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Case Report

Prenatal Diagnosis Of Omphalocele And Soft Markers Of Chromosomal Abnormalities In The First Trimester

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A B S T R A C T

Physiological herniation of the fetal midgut into the umbilical cord occurs during early embryonic development as a result of limited intra-abdominal space and is considered a normal finding until approximately 10-11 weeks of gestation. Persistence of midgut herniation beyond 12 weeks of gestation, or the presence of a herniated mass with a diameter exceeding 7 mm, is regarded as pathological and consistent with a diagnosis of omphalocele. Early identification during the first trimester is of substantial clinical importance, as omphalocele is frequently associated with chromosomal abnormalities, particularly trisomy 18, trisomy 13, and trisomy 21. In this setting, healthcare providers especially midwives working in close collaboration with fetomaternal specialists play a pivotal role in early screening, timely referral, and provision of psychosocial support.

We report the case of a 22-year-old woman, gravida 2 para 1 abortus 0 living 1 (G2P1A0H1), at 11 weeks of gestation, who presented with hyperemesis gravidarum complicated by mild to moderate dehydration and reduced oral intake. Fetomaternal ultrasonography demonstrated a single live intrauterine fetus consistent with gestational age, with a crown– rump length of 39.4 mm (15.3rd percentile) and a regular fetal heart rate of 164 beats per minute. Notable findings included a markedly increased nuchal translucency of 6.8 mm, visualization of the nasal bone, normal ductus venosus Doppler flow, and a 9-mm membranous herniation at the umbilical cord insertion, consistent with omphalocele.

In conclusion, first-trimester ultrasonography enabled early prenatal diagnosis of omphalocele accompanied by chromosomal soft markers. Further management includes cytogenetic analysis and a detailed second-trimester anatomical assessment, supported by comprehensive counseling to facilitate informed clinical decision-making.

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I. INTRODUCTION

Congenital anomalies remain one of the leading causes of neonatal morbidity and mortality worldwide, with abdominal wall defects representing one of the most frequently encountered congenital abnormalities in fetomaternal clinical practice. Among these defects, omphalocele is one of the most common, characterized by a midline defect of the anterior abdominal wall at the base of the umbilical cord, resulting in herniation of intra-abdominal organs through an enlarged umbilical ring into a peritoneal sac. (Al Namat et al., 2025)(Cunningham et al., 2018)

Omphalocele is an embryological developmental disorder caused by the failure of the midgut and other abdominal organs to return to the abdominal cavity following physiological herniation, a process that is normally completed by the end of the first trimester. Population-based studies have reported a prevalence of approximately 3.38 per 10,000 pregnancies, with variation across different national birth registries. (Al Namat *et al.*, 2025)

Prenatal diagnosis during the first trimester is of critical importance, as omphalocele is frequently associated with additional anomalies, particularly chromosomal abnormalities and other structural defects. Contemporary clinical data indicate that more than 30 % of omphalocele cases are associated with chromosomal abnormalities, most commonly trisomy 18, trisomy 13, and other genetic disorders such as Beckwith Wiedemann syndrome .(Que *et al.*, 2023)

Furthermore, omphalocele is often accompanied by other structural abnormalities, including congenital heart defects and central nervous system anomalies. Advances in prenatal ultrasonography and first-trimester screening protocols such as the measurement of nuchal translucency (NT) have significantly enhanced early detection of structural abnormalities, including omphalocele and chromosomal soft markers. This screening is typically performed between 11 and 13 weeks + 6 days of gestation, and increased NT thickness has been strongly associated with an elevated risk of chromosomal abnormalities (Căpitănescu et al., 2025).

Nevertheless, establishing a definitive diagnosis of omphalocele in the first trimester remains challenging, as physiological midgut herniation may still be observed before 11 weeks of gestation and can mimic pathological omphalocele. Accurate early diagnosis therefore requires comprehensive evaluation by trained healthcare professionals and, when indicated, confirmation using advanced diagnostic modalities (Que et al., 2023; Căpitănescu et al., 2025). In addition to diagnostic challenges, maternal comorbidities such as hyperemesis gravidarum (HEG) further complicate clinical management, necessitating a coordinated interdisciplinary approach involving midwives and fetomaternal specialists to ensure maternal stability, diagnostic accuracy, and appropriate family counseling. (Que *et al.*, 2023; Căpitănescu *et al.*, 2025)

This case report aims to document the prenatal diagnostic process of a fetus with omphalocele accompanied by chromosomal soft markers in the first trimester. Furthermore, it highlights the crucial role of midwives in early detection, facilitating collaborative referral pathways, and providing psychosocial support to families facing high-risk pregnancies. The findings of this case report are expected to contribute scientifically to midwifery practice by enhancing the recognition of early signs of congenital anomalies through basic ultrasonographic examination and reinforcing the importance of accurate interpretation of chromosomal soft markers. For primary healthcare facilities, this report may serve as a reference in developing structured referral pathways for early-detected fetal anomalies.

II. CASE REPORT

A 22-year-old woman, gravida 2 para 1 abortus 0 living 1 (G2P1A0H1), presented at 11 weeks of gestation with complaints of severe nausea and vomiting. The patient was diagnosed with hyperemesis gravidarum (HEG) accompanied by mild to moderate dehydration, low oral intake, and suspected fetal abnormalities, including suspected omphalocele with chromosomal soft markers. Fetomaternal ultrasonography revealed a single live intrauterine pregnancy consistent

with 11 weeks of gestation. The fetus was viable with good spontaneous movements. The crown–rump length (CRL) measured 39.4 mm (15.3rd percentile), and the fetal heart rate (FHR) was 164 beats per minute with a regular rhythm. The nuchal translucency (NT) thickness was markedly increased at 6.8 mm. The nasal bone was visualized with a length of 0.3 mm. Doppler assessment of the ductus venosus demonstrated a normal A-wave with a pulsatility index (PI) of 1.3. Amniotic fluid volume was adequate, with a maximum vertical pocket (MVP) of 3.4 cm. Evaluation of craniofacial structures showed normal appearances of the diencephalon, maxilla, and mandible. A protruding mass measuring approximately 9 mm was identified outside the fetal abdominal wall at the base of the umbilical cord, enclosed by a thin membrane, consistent with the sonographic features of omphalocele. The umbilical cord consisted of two arteries and one vein (2a1v), with no evidence of nuchal cord entanglement. Doppler examination of both uterine arteries revealed no notching, with a pulsatility index of 1.6.

Based on these findings, the fetomaternal ultrasound impression was a single live intrauterine pregnancy at 11 weeks of gestation complicated by omphalocele and the presence of chromosomal soft markers.

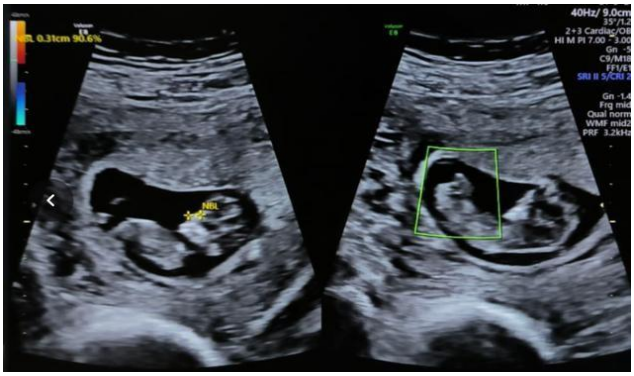


Figure 1. Visualization of the nasal bone with measurements within normal limits.



Figure 2. A. Protruding herniated mass arising from the abdominal wall at the umbilical cord insertion site measuring >7 mm.

B. Three-dimensional (4D) ultrasound image demonstrating omphalocele.



Figure 3. Increased nuchal translucency thickness (6.8 mm), serving as a chromosomal soft marker.

In this case, prenatal diagnostic evaluation using ultrasonography identified a congenital anomaly consistent with omphalocele accompanied by chromosomal soft markers in the first trimester. Further management includes genetic diagnostic testing, such as chromosomal analysis via amniocentesis, and a detailed fetal anatomical survey performed by a fetomaternal medicine specialist at 20–24 weeks of gestation to detect additional structural anomalies. The patient is currently under observation with supportive management for maternal condition. Given the high risk of chromosomal abnormalities, comprehensive counseling and informed consent were provided to the patient and her family to facilitate psychological preparedness in anticipation of potentially adverse pregnancy outcomes.

III. DISCUSSION

First-Trimester Diagnosis of Omphalocele and Its Relationship with Crown-Rump Length The diagnosis of omphalocele in the first trimester remains challenging due to the presence of physiological midgut herniation, which normally persists until approximately 10-12 weeks of gestation. When herniation is still observed beyond this developmental period and is accompanied by additional structural abnormalities, the likelihood of pathological omphalocele increases. In a retrospective study involving 81 fetuses with omphalocele, approximately 71.6% exhibited additional ultrasonographic abnormalities, and chromosomal abnormalities were identified in nearly 30% of cases, predominantly trisomy 18 and trisomy 13, highlighting the strong association between omphalocele and genetic abnormalities. (Căpitănescu *et al.*, 2025)

Clinical Significance of Increased Nuchal Translucency and the Risk of Chromosomal Abnormalities

Increased nuchal translucency (NT) is a well-established marker in first-trimester chromosomal screening. In a study analyzing cases with NT ≥ 6.5 mm, approximately 70.4% of fetuses were found to have abnormal karyotypes, including Turner syndrome, trisomy 18, trisomy 21, and trisomy 13. Structural defects, such as omphalocele, were also frequently observed in this group with markedly increased NT measurements. (Zhou *et al.*, 2023)

Furthermore, increased NT has been consistently associated with other major structural abnormalities, including congenital heart defects and hydrops fetalis, which often coexist with

chromosomal abnormalities. Fetuses with increased NT therefore carry a substantially elevated risk of both genetic and structural anomalies, underscoring the importance of comprehensive follow-up evaluation when NT exceeds 3.5 mm. (Shi *et al.*, 2021)

Association Between Omphalocele and Chromosomal Abnormalities

The literature consistently reports that omphalocele is frequently a manifestation of underlying genetic abnormalities. Prenatal studies have demonstrated that approximately 29.6% of fetuses with omphalocele present with chromosomal abnormalities, most commonly trisomy 18 and trisomy 13. These findings further indicate that non-isolated omphalocele, when accompanied by additional anomalies, is significantly more likely to be associated with aneuploidy compared to isolated omphalocele. (Shi *et al.*, 2021)

The Role of Midwives and Interdisciplinary Collaboration

Cases such as this require a multidisciplinary management approach, particularly in the presence of maternal conditions such as hyperemesis gravidarum (HEG). Core clinical responsibilities of midwives include maternal stabilization and the provision of psychosocial support. Midwives also play a pivotal role in delivering appropriate counseling regarding the potential for adverse pregnancy outcomes and in facilitating the informed consent process for invasive diagnostic procedures such as amniocentesis. Early detection of omphalocele and associated soft markers, such as increased NT, enables timely and comprehensive prenatal planning, including referral to genetic specialists and fetomaternal medicine consultants for advanced genetic evaluation. This approach is particularly important because the combination of structural anomalies and genetic markers (e.g., omphalocele with increased NT) significantly elevates the risk of chromosomal abnormalities prior to the second trimester. Advanced genetic analyses, including karyotyping, chromosomal microarray analysis (CMA), and whole-exome sequencing (WES), may serve as valuable tools in prenatal counseling and individualized pregnancy management. (Shi *et al.*, 2021; Al Namat *et al.*, 2025)

IV. CONCLUSION

The prenatal detection of omphalocele accompanied by markedly increased nuchal translucency (NT) measuring 6.8 mm in the first trimester indicates a serious and potentially adverse prognosis. The coexistence of a major structural anomaly and a significant chromosomal soft marker substantially increases the likelihood of underlying genetic abnormalities and additional fetal structural defects. Based on these findings, the primary clinical recommendation is to proceed with cytogenetic evaluation, including fetal karyotyping and, where available, advanced genetic testing such as chromosomal microarray analysis. In addition, a comprehensive second-trimester detailed fetal anatomical survey performed between 20 and 24 weeks of gestation is strongly advised to assess for associated anomalies, particularly involving the cardiovascular and central nervous systems. Beyond diagnostic and technical considerations, ongoing emotional and psychosocial support remains a critical component of care. Midwives play a central role in providing continuous counseling, facilitating informed and shared clinical decision-making, and supporting the family throughout the diagnostic process. This holistic, interdisciplinary approach is essential to ensure both optimal clinical management and compassionate care for families facing high-risk pregnancies.

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BIOGRAPHY

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