Gastroschisis vs. Omphalocele — Diagnosis and Management

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Omphalocele and gastroschisis are the most common congenital anomalies seen in infants and can be diagnosed prenatally or at birth. Omphaloclees are clearly associated with genetic predisposition and chromosomal abnormalities whereas gastroschisis seems to be caused by complex interactions between genetic predisposition and environmental exposures. The main difference between the two is the gastroschisis has no covering of peritoneum but omphalocele is sealed by peritoneum.

Overview

Omphalocele

Anterior or ventral abdominal defects are the most common congenital malformation diagnosed in live births. **Omphalocele is an anterior abdominal wall defect** that has the intestine protruding but within the intestinal sac. This deformity is believed to result when the normally developing intestine fails to return to the abdominal cavity.

Gastroschisis

Gastroschisis, on the other hand, is **characterized by the protrusion of the intestine from an anterior abdominal wall defect without any overlying sac**. Such a defect
is believed to result from excessive cell death or failure of cellular fusion to close the abdominal cavity during embryogenesis.

**Epidemiology of Omphalocele and Gastroschisis**

**Estimated incidences**

The estimated incidence of omphalocele and gastroschisis combined is 1 per 3500 live births, making them the most common congenital anomaly. Other epidemiological studies estimate the incidence of gastroschisis to be around 4.7 per 1000 neonatal intensive care unit (NICU) discharges and the incidence of omphalocele to be around 1 to 2 cases per 1000 NICU discharges. Large epidemiological studies have shown that the incidence of gastroschisis has been increasing over the last two decades, while the incidence of omphalocele is stable and constant.

**Environmental Exposures**

Omphalocele is reported to have an equal incidence among the different ethnic groups and in both sexes but is clearly associated with genetic predisposition and syndromic malformations. On the other hand, gastroschisis seems to be more common in Latinos and less common in African Americans. Genetic or familial cases of gastroschisis are rarely seen, suggesting that environmental factors might play a more vital role in the pathophysiology of gastroschisis. The incidence of gastroschisis is slightly higher in males compared to females, 1.5:1.

**Prognosis**

The prognosis of an infant with omphalocele is dependent on the presence and severity of the other congenital malformations that can involve any organ system. Infants with gastroschisis usually have less systemic congenital malformations likelihood compared to those with omphaloceles; however, they are more likely to develop intestinal inflammatory dysfunction, intestinal atresia, and malabsorption. Therefore, the prognosis of infants with gastroschisis is dependent on the size of the defect and the presence and severity of these associated complications.

Note: Mortality from omphaloceles and their associated congenital malformations is eight times higher compared to that of gastroschisis.

**Etiology of Omphalocele and Gastroschisis**

The exact etiology of omphaloceles and gastroschisis is poorly understood but several risk factors have been identified. **Folic acid deficiency and hypoxia are the two most commonly studied risk factors for abdominal wall defects** in experimental studies.

Despite our limited understanding of the etiology of these two common congenital malformations, elevated levels of maternal serum alpha-fetoprotein seem to correlate with the presence of ventral abdominal wall defects. Gastroschisis can be associated with polyhydramnios due to intestinal atresia.
Pathophysiology of Omphalocele and Gastroschisis

Pathology of omphaloceles

Babies who develop omphaloceles have a failure of intestinal return into the abdominal cavity. The intestine remains outside the abdominal cavity and confined to the umbilical ring. The exact etiology of omphalocele is unknown, but genetic predisposition is very likely to play a role in the pathology for several reasons:

- Omphaloceles are more commonly seen in trisomies, such as trisomies 13, 18 and 21.
- Omphaloceles have shown a high concordance in monozygotic twins and familial cases of omphalocele are commonly recognized.

Finally, omphaloceles are more common in the offspring of older women where genetic errors are more likely to occur.

Pathology of gastroschisis

The pathology of gastroschisis is a bit different from omphaloceles. The cause of the protrusion of the intestines in gastroschisis is a localized weakness in the abdominal ventral wall due to defective ingrowth, cellular death, or impaired cellular fusion. The intestines are in direct contact with the amniotic fluid which has been shown to play a key role in the long-term complications of gastroschisis, such as malabsorption.

In utero growth retardation, prematurity, and young maternal age are the most common risk factors for gastroschisis. While familial clustering has been suggested in some cases of gastroschisis, genetic predisposition is unlikely to play a key role in the pathology of gastroschisis.

Clinical Presentation of Omphalocele and Gastroschisis

Diagnosis of omphaloceles

Infants with ventral abdominal wall defects, such as omphaloceles or gastroschisis, are usually diagnosed prenatally by ultrasonography. When the diagnosis of omphaloceles is suspected, amniotic fluid examination for the exclusion of chromosomal abnormalities is indicated.

Omphaloceles might be centrally located, or might protrude from the epigastrium or the hypogastrium. Rupture of the omphalocele sac is common and can happen in up to one-fifth of the cases. Syndromic features are commonly seen in cases of omphaloceles. For instance, an infant with a small omphalocele who also has macroglossia, rounded facial features, neonatal hypoglycemia, and genitourinary abnormalities might have Beckwith-Wiedemann syndrome. These infants are also at an increased risk of hepatoblastoma, Wilms tumors, and adrenocortical neoplasms.

Another common association with omphaloceles is what is known as the pentalogy of Cantrell. The five components of this syndrome are an epigastric omphalocele, a cleft
sternum, an absent pericardium, the presence of an anterior diaphragmatic hernia of Morgagni, and the presence of certain cardiac defects, such as ectopia cordis and ventricular septal defects.

Diagnosis of gastroschisis

The abdominal wall defect in gastroschisis is usually right to the umbilical cord insertion and has a diameter that is less than 5 cm. Inflammation of the protruding intestines is common and can be severe. Severe intestinal adhesions might make it impossible to diagnose intestinal atresia.

When gastroschisis is diagnosed prenatally, repeated ultrasonographic follow-up examinations are indicated to exclude the emergence of any new complications such as polyhydramnios. Amniocentesis is indicated to measure fibronectin and monitor lung maturity. Depending on the size of the protrusion and the stage of lung maturity, the time and type of delivery are determined.

Diagnostic Workup for Omphalocele and Gastroschisis

MSAFP levels

The earliest finding of a possible structural fetal anomaly is the presence of an elevated level of maternal serum alpha-fetoprotein (MSAFP). MSAFP levels are elevated in gastroschisis and omphaloceles, but the levels are usually greater in gastroschisis. MSAFP levels are also elevated in neural tube defects. Neural tube defects cause elevated levels of MSAFP, acetylcholinesterase, and pseudocholinesterase, whereas abdominal wall defects cause elevations in the levels of MSAFP alone.

Ultrasonography

Once MSAFP levels are confirmed to be elevated, ultrasonography should be carried out to exclude the presence of any structural defects such as omphaloceles and gastroschisis. When abdominal wall defects are confirmed, an amniocentesis should be performed to exclude chromosomal abnormalities which are commonly seen in cases of omphaloceles. Fetal echocardiography is indicated to exclude the presence of cardiac anomalies, which are more commonly seen with omphaloceles than gastroschisis.

When the diagnosis of gastroschisis is confirmed prenatally, serial ultrasonography scans should be performed to monitor the state of the intestine. If the intestine becomes dilated and thickened, an amniocentesis should be performed to confirm lung maturity so that delivery can be performed as this is an ominous sign of intestinal inflammation.

Treatment of Omphalocele and Gastroschisis

The placement of a warm moist laparotomy pad or a dry pad over the exposed intestine is controversial. Instead, the focus should be directed towards wrapping the baby’s torso to avoid the pulling or kinking of the protruding intestines.

The administration of intravenous fluids and antibiotics should be started as
early as possible in all cases of omphaloceles and gastroschisis. Once the newborn is transferred to a tertiary care center, the decision to go for primary versus staged closure depends on the stage of the prematurity of the infant, the size of the defect, and the co-presence of other congenital malformations.

A small and intact omphalocele should be operated upon as early as possible with primary closure of the abdominal wall defect. Giant omphaloceles usually require a staged procedure because returning the omphalocele into the abdominal cavity in the primary procedure might cause severe elevations in the intra-abdominal pressure. Enteral feeding can be established very early after the closure of the defect.

Infants with gastroschisis usually have extensive inflammation of the protruding intestines and primary closure is usually not possible; therefore, often a silo is created to contain the eviscerated intestine for a limited period until the inflammation subsides. Once the intestines are soft and no longer inflamed, surgical reduction of the extruded viscera might be attempted. When the abdominal wall defect is closed, care must be taken to avoid a too-tight closure as this can impede venous return to the heart. Enteral feeding is usually delayed for up to 6 weeks until the passage of stools occurs, a sign of complete intestinal healing. During this period, total parenteral nutrition is indicated.

References


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