

Management of a Case of Omphalocele in a Primigravida: A Case Report

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Submission Date:

14 May 2025

Acceptance Date:

15 September 2025

Published Date:

doi.org/10.56974/pmjn.284
www.pmjn.org.np

ABSTRACT

Omphalocele is a congenital abdominal wall defect at the umbilicus where abdominal organs are covered by a membrane. It results from abnormal midline development of the abdominal wall during early embryonic weeks (6th to 10th). We present a case of a 29-year-old primigravida, who attended her first antenatal checkup during 24 weeks of gestation. Ultrasound findings for 24 weeks revealed a live intrauterine fetus with normal cardiac activity but also identified a midline abdominal defect consistent with omphalocele, as bowel loops, colon, and liver were herniated into a sac. After clinical consultations, the patient and her family made the decision to terminate the pregnancy. Early detection of omphalocele helps to assess associated abnormalities and prepare for potential interventions after birth and ensure specialized care for the newborn.

Keywords: congenital; omphalocele; prevalence.

INTRODUCTION

Omphalocele (also called exomphalos) is an abdominal wall defect (AWD) at the umbilicus (ulcer) and the viscera of the belly are covered by a membrane.¹ Embryologically, omphalocele is an embryopathy because of abnormal development of the midline abdominal wall in early embryonic development (ED).² It is not fully understood exactly, but it seems the genesis of this defect occurs between the 6th and the 10th gestational week.³ Omphalocele occurs in 17% (out of 1,000,000 per pregnancy).⁴ The estimated survival rate for isolated omphalocele is 50-90% and this decreases significantly with the presence of other anomalies with recurrence rate lower than 1%.⁵⁻⁷ In this case a 29-year-old primigravida presented at 24 weeks gestation with a fetus diagnosed via ultrasound to have an omphalocele. After clinical consultation and counseling, she opted for termination, and she underwent induced premature delivery at 28 weeks.

CASE REPORT

A 29 years of female, primigravida, had presented to general outpatient department for her first antenatal check at 24 weeks of gestation. During the visit, female had not

taken iron, calcium or folic Acid. On abdomen examination, the uterus was around 24 weeks of gestation with present fetal movement and heart sound. Her baseline investigation was done with ultrasound. No other significant gynecological/obstetric history present. Ultrasonography at 24+6 weeks gestation (WOG) showed one live intrauterine fetus corresponding to 25 weeks (0 days) gestational age with normal cardiac activity (Figure 1).

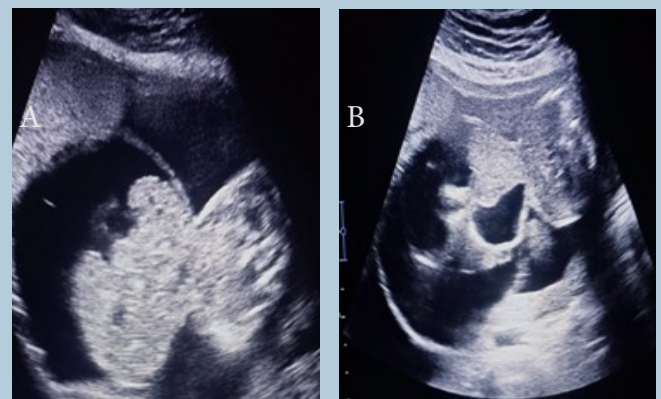


Figure 1. Ultrasonography image of almost 24+6 weeks of gestation showing intra-abdominal fetal content herniated.

The ultrasound report also mentions there was a defect of

size similar to 14.7 mm in midline abdomen through with-in intra-abdominal fetal content (bowel loops, colon and liver is herniated outside within the sac of size 78 x 69 mm) due to small abdominal circumference suggestive of omphalocele and there were no other associated anomalies.

Clinical Consultation with Gynecology, Pediatric and Radiology was done and the patient was advised about the condition of pregnancy and the patient, her family decided not to continue any further. On her visit of check-up and ultrasonography i.e. the 24th week of pregnancy, we as a team counseled her about her pregnancy and the patient took four weeks to decide that she wants to terminate the pregnancy. So, for the abortion for 28 weeks of gestation, the patient was admitted in hospital and underwent induced premature delivery due to omphalocele. Tab Misoprostol 50 µg at posterior fornix 6 hourly after assessment x 3 doses (first dose preferably at 2 pm followed by second dose at 8 pm) was prescribed to her. Post-delivery examination of terminated fetus was male child having body weight of 960 gm with omphalocele and no other associated structural defects were found (Figure 2). Fetus was not sent for autopsy.



Figure 2. Induced premature delivered baby where abdominal organs are protruding outside the body.

DISCUSSION

Omphalocele is a midline defect that develops at the umbilical ring and usually contains the midgut and other organs of the abdomen such as liver, spleen and gonads.⁵ Gender prevalence is in line with literature because in a retrospective cohort study of 12 million births estimated that males had a higher risk of omphalocele.³ There are some known risk factors of omphalocele formation, such as in vitro fertilization, increased maternal age (> 33 years), concurrent anomalies in particular cardiac malformations.^{8,9} Demographic factors include maternal race (African American) and advanced maternal age. Hormonal high dose of vitamin E supplementation, abnormality in the manufacture of vitamin B12, smoking, alcohol con-

sumption and use of drugs such as aspirin and selective serotonin reuptake inhibitors (SSRIs) are involved with risk factors associated with omphalocele.⁶ The genetic risk factor is the most frequent one: firstly, chromosomal aberrations, secondly genetic syndromes and thirdly poly-malformative syndromes. In addition, several genes are related to omphalocele.² No such risk factors is present in our case and genetic testing was not done.

Classification of an omphalocele: The predominant site is epigastric, umbilical or hypogastric. Defect size, sac content and sac integrity should be determined in relation to the classification of an omphalocele. Small omphalocele- A defect 5 cm, with a sac comprising, up to a certain extent, a few loops of the intestines may exist. Giant omphalocele- A defect >5 cm, with a sac containing liver.⁵ But a more commonly used classification is that of Aitken, and the type I (small-sized defects, inferior to 4 cm) is distinguished from other types with good results by survival rate of 90 to 97 per cent, if isolated.² In our case omphalocele was epigastric type and Giant as the size was less than 5 cm but contained liver.

Ultrasound is the first, and probably most common, imaging technique for the prenatal diagnosis of omphaloceles. If the level of maternal alpha-fetoprotein (AFP) is raised during routine obstetrics blood screening, ultrasound investigation should be considered. In addition to obstetric ultrasound assessment, fetal magnetic resonance imaging (MRI) is becoming increasingly useful to better establish the extent of the defect. MRI has also been used to predict postnatal morbidity in neonates with giant omphaloceles by assessment of the lung. Omphalocele size indicates greater likelihood of karyotypic abnormality. In addition to other structural abnormalities, between 30%-70% of cases are associated with chromosomal abnormality or multi-malformation syndromes. The most common of these are inherited trisomy 13, 18, and 21, Beckwith-Wiedemann syndrome (BWS), and Pentalogy (Cantrell).⁶ The abnormalities in terms of prognosis can be classified as: lethal (trisomy 18, anencephaly); very poor (congenital diaphragmatic hernia; large encephalocele); or uncertain (suspicious intestinal atresia, congenital heart defect).¹⁰ In our case report since the first presentation was at late second trimester, ultrasound was confirmatory of the defect and further investigations were not consented.

If other associated anomalies are found there is quite strong association with poor prognosis (i.e. intrauterine fetal death and preoperative neonatal death). As such, after the defects are diagnosed they must be examined for other concurrent anomalies. Every malformation can also be to its anatomical system.^{2,5,8,11} Congenital malformations can be classified according to their affected

anatomical systems, with cardiovascular anomalies such as tetralogy of Fallot and atrial septal defects occurring in 7-47% of cases; central nervous system defects like anencephaly, holoprosencephaly, spina bifida, and primitive orbits in 4-30%; digestive system abnormalities such as Meckel's diverticulum and pyloric stenosis in 3-20%; genitourinary anomalies including micropenis, multicystic kidneys, and thoracic kidney in 6-20%; musculoskeletal defects like limb anomalies, exstrophy of the cloaca, and spinal/vertebral anomalies in 4-25%; and chromosomal abnormalities such as trisomy 13, 18, and 21 in 3-20%. No any other associated anomalies found in the fetus.

As regards management of pregnancy in patients with diagnosed omphalocele: caesarian section vs vaginal delivery has been controversial.⁴ Caesarian section is recommended in the setting of giant omphalocele to prevent abdominal dystocia and the risk of sac disruption or organ damage.⁵ An abdominal wall defect should be closed as soon as possible after delivery.⁴ Two surgical closure methods are proposed: primary (just after birth) for small-sized omphaloceles and delayed (later) for giant omphaloceles.²

Problems of the insanity which may arise from the omphalocele's re-emergence are airways impairment, wound infections, ileus, delayed introduction of oral feeding, gastro-esophageal reflux,⁴ Physical and intellectual development unaffected in children with omphalocele alone. Mor-

tality from omphalocele occurs at a rate more than that from gastroschisis.⁴ After explaining all the possible outcomes our patient and her family choose to abort the fetus.

Fetuses and neonates with isolated omphaloceles should usually have a good prognosis. Longitudinal data on growth and neurodevelopment in children with minor and giant omphalocele are scarce.¹⁰

Prenatal ultrasound might be able to discover most of the defects of the abdominal wall, thus allowing the family to be counseled and prepared for optimal post-natal care. For that reason, especially with large omphaloceles, early prenatal counseling and management of such defects by a team of neonatologists, pediatric surgeons and obstetricians in a tertiary referral center with neonatal intensive care unit and immediate availability of surgical services will reduce morbidity and mortality with this terminal congenital defect.

Conflict of Interest: None.

Consent: Written informed consent was obtained from the patient (or the patient's legal guardian) for the publication of this case report and any accompanying images. Patient anonymity maintained and identifying information has been omitted to the best possible extent to protect the patient's privacy.

REFERENCES

1. Logsdon NT, Gallo CM, Favorito LA, Sampaio FJ. Investigation of a connection between abdominal wall defects and severity of the herniation in fetuses with gastroschisis and omphalocele. *Sci Rep.* 2021 Dec 1;11(1). [[PubMed](#) | [Full Text](#) | [DOI](#)]
2. Poaty H, Pelluard F, Diallo MS, Ondima IPL, André G, Silou-Massamba JF. Omphalocele: a review of common genetic etiologies. Vol. 20, *Egyptian Journal of Medical Human Genetics*. Springer; 2019. [[PubMed](#) | [Full Text](#) | [DOI](#)]
3. Pereira MSV, Vieira DKR, Leite M de FMP, Villar MAM, Farias CVB. Omphalocele: clinical and epidemiological profile of patients born in a tertiary care center in Rio de Janeiro. *BMC Pregnancy Childbirth.* 2023 Dec 1;23(1). [[PubMed](#) | [Full Text](#) | [DOI](#)]
4. Henrich K, Huemmer HP, Reingruber B, Weber PG. Gastroschisis and omphalocele: Treatments and long-term outcomes. *Pediatr Surg Int.* 2008 Feb;24(2):167–
5. Verla MA, Style CC, Olutoye OO. Prenatal diagnosis and management of omphalocele. *Semin Pediatr Surg.* 2019 Apr 1;28(2):84–8. [[PubMed](#) | [Full Text](#) | [DOI](#)]
6. Malhotra R, Malhotra B, Ramteke H. Enhancing Omphalocele Care: Navigating Complications and Innovative Treatment Approaches. *Cureus.* 2023 Oct 25; [[PubMed](#) | [Full Text](#) | [DOI](#)]
7. Hwang PJ, Kousseff BG. Omphalocele and gastroschisis: An 18-year review study. Vol. 6, *Genetics in Medicine.* 2004. p. 232–6. [[PubMed](#) | [Full Text](#) | [DOI](#)]
8. Parata G, Vial Y, Addor MC, Pellegrinelli JM, Wildhaber BE. Anatomic parameters of omphaloceles and their association with anatomic, genetic, or syndromic malformations: a retrospective study. *PediatrSurgInt.* 2024 Dec 1;40(1). [[PubMed](#) | [Full Text](#) | [DOI](#)]
9. Nyberg DA, Fitzsimmons J, Laurence Mack MoA, Hughes M, Pretorius DH, Hickok D, et al. Chromosom-
73. [[PubMed](#) | [Full Text](#) | [DOI](#)]

- al Abnormalities in Fetuses with Omphalocele Significance of Omphalocele Contents. 1989. [[PubMed](#) | [Full Text](#) | [DOI](#)]
10. Hijkoop A, Peters NCJ, Lechner RL, Van Bever Y, Van Gils-Frijters APJM, Tibboel D, et al. Omphalocele: From diagnosis to growth and development at 2 years of age. *Arch Dis Child Fetal Neonatal Ed.* 2019 Jan 1;104(1):F18–23. [[PubMed](#) | [Full Text](#) | [DOI](#)]
 11. Heider AL, Strauss RA, Kuller JA. Omphalocele: Clinical outcomes in cases with normal karyotypes. *Am J Obstet Gynecol.* 2004;190(1):135–41. [[PubMed](#) | [Full Text](#) | [DOI](#)]
 12. REFERENCES
 13. Logsdon NT, Gallo CM, Favorito LA, Sampaio FJ. Investigation of a connection between abdominal wall defects and severity of the herniation in fetuses with gastroschisis and omphalocele. *Sci Rep.* 2021 Dec 1;11(1). [[PubMed](#) | [Full Text](#) | [DOI](#)]
 14. Poaty H, Pelluard F, Diallo MS, Ondima IPL, André G, Si-lou-Massamba JF. Omphalocele: a review of common genetic etiologies. Vol. 20, *Egyptian Journal of Medical Human Genetics*. Springer; 2019. [[PubMed](#) | [Full Text](#) | [DOI](#)]
 15. Pereira MSV, Vieira DKR, Leite M de FMP, Villar MAM, Farias CVB. Omphalocele: clinical and epidemiological profile of patients born in a tertiary care center in Rio de Janeiro. *BMC Pregnancy Childbirth.* 2023 Dec 1;23(1). [[PubMed](#) | [Full Text](#) | [DOI](#)]
 16. Henrich K, Huemmer HP, Reingruber B, Weber PG. Gastroschisis and omphalocele: Treatments and long-term outcomes. *Pediatr Surg Int.* 2008 Feb;24(2):167–73. [[PubMed](#) | [Full Text](#) | [DOI](#)]
 17. Verla MA, Style CC, Olutoye OO. Prenatal diagnosis and management of omphalocele. *Semin Pediatr Surg.* 2019 Apr 1;28(2):84–8. [[PubMed](#) | [Full Text](#) | [DOI](#)]
 18. Malhotra R, Malhotra B, Ramteke H. Enhancing Omphalocele Care: Navigating Complications and Innovative Treatment Approaches. *Cureus.* 2023 Oct 25; [[PubMed](#) | [Full Text](#) | [DOI](#)]
 19. Hwang PJ, Kousseff BG. Omphalocele and gastroschisis: An 18-year review study. Vol. 6, *Genetics in Medicine*. 2004. p. 232–6. [[PubMed](#) | [Full Text](#) | [DOI](#)]
 20. Parata G, Vial Y, Addor MC, Pellegrinelli JM, Wildhaber BE. Anatomic parameters of omphaloceles and their association with anatomic, genetic, or syndromic malformations: a retrospective study. *PediatrSurgInt.* 2024 Dec 1;40(1). [[PubMed](#) | [Full Text](#) | [DOI](#)]
 21. Nyberg DA, Fitzsimmons J, Laurence Mack MoA,

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