Differential Diagnosis of Abdominal Wall Defects - Omphalocele versus Gastroschisis

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Rezumat

Diagnosticul diferenţial al defectelor de perete abdominal – omfalocel versus gastroschizis


Cuvinte cheie: omfalocel, gastroschizis, diagnostic antenatal, management antenatal

Abstract

A newborn with abdominal wall defect is one of the most dramatic cases in neonatology, but also a challenge for pediatric surgeons. This article describes the fundamental concepts of two major abdominal wall defects - gastroschisis and omphalocele - including options and principles of prenatal and postnatal care. Although these birth defects of the abdominal wall are always grouped together, they are two separate and distinct entities, with many differences in terms of pathology and associated anomalies; this explains the different therapeutic approach and results. For a correct management of the newborn with this anomaly, it is essential to understand the similarities and differences between gastroschisis and omphalocele. This article emphasises the similarities between these two parietal defects, highlighting the differences as well.

Key words: omphalocele, gastroschisis, antenatal diagnosis, antenatal management
Definitions

Omphalocele and gastroschisis are congenital malformations of the abdominal wall represented by the extra peritoneal herniation of the abdominal viscera (Fig. 1).

In omphalocele (also known as exomphalos) the parietal wall defect is central, between the inner edges of the rectus abdominis muscle with the umbilical cord inserted to the lower part. The defect can be located at the top, middle or bottom; the size and location have important implications in postnatal management. Abdominal viscera are initially covered by a transparent membrane which becomes whitish, opaque in contact with air. The membrane layers, from outside to the inside, are: amniotic membrane, Wharton jelly and peritoneum. Umbilical vessels insert into the membrane and not the abdominal wall. The abdominal wall defect and the diameter of herniated viscera range from 2-3 cm (the content of an intracondal omphalocele is represented by only a few intestinal loops) to dimensions that almost completely cover the anterior abdominal wall. The omphalocele may contain the small and large intestine, liver, stomach or even the spleen sometimes. The volume of the abdomen is small, being inversely proportional to the volume of herniated viscera (1).

In gastroschisis (also known as laparoschizis) the parietal defect is smaller, about 3-4 cm, located paraumbilically to the right with the umbilical cord normally inserted and intact viscera which float freely into the amniotic fluid. Rarely, the defect can be located paraumbilically to the left. The intestine, which has not undergone rotation, from the duodenum down to the rectum is located outside of the peritoneal cavity floating in the amniotic fluid which has an irritant action on the intestinal wall, causing local perivisceritis (2). The wall of the intestine is thickened, edematous and dark red, changes that are secondary to ischemia and the contact with amniotic fluid. There are many perivisceral adhesions and intestinal loops are covered by a pseudomembrane containing collagen (1). All these cause a shortening of the bowel length. The mesentery is short, thick and presents a high risk of strangulation at the level of the parietal wall defect. The intestine presents a poor peristalsis and significant nutrient absorption imbalance (3).

Embryological development of the abdominal wall and pathology of the omphalocele and gastroschisis

The abdominal wall results through fusion of four folds: cephalic fold which forms the thoracic wall and the epigastric part of the abdomen, caudal fold forming the perineum, bladder and hypogastric region and 2 lateral folds forming the side walls of the abdomen. They meet in the center and merge to form the umbilical ring which is complete in the 4th week of embryonic life. Primitive gut shows a rapid increase in size in the 6th week leading to herniation of the umbilical ring and then goes through the process of rotation and reintegration in the 10th week (4).

An abdominal wall defect occurs due to early interruption in the process of mesenchymal differentiation due to unknown causes (3).

In the case of gastroschisis the most accepted pathogenic theory is intrauterine occlusion of the right umbilical-mesenteric artery associated with infarction, followed by split of the umbilical ring and intestinal eventration (5). The right paraumbilical area has an increased risk because its vascularization is provided by the umbilical-mesenteric artery and right umbilical vein until they involute. If development is disrupted during fetal life, a defect may occur at this level due to ischemia. This theory explains the location mostly on the right of the defect and sometimes the association with intestinal atresia (6,7). Another hypothesis that can explain some cases of gastroschisis is the early break of a hernia sac, at the umbilical cord (8).

Omphalocele etiology is unknown. Various theories have been postulated, including failure of internal organs to return into the abdomen in the 10-12th week and failure of lateral mesodermal abdominal folds to migrate centrally (3). We can note two particular issues in omphalocele (4):

![Gastrochisis](image1.png)  ![Omphalocele](image2.png)

Figure 1. Presentation of gastroschisis and omphalocele
• Cantrell Pentalogy - the child has omphalocele associated with sternal cleft, anterior diaphragmatic hernia, ectopic cords, and cardiac malformations (9).
• Lower midline syndrome – bladder extrophy or cloaca, anal imperforation, colon atresia, intestinal fistula to the bladder, sacral vertebral anomalies, myelomeningocele defect of the caudal fold.

Epidemiology

Internationally, the incidence of gastroschisis is assessed at 1/4000-6000 births, while the incidence of omphalocele is 1.5 - 3/10,000 births. Although in the past omphalocele was the more common of the two disorders, gastroschisis notes a growing incidence (4,10). The etiology of both abdominal wall defects is unknown, most of them are sporadic cases rarely hereditary (possibly genetically determined). There have been maternal risk factors described for abdominal wall defects. Gastroschisis is frequently associated with low maternal age (under 20 years), poverty and social instability (11). In addition, it has also been noted that use of aspirin, ibuprofen and pseudoephedrine during the first trimester of pregnancy can be related to the vascular pathology of the disease, but alcohol, cigarettes and drugs also appear to increase the risk of these birth defects. Association with chromosomal abnormalities and other malformations are rare in gastroschisis. For omphalocele the mother’s living conditions seem to play a minor role, being associated with mothers of older age (mostly older than 30 years) and in 30% of cases there are some chromosomal abnormalities (trisomy 13, 18 and less frequently 21) (12).

In our hospital in the past 10 years, there were 55 cases of omphalocele and 50 of gastroschisis. The gestational age was between 31 and 38 weeks and birth weight between 1500 g and 3300 g. Regarding mortality, there were 19 deaths of infants with gastroschisis and only 5 deaths of infants with omphalocele. The average age of mothers giving birth to children with gastroschisis was 25 years (between ages 16 and 37 years) and the mean age of those who had children with omphalocele was 35 years (ages 27 years and 44 years). In the history of mothers whose children were born with gastroschisis aspirin use was noted in 37 cases, ibuprofen in 28 cases, alcohol in 31 cases, cigarettes in 42 cases and drugs in 2 cases.

Associated anomalies

Infants with congenital abdominal wall defects have an increased risk of additional abnormalities. The relative risk and the type of associated anomalies is a major difference between gastroschisis and omphalocele. These differences are important in the clinical management and prognosis. In gastroschisis, the incidence of associated anomalies is between 10% and 20%, and most of the defects are at the level of the gastrointestinal tract (13). Approximately 10% of infants with gastroschisis associate intestinal stenosis or atresia as a result of insufficient arterial blood supply to the small bowel during development of gastroschisis or, more commonly, by volvulus and mesenteric vascular pedicle compression in the narrow parietal defect (14). Other less common associated anomalies include undescended testes, Meckel's diverticulum and intestinal duplications. Severe anomalies such as chromosomal abnormalities are extremely rare.

Among infants with gastroschisis, 5 cases were associated with intestinal atresia and one case with ventricular septal defect who died 4 days after surgery due to subarachnoid hemorrhage.

Infants with omphalocele have a high incidence of associated anomalies (50-88%). The severity of these anomalies determines the clinical prognosis. It is believed that they are more common when there is polyhydramnios or oligohydramnios, which complicate the pregnancy. Chromosomal abnormalities are found in 40-60% of cases and include trisomies 18, 13 and 21 also Turner syndrome, Klinefelter and triploidy. Heart defects are common (30-50%) and include ventricular and atrial septal defects, tetralogy of Fallot, pulmonary artery stenosis, pulmonary hypoplasia, double right ventricle, bicuspid aortic valve syndrome, transposition of the great vessels, coarctation of the aorta, ectopic cords, absence of the inferior vena cava. Respiratory failure is present in cases of giant omphalocele. Genitourinary anomalies (40%) include bladder extrophy, imperforate anus, spinal anomalies, ureteropelvic junction obstruction, kidney malposition, cloacal extrophy. Anomalies of the neural tube, head and neck include: neural tube defects, holoprosencephaly, encephalocele, cerebellar hypoplasia, cleft lip, facial cleft, micrognathia, cystic hygroma. Among the digestive anomalies (40%) are diaphragmatic hernias, malrotation, duplicated intestine, atresia, absence of gallbladder, abnormal liver fixation, transesophageal fistula, imperforate anus (15). Musculoskeletal abnormalities (10-30%) include: scoliosis, hemivertebra, dwarfism, syndactyly, leg varus deformity. Beckwith-Wiedemann syndrome (5-10%) comprises a group of anomalies involving omphalocele, macroGLOSSIA, visceromegaly, hyperinsulinemia and an increased risk for Wilms tumor, hepatoblastoma, neuroblastoma developing later in childhood. Omphalocele is also associated with Reiger syndrome and Prune-Belly Syndrome especially in large parietal defects such as Cantrell pentalogy and lower median line syndrome (3).

In our clinic, the recorded malformations associated with omphalocele were: 2 cases with Fallot tetralogy, 2 cases with ventricular septal defect, 1 case with agenesis of the left atrium, ventricular septal defect, coarctation of the aorta and anorectal agenesis, which died day 1 postoperatively. Another case was associated with transposition of the great arteries, ventricular septal defect and intestinal atresia who died 2 months postoperatively due to Pseudomonas sepsis. Other associated anomalies included: two cases of omphalocele associated with diaphragmatic hernia and chromosomal abnormalities, 1 case with Down syndrome and one case with trisomy 18.

Antenatal diagnosis

We can no longer accept incidentally discovered conditions of this type considering the complications and difficult treatment.
Elevated serum levels of AFP in the second trimester are useful as screening test for both gastroschisis and omphalocele. Alpha-fetoprotein is a protein produced by fetal tissues, secreted in fetal and maternal serum. The maximum concentration is reached in the 14th week of pregnancy. AFP is also found in amniotic fluid, urine and cerebrospinal liquid. Its role is to carry hormones necessary for normal development, as well as other substances necessary for fetal physiological development. Because herniated viscera float freely into the amniotic fluid, the level of alpha-fetoprotein (AFP) is increased in both maternal blood and amniotic fluid. It should be noted that the level of AFP for omphalocele increases but does not reach levels as high as in gastroschisis. Gastroschisis can be suspected as a result of high maternal AFP, found in 77-100% of cases, and 60% of cases in omphalocele (4,16).

Acetylcholinesterase is an enzyme that can also be increased in the amniotic fluid in the presence of omphalocele. Acetycholinesterase can be detected by chromatography. Chromosomal tests are indicated to those who associate other malformations of the digestive system or other organs and systems.

Diagnosis of parietal wall defect is done using fetal morphology ultrasound starting in the second trimester of pregnancy. Using transvaginal ultrasound, the diagnosis of abdominal wall defects can be made as early as 12 weeks of gestation (17,18,19,20).

Fetal ultrasound is the imaging technique of first choice used during pregnancy because it is noninvasive, fast, allows real-time fetal examination and can be repeated without any risk for mother and fetus (Fig. 2). To identify gastrochisis and omphalocele using ultrasound, specificity is high (over 95%) but the sensitivity is only 75% to 77.3% for gastrochisis and omphalocele (19,21). The aspect of anterior abdominal wall and umbilical cord insertion can be assessed using antenatal ultrasound because the abdominal wall provides a direct interface between it and the amniotic fluid. Anterior abdominal wall is best represented in axial section. Its evaluation is occasionally complicated by flexed fetal limbs. The interior of the anterior abdominal wall is difficult to observe because the density is the same as the rest of the abdominal viscera, with the exception of the case in which the fetus has ascites (22).

**Sonographic features suggestive of gastrochisis**

Prenatal ultrasound is the imaging examination of choice for gastrochisis with a detection rate of 70-72%. The findings include bowel herniation in relation to the anterior abdominal wall, wall thickening, intestinal loops floating freely in the amniotic fluid. The bowel can be identified by its characteristic pattern. Because the intestines are not covered by any membrane in gastrochisis they appear as a mass with irregular edges. Usually the herniated organs are: small bowel and colon, but occasionally may include the stomach, liver, gall bladder, spleen, uterus, ovary, ectopic testis and bladder. Sometimes there can be described signs of bowel obstruction: dilated loops (both intraperitoneal and extraperitoneal), with diameter greater than 17 mm and increased peristalsis. Polyhydramnios may result if high intestinal obstruction occurs. A diameter greater than 17 mm suggests a significant distension and diameter over 11 mm is usually associated with a higher risk of postnatal complications. The right paraumbilical parietal defect is usually 2-5 cm. Insertion of the umbilical cord is normal (Fig. 3). Usually we do not observe ascites. Intestinal perforation can cause calcification and intramesenteric extra abdominal pseudocyst. Unlike omphalocele, associated anomalies are uncommon, but if present, most of them can be detected by prenatal ultrasound. In the diagnosis of gastrochisis color Doppler and Doppler velocimetry for the evaluation of the mesenteric circulation have also been used (23). 3-D ultrasound in patients with abdominal wall defects can help planning postnatal therapy (19).

From our 55 cases of gastrochisis, 34 cases were diagnosed antenatally using ultrasound and the diagnosis was confirmed at birth. The child was delivered in all these cases by cesarean section. The remaining 21 cases were delivered vaginally due to lack of antenatal diagnosis.
Sonographic features suggestive of omphalocele

Definitive diagnosis of omphalocele is possible after 12 weeks of pregnancy, when no confusion can be made with physiologic bowel herniation. The ultrasound demonstrates an abdominal mass which exceeds the abdominal midline. The mass consists of internal organs herniated through a defect at the base of the umbilical ring. The diameter is of 2.5-5 cm. The mass is smooth and contains the abdominal viscera: liver, spleen, small bowel or stomach (Figs. 4, 5). The peritoneum that covers it may break. It is not always visible. Wharton’s jelly can be detected around the peritoneum. The umbilical cord is attached to the tip of the herniated mass, where the umbilical vein can be seen. The umbilical cord can be enlarged at junction with the protruding mass. Fetal ascites is common and is seen in the herniated sac. Rarely oligo-hydraminos and polyhydraminos may be present. When omphalocele diameter is enlarged by 60% compared to the transverse abdominal diameter, it contains the liver.

Of the 50 cases of newborns with omphalocele, 28 of them were diagnosed antenatally and were delivered by caesarean section, thus avoiding complications. Antenatal diagnosis was confirmed at birth in all cases. The remaining 22 infants were from unsupervised pregnancies and were delivered vaginally causing rupture of the omphalocele in 8 cases.

Limitations of ultrasound technique

Ultrasound result depends very much on the examiner, and artefacts can also represent a problem. Despite the clarity of the anomaly, diagnosis of gastroschisis and omphalocele can sometimes be missed.

Misdiagnosed omphalocele being interpreted as gastroschisis has happened in 5% of patients. This misdiagnosis has serious implications because the omphalocele is often associated with chromosomal abnormalities and other serious birth defects. Amniocentesis may be required in cases when one cannot differentiate omphalocele from gastroschisis during antenatal ultrasound. It is also beneficial to perform the karyotype if gastroschisis is associated with other defects.

Ultrasound parameters to follow are: the size of the abdominal defect, presence or absence of intestinal peristalsis, the presence of Doppler flow at the superior mesenteric artery, fetal biometry, amniotic fluid quantity and quality (hydramnios may be a sign of intestinal obstruction). Hypoecho and fetal distress, intestinal loops’ appearance (more than 3 mm thick wall with hyper echoic appearance) and the quality of mesenteric vasculature are important factors that could indicate induction of preterm birth for improved prognosis.

Nuclear Magnetic Resonance

In general, magnetic resonance imaging (MRI) is not frequently used in the diagnosis of abdominal wall malformations, it is expensive and not always available. MRI can be used in addition to ultrasound for cases where ultrasound diagnosis is uncertain due to oligohydraminos or obesity. MRI can detect birth defects and other fetal organs and systems associated with omphalocele, such as cardiovascular and neural tube

Figure 4. 2D ultrasound of omphalocele containing small bowel and liver - sagittal section (A) and cross section (B)

Figure 5. Doppler ultrasound, sagittal section - umbilical vein in the omphalocele
defects (Figs. 6,7). Its diagnostic accuracy is superior to ultrasound in some cases of brain malformations due to high soft tissue resolution and global viewing of complex fetal defects. MRI is considered safe for fetal development. There are no studies to demonstrate teratogenicity or other adverse effects during pregnancy (24,25,26,27).

**Differential diagnosis**

The most common differential diagnosis of gastrochisis must be done with ruptured omphalocele. There are enough elements to the make the correct diagnosis: examination of viscera, the paraumbilical opening, normal umbilical insertion, other associated anomalies (28,29). One should also make differential diagnosis with: physiologic bowel herniation, umbilical hernia, bladder extrophy, cloaca extrophy.

**Antenatal management**

Prenatal diagnosis offers the possibility to choose the location, type of birth (natural / caesarean) and optimal gestational age delivery to minimize all possible complications. The type of birth of fetuses with such congenital malformations is a debated topic. Proponents of routine caesarean birth state that vaginal delivery may affect the exposed viscera (30). This is not supported by published data, so that no difference between vaginal delivery and cesarean delivery could be seen (31,32). It is important to identify signs of early antenatal complications and fetal distress that would allow a better determination of the indications for cesarean delivery (33). Therefore, the method of birth of the fetus with gastrochisis should remain at the discretion of the obstetrician and mother.

The moment of birth is also controversial. Birth before term
was proposed in order to limit intestinal lesions through prolonged contact with amniotic fluid. This is supported by the role of cytokines in amniotic fluid and of pro-inflammatory mediators (including interleukin-6 and interleukin-8) and the damage to the myenteric plexus nerve and Cajal interstitial cells (34,35). Since intestinal edema and inflammation increase during the course of pregnancy, preterm birth is considered by some as a solution to mitigate these effects. However, the data in the literature is divided on the benefits of premature labor. Birth can be induced successfully in a number of cases at 36-37 weeks of gestation, probably due to the inherent tendency toward premature birth (36). The arguments against it are that a preterm low birth weight negatively influences the outcome, infants weighing less than 2 kg accumulating a longer period of enteral nutrition, the number of days on ventilation and the duration of parenteral nutrition (37). Some authors support premature birth based on appearance of distended bowel and its thickness measured by prenatal ultrasound. The presence of bowel distension correlates with poor outcomes, including fetal distress, even fetal death in some cases (38).

It is very important that birth should take place in a tertiary center to provide immediate access to neonatal intensive care and pediatric surgery (Fig. 8 A, B). It is essential to reduce the time between birth and reintegration of viscera. A recent review found that birth in a tertiary center was associated with an overall reduction in the risk of morbidity compared to birth in a hospital without such facilities (39,40).

Conclusions
Mandatory screening of pregnant women at 10-12, 20-22 or 32-34 weeks of gestation is necessary for the study of fetal morphology. This can accurately diagnose these birth defects of the anterior abdominal wall and can determine a proper course of treatment and proper advice for parents from a complex team (obstetrician, neonatologist, pediatric surgeon, and psychologist) in terms of prognosis and possible associated malformation and planning birth in a tertiary center.

Contributing authors
All the authors have the same contribution.

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