



OMPHALOCELE MAJOR IN A NEONATE – A CASE REPORT IN TERTIARY CARE CENTRE, KOLAR.

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ABSTRACT

Introduction: Omphalocele is a congenital abdominal wall defect characterized by the herniation of abdominal organs through the base of the umbilical cord, enclosed in a membranous sac composed of peritoneum and amnion. **Case Report:** A 22-week antenatal diagnosis of an 8 mm omphalocele in a G2P1L1 mother. **Conclusion:** Omphalocele is a significant neonatal abdominal defect with variable clinical presentations and outcomes. Early prenatal diagnosis, thorough evaluation for associated anomalies, and a coordinated multidisciplinary approach are crucial for optimizing patient care.

KEYWORDS : Omphalocele, newborn, abdominal wall defect.

INTRODUCTION:

An omphalocele is a congenital midline defect of the ventral abdominal wall, characterized by the herniation of abdominal organs into the base of the umbilical cord, enclosed within a sac composed of an outer amniotic layer and an inner peritoneal layer. This condition arises when the physiologically herniated intestines fail to return to the abdominal cavity between the 6th and 10th weeks of gestation. Although it can be detected in the later part of the first trimester, omphalocele is most commonly diagnosed during the routine anomaly scan performed around 18 to 20 weeks of gestation.¹ Omphalocele major (OM), also referred to as giant exomphalos or hepato-omphalocele, is characterized by the presence of the liver as the predominant organ herniated outside the abdominal cavity.² large omphalocele, the prevalence is one out of every 10,000 births and OM along with gall bladder defect occurs in 1/1,00,000.³ A "giant omphalocele" is defined as an abdominal wall defect measuring 4 cm or more in diameter, typically containing part or all of the liver, and in many cases, the gallbladder as well.⁴

Case Report:

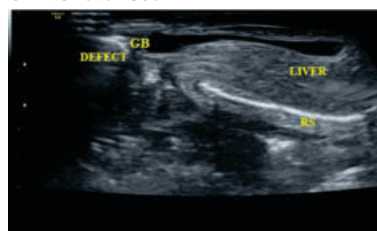
A 22-week antenatal scan of a G2P1L1 mother revealed an omphalocele. A pediatric surgical consultation was obtained, and counseling was provided regarding the identified birth defect. As the defect measured 8 mm, the decision was made to continue the pregnancy to term with regular follow-up. On 06/09/2024 at 2:07 PM, a late preterm female infant was delivered via lower segment cesarean section (LSCS) at Sri Devaraj Medical College, Kolar, due to leaking per vaginam. Postnatal examination confirmed the presence of an omphalocele.



B)
A & B Neonate With Omphalocele



C)
C) 22 Weeks Antenatal Scan



D)
D) USG Abdomen Of The Baby



A)

Management: On day 1 of life, an abdominal ultrasound revealed a defect measuring 8 to 10 mm with herniation of the left lobe of the liver and gallbladder into the sac, both demonstrating normal vascularity. A 2D echocardiogram

showed anterior mitral leaflet prolapse with moderate mitral regurgitation, a 5 mm ostium secundum atrial septal defect (ASD) with a left-to-right shunt, and normal biventricular function. Chromosomal analysis and genetic testing was sent and revealed to be normal. On day 3 of life baby had exploratory laparotomy with reduction of contents with primary closure. Post op follow up was uneventful and baby was discharged on post op day 7.

DISCUSSION:

In clinical practice, omphalocele and gastroschisis are most commonly encountered anterior abdominal wall defects.⁶ Abdominal wall develops from the fusion of cranial, caudal and lateral folds of ectomesoderm⁶ failure of which results in omphalocele. It is often associated with other congenital anomalies and genetic syndromes, such as trisomy 13, 18, and Beckwith-Wiedemann syndrome, making early diagnosis and multidisciplinary management essential. Classical omphalocele is a lateral fold fusion defect with a mid-abdominal defect. Epigastric omphalocele commonly found in the Pentalogy of Cantrell is due to Cranial fold failure. Hypogastric omphalocele is due to Caudal fold failure⁶. Omphalocele and ventral abdominal wall defects have an Incidence of about 2.2 and 2.6 per ten thousand live births in India omphalocele respectively⁵. Maternal Age above 35 years have 1.8 times greater risk of having a newborn affected with omphalocele. Anterior abdominal wall defects can be detected as early as 10-12 weeks of gestation using transvaginal sonography, but the detection rate of omphalocele in comparison with gastroschisis is less because the abdominal cavity is covered by an intact the membrane in omphalocele, but intestines are directly exposed to amniotic fluid in gastroschisis⁵. MRI for assessment of total lung volume is required at 32-34 weeks of gestation as nearly half of the cases with giant omphalocele have pulmonary hypertension and hypoplasia which affects the neonate outcome⁵.

CONCLUSION:

Prognosis of the omphalocele is fatal only if it is associated with chromosomal and structural anomalies. But all cases with omphalocele need not be terminated. Early detection and intervention help in a better outcome. While advances in surgical techniques and neonatal intensive care have improved survival rates, the prognosis is still closely linked to the size of the defect and the presence of concurrent anomalies or genetic syndromes. Continued research and improvements in prenatal screening, genetic counseling, and postnatal management are essential to enhance outcomes and quality of life for affected infants.

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REFERENCES:

1. Zahouani T, Mendez MD. Omphalocele [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan.
2. Mack AJ, Rogdo B. Giant omphalocele: current perspectives. *Res Rep Neonatol*. 2016 Jan 1;6:33-9.
3. Bedi NK, Grewal AG. A simple technique for the repair of a complex case of exomphalos major. *Int Surg J* 2014;1:102-7.
4. Valiollah Mehrabi, Arianeb Mehrabi, Maliheh Kadivar, Mehrdad Soleimani, Azadeh Fallahi, and Nazila Khalilzadeh. Staged Repair of Giant Recurrent Omphalocele and Gastroschisis "Camel-Litter Method"-A New Technique. *Acta Medica Iranica* 2012; 50 (6): 388-394.
5. Porter A, Benson CB, Hawley P, et al. Outcome of fetuses with a prenatal ultrasound diagnosis of isolated omphalocele. *Prenatal Diagn*. 2009;29:668-73.
6. Weber T, Au-fliegner M, Downard C, Fishman S. Abdominal wall defects. *CurroPinPaediatr* 2002; 14(4): 491-497.