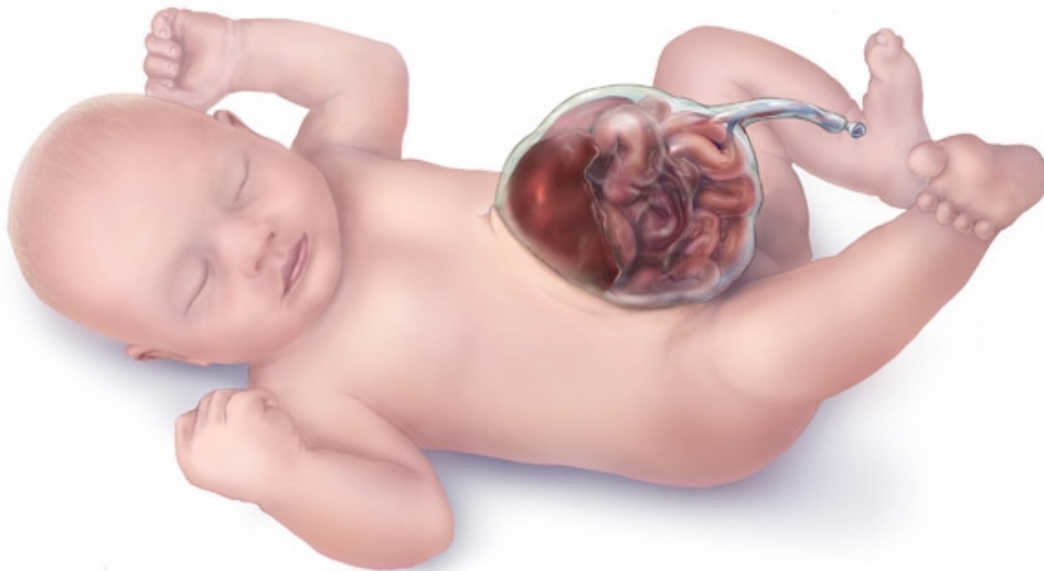


# Gastroschisis and Omphalocele: Diagnosis and Management

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**Omphalocele and gastroschisis are the most common congenital anomalies seen in infants and can be diagnosed prenatally or at birth. Omphaloceles 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** in which the intestine protrudes but remains within the intestinal sac. It is believed to occur when the normally developing intestine fails to return to the abdominal cavity.

### Gastroschisis

Gastroschisis refers to the **intestine protrusion without an overlying sac**. It is also an anterior abdominal wall defect and is believed to result from excessive cell death or failure of cellular fusion to close the abdominal cavity during embryogenesis.

# Epidemiology

## Estimated Incidence

**The combined estimated incidence of omphalocele and gastroschisis is 1 in 3,500 live births, making these conditions the most common congenital anomaly.** Other epidemiological studies estimate the incidence of gastroschisis to be around 4.7 per 1,000 neonatal intensive care unit (NICU) discharges and the incidence of omphalocele to be around 1–2 cases per 1,000 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.

## Environmental Exposures

**Omphalocele is reported to be equally common among different ethnic groups and in both sexes. However, it is clearly associated with genetic predisposition and syndromic malformations.** Gastroschisis seems to be more common in Latinos and less common in African Americans. Genetic or familial cases of gastroschisis are rare, suggesting that environmental factors have a greater influence on gastroschisis pathophysiology. The incidence of gastroschisis is slightly higher in males than females, at 1.5:1.

## Prognosis

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

Mortality from omphaloceles and associated congenital malformations is eight times higher than gastroschisis.

## Etiology

Although the exact etiology of omphaloceles and gastroschisis is poorly understood, experimental studies indicate risk factors for abdominal wall defects include **folic acid deficiency and hypoxia.**

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

## Pathophysiology

### Omphaloceles

**Babies who develop omphaloceles experience failure of intestinal return into**

**the abdominal cavity.** The intestine remains outside the abdominal cavity and confined to the umbilical ring. Genetic predisposition may play a role in the pathology for several reasons:

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

Omphaloceles are also more common in babies born to older women because genetic errors are more likely to occur due to maternal age.

## Gastroschisis

The pathology of gastroschisis is somewhat different from omphaloceles. **The cause of gastroschisis is a localized weakness in the abdominal ventral wall** due to defective growth, cellular death, or impaired cellular fusion. The intestines are in direct contact with the amniotic fluid, which plays 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 was indicated in some cases of gastroschisis, genetic predisposition is unlikely to play a key role in this condition's pathology.

## Clinical Presentation

### Omphaloceles

**Infants with ventral abdominal wall defects, such as omphaloceles or gastroschisis, are usually diagnosed prenatally by ultrasonography.** When omphaloceles is suspected, the amniotic fluid should be examined to rule out chromosomal abnormalities.

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

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

### Gastroschisis

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

**When gastroschisis is diagnosed prenatally, repeated ultrasonographic follow-**

**up examinations are indicated to rule out any new complications**, such as polyhydramnios. Amniocentesis is indicated to measure fibronectin and monitor lung maturity. Depending on the protrusion size and the stage of lung maturity, the time and type of delivery may need to be determined.

## Diagnostic Work-up

### Maternal Serum Alpha-Fetoprotein Levels

**The earliest indication of a possible structural fetal anomaly is an elevated level of maternal serum alpha-fetoprotein (MSAFP).** Although MSAFP levels are elevated in both gastroschisis and omphaloceles, they are usually higher in gastroschisis. MSAFP levels are also elevated in neural tube defects, which cause elevated levels of acetylcholinesterase and pseudocholinesterase. Abdominal wall defects cause elevations in the MSAFP levels 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 common in omphaloceles than gastroschisis.

**When gastroschisis is diagnosed 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; this is an ominous sign of intestinal inflammation.

## Treatment

Whether to place a warm moist or dry laparotomy pad over the exposed intestine is controversial. Instead, attention should be directed toward wrapping the baby's torso to avoid pulling or kinking the protruding intestines.

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

**A small and intact omphalocele should be operated on as early as possible** with primary closure of the abdominal wall defect. **Giant omphaloceles usually require a staged procedure;** returning the omphalocele to the abdominal cavity in the primary procedure can elevate the intra-abdominal pressure. Enteral feeding can be established very early after the closure of the defect.

As infants with gastroschisis usually have extensive inflammation of the protruding intestines, so primary closure is usually impossible. Therefore, **a silo is often 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 may be attempted. When the abdominal wall defect is closed, care must be taken to avoid a too-tight closure, which could impede venous return to the heart. Enteral feeding is usually delayed for up to six weeks until the infant can pass stools, a sign of complete intestinal healing. During this period, total parenteral nutrition is indicated.

## References

Glasser J. Pediatric Omphalocele and Gastroschisis. Background, Pathophysiology, Etiology. <http://emedicine.medscape.com/article/975583>. Published March 9, 2017. Accessed April 30, 2017.

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