Content available at: https://www.ipinnovative.com/open-access-journals

Indian Journal of Clinical Anatomy and Physiology

Journal homepage: https://www.ijcap.org/

Case Report

Omphalocele major with spine deformity, a congenital anomaly

Swetha B[1,*], **Hema N**[2]

[1]*Dept. of Anatomy, BGS Global Institute of Medical Sciences, Bengaluru, Karnataka, India*
[2]*Dept. of Anatomy, ESI & RC, Bengaluru, Karnataka, India*

ARTICLE INFO

Article history:
Received 01-02-2023
Accepted 12-02-2023
Available online 19-04-2023

Keywords:
Scoliosis
Congenital anomaly

ABSTRACT

Multigravida, 26 years old lady with 21 weeks gestation came with complaint of abdominal pain diagnosed as Omphalocele major with spine deformity. Due to associated anomalies, the pregnancy was terminated with parental consent. On dissection, it was observed that liver and small intestine protruded from right and left side respectively through the defect. Acute angulation of the Spine was observed. There was a privilege of intervention of omphalocele of various clinical presentations. Here is an attempt made to describe in this case which will be helpful to paediatric surgeons for interventions.

1. Introduction

Omphalocele is a rare congenital anomaly with the prevalence of around 3.38 per 10000 pregnancies.[1] The incidence is 17 per 100000.[2] It appears to develop frequently in women of extreme reproductive age, less than 20yrs and more than 40 yrs age.

This defect also known as exomphalos,[3] where anterior abdominal wall is not closed leading to protrusion of the abdominal contents. During Organogenesis, at 6^{th} week, due to lack of space inside the abdominal cavity and associated enlargement of the abdominal organs, they protrude at the base of the umbilical cord. This is physiological midgut herniation where we see only midgut loops which will reduce by 11 weeks. By 12 weeks of gestation, if the hernia persists, it is considered as pathological. The clinical presentation of Omphalocele may be isolated, but frequently it is associated with congenital anomalies. If it is simple, isolated survival rate will be close to 80% which reflects the diagnosis and treatment efficacy.[3]

2. Case Report

2.1. Patient information

26 yrs old lady, 3^{rd} pregnancy of 21weeks gestation with complaint of mild pain abdomen for a day. Diagnosed to have Omphalocele major with spinal deformity by scan and no other anomalies observed. Lab findings were Normal. Non consanguineous marriage, obstetric history of 1^{st} pregnancy-previous full term normal vaginal delivery, 2^{nd} pregnancy- miscarriage at 8 weeks of gestation. Undergone MTP with parents informed consent. Present pregnancy scanned at 8 weeks showed small live intrauterine growth foetus and no genetic history in the family were observed. Pregnancy was terminated after taking parent's consent and the infant was donated for an education purpose. On observation, it was found that the omphalocele swelling was covered with thin membrane (Figure 1). The dissection revealed that, liver was found to be protruding from the defect on right side as well as the small intestine loops were protruded from the left side (Figure 1). Enlarged normal thymus and Immature small lungs were observed whose growth was normal for the gestational age. Spine scoliosis

* *Corresponding author*.
E-mail address: swethadhananjay90@gmail.com (Swetha B).

https://doi.org/10.18231/j.ijcap.2023.011

with acute angulation of the spine was observed from T10 level to L2 (Figure 2).

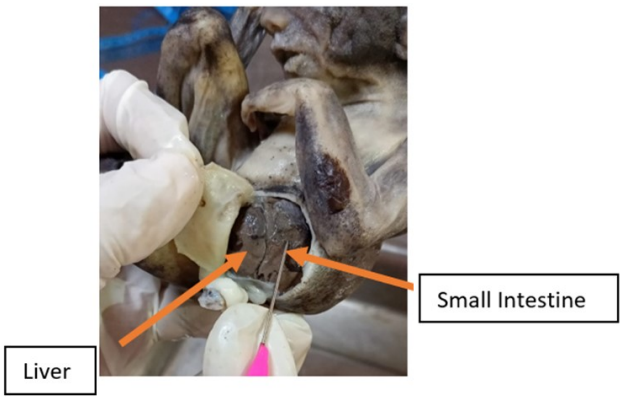

Fig. 1: Thin layered sac, protrusion of liver (right side), and small intestine (left side)

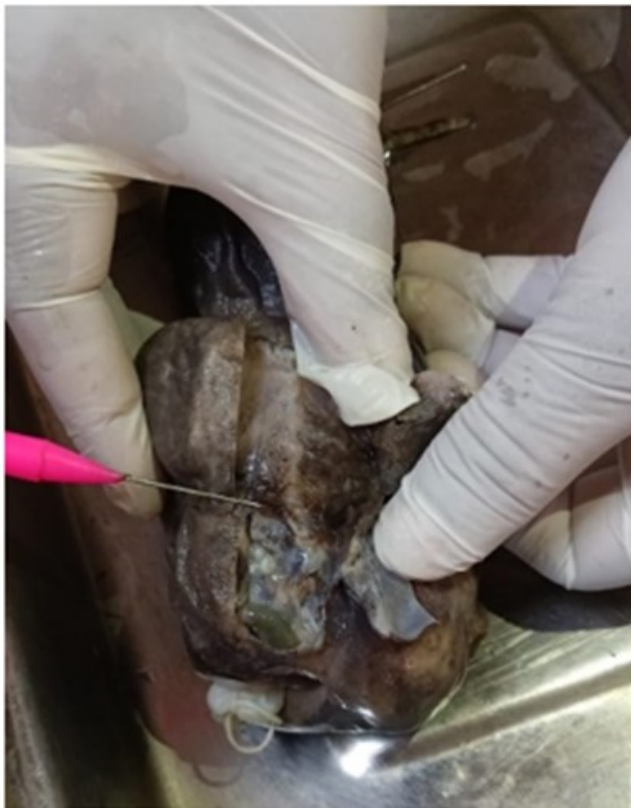

Fig. 2: Acute angulation of the vertebra

3. Discussion

In this present case, multigravida mother aged 26 yrs (normal range) without family/ genetic history had a infant with Omphalocele anamoly. The defect was covered by normal thin sac as described in literature which was made up of fused membranes of amnion externally and peritoneum internally with embedded wharton's jelly between two layers.[3–6] Omphalocele survival rate directly depends on its severity and its associated anomalies. Infants with isolated Omphalocele have a higher survival rate of 90%,[7] Omphalocele is classified as small if there is protrusion of intestinal loops through the defect, Large -if abdominal organs are noticed, giant- if the defect size is greater or equal to 5 cms with liver protruding.[1] Large Omphalocele with liver protrusion through the defect with spinal deformity will have poorer prognosis especially in the presence of genetic abnormalities.[8] Proper diagnosis at the right time reflects the efficacy of prenatal diagnosis and the decision taken by the families to terminate pregnancies with severe anomalies.[1] Omphalocele can be associated with several syndromes like Beckwith Wiedemann syndrome, congenital heart disease, Meckels Diverticulum. Other syndromes associated are Trisomy 13, trisomy 18, Trisomy 21, Pentalogy of Cantrell, Charge syndrome.

Evaluation: The early diagnosis is possible through foetal ultrasound screening (67.2% of cases in first & second trimester)[9] and also through resonance magnetic imaging (RMI), computed tomography scan. By the end of first trimester, it can be detected by elevated maternal serum alpha fetoprotein and acetylcholinesterase in the amniotic fluid.[10]

Treatment of the Omphalocele complications depends on the time of occurrence. Elective surgical interventions are the treatments preferred for neonate with omphalocele.[11] Many articles have mentioned rupture of the Omphalocele can happen prenatally or during delivery. In case of giant Omphalocele, the liver may be injured. The majority of infants with omphalocele have a small thorax with varying degrees of pulmonary hypoplasia. Here mechanical ventilation was assisted for few weeks or even months until the lungs mature. Tracheostomies are often needed.

Parental nutrition is must for the baby with omphalocele, nevertheless it leads to complications like cholestasis and hepatomegaly. Large omphaloceles need a staged repair.

4. Conclusion

Omphalocele still remains one of the congenital anomalies which can be repaired in most of the cases. This report helps paediatric surgeon to have thorough knowledge on Omphalocele.

5. Source of Funding

None.

6. Conflict of Interest

None.

Acknowledgment

My sincere thanks to Dr Usha, Gynaecologist for her support for conducting this study.

References

1. Zahouani T, Mendez MD. Omphalocele. Treasure Island (FL): StatPearls Publishing; 2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK519010/.
2. Calzolari E, Bianchi F, Dolk H, Milan M. Omphalocele and gastroschisis in Europe: a survey of 3 million births 1980-1990. *Am J Med Genet*. 1995;58(2):187–94.
3. Copel J, D'Alton ME, Feltovich H, Gratacos E, Odibo AO, Platt L, et al. Obstetric Imaging: Fetal Diagnosis and Care. 2nd ed. Philadelphia: Elsevier; 2018. p. 85–91.
4. Kominiarek MA, Zork N, Pierce SM, Zollinger T. Perinatal outcome in the liveborn infant with prenatally diagnosed omphalocele. *Am J Perinatol*. 2011;28(8):627–34.
5. Rattan KN, Singh J, Jakhar R, Dalal P, Sonika P. Omphalocele: 15-years experience from a single center in developing country. *J Clin Neonatol*. 2018;7(3):125–9.
6. Brantberg A, Blaas HGK, Haugen SE, Eik-Nes SH. Characteristics and outcome of 90 cases of fetal omphalocele. *Ultrasound Obstet Gynecol*. 2005;26(5):527–37.
7. Hijkoop A, Peters NCJ, Lechner RL, Bever YV, vanGils Frijters A, Tibboel D, et al. Omphalocele: from diagnosis to growth and development at 2 years of age. *Arch Dis Child Fetal Neonatal Ed*. 2019;104(1):18–23.
8. Marshall J, Salemi JL, Tanner JP, Ramakrishnan R, Feldkamp ML, Marengo LK, et al. Prevalence, Correlates, and Outcomes of Omphalocele in the United States, 1995-2005. *Obstet Gynecol*. 1995;126(2):284–93.
9. Poaty H, Pelluard F, Diallo MS, Ondima IPL, André G, Silou-Massamba JF. Omphalocele: a review of common genetic etiologies. *Egypt J Med Hum Genet*. 2019;20:37. doi:10.1186/s43042-019-0040-3.
10. Grande M, Arigita M, Borobio V, Jimenez JM, Fernandez S, Borrell A. First-trimester detection of structural abnormalities and the role of aneuploidy markers. *Ultrasound Obstet Gynecol*. 2012;39(2):157–63.
11. Mack AJ, Rogdo B. Giant omphalocele: Current perspectives. *Res Rep Neonatol*. 2016;6:33–9.

Author biography

Swetha B, Associate Professor https://orcid.org/0000-0002-1356-8987

Hema N, Associate Professor https://orcid.org/0000-0002-1782-9785

Cite this article: Swetha B, Hema N. Omphalocele major with spine deformity, a congenital anomaly. *Indian J Clin Anat Physiol* 2023;10(1):52-54.