

## CASE REPORT

### Omphalocele: a multidisciplinary approach is of paramount importance

**Short title:** *Omphalocele management*

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## ABSTRACT

**Background.** Omphalocele is one of the most common congenital abdominal wall defects, characterized by the absence of abdominal muscles, fascia, and skin and requiring urgent surgical intervention. The characteristic ultrasound appearance includes a midline defect with herniation of the abdominal contents at the base of the umbilical cord after the 12<sup>th</sup> week of gestation.

**Case presentation.** In the reported case, the omphalocele was diagnosed at 17<sup>th</sup> week of gestation. The karyotype showed a 46XX karyotype and after careful counseling, the woman decided to continue the pregnancy. The pregnancy was carried to term at 39<sup>th</sup> week of pregnancy by elective cesarean section.

The treatment to which the newborn was submitted was in two stages: the first involved suspension of the hernia sac with mercurochrome solution; the second involved an operative approach with reduction of the hernia sac. The child was followed until the age of two years.

**Conclusions.** The multidisciplinary approach in the management of omphalocele and conversation with parents from detection of a malformation through the potential of associated anomalies to the operative and non-operative procedures available to solve the condition are of utmost importance in such cases.

**Key words:** omphalocele; prenatal ultrasound diagnostics; abdominal wall defect; congenital malformation; neonates.

## INTRODUCTION

Omphalocele, is a rare but severe congenital malformation of the abdominal wall that affects 1 per 4000 to 10000 live births, or even 1 per 3000 to 4000 if taking abortions and stillbirths in account [1].

During the 6<sup>th</sup> week of fetal growth, the abdominal cavity becomes temporarily too small to accommodate all the pertaining organs, resulting in abdominal organ protrusion into the residual coelom at the base of the umbilical cord. This temporary herniation is physiologic and is detected by ultrasound (US) from the 9<sup>th</sup> to 11<sup>th</sup> gestational week following last menstruation (by the crown-rump length of 45 mm). Hernia reduction occurs by 12<sup>th</sup> week of last menstruation, i.e. herniation present after 12<sup>th</sup> week is not physiologic anymore [2]. Simple central omphalocele occurs when the extraembryonic part of the intestine fails to return to the abdominal cavity, rotating 270° degrees counterclockwise back to the abdomen. Thus, omphalocele is considered a rotational anomaly [3].

Moreover, other associated anomalies occur in 30% to 70%, and chromosomal abnormalities in 10% to 30% of cases. That is why a finding of omphalocele is an absolute indication for fetus karyotyping [4].

The rate of perinatal mortality is almost 30%. Associated anomalies and abnormal karyotype predict increased mortality. With improvement in neonatal care and surgical techniques, the prognosis is good in live births without chromosomal abnormalities or severe associated anomalies.

To date, there are scarce data on the mode of delivery in these cases, and postnatal outcomes also depend on the type of surgical management and closure.

## CASE PRESENTATION

A 31-year-old pregnant woman in the 17<sup>th</sup> gestational week was referred by her primary care gynecologist to the Department of Gynecology and Obstetrics, Split University Hospital Center, for suspected fetal malformation.

The patient was in her second pregnancy, with a first pregnancy terminated spontaneously at 12<sup>th</sup> week of gestation, after which she underwent dilatation and curettage at the department in 2016.

Neither the woman nor the father had any serious chronic diseases. On initial examination at the Department, US fetal measurements corresponded consistently to the 17<sup>th</sup> gestational week, fetal heart beats and fetal movements were positive, and the amount of amniotic fluid was normal. US study at the level of umbilical cord insertion showed a hyperechoic mass, 15 mm in diameter, and two minor cord cysts, indicating the diagnosis of omphalocele. US study of the neurocranium revealed a cyst of the choroid plexus (Figure 1). No data are available on first trimester screening, as the patient did not have it by her own choice. Considering the ultrasound diagnosis of omphalocele and the possible correlation with other congenital malformations and abnormal karyotypes, fetal karyotyping by amniocentesis was indicated.

Upon obtaining normal female karyogram (46XX), the options were explained to the patient; these included the pregnancy termination and surgeon pediatric consultation as well as a neonatologist one. After receiving the necessary information, the woman decided to continue the pregnancy. The patient was regularly followed-up at the Department and admitted to the hospital in the 37<sup>th</sup> week of gestation for delivery. Because of the US assessed macrosomic fetus and omphalocele, a decision was made to terminate pregnancy operatively in the 38<sup>+1</sup> gestational weeks.

Cesarean delivery was performed without complications, and a live baby girl was born with a birth weight of 4700 g and birth length of 50 cm. On neonatal examination, the baby girl scored an Apgar 1st minute of 9; objective examination showed normal vital functions, a 7-cm diameter omphalocele with two edematous cord cysts, and the presence of lower extremity edema (Figure 2).

The baby was admitted to the Neonatology Unit of the University Hospital Center in Split, and after consultation with the pediatric surgery team, a planned two-stage treatment was proposed to the parents by mutual agreement. In our case, in consultation with a neonatologist and pediatric surgeon, a decision was made on delayed closure due to the defect size, as well as good team experience with this method of treatment. In the first stage of treatment the infant was placed in a warm cradle, and the umbilical cord was clamped as close to the omphalocele as possible, smeared and disinfected with a mercurochrome solution, and placed in suspension (Figure 3). This antiseptic procedure was performed for several days until the amniotic tissue became necrotized and covered with a thin scaly crust that took on the role of protecting the contents of the sac from infection and perforation. Contextually, regression of lower extremity edema occurred.

When the viscera contained in the herniary sac returned to the child's abdomen and the omphalocele decreased in volume (Figure 4), the pediatric surgeon performed surgical repair as the second stage of treatment. The surgery was completed without complications (Figures 5 and 6). The child was discharged home on the 10<sup>th</sup> day after surgery. The total length of hospital stay was 30 days. The child is in her second year of follow-up; to date, normal neuro-motor development, regular growth, and the absence of other congenital malformations have been found.

## DISCUSSION

Omphalocele is the most common congenital defect of the abdominal wall, caused by its incomplete closure. It is associated with other anomalies in 30% to 70%, and chromosomal abnormalities in 10% to 30% of cases [1]. Some authors report the incidence of associated anomalies with omphalocele of 50%-70%, and even 80%-90% in some studies. These anomalies are not exclusively related to the gastrointestinal system but may involve heart (up to 40%), neural tube, mouth and palate, or cloaca [5–10]. Some studies suggest that minor defects (less than 4 cm) are associated with gastrointestinal deficiency, whereas major defects are usually associated with cardiac irregularities. Chromosomal abnormalities, in particular aneuploidies such as trisomy 18, 21 or 13, are present in 40%-60% of fetuses with omphaloceles not containing liver. The possible presence of associated trisomy (e.g., trisomy 13, 18, 21) can be detected by first trimester nuchal ultrasound [11]. First trimester screening also allows calculation of the risk of developing pregnancy-related hypertensive disorders, such as preeclampsia [12].

As already stated, the rate of perinatal mortality is almost 30% and associated anomalies and abnormal karyotype predict increased mortality. Intracorporeal liver and multiple anomalies are associated with abnormal karyotype [13]. With improvement in neonatal care and surgical techniques, the prognosis is good in live births without chromosomal abnormalities or severe associated anomalies, as in our case [1].

The rate of women opting for abortion is relatively high (omphalocele 37% and gastroschisis 29%) in spite of having been informed on the generally favorable outcome following operative correction of such anomalies [5].

Respiratory and feeding problems are the most common neonatal and later medical issues. Prior to childbirth, the parents need to receive consultation about the possibilities and multiple operations, as well as long-term postnatal hospitalization of their child [14].

Ultrasound can be useful in predicting aneuploidy cases with omphalocele. Like other previous studies, Zork et al. [14] showed the presence of additional anomalies to increase the probability of abnormal karyotype. They point to the fact that increase in maternal age, bone and central nervous system anomalies along with another two associated malformations increase the probability of abnormal karyotype in a fetus with omphalocele. On the other hand, isolated omphalocele need not imply a normal karyotype. These data can be used in consultation of women, pointing to the role of invasive testing because aneuploidies and/or multiple anomalies are associated with poor neonatal outcomes [15]. The newborns with omphalocele are likely to have other anomalies, pulmonary hypertension, and higher mortality as compared with the newborns with gastroschisis, as reported by Corey et al. [16].

According to Kominiarek et al. [17], it is not surprising that a twofold greater number of neonates were diagnosed with additional anomalies or syndromes in the non-isolated group, while one-third of the presumed isolated cases of omphalocele on US still had additional diagnoses confirmed postnatally. Similarly, all prenatally recorded anomalies (cardiac septal defects in particular) were not confirmed postnatally in the non-isolated group, suggesting that not all abnormalities can be diagnosed on US examination.

Data on the mode of delivery in these cases are not enough. Available data indicate that the Caesarean section does not ensure the positive outcome of delivery, contrary it may increase maternal morbidity [4]. Although most literature reports shows that Caesarean section should not be preferred, it remains the predominant delivery method accounting for 80% of cases, of which 50% with omphalocele as the indication. This may be due to poor coordination with

neonatologists and postnatal care providers. However, there is no definitive evidence for cesarean section as a method of pregnancy termination to be favorable for fetuses with omphalocele [17].

According to Chen et al. [18], cesarean section is recommendable in cases where omphalocele is associated with a large umbilical cord cyst and normal karyotype, in order to avoid the risk of intrauterine vascular compression of blood flow during vaginal delivery. In our case, pregnancy was terminated by cesarean section, primarily due to macrosomia; otherwise, most physicians decide on cesarean section as the mode of pregnancy termination, even when only omphalocele is present.

In isolated cases, postnatal outcome depends on the type of surgical management and closure (primary or delayed). Primary closure is defined as healing right after birth. Delayed closure is defined as late or even post-neonatal healing after having allowed the omphalocele to desiccate, contract and epithelialize, with closure of ventral hernia at a later stage. Decision on the mode of surgical closure is made according to the size of the defect, liver evisceration, intra-abdominal pressure, time of mechanical ventilation, inspiratory oxygen fraction, and clinical presentation of pulmonary hypoplasia. Delayed closure is associated with increased comorbidity and prolonged hospital stay [19]. In our case, in consultation with a neonatologist and pediatric surgeon, a decision was made on delayed closure due to the defect size, as well as good team experience with this method of treatment. Although prolonging the treatment, we considered this approach to be less aggressive. Inconsistent outcomes have been reported for liver location, neonatal morbidity and mortality [17].

## **CONCLUSIONS**

Although gynecologists can confirm the diagnosis and determine the prognosis during the neonatal period, they are only familiar with the prenatal US characteristics used on consultation of pregnant women about the outcome of a fetus with omphalocele.

When consulting women about fetal omphalocele, emphasis should be put on US limitations in pregnancy, importance of US follow-up, role of liver location in predicting neonatal outcome, and role of neonatal evaluation such as fetal karyotyping following amniocentesis. The women should be referred to consultation with a neonatologist and pediatric surgeon about neonatal care, and management of omphalocele and possible associated anomalies. A team and multidisciplinary approach to both medical care of a newborn with omphalocele and previous parental consultation is of utmost importance.

## **COMPLIANCE WITH ETHICAL STANDARDS**

### ***Authors contribution***

AD and JM were responsible for the acquisition, analysis, and interpretation of the data. AE and AD were responsible for drafting the work. VC was responsible for revising the work critically for important intellectual content. MM, IK, KPM and GR gave final approval of the version to be published. AD and JM agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All authors meet the ICMJE criteria for authorship, have read and agreed to the current version of the manuscript.

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### **Study registration**

N/A.

### **Disclosure of interests**

The authors have no conflicts of interest.

### **Ethical approval**

Obtained

### **Informed consent**

The patient signed informed consent to allow publication of the case.

### **Data sharing**

Data are available under reasonable request to the corresponding author.

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**Figure 1.** Ultrasound images confirming the diagnosis of omphalocele.





**Figure 2.** Postnatal condition.



**Figure 3.** Treatment phase 1 (Suspension of the herniary sac).

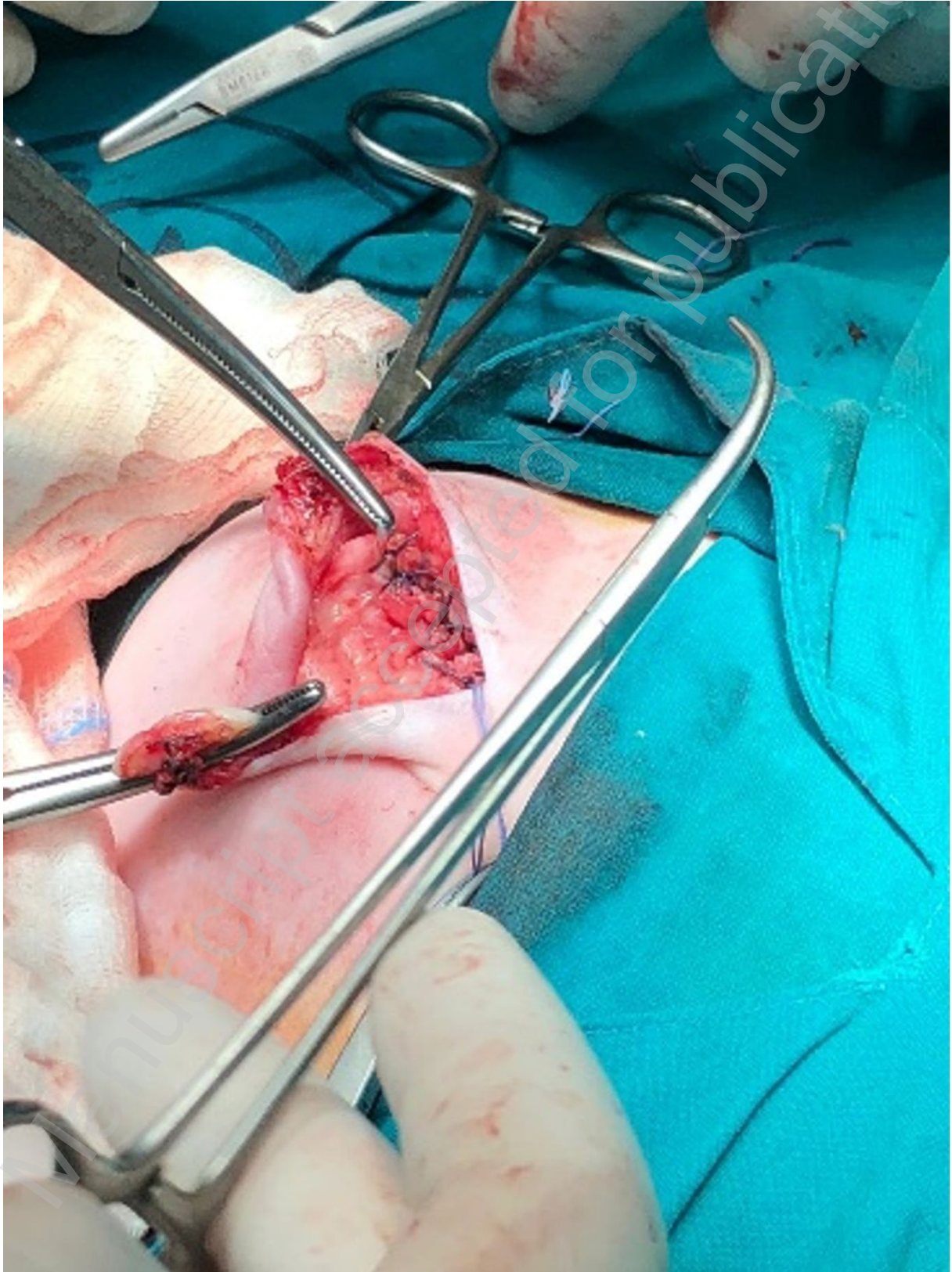


**Figure 4.** Treatment phase 1 (dried herniaria sac).





**Figure 5.** Treatment phase 2 – operative approach.





**Figure 6.** Treatment phase 2 completed.

