Gastroschisis and Omphalocele

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The newborn who has an abdominal wall defect is one of the most dramatic presentations in medicine and offers many challenging problems to the pediatric surgeon. This article presents the basics of the two most common abdominal wall defects—gastroschisis and omphalocele—including principles and options of prenatal, postnatal, and surgical management. Although textbooks group the two entities together, they are separate and distinct and have many important differences in pathology and associated conditions that explain the differences in treatment plans and outcomes. Understanding the similarities and differences between gastroschisis and omphalocele is essential for patient management; therefore, the following sections first acknowledge the similarities and then emphasize the differences.

Definitions

Gastroschisis is a full-thickness defect in the abdominal wall usually just to the right of a normal insertion of the umbilical cord into the body wall. Rarely it is located in a mirror-image position to the left of the umbilical cord. A variable amount of intestine and occasionally parts of other abdominal organs are herniated outside the abdominal wall with no covering membrane or sac.

An omphalocele (also known as exomphalos) is a midline abdominal wall defect of variable size, with the herniated viscera covered by a membrane consisting of peritoneum on the inner surface, amnion on the outer surface, and Wharton’s jelly between the layers. The umbilical vessels insert into the membrane and not the body wall. The hernia contents include a variable
amount of intestine, often parts of the liver, and occasionally other organs. The defect may be centered in the upper, mid, or lower abdomen and its size and location have important implications for management.

**Abdominal wall development and the pathology of omphalocele and gastroschisis**

The abdominal wall is formed by infolding of the cranial, caudal, and two lateral embryonic folds. As the abdominal wall is forming, the rapid growth of the intestinal tract leads to its migration outside the abdominal cavity through the umbilical ring and into the umbilical cord during the sixth week of gestation. By the 10th to 12th week, the abdominal wall is well formed and the intestine returns to the abdominal cavity in a stereotypical pattern that results in normal intestinal rotation and later fixation [1].

Gastroschisis is thought to result from an ischemic insult to the developing body wall. The right paraumbilical area is an area at risk because it is supplied by the right umbilical vein and right omphalomesenteric artery until they involute. If this ordered development and involution is disturbed in degree or timing, then a body wall defect could result from the resulting body wall ischemia [2,3]. An alternative hypothesis that may account for some cases of gastroschisis is that the defect results from an early rupture of a hernia of the umbilical cord [4].

In omphalocele, the bowel does not return to the abdomen but remains out in the umbilical cord. The exact sequence of events are not known but are presumed to involve a failure of abdominal wall infolding [1]. A variable amount of midgut and other intra-abdominal organs are herniated out of the defect depending on its size and relative location on the abdominal wall. Cranial fold deficits predominately result in epigastric omphaloceles that may be associated with additional cranial fold abnormalities such as anterior diaphragmatic hernia, sternal clefts, pericardial defects, and cardiac defects. When these elements occur together, they are known as the pentalogy of Cantrell [5]. When the infolding deficit involves the caudal fold, the omphalocele may be associated with bladder or cloacal exstrophy.

**Epidemiology**

There are regional differences in the incidence of abdominal wall defects and the relative proportions of gastroschisis and omphalocele; however, a rough estimate is that worldwide, the incidence of gastroschisis ranges between 0.4 and 3 per 10,000 births and seems to be increasing, whereas the incidence of omphalocele ranges between 1.5 and 3 per 10,000 births and is stable [6–8]. The etiology of both abdominal wall defects is unknown and most are sporadic, but there are rare familial (and possibly genetically determined) cases of gastroschisis and omphalocele. There are distinctive
maternal risk factors for the different abdominal wall defects. Gastroschisis has a very strong association with young maternal age, with most of these mothers being age 20 years or younger [6]. In addition, gastroschisis has been linked to maternal exposure to cigarette smoking, illicit drugs, vasoactive over-the-counter drugs (such as pseudoephedrine), and environmental toxins [6]. These associations are consistent with the vascular insufficiency of the abdominal wall theories of the etiology of gastroschisis. In contrast, omphalocele is associated with advancing maternal age, with most of these mothers being over 30 years old [7].

**Associated anomalies**

Like all babies who have birth defects, children who have abdominal wall defects are at an increased risk for additional anomalies, but the relative risk and pattern of associated anomalies is a major difference between gastroschisis and omphalocele. These differences are very important in clinical management and long-term prognosis. In gastroschisis, the incidence of associated anomalies is between 10% and 20%, and most of the significant anomalies are in the gastrointestinal tract [9]. About 10% of babies who have gastroschisis have intestinal stenosis or atresia that results from vascular insufficiency to the bowel at the time of gastroschisis development or, more commonly, from later volvulus or compression of the mesenteric vascular pedicle by a narrowing abdominal wall ring [10]. Other less common associated anomalies include undescended testes, Meckel’s diverticulum, and intestinal duplications. Serious associated anomalies outside the abdomen or gastrointestinal tract, such as chromosomal abnormalities, are unusual.

In contrast to the relatively low risk of associated anomalies mainly localized to the gastrointestinal tract seen in patients who have gastroschisis, patients who have omphalocele have a very high (up to 50%–70%) incidence of associated anomalies. The incidence of associated anomalies is lower in liveborn patients because those who have multiple and serious anomalies are more likely to be stillborn [11]. Chromosome anomalies, notably trisomy 13, 14, 15, 18, and 21, are present in up to 30% of cases. Cardiac defects are also common, being present in 30% to 50% of cases. Multiple anomalies are frequent and may be clustered in syndromic patterns. One important pattern is the Beckwith-Wiedemann syndrome that may be present in up to 10% of cases [12]. Beckwith-Wiedemann syndrome is marked by macroglossia, organomegaly, early hypoglycemia (from pancreatic hyperplasia and excess insulin), and an increased risk of Wilms’ tumor, hepatoblastoma, and neuroblastoma developing later in childhood. The size of the abdominal wall defect in omphalocele does not directly correlate with the presence of other anomalies, as demonstrated by the finding that small defects found on prenatal ultrasound have a higher risk of associated chromosomal abnormalities and cardiac defects [13].
Prenatal diagnosis

Abdominal wall defects are often diagnosed by prenatal ultrasound done for routine screening or for obstetric indications such as evaluating an elevated maternal serum alpha fetoprotein (AFP). AFP is the fetal analog of albumin, and maternal serum AFP reflects the level of AFP in amniotic fluid. The testing was developed to evaluate the fetus for chromosomal abnormalities and neural tube defects, but AFP is also usually elevated with abdominal wall defects. The magnitude and likelihood of AFP elevation varies between gastroschisis and omphalocele [14]. In gastroschisis, maternal serum AFP is usually markedly abnormal, with an average elevation of more than nine multiples of the mean (MoM). In contrast, in omphalocele, AFP is elevated by an average of only four MoM, with a much wider range. This different pattern results in a lower sensitivity of maternal serum AFP for omphalocele than for gastroschisis. Like many screening tests, the sensitivity depends on the cutoff value chosen. For example, if abnormal is defined as greater than 3 MoM, then 96% of gastroschisis would be detected but only 65% of omphalocles [15].

Prenatal ultrasound is done in most pregnancies in the United States and, when done, could potentially identify the overwhelming majority of abdominal wall defects and accurately distinguish omphalocele from gastroschisis. This identification would permit an opportunity to counsel the family and to prepare for optimal postnatal care. It is unfortunate, however, that the accuracy of prenatal ultrasound for diagnosing abdominal wall defects is affected by the timing and goals of the study, fetal position, and the experience and expertise of the operator. The specificity is high (more than 95%), but the sensitivity is only 60% to 75% for identifying gastroschisis and omphalocele [8,16]. Diagnostic errors may result because of (1) confusion with other rare abdominal defects (often away from the umbilicus, not covered by a membrane, and fatal); (2) ruptured omphalocles that mimic a gastroschisis because of the lack of a covering membrane; or (3) rare cases of gastroschisis that start out as a covered defect and later rupture but are likely and more commonly simply missed during studies done for reasons other than screening for structural defects.

Prenatal management

A fetus with an abdominal wall defect is a high-risk pregnancy on many levels. For gastroschisis and omphalocele, there is an increased risk of intrauterine growth retardation (IUGR), fetal death, and premature delivery, so careful obstetric follow-up with serial ultrasounds and other tests of fetal well-being are indicated. In both cases, there is some controversy regarding the timing and mode of delivery.

In gastroschisis, the diagnosis of IUGR can be problematic because of the difficulty measuring the torso, but it probably affects 30% to 70% of
fetuses. The cause of fetal growth failure in gastroschisis is unknown but presumed to be due to increased losses of protein from the exposed viscera, although inadequate supply of fetal nutrients is an alternative hypothesis. The exposed bowel is vulnerable to injury. The injury can range in severity from volvulus and loss of the entire midgut, to a more localized intestinal atresia and stenosis, to widespread inflammatory “peel” or serositis that can make the bowel loops indistinguishable from one another. The inflammatory peel develops after 30 weeks’ gestation and is presumed to be due to bowel wall exposure to amniotic fluid or to intestinal lymphatic obstruction. The degree of the inflammatory peel is difficult to quantify on prenatal ultrasound and after delivery, so it has been difficult to correlate with clinical outcome variables [17]. Because bowel injury is a major predictor of postnatal mortality and morbidity, improved understanding and predictive testing would point the way toward potential interventions. Oligohydramnios is also common in gastroschisis, being present in up to 25% of cases. The cause is unknown and it is usually of moderate severity and associated with IUGR, fetal distress, and birth asphyxia. Cases of gastroschisis associated with oligohydramnios prompted investigation of amniotic fluid replacement with saline. Observations from these trials and experimental models supported the hypothesis that amniotic fluid was responsible for the inflammatory peel, and this has been investigated by amniotic fluid exchange transfusion. Preliminary reports have been promising, but additional experience is needed to confirm these results because of the previously noted difficulties in measuring the degree of bowel injury pre- and postnatally [18]. The most devastating prenatal complication with gastroschisis is the uncommon but unpredictable fetal death. It may be caused by an in utero midgut volvulus or probably more commonly by an acute compromise of umbilical blood flow by the eviscerated bowel. It is unfortunate that there are no reliable predictors of this complication [19]. It has been presumed that early ultrasound signs of bowel obstruction, such as increased bowel diameter, indicate a high-risk fetus, but whether it is a higher risk for fetal death and intestinal loss or only a higher risk for intestinal stenosis and atresia or inflammatory peel is not known. The uncommon but tragic fetal death or the patient who has major intestinal loss has been a strong motivating force for the early delivery of the fetus with gastroschisis before such complications happen [20]. It is still unclear whether a fetus with a high risk of prenatal complications can be reliably identified and whether the benefits outweigh the risks of early delivery [21,22].

Omphalocele also has an increased incidence of IUGR (5%–35%), fetal death (usually attributed to related to severe associated anomalies), and premature labor (5%–60%) [23]. Further prenatal diagnostic studies such as high-resolution ultrasound looking for structural defects (especially cardiac defects) and chromosomal studies are often done to diagnose associated anomalies and to help predict prognosis. Unlike gastroschisis, there is usually no reason to consider early delivery, although cesarean section is often
done with giant omphaloceles to prevent rupture or dystocia during labor [24].

Newborn management

The initial management of newborns who have abdominal wall defects starts with the ABCs of resuscitation, and after these have been assessed and stabilized, attention is turned to the abdominal wall defect. Heat loss is an important problem, so care must be taken to dry the baby and maintain a warm environment while protecting the exposed viscera. Premature birth is frequently associated with both conditions and must be considered during the evaluation and treatment. Checking and maintaining serum glucose levels is part of any neonatal resuscitation but especially important in babies who have abdominal wall defects because of the associated prematurity, IUGR, and in omphaloceles, the possibility of Beckwith-Wiedemann syndrome. Prematurity, associated pulmonary hypoplasia, or the significant heart defects seen in omphalocele may lead to early intubation and mechanical ventilation [25]. Gastric decompression is important to prevent distention of the gastrointestinal tract and possible aspiration. Vascular access is obtained for intravenous fluids and broad-spectrum prophylactic antibiotics. Babies who have gastroschisis in particular have high fluid losses from evaporation and third-space losses and may require twice the maintenance volumes of fluids to maintain an adequate intravascular volume. A bladder catheter is useful to closely monitor urine output and guide the resuscitation. The umbilical artery and vein may be cannulated if needed during resuscitation but in omphalocele, placement may be difficult because of the abnormal insertion and course of the vessels. Even if umbilical cannulas are successfully placed, they may need to be removed during the repair of the defect.

When the ABCs have been accomplished, the abdominal wall defect can be assessed and treated. This process involves different considerations in gastroschisis and omphalocele. In gastroschisis, the exposed viscera are inspected and care is taken to avoid twisting of the mesenteric vascular pedicle. If there is vascular compromise because the abdominal wall opening is too small, then the defect should immediately be surgically enlarged, with care taken to avoid the adjacent umbilical vessels and mesentery. The exposed bowel needs to be protected and fluid and heat losses minimized. The easiest method is to place the exposed viscera and entire lower half the baby into a transparent plastic bowel bag. This is fast, requires no special skills or experience, and allows for ongoing assessment of bowel perfusion. Alternatively, the bowel alone can be wrapped with clear plastic wrapping but this can be technically more difficult. Finally, moist dressings over the bowel covered with a clear plastic wrapping is another strategy but requires judgment on how tight to wrap and it hides the bowel from view. Moist dressings alone should be avoided because of the increased
evaporative heat losses. After the exposed bowel is covered, the entire mass is stabilized by placing the baby with its right side down to prevent kinking of the mesenteric pedicle [26].

With omphalocele, the care of the defect and its contents is different. The defect is inspected to make sure that the covering membrane is intact, and nonadherent dressings are applied and stabilized to prevent trauma to the sac. If the omphalocele sac is ruptured, then exposed bowel should be treated as it is for gastroschisis.

Surgical management

In gastroschisis and omphalocele, the ultimate goals are straightforward: to reduce the herniated viscera into the abdomen and to close the fascia and skin to create a solid abdominal wall with a relatively normal umbilicus while minimizing risks to the baby. To achieve these goals, many different techniques have been described. Treatment often varies depending on the size and type of the defect, the size of the baby, and the associated problems. Because there is little hard scientific evidence to favor one method over another, there is considerable variation in the surgical approach. What follows is the author’s personal approach to gastroschisis and omphalocele, with discussion of some but not all of the alternative methods.

In gastroschisis, the ongoing fluid and heat losses of exposed bowel and the subsequent metabolic derangements make rapid coverage a high priority. During the initial resuscitation at delivery or as soon as possible thereafter, a prefabricated, spring-loaded Silastic silo is placed in the defect to cover the exposed bowel. This practice minimizes evaporative losses, prevents additional trauma, and allows for an ongoing assessment of bowel perfusion. These devices can be placed in the delivery room or at the bedside without anesthesia [27]. If the abdominal wall defect is too small to accommodate the device, then the defect can be enlarged under local anesthesia and sedation. If this device cannot be placed at the bedside, then as soon as possible after the initial resuscitation and stabilization, the baby is taken to the operating room for primary closure or silo placement. Formal closure in the delivery room is an intriguing concept that minimizes time and preoperative trauma but is only possible with planned delivery of a known defect and requires a significant commitment of resources [28]. Immediate primary repair without anesthesia has been reported for selected cases and may be the most dramatic example of minimally invasive and minimally traumatic surgery [29]. It is not known how often this approach is feasible.

After the placement of the spring-loaded silo, the baby is evaluated further and cared for in the ICU. With spontaneous diuresis, gastrointestinal tract decompression from above and below, and resolution of bowel wall edema, the volume of the exposed bowel in the bag markedly decreases in a short period of time. When the baby is otherwise stable and the spontaneous reduction of bowel into the abdomen has reached a plateau, the baby is
taken to the operating room for an attempt at delayed primary closure. Serial reduction of the device at the bedside has been advocated, but the risk of displacing the coverage device makes this plan less appealing.

In the operating room, if the bowel can be reduced into the abdomen and the defect closed primarily (or by delayed primary repair), then it should be done. The decision of whether a baby can tolerate reduction and repair can be difficult and can be aided by measuring the intragastric pressure during attempted closure. A pressure of less than 20 mm Hg predicts successful closure without complications of excessive intra-abdominal pressure [30]. Other methods reported to help in the decision to close or not to close are measuring changes in central venous pressure, in ventilatory pressures, and in end-tidal carbon dioxide. If the baby is stable when the fascia is closed, then an umbilicus should be constructed at the level of the posterior iliac crest during the skin closure. Several different methods have been described that achieve an acceptable long-term contour by avoiding the long-term tendency of surgically sutured skin folds to flatten [31]. Creation of an umbilicus can always be deferred to a later time. If primary repair is not possible, then a formal Silastic silo is sutured to the fascia and serial reduction is done postoperatively. Several different methods of serial reduction have been described, but the author’s preference is to use a specially designed wringer clamp with a guard that allows the Silastic sheets to be approximated while pushing the bowel down and away from the roller mechanism [32]. The incremental reduction steps are quick, easy, and easily reversible and can be done multiple times during the day and permit a gradual reclamation of abdominal domain. The reduction is done as tolerated over several days, with the goal to finish it within a week to 10 days to avoid the serious complications of wound infection and necrosis that result in separation of the Silastic sheets away from the fascia, creating an open abdomen. After the viscera have been reduced to the level of the abdominal wall, the baby is returned to the operating room for removal of the silo and closure.

Babies who have gastroschisis and associated intestinal atresia pose a serious challenge. If the bowel is in good condition and the abdomen can be closed without difficulty, then combined primary repair of both defects is possible, but such ideal circumstances are unusual. Therefore, when an atresia is present or suspected, the first priority is to close the abdomen by primary, delayed primary, or staged silo repair. The baby is maintained with gastric decompression and parenteral nutrition for several weeks until repeat laparotomy and repair of the intestinal atresia [10]. This staging of the repairs allows the inflammatory peel to resolve and the herniated contents to gain domain of the abdomen before opening the bowel and creating a vulnerable anastomosis.

For omphalocele, the strategy is markedly different. First, if the covering sac is intact, then there is no urgency to perform operative closure. So long as the viscera are covered with the membrane, a complete evaluation for associated defects can be done and other problems treated. When the baby is
otherwise stable and if the defect is relatively small, then a primary repair can be done by excising the omphalocele membrane, reducing the herniated viscera, and closing the fascia and skin. Membrane overlying the liver that might be injured during excision can be left in place. When primary closure is not likely to be possible, there are many options, but the author’s preference is to treat the omphalocele sac with topical silver sulfadiazine and allow it to epithelialize over the ensuing weeks to months [33,34]. Enteral feedings are usually tolerated after the baby recovers from any systemic problems. After the associated problems have been addressed, the family can be taught the relatively easy wound care, and the baby is followed in the outpatient clinic. When the sac is epithelialized or otherwise sturdy enough to withstand external pressure, compression is done with elastic bandages and serially increased until the abdominal contents are reduced. When the abdominal contents are reduced, the membrane is epithelialized, and the baby is well, a ventral hernia repair is done. This can usually be accomplished within 6 to 12 months, but there is little risk in waiting even longer. The fascial defect remains roughly the same size while the baby grows around it. This permits a relatively easy late closure of initially giant omphalocoeles. This strategy was initially adopted for only those patients who had giant omphalocoeles or serious associated problems but has worked so well in those difficult cases that it is now the author’s preferred method for any defect that cannot be closed. It avoids problems of pulmonary compromise, wound breakdown, infection, and delayed enteral feedings that are encountered with a major operation in a vulnerable newborn. It is also especially useful for obtaining fascial closure in the epigastric region of large omphalocoeles. Many alternative strategies for omphalocele closure have been described, including skin coverage only, staged Silastic silo reduction and repair, reduction within the omphalocele membrane, amnion inversion, and fascial patches. Each technique may have utility in certain circumstances, but in the author’s experience, topical treatment with silver sulfadiazine and delayed repair of the epithelialized ventral hernia has been safe and effective.

Outcomes

The outcome of patients who have gastroschisis depends largely on the condition of the vulnerable bowel, whereas the outcome of patients who have omphalocele depends largely on the associated anomalies and medical conditions. Overall, patients who have gastroschisis have an excellent prognosis. Survival is at least 90% to 95%, with most of the deaths in patients who have catastrophic bowel loss, sepsis, and the long-term complications of short bowel syndrome. Patients who have atresia and relative short bowel syndrome may eventually do well despite a long initial hospitalization and prolonged dependence on hyperalimentation. Even babies who have intact intestinal tracts may require a prolonged hospitalization of weeks to months
because of slow tolerance of enteral feeding [9]. A form of necrotizing enterocolitis manifested by pneumatosis intestinalis on abdominal radiograph is a unique form of intestinal injury that occurs in the postoperative period after gastroschisis repair when feedings are being advanced. Feedings are often complicated by gastroesophageal reflux that may be severe [35]. Long-term gastrointestinal function is usually good, although there is a 5% to 10% long-term risk of adhesive obstruction. Occasional patients have inexplicable long-term intolerance of enteral feedings. The outcome of babies who have omphalocele is much harder to generalize, but most mortality and morbidity is related to associated anomalies rather than the abdominal wall defect.

Summary

Although gastroschisis and omphalocele are distinct abdominal wall defects with many important differences, an understanding of the normal development of the abdominal wall and principles of intra-abdominal pressure evaluation and management and abdominal wall reconstruction are important for both conditions. Gastroschisis is likely the result of a discrete abdominal wall ischemic event and most of the morbidity is a result of acquired in utero bowel injury. The correction of gastroschisis may be accomplished by primary, delayed primary, or staged repair; the exact method is largely determined by the baby’s general medical condition and capacity of the abdominal cavity relative to the herniated viscera. The long-term prognosis is excellent. Omphalocele is likely the result of a more general defect of body wall development and is associated with a much higher incidence of serious anomalies. The surgical correction of small defects is straightforward; for larger defects, a strategy of topical treatment of the membrane with silver sulfadiazine and late repair after epithelialization and external reduction of the hernia contents has simplified management and optimized outcomes. Rather than being related to the abdominal wall defect, the long-term outcome in gastroschisis is mainly related to the degree of associated intestinal injury, and the long-term outcome in omphalocele is mainly related to the associated anomalies.

References


