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Abdominal wall defects: Prenatal diagnosis, newborn management, and long-term outcomes

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ABSTRACT

Omphalocele and gastroschisis represent the most frequent congenital abdominal wall defects a pediatric surgeon is called to treat. There has been an increased reported incidence in the past 10 years mainly due to the diffuse use of prenatal ultrasound. The early detection of these malformations, and related associated anomalies, allows a multidisciplinary counseling and planning of delivery in a center equipped with high-risk pregnancy assistance, pediatric surgery, and neonatology. At present times, closure of defects, even in multiple stages, is always possible as well as management of most of cardiac-, urinary-, and gastrointestinal-associated malformations. The progress, herein discussed, in the care of newborns with abdominal wall defects assures most of them survive and reach adulthood. Some aspects of transition of medical care will also be considered, including fertility and cosmesis.

Introduction

Omphalocele and gastroschisis are the two most common congenital abdominal wall defects, and their repair and management need multidisciplinary expertise. The increased possibility to detect them at early stages during pregnancy allows planning the best prenatal and postnatal management in a center with pediatric surgery, neonatology, and high-risk pregnancy assistance. The common goal of closure of defect, either primary or staged, is herein discussed together with the dissimilar and peculiar aspects each malformation has. Finally, early and long-term outcomes are considered, in light of the general favorable prognosis and survival into adulthood.

Omphalocele (exomphalos)

Definition and pathogenesis

Omphalocele (from ancient Greek: *omphalos* = umbilicus; *cele* = hernia) is a congenital defect of the abdominal wall, located by the umbilical ring, consisting of an evisceration of internal organs in a sac covered by a three-layered membrane made of peritoneum, Wharton's jelly, and amnion. The sac usually contains the small intestine, liver, spleen, colon, and sometimes the gonads. Exomphalos results from the failure of bowel loops that do not return to the abdominal cavity following the physiological herniation of the umbilical cord between the 6th and 10–11th week of development. The small bowel is usually malrotated or non-rotated, although usually it is morphologically and functionally normal. An omphalocele containing the liver of at least 4.5–5 cm in diameter is defined as a giant omphalocele (GOC). In these cases, the thoracic cavity may also be abnormally shaped and reduced in size. GOC is associated with an underdeveloped abdominal cavity and with a significant visceral–abdominal disproportion, which prevents a safe primary closure. Pulmonary hypoplasia may also be present, associated with pulmonary distress and narrow thorax in chest radiographs. Children with an omphalocele generally have essentially intact abdominal wall muscles, but hypoplastic and laterally displaced rectal abdominal muscles[1] (Table 1).

Prevalence

The prevalence of omphalocele has been estimated to be 1 per 3000–4000 births in Western countries.[2] In Japan, the prevalence during the period 1997–2006 was reported to be 1 per 2500 births by the Japan Association of Obstetricians and Gynecologists Program of Birth Defects Monitoring.[3] At around 14 to 18 weeks of gestation the incidence is 1 case out of 1100 fetuses. A recent study reported a prevalence of one in 381 at 11–14 weeks of gestation.[4] There is a very high rate of termination of pregnancy (30–52%) due to the presence of associated anomalies, and spontaneous abortion.

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Table 1
Type of defect, contents, and modality of delivery.

Hernia of the cord		Small bowel/Meckel's diverticulum/omphalomesenteric duct remnant	Vaginal at term	
Minor/medium defect	2–4 cm	Small bowel/liver/stomach	Vaginal/cesarian at term	Intact/ruptured sac
Large defect (GOC)	> 4 cm	Liver, spleen, and small bowel	Cesarian at term	Intact/ruptured sac

A recent article from EUROCAT reports an increasing prevalence of abdominal wall defects that, however, seems to involve only instances of gastroschisis[5] (Table 2).

Prenatal diagnosis, associated anomalies, and delivery

Prenatal diagnosis of omphaloceles is usually made by ultrasound screening in the late first trimester to mid-second trimester. In some cases, an elevated maternal alpha fetoprotein level may be detected. Congenital omphaloceles are often associated with other anomalies; chromosomal defects (trisomy 13, 18, or 21) are observed in 30–40% of cases, and multiple anomalies including chromosomal aberrations are observed in 67–88% of cases. Interestingly, multiple associated anomalies appear to be more common with minor omphalocele (< 4 cm) than giant omphalocele (55% vs. 36%).[6] Associated syndromes such as cloacal exstrophy, Donnai–Barrow syndrome, pentalogy of Cantrell, and Beckwith–Wiedemann syndrome (12%) can also be suspected or identified through a fetal ultrasound. In a study from the Netherlands, only 14% of omphaloceles were effectively isolated lesions. Prenatal screening in omphalocele fetuses needs to have a detailed evaluation of the cardiac system (14–47% of associated anomalies) and of the central nervous system (3–33% of associated anomalies), as severe defects strongly encourage a termination of pregnancy. Nonetheless, one-third of the fetuses considered to present isolated cases end up having multiple associated defects that are only detected postnatally.[7,8] This is an important evidence for high-risk pregnancy teams to discuss with prospective parents of an omphaloceles fetus. Reliable sonographic data and/or MRI evaluation to predict postnatal morbidity and survival rates are being developed.[9,10] A measurement of the lung/thorax transverse area ratio (L/T) may be useful to predict associated pulmonary hypoplasia in giant omphaloceles.[9] Some researchers have studied the ratios between the biggest omphalocele diameter compared with abdominal circumference (O/AC), femur length (O/FL), and head circumference (O/HC), in an attempt to correlate it with postnatal morbidity and mortality. The most useful parameter to predict the need for staged versus primary closure and respiratory insufficiency is probably that of O/HC, as 0.21 or more had 84% sensitivity and 58% specificity in predicting challenging cases.[1]

In the meantime, other studies support the thesis that, in the first trimester, isolated fetal exomphalos with normal nuchal translucency (NT) may be considered "physiological." The midgut is normally herniated within the umbilicus in early embryogenesis but resolves by the 12th week of gestation. Therefore, before this time, a "physiological" exomphalos may be detected in all fetuses. In most cases, the exomphalos will resolve spontaneously as pregnancy progresses, with no adverse sequelae; this is particularly true for exomphalos containing only the bowel.[4]

Currently, there are no specific fetal interventions performed either experimentally or on people for omphaloceles, other than karyotype amniocentesis.

The timing of delivery of an omphalocele is not controversial; most infants are indeed born at term, unless there are complicating features such as polyhydramnios. Preterm delivery is in fact not recommended. In contrast, the route of delivery is not yet a settled issue, with a dispute between vaginal birth and caesarian section proponents. Generally, when the defect is an extra-abdominal liver, the concern for hepatic injury during vaginal delivery encourages resorting to a caesarian section.[1]

Table 2
Prevalence of abdominal wall defects in the last 50 years.

	Omphalocele	Gastroschisis
Sweden, 1965–1976	1/5000	1/15,400
California, 1968–1977	1/4000	1/20,000
Spain, 1976–1981	1/6600	1/25,000
Finland, 1970–1979	1/5100	1/9200
Literature review, 1985–2003	1/4–7000	1/10–20,000
EUROCAT, 1999–2008	2.19/10,000	2.98/10,000

Treatment

Perinatal care

In the delivery room, the initial management of an infant with omphalocele requires a careful assessment of the cardiopulmonary condition, since these children may have unsuspected pulmonary hypoplasia that requires immediate intubation and ventilation. A thorough search for associated anomalies should then be undertaken. The high risk of associated cardiac defects demands a directed cardiac evaluation, including auscultation, four-limb blood pressures, and peripheral pulse examination. Once stabilized, a more detailed evaluation can be pursued with an echocardiography. Likewise, an abdominal ultrasound should be obtained to evaluate the possibility of associated renal anomalies. Neonatal hypoglycemia should alert the physician to the possibility of Beckwith–Wiedemann syndrome (EMG syndrome: exomphalos, macroglossia, and gigantism), which occurs in 12% of omphaloceles.[6] When planning the delivery of infants with omphalocele, risks arising from associated anomalies have to be properly taken into consideration. Adequate intravenous access should be obtained in order to begin fluid resuscitation. Infants with omphalocele with an intact sac do not have as significant fluid and temperature losses as those with gastroschisis; nonetheless, losses are higher than for infants with an intact abdominal wall. To minimize losses, the omphalocele itself can be dressed with saline-soaked gauze and an impervious dressing. A nasogastric or orogastric tube should be inserted and placed on suction or gravity drainage. In case of a ruptured omphalocele, the initial management of the viscera should be the same as that recommended for infants with gastroschisis.

Surgical treatment: General considerations

Over the past five decades, the surgical management of omphalocele has evolved, to the point that now the ultimate goal of surgical intervention is to provide complete fascial and skin closure without causing excessive intra-abdominal pressure or abdominal wall tension. The improved safety of techniques and of supportive care is now allowing us to make a step further from the modern ideal of a single-stage reduction and defect closure adding to it an enhanced cosmetic outcome. The levels of safety of reduction and closure techniques are related to the level of intra-abdominal pressure (IAP) and therefore to the degree of visceral–abdominal disproportion, which in turn, influences the risk of complications and thus the ultimate outcome of the intervention. Therefore, it is important to minimize intra-abdominal hypertension (IAH) in order