Giant omphalocele - prenatal diagnostics, pregnancy evaluation and postnatal treatment

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Summary

This study describes prenatal diagnostics of fetal omphalocele by ultrasonographic examination, planned childbirth by cesarean section and surgical correction of the anomaly in an older primipara who became pregnant through a spontaneous menstrual cycle after a five-year long medical examination and treatment of infertility. Pregnancy was carried out to full term. Planned cesarean section performed at 40 weeks of pregnancy gave birth to an infant with a giant omphalocele 8-9 cm in size with an abdominal wall defect approximately 5 cm in size. The same day the newborn was subjected to surgery during which the hernial sac containing intestine was repositioned inside the child's abdomen and a paraumbilical defect in the abdominal wall was sutured. The newborn spent seven days in the intensive care unit on assisted ventilation in order to maintain a constant level of intraabdominal pressure. The postoperative period was complication-free. The infant was released from the hospital after 14 days completely adapted and ready for breastfeeding. The rest of the neonatal period remained without complications.

Key words: Omphalocele; Ultrasonographic diagnostics; 2D/3D ultrasound.

Introduction

Omphalocele is a defect in the umbilical ring from which a sac protrudes covered with amnion and peritoneum and into which abdominal contents have typically herniated. The incidence of omphalocele is approximately one in every 3,000-4,000 births [1].

The exact etiology of omphalocele is still not well known [2]. The most interesting particularity about this malformation is that it is associated with other structural anomalies in a high precentage of cases (ranging from 25-70%). Congenital heart defects, urogenital tract anomalies and central nervous system anomalies are among the most frequent. References even cite a case of omphalocele associated with dextrocardia, a very rare anomaly [3]. A case of omphalocele associated with alveolar capillary dysplasia was also described [4]. Genetic anomalies, i.e., chromosome aberrations, are not rare either, trisomy 13 and 18 being the most frequent. As early as 1987, Gilbert and Nicolaides performed karyotyping in 35 fetuses with omphalocele at 16 to 36 weeks. They reported that 54% had chromosomal abnormalities [5]. Getachew and associates reported in 1991 that 87% of fetuses with an omphalocele containing only intestine also had abnormal karyotypes, compared to only 95% of cases where the sac contained liver as well [6].

Ultrasonographic diagnosis is made in the early part of the second trimester by ultrasound (US) visualization of an ovoid mass beginning at the front abdominal wall and representing visceral hernia. The size of this centrally located defect ranges from 2 to 12 cm.

Omphalocele prognosis depends on anomalies associated with it. Considering that associated abnormalities are present in a high percentage, additional examinations (invasive diagnostics, most frequently amniocentesis with karyotype determination, fetal echocardiography, 3D/4D color Doppler, etc.) are necessary. Only after all examinations and detection of eventual presence or absence of other associated anomalies are performed can an opinion be given to parents.

Preterm labor and delivery complicates over half of all pregnancies associated with a fetal abdominal-wall defect. Corresponding mortality rate is 60% for omphalocele. The prognosis is good for the fetus weighing more than 1,500 g and has no associated anomalies, provided surgical correction is achieved rapidly. There is no evidence that cesarean delivery improves survival. Fitzsimmons and colleagues emphasized that elective timing of delivery optimizes neonatal surgical care [7].

Even though postnatal corrective surgery is not complicated and most frequently has a positive outcome, it poses a challenge for every surgeon not only when it comes to selecting the right surgical treatment but especially in selecting the optimal moment for surgical treatment and postoperative care of the newborn.

Case Report

A case of pregnancy in which prenatal US examination at 18 weeks gestation detected an omphalocele is described.

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The patient, age 38, achieved pregnancy through a spontaneous cycle after a five-year long period of infertility examinations and treatment. She previously had had one miscarriage at eight weeks of gestation.

Monitoring of fetal development and pregnancy control according to the standard protocol was continued after US diagnostics of pregnancy. At 16 weeks of gestation invasive diagnostics was performed by amniocentesis, because of the pregnant woman's age. Fetal karyotype 46XY confirmed a genetically healthy male fetus. At 18 weeks of gestation an omphalocele, paraumbilical defect through which intestinal convolutes protruded into the umbilical sac, was visualized by US. It contained convolutions of small intestine, colon and mesenterium.

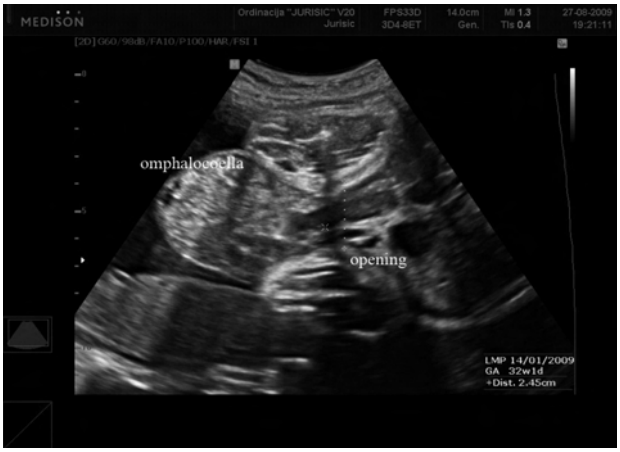

Figure 1. — 2D Ultrasonographic view of the omphalocele.

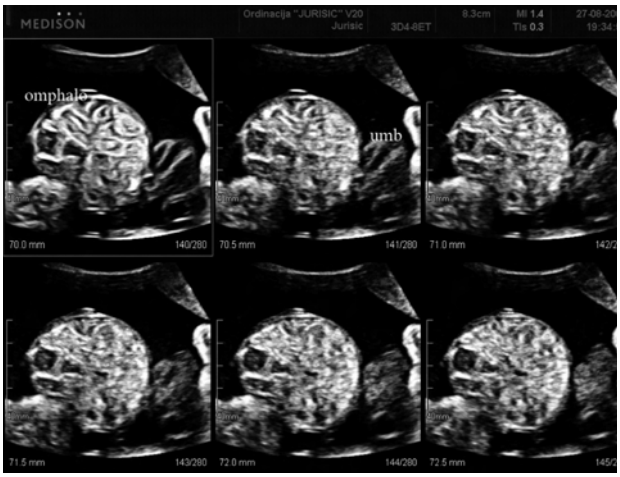

Figure 2. — Omphalocele – 2D/3D multislice ultrasound.

The parents were informed that, at that time, pregnancy became high-risk and they were given necessary explanations about childbirth, early neonatal outcome as well as the necessity of subjecting the infant to surgery in the early neonatal period. Regardless of unpredictable development and growth of the omphalocele during the course of pregnancy, i.e., growth of the fetus, considering the possibility of surgical correction in the early postnatal period, it was decided not to terminate the pregnancy but instead to continue monitoring until term, and to

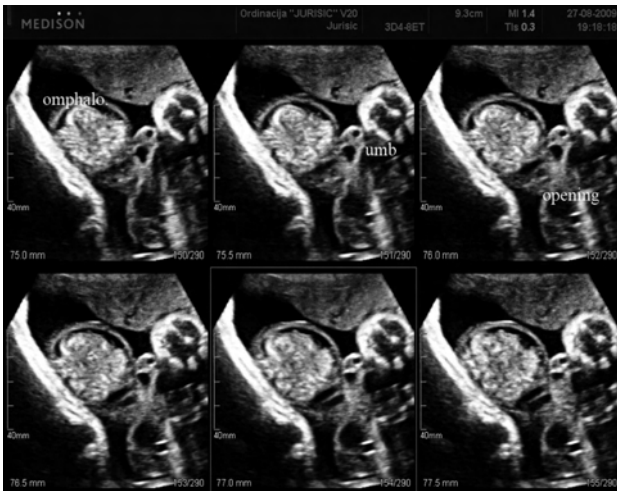

Figure 3. — Ultrasound view of the omphalocele, paraumbilical abdominal wall defect and umbilical cord.

deliver by elective cesarean section, while at the same time planning the surgical treatment of the infant after birth.

US examinations that followed indicated normal development of the fetus. Fetal echocardiography verified regular development of all cardiac chambers and normal position of the large vessels. Development of fetal lungs was regular. Morphology of abdominal organs was also regular, with normoposition of the liver, spleen, and stomach. Fetal kidneys were without hydronephrosis, i.e., normal in structure and normal renal vascularization. Visualization of the fetal bladder showed it filled and discharged normally. US visualization showed the spinal column to be normal in structure. 3D morphological analysis of the fetal head and brain was regular, with a face of normal morphology. Considering the length of gestation, fetal extremities were also normal in morphology and biometry. Fetal biometry and the biophysical profile showed that development of the fetus was proceeding regularly. Hemodynamic parameters in both uteroplacental and fetal circulation were normal. Three blood vessels with normal flow velocity and vascular indices were visualized in the umbilical cord.

Dimensions of the omphalocele at 28 weeks of gestation were 60 x 50 x 60 mm. The paraumbilical defect on the front abdominal wall was 25 mm in size. At 38 weeks, omphalocele dimensions were 66 x 55 x 60 mm while the paraumbilical defect was 29 mm in size.

Cesarean section was performed at 40 weeks of gestation. A live male infant was born weighing 3,550 g and 52 cm in length. The paraumbilical defect in the abdominal wall at birth was approximately 5 cm in diameter.

The infant was subjected to surgery on the first day of life. Initial treatment included gentle compression of the hernial sac and slow repositioning of hernial contents back into the infant's abdomen. The paraumbilical defect of the front abdominal wall was surgically closed 12 hours after delivery. After surgery, the newborn was monitored in the intensive care unit; he spent seven days under assisted mechanical ventilation in order to prevent variations, i.e., increase in intraabdominal pressure when crying, breast-feeding, etc. The postoperative period went by without complications. The infant was released from hospital 14 days after surgery completely adapted and ready for breastfeeding. The rest of the neonatal period was without complications.

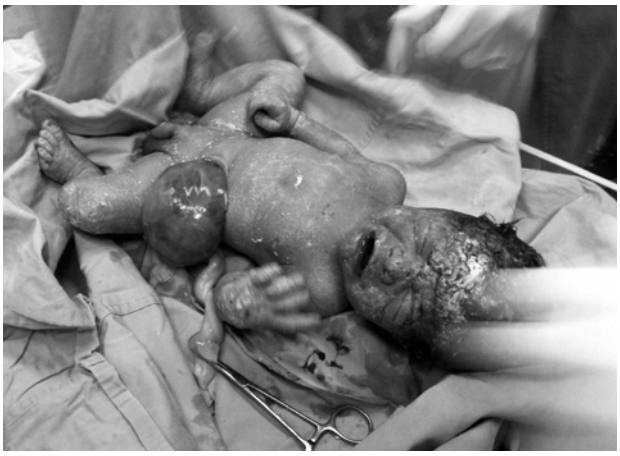

Figure 4. — Omphalocele at birth.

Discussion

Surgical treatment, decisions on surgical technique and optimal time for closure of a paraumbilical defect of giant omphaloceles always pose a challenge to surgeons. Surgical techniques differ. They depend on the size of omphalocele, size of abdominal wall defect, content of hernial sac, eventual presence of other anomalies, previously detected or diagnosed postnatally, infant maturity, body weight at birth, etc. Surgical treatment possibilities are: immediate closure, staged closure or delayed closure after epithelialization. Reference data, however, imply that no precise definitions exist about which omphaloceles should be surgically resolved immediately after birth, which are to be resolved within a certain time period after birth, i.e., which is the optimal time period between birth and closing of the paraumbilical defect [8].

Omphaloceles that cannot be operated imediately after childbirth must initially be subjected to conservative treatment by medications that speed up epithelialization in order to be able to conduct surgical treatment later on [9]. According to a study performed by Mitanchez et al., giant omphaloceles consist of paraumbilical defects larger than 5 cm and/or contain liver as well [10]. Initial treatment of described giant omphaloceles consists of gentle compression aimed at pushing hernial contents towards the abdominal cavity, thus partially squeezing hernial contents back in. Duration of such an initial treatment may vary – from seven days to one month or longer. After complete repositioning of the abdominal organs from the hernial sac into the abdomen, surgical closing of the defect on fascia and skin of the front abdominal wall is performed. Some authors described cases of surgical resolution of giant omphaloceles in 14 newborns. The average gestational age of children born by elective cesarean section was 39 weeks while average weight at birth was 3,100 g. Average duration of gentle repositioning of hernial contents into the infant's abdomen was six days (0-20 days). Full enteral feeding was conducted after an average of 33 days (8-82 days). These infants spent approximately 24 days in an Intensive Care Unit (11-85 days). However, more than half of the babies had postoperative courses complicated by development of sepsis (9 out of 14 children).

Even though the infant described in this study had a giant omphalocele with hernial opening of approximately 5 cm, it was subjected to surgery on the first day of life. Compression of hernial contents and the gradual repositioning into the abdominal cavity was started immediately after birth. The child was operated on under general anesthesia after 12 hours. The paraumbilical defect was sutured in layers – fascia and skin. Problems in initial repositioning can be related to the size of paraumbilical defect; it is easier to perform when paraumbilical defects are larger while it is more difficult and more complicated when defects are narrow. Closing of the abdominal wall defect, however, is easier if the opening is smaller and more complicated if the opening is large. Large paraumbilical hernial openings sometimes require closing by skin grafts [11]. Temporary increase of intraabdominal pressure due to crying, feeding, etc., represents a particular problem related to repositioning of hernial contents into the abdomen and closing of the abdominal defect. That is why the infant described in this study had to be under assisted mechanical ventilation accompanied by adequate sedation by medication. Antibiotic prophylaxis for infection had to be administered during the postoperative period. During this 7-day period, the infant was on parenteral nutrition. After seven days, when it was assessed that the front abdominal wall defect was adequately treated and that the skin wound was healing per primam, the child was extubated and adaptation to enteral feeding started. Full enteral feeding by mother's milk was possible starting from the 14th day after birth.

Discussions are often found in references about the type of delivery, i.e., is it indicated to do elective cesarean section in cases of a fetus with an omphalocele without other visible morphological and genetic malformations? Although opinions are divided, there is a real danger of hernial sac rupture in spontaneous births regardless of the mode of delivery [11].

References also cite discussions about the use of povidone-iodine, i.e., iodine solution, for conservative treatment of omphaloceles, for epithelialiazation of an omphalocele sac as well as even the use of povidone iodine for disinfection of a surgical lesion on the front abdominal wall. This is due to resorption of iodine through the skin and mucous membranes and effects this might have on thyroid gland function. Whitehouse *et al*. believe that topical povidone-iodine promotes epithelialization of the omphalocele sac; systemic effects of iodine are minimal and thyroid supplementation is not necessary. Topical povidone-iodine is an effective initial strategy for giant omphaloceles and does not produce clinically significant hypothyroidism [12].

Advances in US diagnostics of fetal anomalies and their early detection have made it possible to monitor pregnancies with diagnosed anomalies, to start fetal therapy as early as inside the uterus, and to perform birth planning

as well as planning of surgical correction of anomalies after birth. Thus the number of terminations of pregnancy due to anomalies that can be surgically corrected, either prenatally or postnatally, is diminished even though, until recently, such anomalies represented an indication for pregnancy termination.

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