Case
Report

# Omphalocele in Both Twins: Revealing Developmental Complexities: A Case Report

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**ABSTRACT**

Omphalocele is a rare congenital anomaly with eviscerated abdominal organs in the proximal segment of the umbilical cord covered by a membranous sac. This case presents omphalocele in both fetuses in a twin pregnancy, discussing the embryological basis, the importance of early detection, and effective management. A 34-year-old primigravida was confirmed to have dichorionic diamniotic twins at 6 weeks of gestation. At 14 weeks, a routine ultrasound revealed omphalocele in both fetuses, complicated by early amniotic rupture, following an uncomplicated preconception period. Omphalocele is a rare occurrence, simultaneously in a twin pregnancy. Among the various theories, this presentation favors the theory based on malformation of the ventral body wall. The exact embryological explanation is yet to be identified.

**Keywords:** Case report; First-trimester ultrasound scan; Omphalocele in twin pregnancy

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## Introduction

Omphalocele is a congenital anomaly with eviscerated abdominal organs; bowel with or without liver in the proximal segment of the umbilical cord covered by a membranous sac (1). The earliest detection of omphalocele at 12 weeks by ultrasonography is a possible guide to determine the outcome of the pregnancy (2) A rare occurrence of Omphalocele involving both twins is presented in this case (3).

## Case presentation

A 34-year-old woman, who was in her first pregnancy, was undergoing routine obstetric follow-up and was diagnosed with a dichorionic diamniotic twin pregnancy (Figure 1). It was a planned pregnancy following intrauterine insemination after 6 years of primary subfertility. She had a non-consanguineous marriage with no family history of congenital anomalies. She was not exposed to chemicals, radiation, or infections during the periconceptional period or pregnancy.

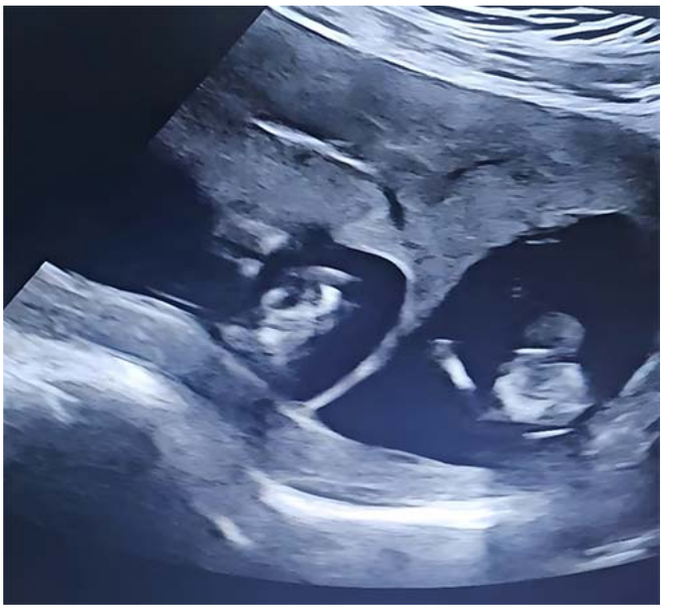

***Figure 1:*** *Dichorionic diamniotic twins at 14 weeks of gestation*

During a routine ultrasound examination at 14 weeks and 3 days of gestation, the dates were confirmed, and omphalocele was revealed in both twins. The omphalocele was associated with scoliosis. The first twin was detected to have an increased nuchal translucency, while the second twin had a normal nuchal translucency (Figures 2 and 3). Subsequently, after two days, an ultrasound scan revealed the absence of heart sounds in the first fetus. The mother was informed that the other fetus was likely to develop severe disabilities if it sur-

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vived until birth, and she opted for a termination of pregnancy. The chromosomal analysis by karyotyping was suggested for further evaluation, but she declined.

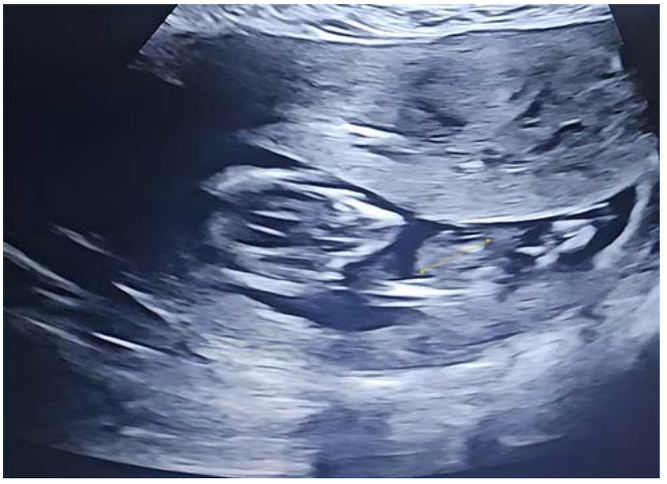

***Figure 2:*** *An ultrasonography of the first twin with omphalocele at 14 weeks of gestation*

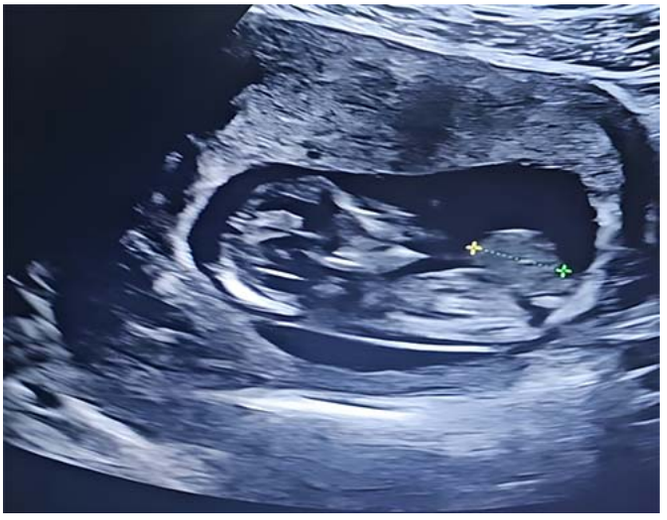

***Figure 3:*** *Ultrasonography of the second twin*

## Discussion

Twin pregnancies have a 1.5 to 3 times higher risk of congenital anomalies than singleton pregnancies, mostly in monochorionic twins with discordant phenotypes (1). Omphalocele is a rare condition but one of the most common congenital abdominal wall defects detected in 1-3.8/10,000 pregnancies (3). Omphalocele is commonly associated with atypical karyotypes, which determine the mortality as isolated omphalocele has a higher (90%) chance of survival (1). Omphaloceles involving both twins are rarely reported in the existing literature and the exact prevalence is not yet identified. The literature survey revealed one case reported in 1968 (4). Here, we present a rare case of dichorionic diamniotic twins with omphalocele detected by prenatal ultrasound scan at 14 weeks.

Anterior abdominal wall defects can be classified into three types: omphalocele, gastroschisis, and body stalk abnormalities (5). Omphalocele is a congenital anomaly with encapsulated, eviscerated abdominal organs; bowel with or without liver in the proximal segment of the umbilical cord (1,6).

Various theories have been proposed in the literature, mainly based on clinical observation. The exact cause of omphalocele has not yet been identified, which is most likely to occur as an interaction of genetic and environmental factors based on the degree of involvement and timing during fetal development (2). The combination of embryonic dysplasia theory and malfunction of the ectodermal placodes is widely accepted. Embryonic dysplasia theory, proposed by Streeter and later by Herva, describes defects in the germinal disc during early development that manifest later as abdominal wall defects. Ectodermal placodes serve as a transition zone for meso-ectodermal cells, which form the ventral body wall. Abnormal deposition of mesodermal cells can result in an enlarged umbilical ring, which allows omphalocele to occur due to the underdevelopment of the ventral body wall, disrupting the body folding process as proposed by Hartwig et al. and Russo et al (7).

Other theories include the failure of lateral mesodermal body folds to migrate centrally, incomplete midgut rotation, and the persistence of the body stalk beyond 12 weeks of gestation (2,8). Physiological rotation of the midgut occurs at the 8th week of gestation, with the herniation of the fetal midgut at the base of the umbilical cord due to relatively rapid growth. The bowel undergoes a 270-degree anticlockwise rotation in two stages around the axis of the superior mesenteric artery and returns to the abdominal cavity by 12 weeks (2). Incomplete midgut rotation failing to return to the abdominal cavity is one of the theories identified as the basis for omphalocele (6). However, the simultaneous presentation in both twins favors the theory based on malformation of the ventral body wall, though the exact embryological explanation is yet to be identified (7).

Maternal and fetal risk factors have been identified as being associated with omphalocele. Extremes of maternal age and obesity are some of the maternal risk factors. Neonatal risk factors include genetic variations and chromosomal abnormalities (1). Chromosomal defects; trisomy 13 (Patau syndrome), trisomy 18 (Edward syndrome), and trisomy 21 can be associated with omphalocele (2,5). Abnormalities involving other organs can coexist with omphalocele, including congenital heart, genitourinary, musculoskeletal, gastrointestinal, and neural tube defects (6).

An omphalocele can be detected earliest at 12 weeks of gestation by ultrasonography. A fetal anterior midline mass consisting of abdominal contents at the base of the umbilical cord can be detected sonographically as an omphalocele, which can be differentiated from physiological midgut herni-

ation (2). This differentiation is based on the confirmation of gestational age, as physiological herniation rarely persists after 12 weeks of gestation, and based on the size, echogenicity, and associated other abnormalities. Omphalocele can be associated with other sonographically identifiable abnormalities such as increased nuchal translucency and intrauterine growth restriction. Early detection of anatomical abnormalities is important for timely interventions, early recommendations for chromosomal screening analysis, and informed decision-making for expectant parents (6).

*Declarations*

*Conflict of interest: There are no conflicts of interest.*

*Consent for publication: Informed written consent was obtained from the patient to publish the case and photographs.*

*Competing interests: The authors declare that they have no competing interests.*

*Authors' contributions: SG contributed to the patient management and drafting of the manuscript. AK was involved in manuscript writing and editing*

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