Overview of Gastrointestinal Embryology
The Primitive Gut

- The primitive gut forms during the 4th week of gestation when the flat embryonic disc folds in median and horizontal planes to form a tubular structure that incorporates part of the yolk sac into the embryo.

- Ventral folding of lateral sides forms the midgut.

- Ventral folding of cranial and caudal ends (head and tail folds) form the foregut and the hindgut.
Folding of the Embryonic Disc

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Formation of human gastrointestinal tract (A)

Image by MIT OCW.
The Foregut

• The foregut gives rise to the:
  – Pharynx
  – Lower respiratory system
  – Esophagus
  – Stomach
  – Proximal duodenum
  – Liver and the biliary tree
  – Pancreas
Partitioning of the foregut by the tracheoesophageal septum

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Errors of the Foregut Development

Errors in partitioning of the laryngo-tracheal tube from the esophagus by the tracheo-esophageal septum result in various forms of esophageal atresia and tracheo-esophageal fistulas or EA/TEF (1 in 3000-4500 live births, M>F)
Tracheoesophageal fistula

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Development of the Lumina

Epithelial Plug → Vacuolization → Recanalization
Abnormal Development of the Lumina

Epithelial Plug → Vacuolization → Recanalization

- Atresia
- Stenosis
- Duplication, Cyst or Septum
Jejunoileal atresia

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Congenital Hypertrophic Pyloric Stenosis
Errors of the Foregut Development

- **Congenital Hypertrophic Pyloric Stenosis** is the most common congenital anomaly of the stomach and occurs in 1-8:1000 live births with a 4-6:1 M:F ratio.

- Pyloric stenosis is a multifactorial and progressive disease that classically presents with non-bilious projectile vomiting in the first few weeks of life.
The Liver and the Pancreas

Figure removed due to copyright reasons. Please see:

Errors in Pancreatic Development

- Annular pancreas
- Pancreas divisum
- Ectopic pancreatic tissue
The Midgut

- The midgut gives rise to:
  - Distal duodenum
  - Jejunum and ileum
  - Appendix
  - Ascending colon
  - Proximal transverse colon
Epithelial cytodifferentiation

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Intestinal epithelial differentiation

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The Midgut Rotation

- The midgut forms a U-shaped loop that herniates into the umbilical cord during the 6th weeks of gestation.

- While in the umbilical cord, the midgut loop rotates 90 degrees.

- During the 10th week of gestation, the midgut loop returns to the abdomen, rotating an additional 180 degrees.
Errors in Midgut Rotation

Anything can happen, but it usually doesn’t!
Errors in Midgut Development

- *Omphaloceles* result from failure of the intestines to return to the abdominal cavity

- *Umbilical hernias* occur when intestines do return to the abdomen, but later herniate *through the umbilicus*

- *Gastroschisis* is a linear defect of the abdominal wall that permits extrusion of the viscera *without involving the umbilicus*
Infant with gastroschisis

Image removed due to copyright reasons.
Infant with omphalocele

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<table>
<thead>
<tr>
<th></th>
<th>Gastroschisis</th>
<th>Omphalocele</th>
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</thead>
<tbody>
<tr>
<td>Maternal age</td>
<td>Younger</td>
<td>Older</td>
</tr>
<tr>
<td>Associated anomalies</td>
<td>10% (usually intestinal atresia)</td>
<td>50% (structural and chromosomal)</td>
</tr>
<tr>
<td>Sac covering abdominal contents</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>Liver out through abdominal wall defect</td>
<td>No</td>
<td>Often</td>
</tr>
<tr>
<td>Intestinal dysfunction (hypomotility and malabsorption)</td>
<td>Yes</td>
<td>No</td>
</tr>
</tbody>
</table>
Remnants of the omphalomesenteric duct (yolk stalk) are found in 1-4% of infants, making them the most common congenital anomaly of the GI tract.

Figure removed due to copyright reasons. Please see:

Meckel’s Diverticulum

- Meckel’s or ileal diverticulum accounts for up to 80% of omphalomesenteric remnants

- Typical Meckel’s diverticulum measures 3-5 cm, and is located in the anti-mesenteric wall of the ileum 40-50 cm from the ileocecal valve

- Most symptomatic cases present in childhood

- The M:F incidence ratio is ~1, but there is a 3:1 M:F ratio in clinically symptomatic cases
The Hindgut

• The hindgut gives rise to:
  – Distal transverse colon
  – Descending colon, sigmoid, and rectum
  – Proximal anal canal (superior to the pectinate line)

• The caudal part of the hindgut known as the *cloaca*, is divided by the urorectal septum into the urogenital sinus and the rectum
Partitioning of the Cloaca by the Urorectal Septum

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Errors in Hindgut Development

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Hirschsprung’s Disease

• HD is the partial or total absence of autonomic ganglia resulting from failure of migration of the neural crest cells into the colonic wall during 5th-7th week of gestation

• With an incidence of 1 in 5000 liver births, HD is the most common cause of neonatal colonic obstruction, and can result in *congenital megacolon*

• HD has been associated with several genetic abnormalities including Trisomy 21, mutations of the RET gene and the endothelin receptor type B gene
Intraoperative photograph of Hirschsprung's disease

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Hirschsprung’s Disease

Figure removed due to copyright reasons. Please see:

Hirschsprung’s- submucosal plexus

Image removed due to copyright reasons.
Three common operations for Hirschsprung's disease

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