and pulmonary hypertension may complicate the neonatal course in nearly 50% of the cases having giant omphalocoele. Hence, routine MRI for total lung volume assessment is recommended at 32-34 weeks.

The location and mode of delivery has been controversial hence the goal of management is delivery of a term fetus as far as possible preferably in a well equipped hospital.

Corner stone of treatment is by primary fascial closure.

Prognosis of infant with omphalocoele can be fatal if it is related with structural and chromosomal anomalies.

In this case, baby was salvaged and treated successfully. Hence every case of Omphalocoele need not be terminated.

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