# Congenital GI Disorders

## Developmental Remnants/Heterotopic Tissue

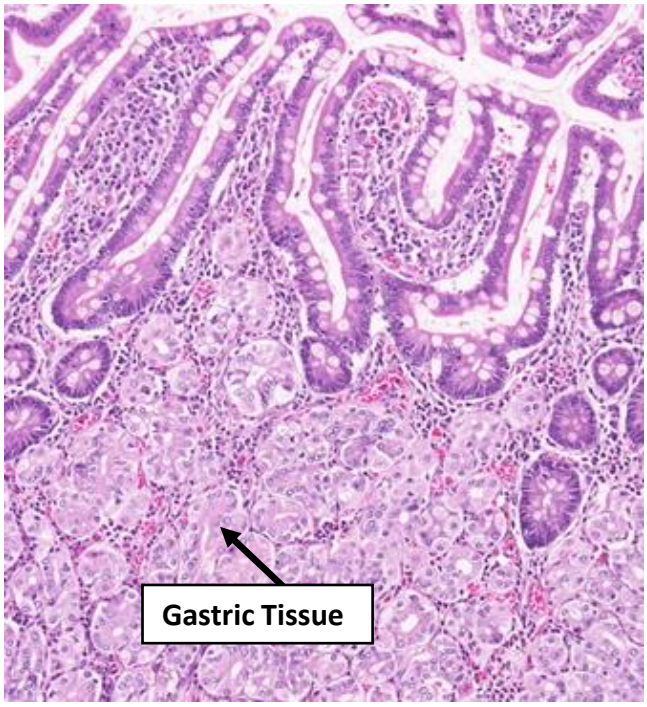
### Meckel's Diverticulum

**Omphalomesenteric (vitelline) duct remnant**
In **Ileum**. Contains all layers of bowel

*"Rule of 2's"*
~2% of population
~2 inches in length
~2 feet from the ileocecal valve
2 types of common heterotopia (gastric and pancreatic)
Most common presentation is ≤ 2 yrs, usually with GI bleeding→ "currant jelly stool" (but can be any age!)

Can diagnose with a "Meckel's [nuclear medicine] Scan" (Technetium-99m pertechnetate scintigraphy)

Can cause issues through different mechanisms:
-Lead point for intussusception or volvulus
-Ectopic gastric mucosa acid→ ulcers; Diverticulitis
-Rarely develop malignancy




***Umbilical polyp***—rarer vitelline duct remnant with intestinal mucosa in the soft tissue of the umbilicus

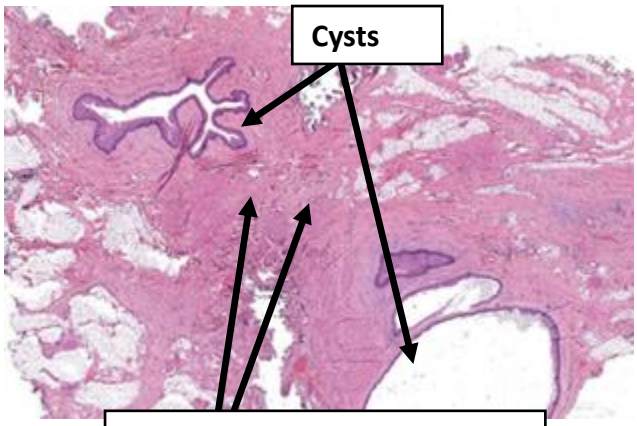
### Tailgut Cyst

*aka "Retrorectal cystic hamartoma"*

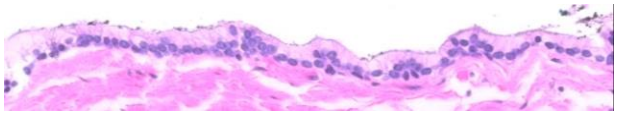
Remnants of embryonic **postanal gut**
Cystic lesion in presacral (retrorectal) potential space
Multilocular, well-circumscribed

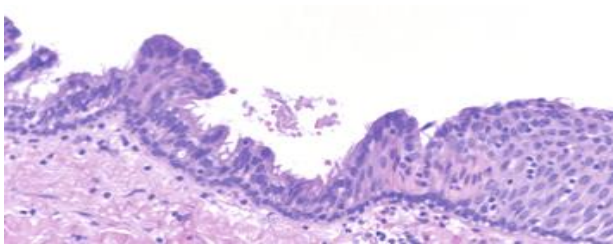
**Lined by *any* GI tract or transitional epithelium**
(most common = squamous)
**Disorganized, smooth muscle bundles**
(No well-organized muscle or nerves)

*May* cause symptoms from mass effect
*May* undergo malignant transformation, rarely.

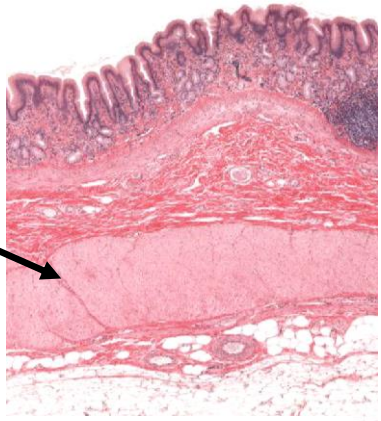



**DDX:**
*Cystic sacrococcygeal teratoma*→ should have ≥ 2 germ layers. Newborns.
*Rectal/enteric duplication cyst*→ Well-formed, double muscle layer and nerve plexus (like normal gut)
*Epidermoid/dermoid cyst*→ Squamous lining, No smooth muscle.



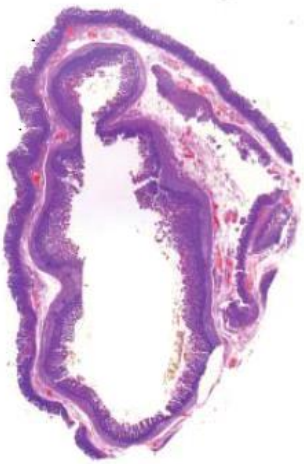


# Duplication Cysts



Usually single
Can be Cysts (no communication with lumen) or Tubular (communicate with lumen, often running parallel, a "true" duplication)
**Well-developed, double smooth muscle layer with nerve plexus**
*(Think: normal bowel…just extra)*

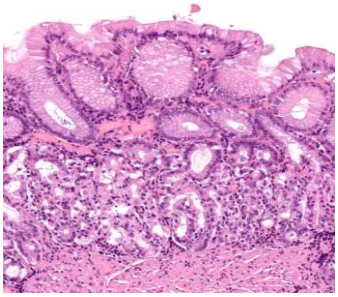
**Esophageal Duplication Cyst**
Located within or attached to esophagus.
Can be lined by gastric, squamous, intestinal, pancreatic, or respiratory mucosa
*vs Bronchogenic cyst*→ respiratory epithelium, cartilage, seromucinous glands
Can just call "Foregut cyst" if it's unclear esophageal vs bronchogenic (they're embryologically related)



**Small Intestine Duplication Cyst**
Located on the mesenteric side of small bowel
Usually enteric mucosa, but may have gastric or pancreatic

**Colonic Duplication Cyst**
Least common. Usually colon lining, but can have heterotopia.

# Inlet Patch/Heterotopic gastric mucosa

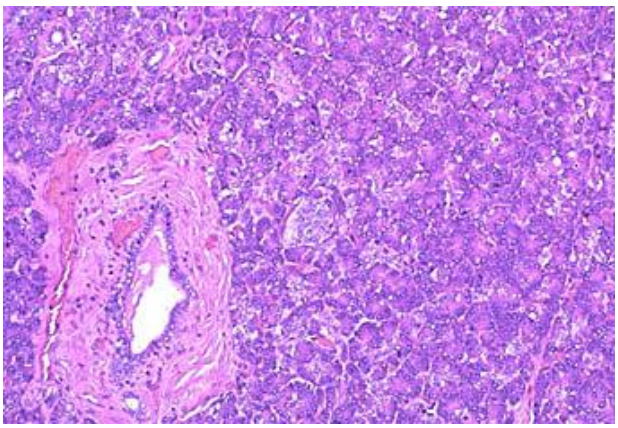


**Inlet Patch→ Heterotopic gastric columnar mucosa in cervical esophagus**
Thought to be incomplete replacement by squamous epithelium
Asymptomatic usually. Usually fundic-type mucosa with Parietal and Chief cells.
Requires endoscopic correlation
*vs Barrett's mucosa*→ at GEJ with intestinal metaplasia.

Most common site of heterotopic gastric mucosa is duodenal bulb (appears polypoid)
*vs Peptic duodenitis/foveolar metaplasia*→ lacks parietal and chief cells (just surface foveolar epithelium)

**Note: You can see heterotopic stomach in pretty much *any* part of the GI tract, including the colon!**

# Ectopic Pancreas



**Pancreatic tissue ductal and/or acinar tissue** that has no connection to the pancreas
Most common sites: **Stomach**, Duodenum, Jejunum
**Submucosal mass** (mistake for GIST clinically)

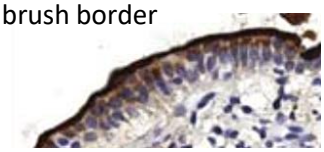
Usually asymptomatic, incidental finding.
Rarely, can develop abscess, pseudocyst, tumor, etc…
If in second part of duodenum, may represent minor papilla!

# Congenital Enteropathies

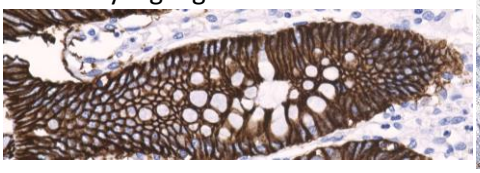
Present with **chronic, intractable diarrhea** and failure to thrive, usually within first days/months of life. Based on duodenal biopsies. Great algorithm with recommended IHC panel PMID: 25188866

**Recommended IHC Panel: BerEP4, CD10, Chromogranin**

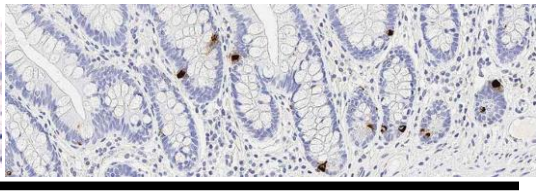
**CD10**
Normally highlights brush border



**BerEP4**
Normally highlights membrane

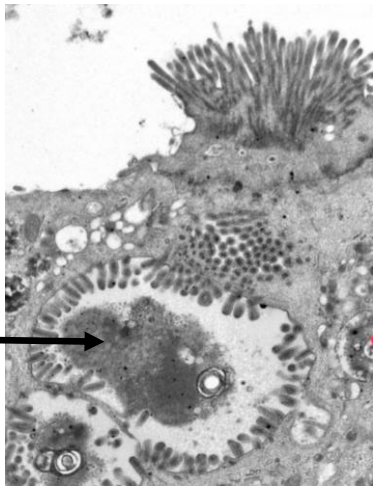


**Chromogranin**
Normally highlights scattered NE cells



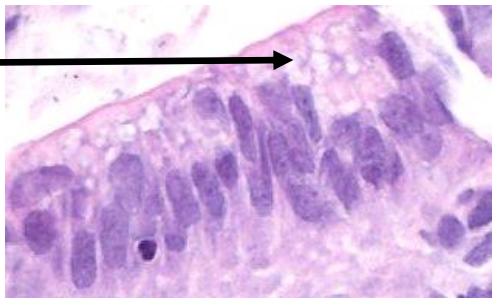
## Microvillous Inclusion Disease

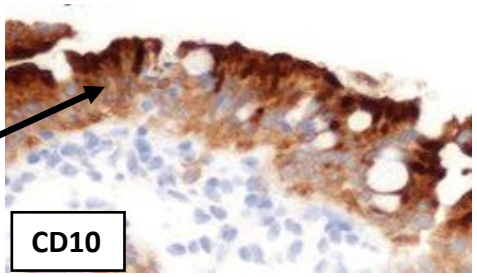
Mutation in **MYO5B gene** (Autosomal Recessive)
→ dysfunctional myosin motor→ abnormal vesicle trafficking.
**Atrophic microvilli with microvillous inclusions in cytoplasm**
→ Poor absorption→ **severe diarrhea from birth**→ often require TPN
Abnormal vesicle trafficking also associated with Meckel's diverticulum, renal dysplasia, etc…



Electron microscopy: Shows microvillous inclusions.

**Small cytoplasmic vacuoles in cytoplasm of enterocytes**.
Variable villus atrophy and inflammation.



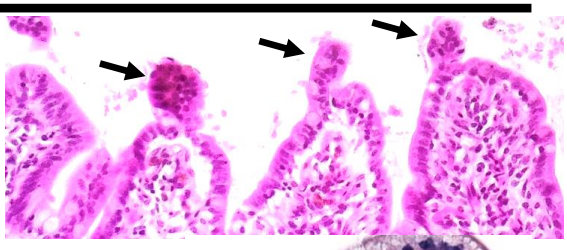


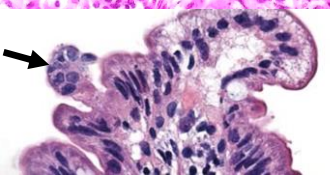

Stains: **CD10** (and PAS) Double band at brush border with **apical inclusion staining** (Normal = brush border only)

## Tufting Enteropathy

**Mutation in EpCAM** (Epithelial Cell Adhesion Molecule—involved in tight junctions)

**Villous atrophy with "Tufts" of rounded, teardrop-shaped enterocytes that appear to shed into the lumen**





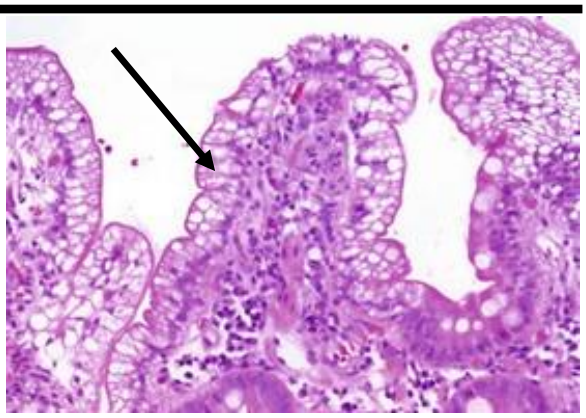
IHC: ***Loss*** **of BerEP4** (and MOC31) staining (recognize EpCAM)

## Endocrine Cell Dysgenesis

**NEUROG3 mutation** (Autosomal recessive)
**Absence of enteroendocrine cells→ IHC: *Negative for Chromogranin A***
Normal to mildly blunted villi

# Other Bowel Diseases

## Lipid Trafficking Disorders

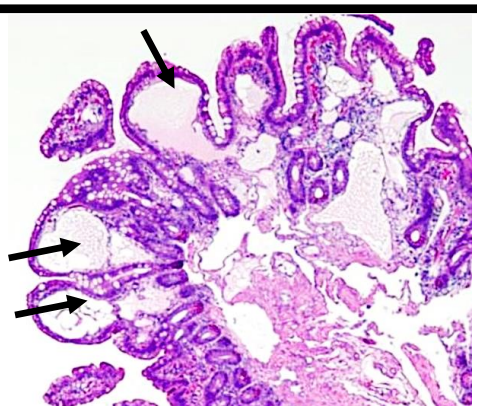


Mutations cause impaired lipid trafficking from lumen through enterocytes to chylomicrons in blood→ Lipids build up in enterocytes→ **Characteristic vacuolization of enterocytes** and **fat malabsorption/diarrhea**

Examples: Abetalipoproteinemia, Hypobetalipoproteinemia, Chylomicron Retention Disease

**Note**: *Some* enterocyte vacuolization can be seen in infants normally, particularly after recent feeds, but it should not be too dramatic.

## Lymphangiectasia



**Dilated lacteals in mucosa.**

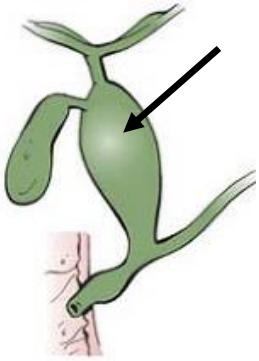
If primary, Congenital lymphatic obstruction/malformation (can also be secondary to cardiac issues or localized fibrosis, etc…)

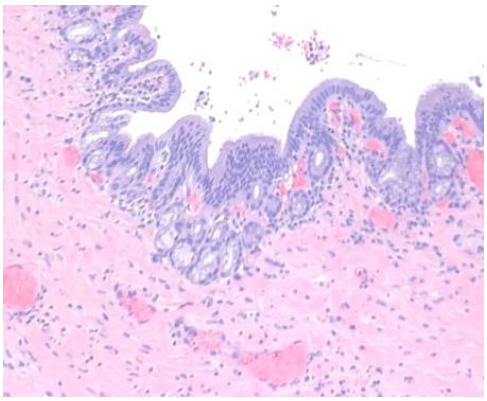
Present with: **Diarrhea, Protein losing enteropathy** (PLE), **Hypoalbuminemia**, Hypogammaglobulinemia, and lymphopenia (resulting in **immunosuppression→ infections**)

Endoscopically can see white dots (lacteals)

# Biliary Diseases

## Choledochal Cyst





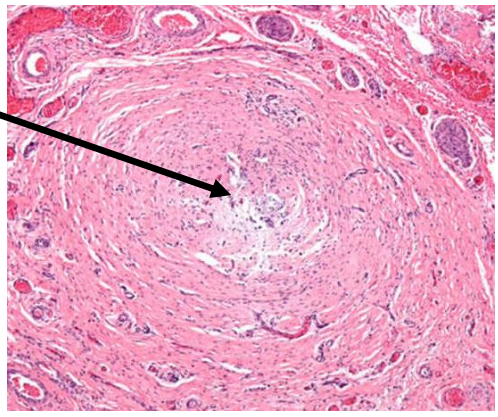
**Dilation of the biliary tree**

Anomalous pancreatobiliary junction→ reflux of pancreas secretions up biliary tree→ dilation → obstructive jaundice and mass

Cyst wall: Fibrous tissue, biliary lining, inflammation

Treat with surgery to prevent obstruction (and risk of cancer)

## (Extrahepatic) Biliary Atresia



**Fibroinflammatory destruction of the extrahepatic bile ducts** → cholestasis → biliary cirrhosis

Treat first with Kasai procedure (hepatic portoenterostomy) as a bridge to transplantation often. Most common indication for liver transplantation in infants.

Liver biopsies demonstrate finding consistent with large duct obstruction (cholestasis, bile duct reaction, portal edema, acute inflammation)