GASTROSCHESES

Gastroschisis refers to a full-thickness paraumbilical abdominal wall defect associated with evisceration of fetal intestine. Widespread use of antenatal ultrasound examination and maternal Quad Screening has enhanced rates of antenatal diagnosis and brought diagnosis into the first trimester in many cases.

The incidence of gastroschisis varies from 1 to 5 per 10,000 live births and is similar in male and female fetuses.

Several hypotheses have been proposed to explain the pathogenesis of the gastroschisis; all involve defective formation or disruption of the body wall in the embryonic period, with subsequent herniation of bowel:

- Failure of mesoderm to form in the body wall
- Rupture of the amnion around the umbilical ring
- Abnormal involution of the right umbilical vein leading to weakening of the body wall
- Disruption of the right vitelline artery with subsequent body wall damage
- Abnormal folding of the body wall resulting in a ventral body wall defect

Almost all cases of gastroschisis are associated with an elevated maternal serum alpha fetoprotein (MSAFP) level, thus, the possibility of gastroschisis, as well as other fetal anomalies, should be considered in pregnancies with elevated second trimester MSAFP on Quad Screening. The adjacent umbilical cord insertion site should be normal. The intestine usually is the only herniated organ; other intra-abdominal organs rarely pass through the defect. The intestinal mass lacks a covering sac and floats freely in the amniotic fluid.

In contrast to omphalocele, gastroschisis is not generally associated with malformations outside of the gastrointestinal tract. A study of pooled data from 24 international birth defects registries including over 3300 cases of gastroschisis concluded that about 10 percent were associated with major unrelated defects. About 2 percent of cases were part of a recognized syndrome. Cardiac defects were detected in 2 to 3 percent of cases, however, this may be due to ascertainment bias.

Additional gastrointestinal problems (eg, malrotation, atresia, stenosis) are present in up to 25 percent of cases and are probably related to vascular disruption caused by herniated bowel. As an example, disruption of the superior mesenteric artery may lead to volvulus or to "apple peel" jejunal-ileal lesions. Meckel's diverticulum and gallbladder atresia also occur, but are less common.
The prevalence of chromosomal abnormalities in fetuses with isolated gastroschisis is not increased above the baseline population risk. But chromosomal abnormalities rise in cases with other associated structural abnormalities.

There is an increased risk of spontaneous preterm delivery; nevertheless, over 70 percent of pregnancies complicated by gastroschisis complete 37 weeks of gestation.