DEVELOPMENTAL PROBLEMS ASSOCIATED WITH THE GI SYSTEM

Omphalocele

This term refers to the presence of intestine in the umbilical cord at birth. It results from failure of the midgut to return from its physiologic umbilical herniation. The herniated gut is covered by the thin outer layer of the umbilical cord, which is more like amniotic membrane than skin. As a consequence, during the pregnancy there is an abnormally long period during which certain internal organs of the fetus are not separated from amniotic fluid by skin. This allows alpha-fetoprotein to leak into the amniotic fluid in sufficient quantity to be detected in the maternal blood serum. However, omphalocele is also detectable by ultrasound imaging of the fetus.

Omphalocele is often associated with cardiac and renal anomalies, and for this reason it has a poor prognosis.

Congenital Umbilical Hernia

This term refers to herniation of some part of the bowel through the umbilicus, differing from omphalocele in that the herniated structures are covered by skin, suggesting that the midgut had returned normally but some part of the bowel later herniated. A congenital umbilical hernia is usually smaller than an omphalocele. It needs surgical correction but is not as ominous as omphalocele.

Gastroschisis

This condition arises when, during the period of physiologic midgut herniation, the umbilical cord ruptures and the midgut become exposed directly to amniotic fluid. The umbilical cord will heal, but at birth part of the bowel protrudes through a gap in the anterior abdominal wall. As with omphalocele, alpha-fetoprotein may leak into the amniotic fluid in sufficient quantity to be detected in the maternal blood serum, but the condition is also detectable by ultrasound imaging of the fetus.

From its description, gastroschisis sounds like a very serious condition, but it is less serious than omphalocele because it is not associated with other developmental anomalies. It is true that in gastroschisis the portion of the bowel exposed to amniotic fluid becomes inflamed and will not exhibit normal peristalsis until it is replaced in the abdomen and the inflammation subsides. In some cases, the involved segment of bowel may need to be excised.

## Meckel's Diverticulum / Umbilico-ileal fistula

Normally the vitelline duct and yolk sac completely degenerate. Sometimes the part of the vitelline duct closest to the midgut persists as a blind outpocketing (usually 1 -2 inches long) from the antimesenteric border of ileum 2 feet proximal to the ileocecal valve This blind outpocket is called a Meckel's diverticulum. Meckel's diverticulum is asymptomatic more than 90% of the time. However, if it can become infected, like an appendix, and Meckel's diverticultis may be misdiagnosed as appendicitis. A Meckel's diverticulum may also contain aberrant gastric and/or pancreatic tissue. The secretions of such tissues can irritate the adjacent ileum and lead to GI bleeding in early childhood.

If more of the vitelline duct persists and stays patent, it may form a tube that reaches all the way to the umbilicus, where it opens onto the skin of the anterior abdominal wall as an umbilico-ileal fistula.

Atresias and Stenoses

I don't know why, but in normal development the epithelial (i.e., endodermal) lining of the gut tube proliferates to fill its lumen, then a new lumen forms through a process called recanalization. Improper recanalization can lead to atresia (absence or closure of a normal opening or tube) or stenosis (narrowing of the lumen of a tube). Some people think atresias of the jejunum and ileum are not due to a recanalization problem, but are caused by localized regions of vascular insufficiency. The distal ileum is the most common site of small bowel atresia.

Although an esophageal atresia can exist as an independent entity, most are associated with an open connection (fistula) between the abnormal esophagus and the trachea. The most common type of tracheo-esophageal fistula is illustrated below.

Modified from
Larsen WJ (1993)
"Human Embryology"
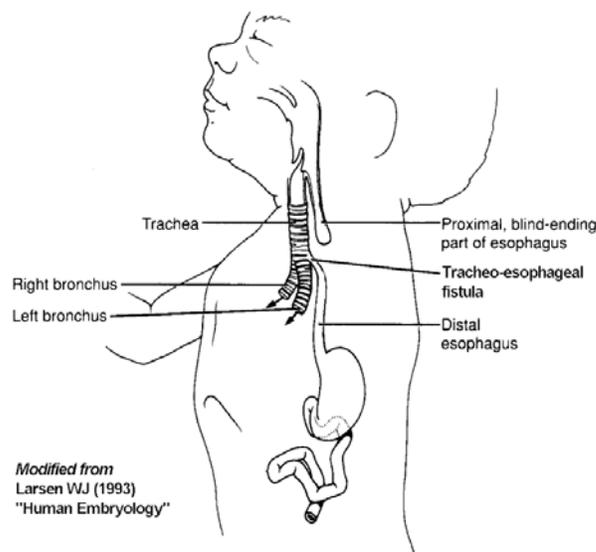
As you may already have read, esophageal atresias are associated with polyhydramnios. Poly-hydramnios occurs 60% of time in duodenal and proximal jejunal atresias. In severe congenital duodenal stenosis (not as serious as atresia), the infant will be feeding-intolerant. Milder cases may go undetected during the postnatal period when milk is the sole source of food, and only become apparent when solid food becomes a part of the infant's diet.

Malrotations

Improper rotations of the bowel segments may be associated with anomalous mesenteric bands (Ladd's bands) that can obstruct the bowel. More serious is the possibility of volvulus (twisting of the bowel upon itself) with vascular obstruction.

Subhepatic Cecum

The cecum may fail to separate from the liver with the result that there is no transformation of the oblique colon into ascending and transverse colons. The major significance of a subhepatic cecum is that it gives an unusual location of symptoms if you get appendicitis.

Annular Pancreas

This condition arises when the ventral pancreatic diverticulum abnormally bifurcates, with one branch passing behind the duodenum, as is normal, but the other branch passing anterior to the duodenum. The two portions fuse, with the result that the head of the pancreas encircles the second part of the duodenum and produces a duodenal obstruction that must be surgically corrected by a duodenoduodenostomy.

Pancreas Divisum

This term refers to failure of fusion of the ventral and dorsal pancreatic ducts. Standard teaching is that it occurs about 10% of the time and is associated with a higher incidence of pancreatitis. ERCP (endoscopic retrograde cholangiopancreatography) has shown it to be more common than previously thought, casting doubt on its association with any pathology.

Hirschsprung's Disease (Congenital Megacolon) - 1/5000 births, 80% in males

Hirschsprung's disease arises when a portion of the large intestine is not invaded by neural crest cells that form the parasympathetic postganglionic neurons normally residing in the wall of the bowel. Hence, a portion of the large intestine is aganglionic. No matter where the aganglionic segment starts, it always extends to the anus. Ninety-five percent of the time it starts distal to the splenic flexure, and in the vast majority of these cases (85% of the time overall) it is confined to the sigmoid colon and rectum. The aganglionic portion of the colon is aperistaltic, producing the following symptoms in the infant: abdominal distension (because of dilatation of normal colon proximal to the obstruction posed by the involved segment), delayed passage of meconium, constipation, and painful defecation. If the aganglionic area is short, the condition may not be diagnosed until the child is older. Definitive diagnosis is made by rectal biopsy. Hirschsprung's disease is treated surgically by excision of the aganglionic segment.