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Diagnosis and management of fetal omphalocele: a case report

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Summary

Omphalocele is the commonest fetal abdominal wall defect and can represent a part of a syndrome or an isolated defect. The eviscerated abdominal organs are covered by the peritoneum and amnios. Obstetric prenatal ultrasound should be integrated by fetal echocadiography because of high incidence of associated cardiac anomalies. The authors describe the diagnosis and management of a 41 years primigravida woman with first trimester ultrasound diagnosis of omphalocele.

Key words: Omphalocele; Gastroschisis; Prenatal diagnosis.

Introduction

Omphalocele is a congenital anomaly of abdominal wall closure due to failure of infolding of the body wall. The eviscerated abdominal organs are covered by the parietal peritoneum and the amniotic membrane. The incidence of the disease is about 1:5000 live births [1]. Omphalocele might contain the small bowel, colon, stomach and liver. It can present as a part of a syndrome or as an isolated defect. The omphalocele in 10% to 40% of cases is associated with polimalformative syndromes (such as Pentalogy of Cantrell: omphalocele, defects of the sternum, anterior diaphragm, and diaphragmatic pericardium with intracardiac lesions) or occurs in fetuses with chromosomal anomalies (Trisomy 13, 18) and/or genetic (Beckwith-Wiedemann syndrome: gigantism, macroglossia, omphalocele) [2-4]. They are frequently associated with omphalocele defects of: central nervous system, skeletal abnormalities, and heart disease [5]. Ultrasound examination is therefore mandatory for other morphological anomalies if there is an ultrasound diagnosis of omphalocele.

The sonographic diagnosis is based on the demonstration of abdominal viscera contained by a sac into the basis of umbilical cord. Herniated organs are covered by a membrane, formed by the peritoneum and amnios [6-10]. Gastroschisis, on the contrary, is an abdominal wall defect (as well as omphalocele), but at ultrasound the absence of a sac covering the prolapsed organs is typical (mostly to the right of the umbilicus).

Both in the presence of polyhydramnios and oligohydramnios fetal prognosis worsens significantly. Fetal karyotype is recommended in case of a fetus with an ultrasound diagnosis of omphalocele [9].

Case Report

The authors report the case of a patient at 16 weeks of gestation that was diagnosed with fetus suffering from omphalocele. The patient was admitted at the Department of Gynaecology and Obstetrics of the University of Sassari in September 2015. The patient was a 41-year-old woman, healthy, nulliparous with a medical and surgical history negative for pathologies. The patient denied use of alcohol during pregnancy and the use of drugs. She has not been exposed to environmental pollutants and had spontaneous conception. Cases of malformations and chromosomal disorders were not present within her family.

The first trimester combined test showed the presence of omphalocele in which it appeared clear and the presence of liver in the hernial sac excluded intestinal parts. The stomach was found to be visible subdiaphragmatic and at intra-abdominal level. The amniotic fluid was normal. Nuchal translucency corresponded to 1.0 mm associated with the presence of nasal bone. The patient had a trisomy 21 adjusted risk of 1:313, trisomy 18 1:1, and 1.447 of trisomy 13. Determination of alpha-fetoprotein was on the upper limit of normal. She refused to perform fetal cariotype.

At the 16 week of gestation, ultrasound examination confirmed the diagnosis done during the first trimester combined test. There were no major abnormalities compatible. The cerebellum, the orbits, as well as the cisterna magna were found to be regular. The ends were all present and well represented. Fetal echocardiography showed no macroscopic abnormalities. The umbilical cord was located on the apex of the hernia. The measure was 33×31×25 mm. The presence of liver parenchyma and part of bowel loops was suspected (Figure 1). The presence of liver in the hernia sac was highlighted with the intrahepatic vessels research made visible by Doppler ultrasound.

The patient decided to terminate the pregnancy and expelled the fetus the next day hospitalization after a cycle of prostaglandins, one vaginal ovule every three hours within 24 hours.

Anatomical dissection the fetus presented a voluminous umbilical hernia (30×40×30 mm diameters) associated with a non-completion of the abdominal wall (length 23 mm), at the

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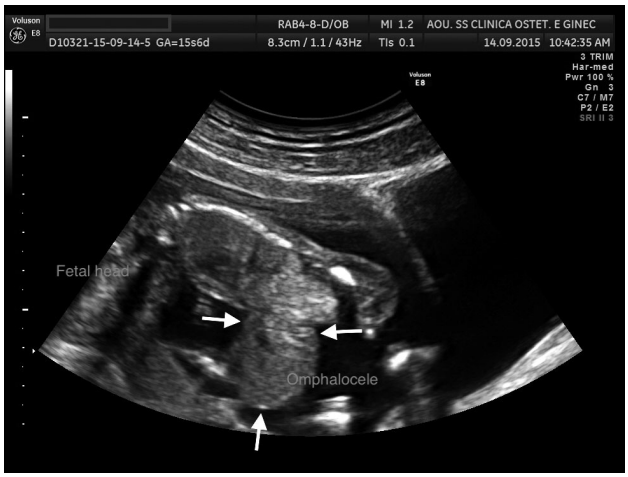

Figure 1. — Ultrasound showing omphalocele (within white arrows)

supraumbilical portion of the linea alba (Figure 2). The hernia completely contained the liver, chest, skull, organs of the abdomen, and pelvis did not show macroscopic pathologies. The weight was 130 grams with a female phenotype. The karyotype was found to be normal.

Discussion

Diagnosis of omphalocele is performed by ultrasound investigation after the 12th week of gestation, or earlier, but up to 12 weeks of gestation the greater part of the developing bowel is physiologically located outside the abdominal cavity (physiological omphalocele). The reposition of the bowel into the abdominal cavity should be terminated by the 12th week of gestation. Mandatory is to program fetal karyotype analysis to determine the possible combination of genetic and/or chromosomal abnormalities involving the fetus. Determinant is the differential diagnosis with gastroschisis (absence of a sac covering the prolapsed organs). Following the genetic assessment is good to evaluate the extent of abdominal wall defect and check which abdominal viscera are herniated outside the wall. Obstetric prenatal ultrasound should be integrated by fetal echocardiography because of high incidence of associated cardiac anomalies [10].

The collaboration with a multidisciplinary team of obstetricians, neonatologists, and pediatric surgeons will make a more accurate assessment methods of childbirth and the life expectancy of the fetus.

It is crucial to carry out the delivery in a Center equipped with neonatal resuscitation service where you can also schedule a neonatal and obstetric care appropriate for the degree of malformation. Also to be considered is the possibility that the baby can be affected by other abnormalities not detected prenatally (such as chromosomal and genetic abnormalities).

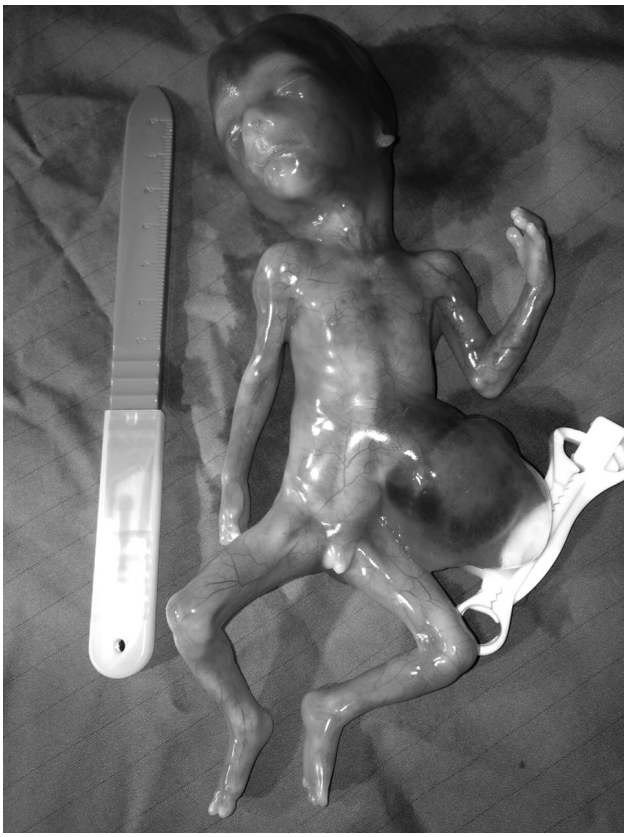

Figure 2. — Fetus after voluntary abortion showing omphalocele.

Programming the mode of delivery is influenced by the size of the hernia sac and the organs contained within it. High volume and/or liver inside direct you to a birth through elective caesarean section.

Prenatal diagnosis (3rd trimester) does not affect the neonatal prognosis and treatment options if the birth takes place in third level Centres [11]. An important negative prognostic factor is represented by prenatal diagnosis early (before 25th week) as associated with the extent of abdominal wall defect and with the severity of the condition. The presence of ascites in liver hernial sac argues for a poor prognosis because of liver pathology. There are prenatal parameters that allow us to establish with certainty the postnatal prognosis in individual cases [9]. The survival of a baby suffering from omphalocele isolated, not associated with other abnormalities malformation is 60-70% [1].

Prenatal diagnosis of omphalocele should influence timing, mode, and Center of delivery. Neonatal intensive care unit (NICU) support should be offered to women who decide not to terminate pregnancy. On the contrary, the woman in the present case decided voluntarily to terminate the pregnancy at 16 weeks of gestation.

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