

CdLS Foundation

GI Challenges in CdLS

Carol Potter, MD

Nationwide Children's Hospital

July 31, 2019



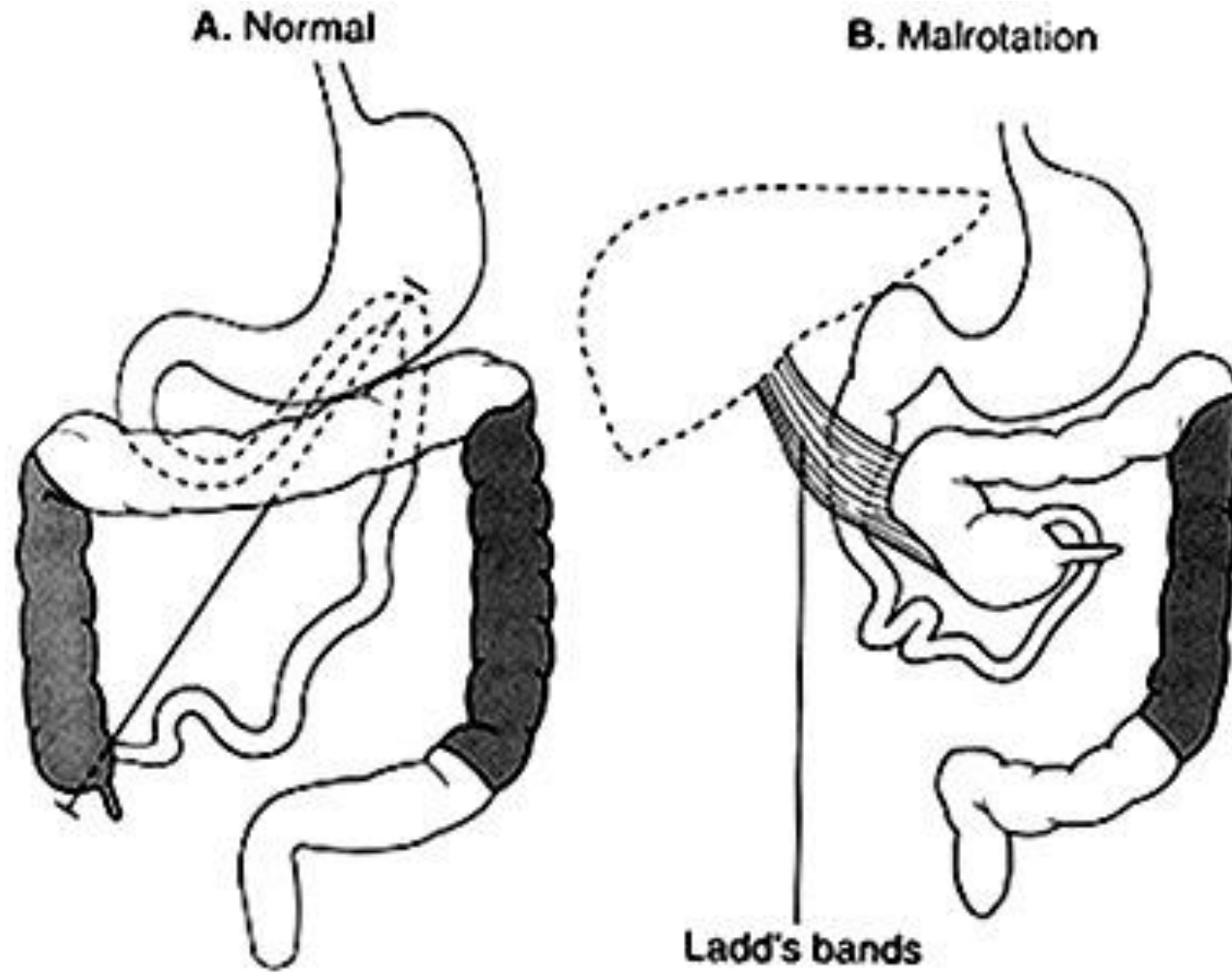
CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

Objectives

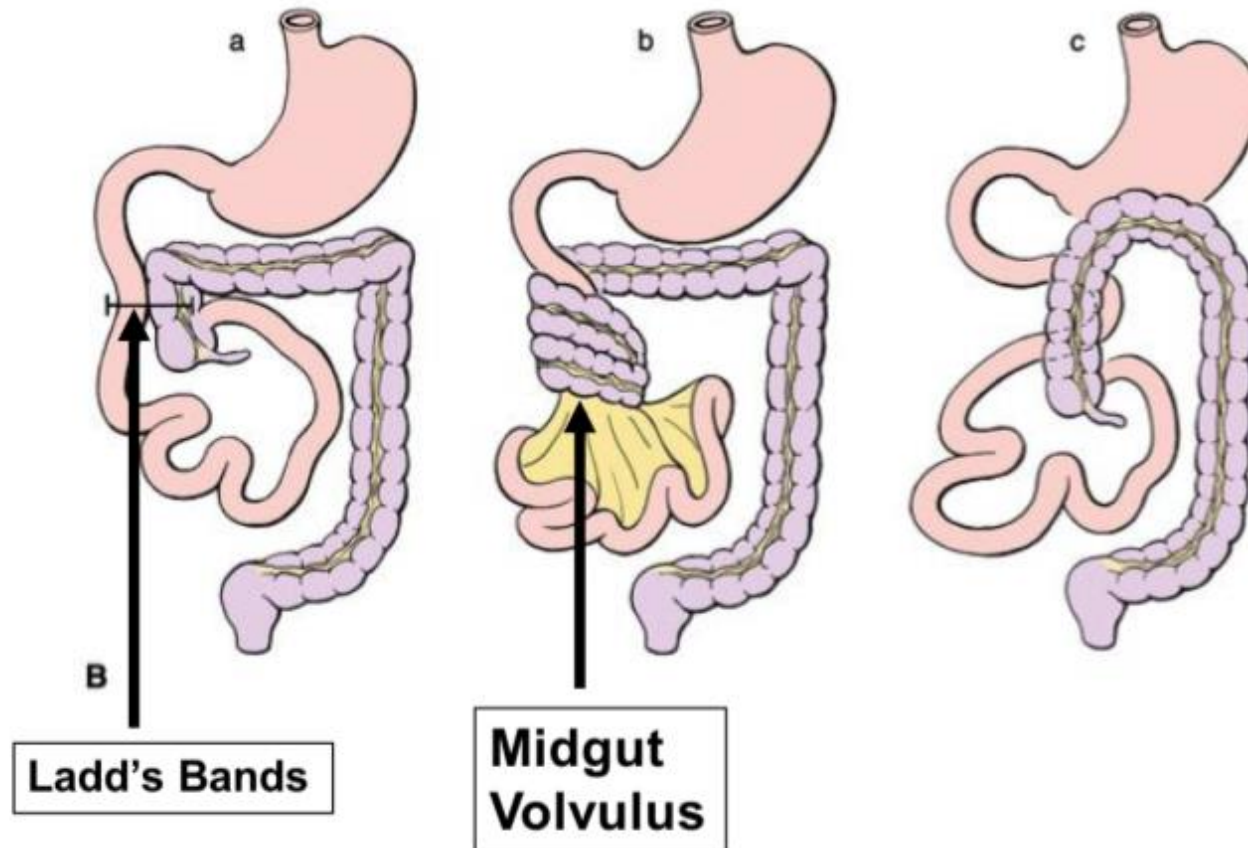
- Malrotation
- Bowel Obstruction
- Constipation
- Pancreatitis

Malrotation

- Abnormal attachment of the intestine to the back abdominal wall
- Allows twisting of the small intestine around the artery and bowel
- Present in 5% of patients with CdLS



MALROTATION AND MIDGUT VOLVULUS



Chronic Volvulus

- Twisting of the bowel enough to cause partial obstruction and vomiting
- Intestine is still viable
- May wax and wane
- Should be immediately fixed

Acute Volvulus

- Complete obstruction of the bowel with ischemia
- Medical Emergency
- Pain, vomiting, very ill
- May result in loss of small intestine



Diagnosis of Malrotation

- Barium studies show malposition of the first part of the small intestine
- Some benign variants may mimic malrotation on UGI
- Most vomiting in CdLS is not going to be malrotation
- You have malrotation or you don't---it doesn't develop



Therapy for Malrotation

- Varies by country
- If associated with vomiting and volvulus on UGI
 - Urgent surgery
- If no vomiting and no volvulus
 - Some elect to wait
- If acutely ill
 - Surgical emergency with out UGI

Questions?

Bowel Obstruction

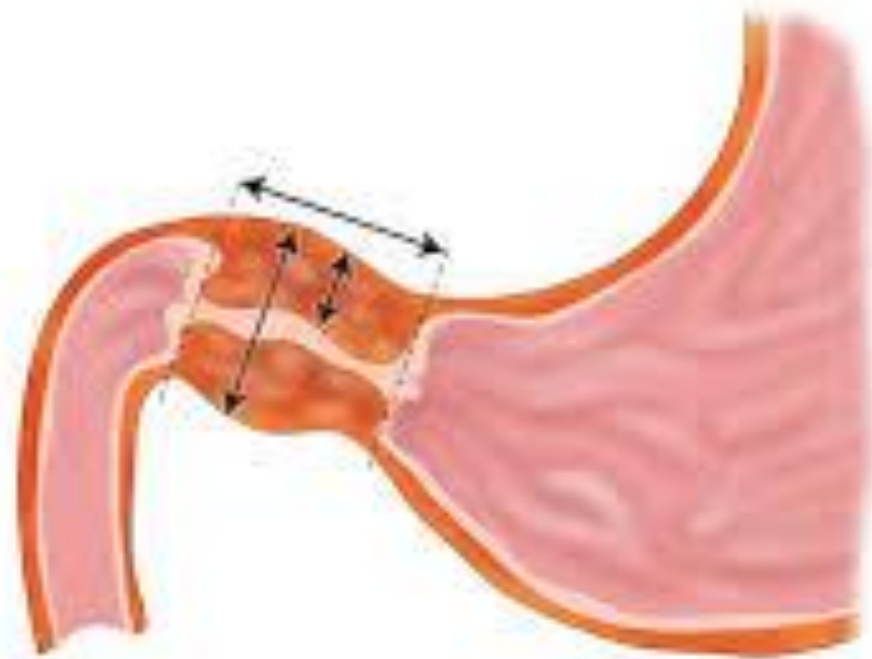
- Congenital Malformations
- Post surgical complications
- Tube related obstruction
- Constipation

Congenital GI Malformations

- More common in CdLS
- May present with vomiting and feeding intolerance
- Radiology often helpful
- High Index of Suspicion

Pyloric Stenosis

Pyloric stenosis



Normal values *

- Length < 1.5cm
- Single muscle thickness < 0.3cm
- Pyloric width < 1.5cm

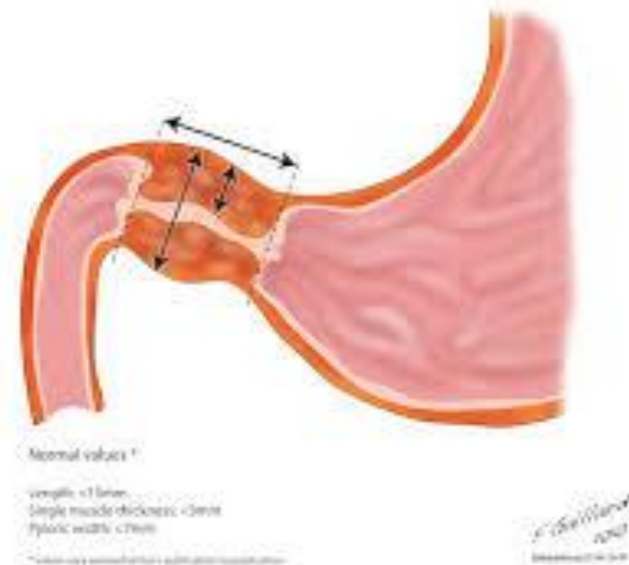
*Values vary somewhat between public and private institutions

© Williams & Wilkins

Pyloric stenosis

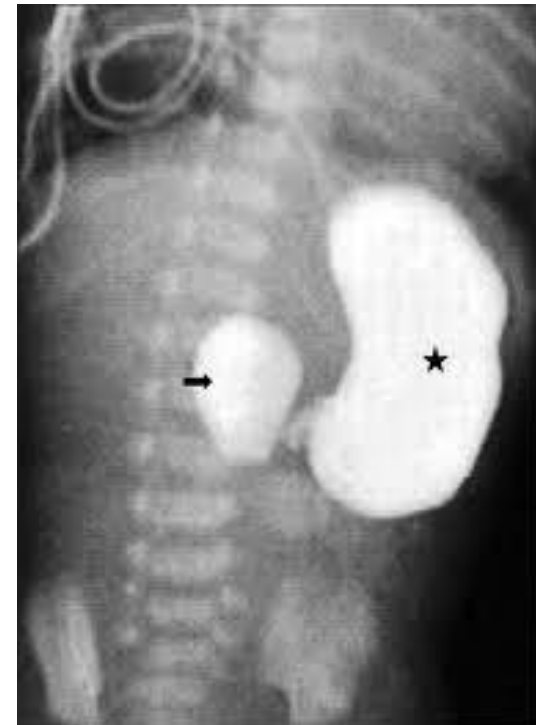
- Hypertrophy of muscle
- In very young babies
- Relentless vomiting
- Diagnosis with US
- Treated with surgery

Pyloric stenosis



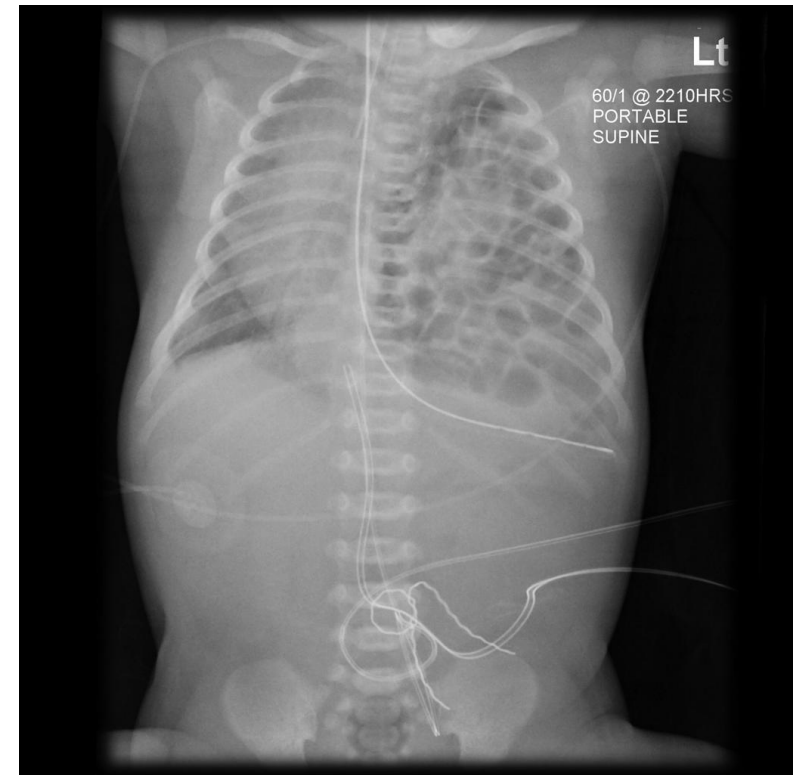
Duodenal Atresia

- Presents at birth
- May be associated with pancreas abnormalities
- Requires surgical intervention



Diaphragmatic Hernia

- Bowel is in the chest
- Small ones may not be obvious at birth



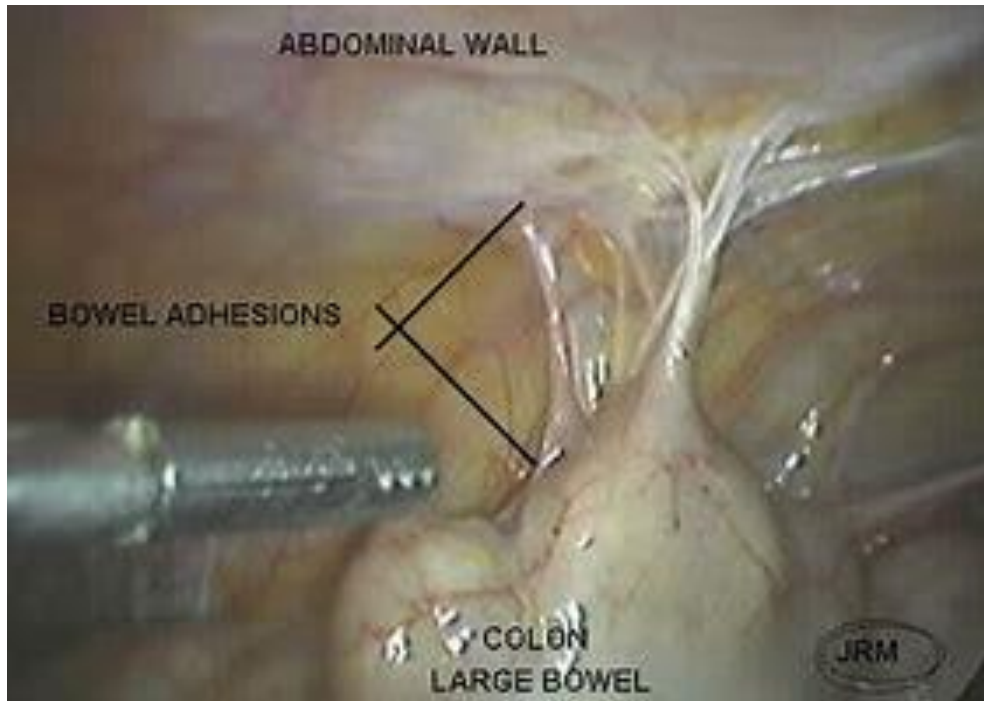
Cecal Volvulus



Post Surgical Complications

- Adhesions
 - Scar from surgery
 - May block bowel
 - The more you fix it the worse it is
- Hernia
- Infections
- Perforations
- Tube Complications

Adhesions



Hernia

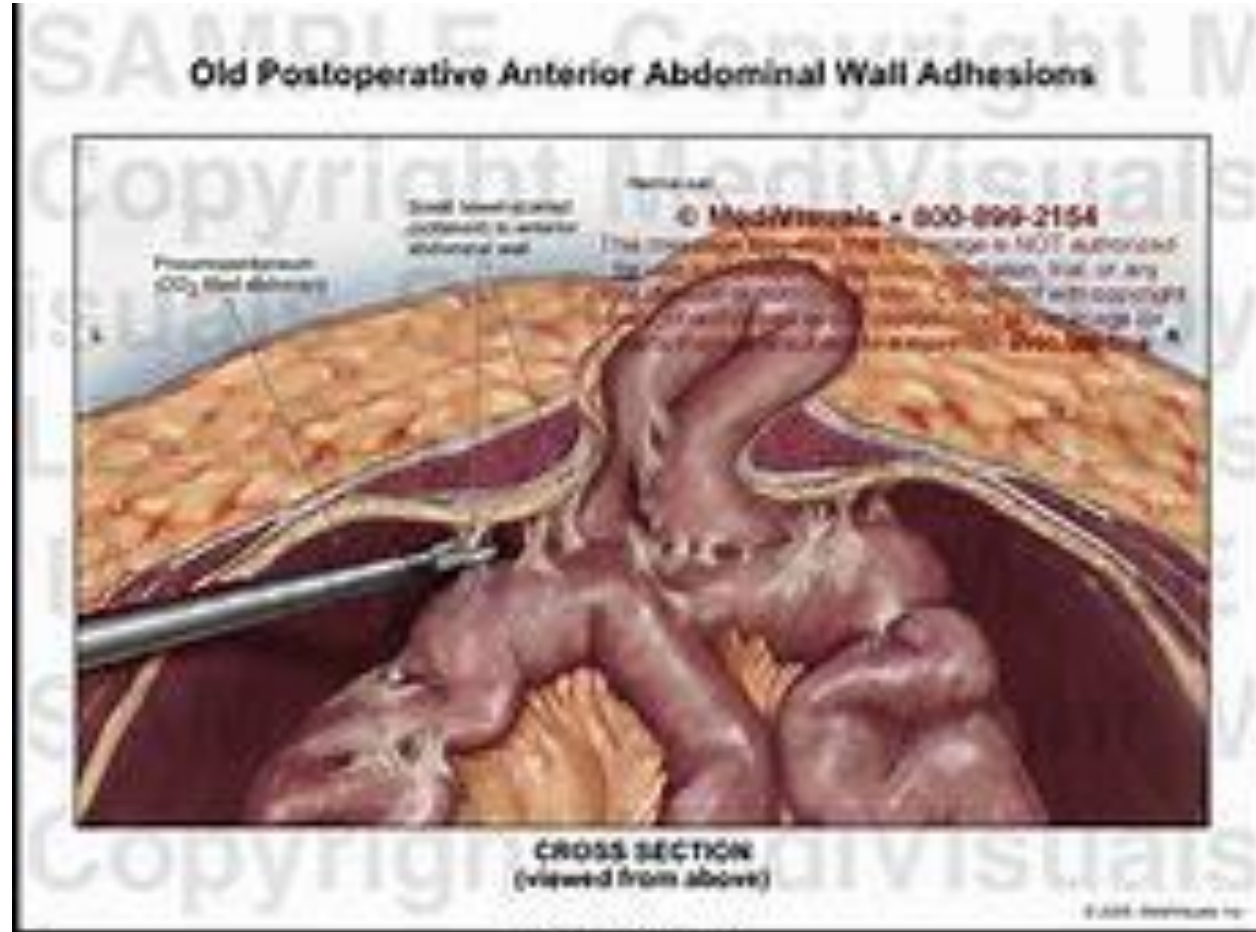
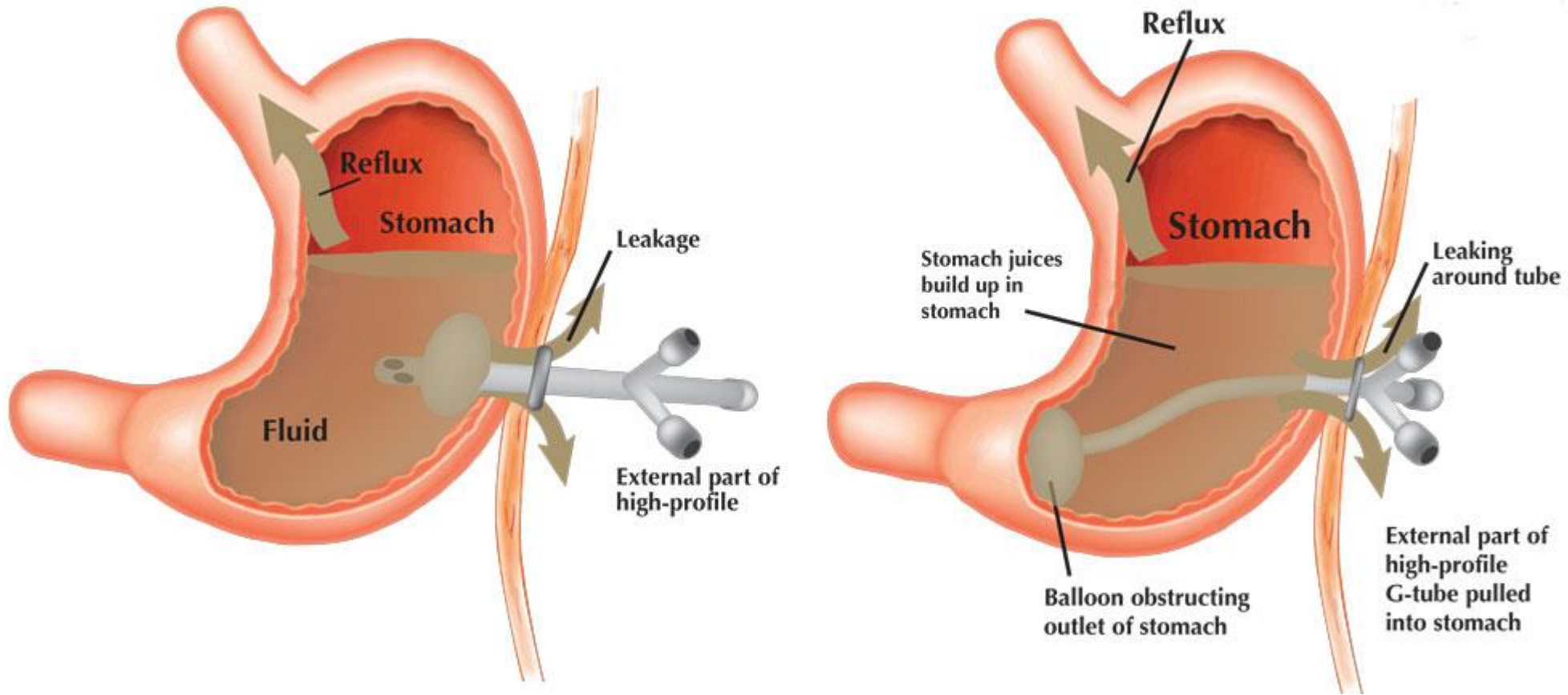


Exhibit 405231-02X

Tube Migration



Questions?

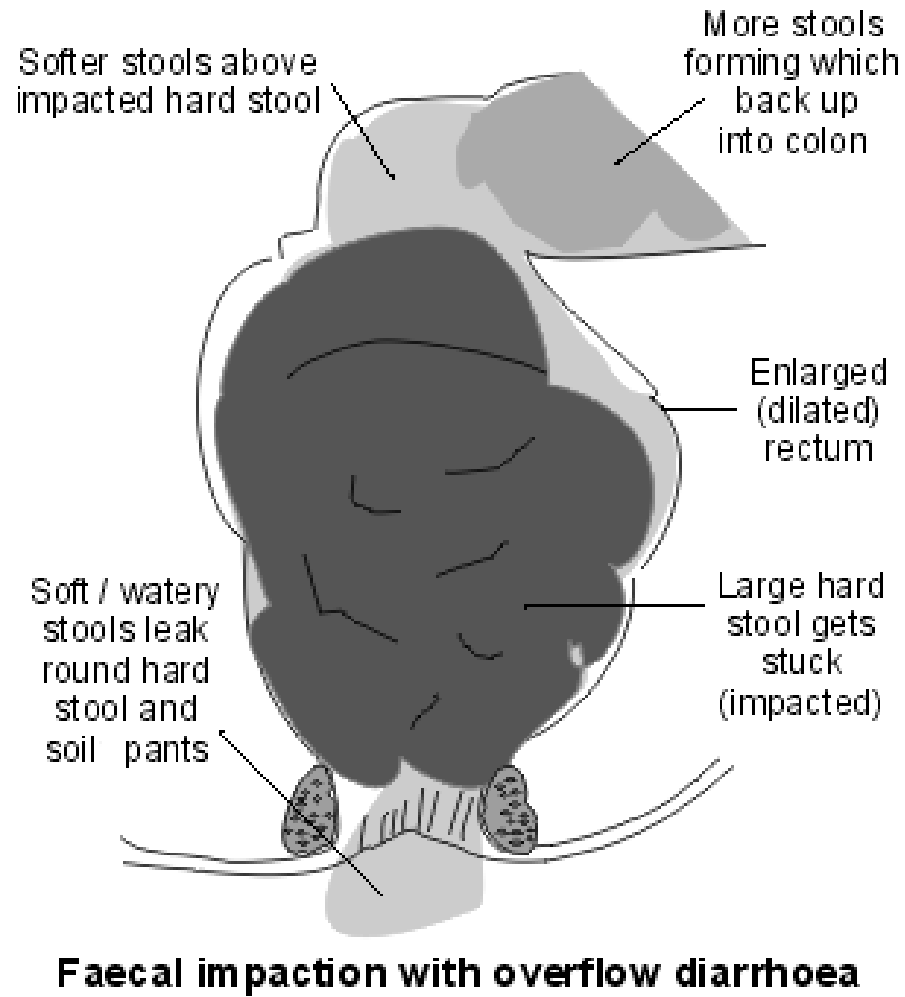
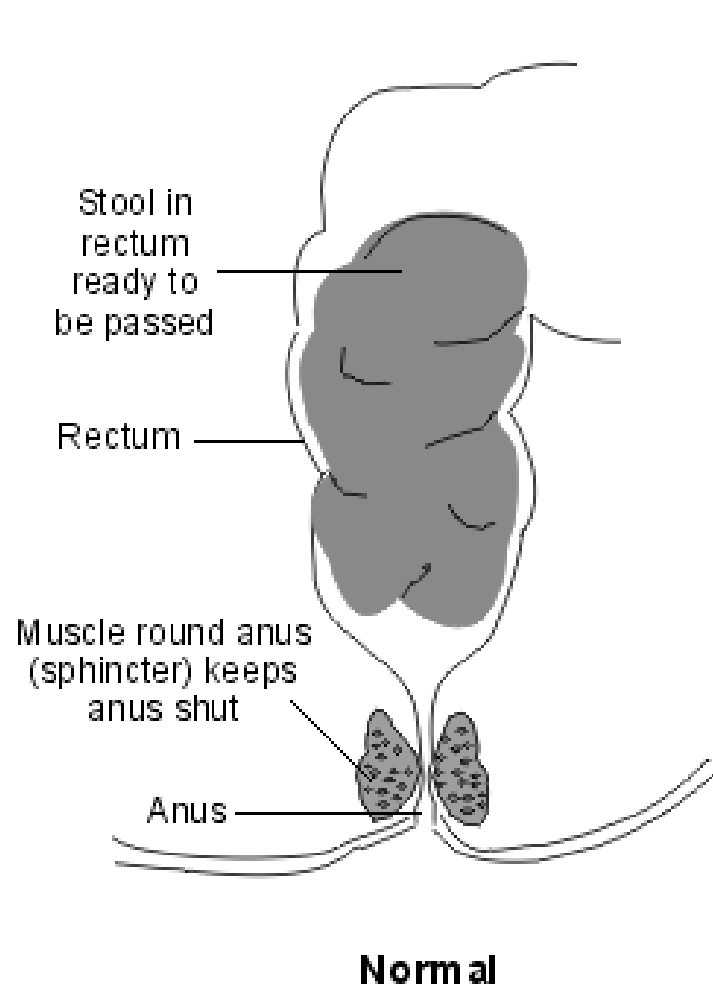
Constipation

- Very common in CdLS
- They may be having a bowel movement everyday
- They have an underlying dysmotility
- Chronic fecal burden leads to stretching and loss of muscle tone



BIGSTOCK

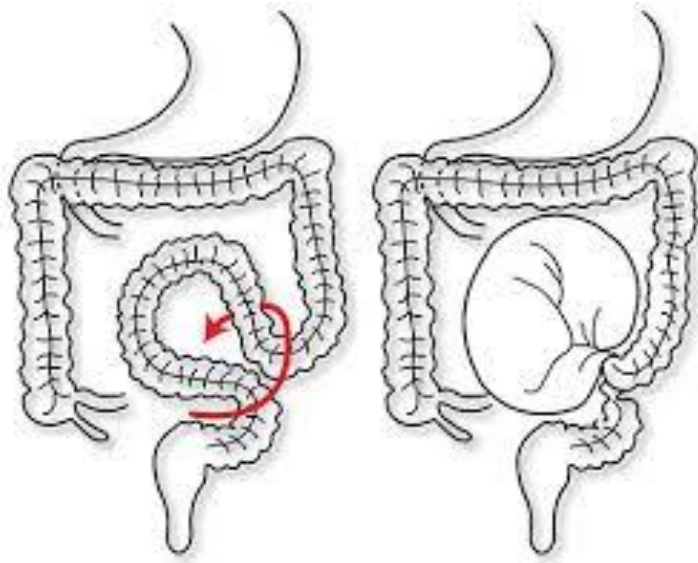
Image ID: 37185901
bigstock.com



Sigmoid Volvulus


- Chronic constipation stretches the colon
- The stool filled colon falls over on itself and twists off the blood supply
- Insidious presentation with abdominal pain, and distension that then becomes fulminant
- Diagnosis with CT scan
- Treated with a scope or surgery
- May reoccur


Sigmoid Volvulus








Bristol Stool Chart


TYPE 1  Separate hard lumps, like nuts - difficult to pass, indicates constipation


TYPE 2  Sausage-shaped but lumpy, indicates constipation

TYPE 3  Sausage-shaped with surface cracks, ideal stools as easier to pass

TYPE 4  Sausage shaped, smooth and soft, ideal stools as easier to pass

TYPE 5  Soft blobs with clear-cut edges, may indicate diarrhoea and urgency

TYPE 6  Fluffy pieces with ragged edges, a mushy stool, may indicate diarrhoea and urgency

TYPE 7  Watery, no solids, entirely liquid, may indicate diarrhoea and urgency





Constipation

- Many ways to treat it
- Don't under treat it

Constipation Therapy

- Pulling more water into the stool
 - Miralax
 - Milk of Magnesia
 - Fiber
- Drinking more water won't help
 - Colon just absorbs it

Stimulants

- May be needed due to dysmotility
- Senna
 - Liquid, pills, and chews
- Bisacodyl
 - Tablets and suppositories

Fermentation/Gas Production

- Lactulose
- Juice

Questions?

Pancreatitis

- Inflammation of the pancreas
- Causes include
 - Infection
 - Trauma
 - Drug
 - Malformation
- Symptoms
 - Pain and Vomiting
 - Can be very sick
- Diagnosis
 - US
 - Lab work

Pancreatitis-Treatment

- Vigorous hydration (IV)
- Supportive
- Nutrition
- Watch for complications

Surgical Complication

- Adhesions
- Hernias
- May be hard to diagnose
- Fixing them may cause more problems than it solves

Questions?



Thank you for attending!

We hope to see you on the
next webinar.

www.CdLSusa.org