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MEDICINE

# Digestive Woes in Myotonic Dystrophy

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# Overview

- GI symptoms in DM
- Diagnostic testing
- Treatment options
  - General disease specific therapies
  - DM specific treatments (if available)

# GI Symptoms in Muscular Dystrophy

- GI symptoms present in approximately 30-60% of patients
- GI symptoms may precede diagnosis of muscular dystrophy
  - IBS like symptoms: abdominal pain/cramping
- 25% felt GI symptoms most disabling

# Common GI Problems

Symptoms	Clinical Conditions
Difficulty Chewing, Swallowing or Coughing while eating (52-62%)	<ul style="list-style-type: none"><li>- Oropharyngeal dysphagia</li><li>- Esophageal dysmotility</li><li>- Acid reflux</li></ul>
Nausea and Vomiting	<ul style="list-style-type: none"><li>- Gastroparesis</li><li>- Acid reflux</li></ul>
Abdominal pain (45-62%)	<ul style="list-style-type: none"><li>- Gastroparesis/Pseudoobstruction</li><li>- Gallstones</li><li>- Sphincter of Oddi dysfunction</li></ul>
Constipation (55-62%)	<ul style="list-style-type: none"><li>- Slow transit constipation</li><li>- Anal spasm</li><li>- Megacolon</li></ul>
Diarrhea	<ul style="list-style-type: none"><li>- Bacterial overgrowth</li><li>- Bile salt malabsorption</li></ul>
Fecal incontinence	<ul style="list-style-type: none"><li>- Weak anal sphincter</li></ul>

# Dysphagia

- Difficulty swallowing/choking
- Most commonly reported symptom
- No correlation between peripheral muscle symptom severity and esophageal symptoms

## Universal Choking Sign

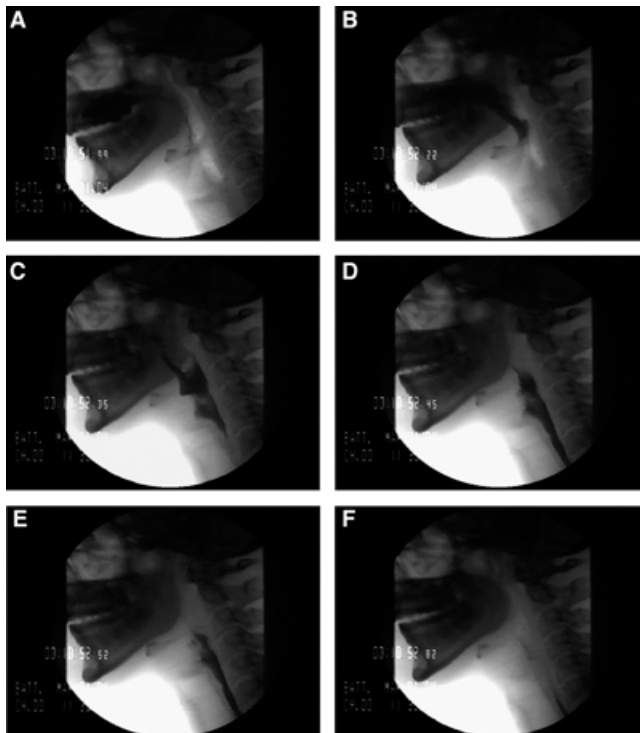


# Symptoms

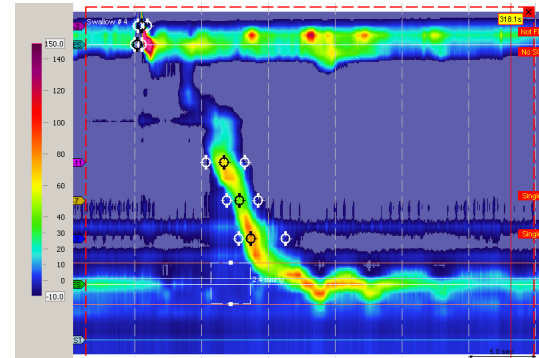
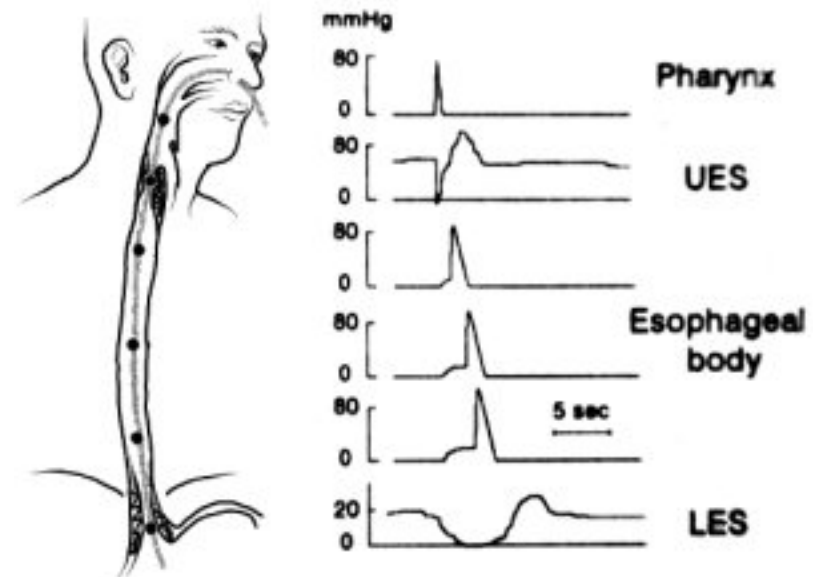
- Difficulty swallowing: Food getting stuck
  - Myotonia of the face, tongue, jaw
  - Pharyngeal weakness (Weak swallow)
  - Esophageal stricture/narrowing (Complication of acid reflux)
  - Muscle spasms of the lower esophagus
- Aspiration: Coughing/Pneumonia
  - Pharyngeal weakness (weak swallow)
  - Weak Upper esophageal sphincter
  - Acid Reflux
- Chest pain

# Esophageal Testing

- Video fluoroscopy (Swallow Study)

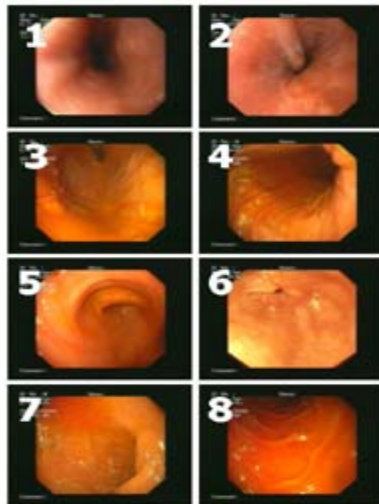
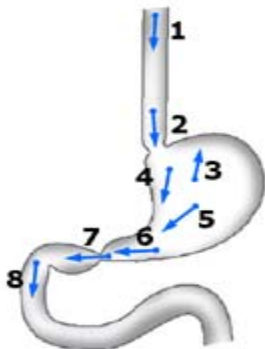
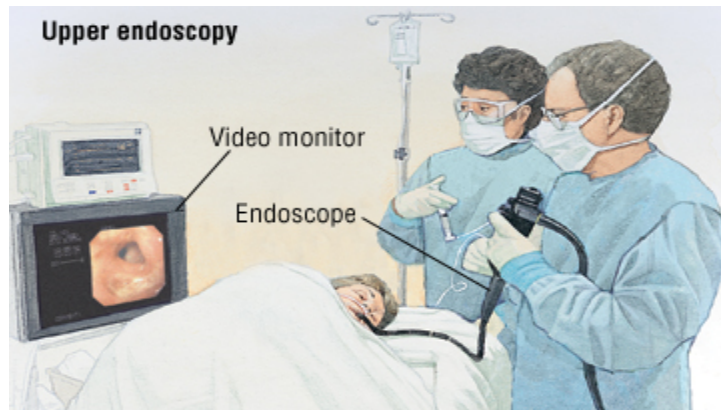


- Esophageal Manometry

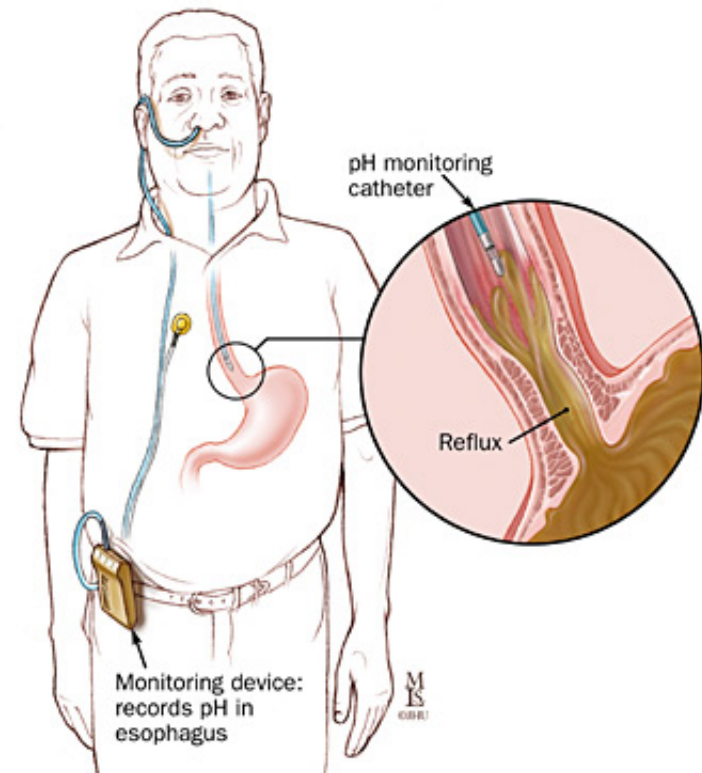


# Esophageal Testing

- Endoscopy



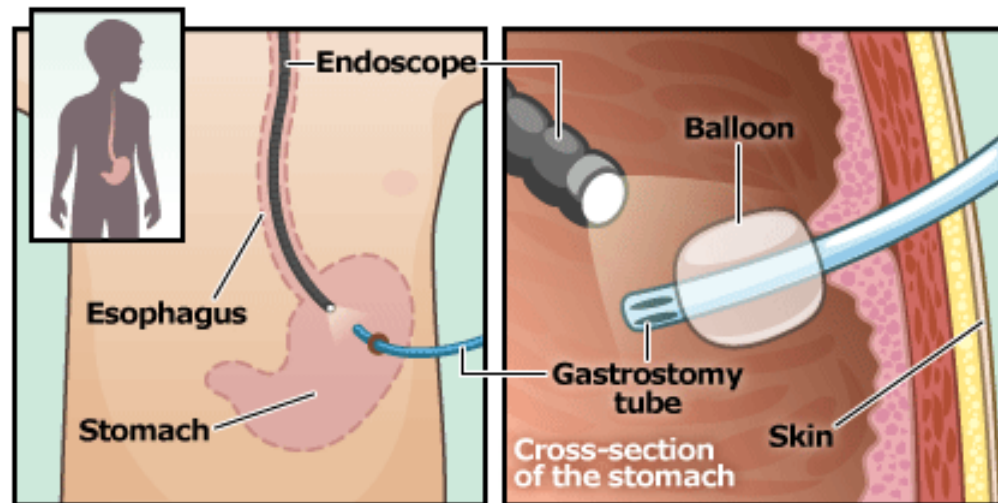
- Esophageal pH testing





# Treatment of Swallowing Problems

- Speech therapy
- Dietary changes: mechanical chopped, soft, thick liquids
- Feeding tube (especially if aspirating, weight loss)



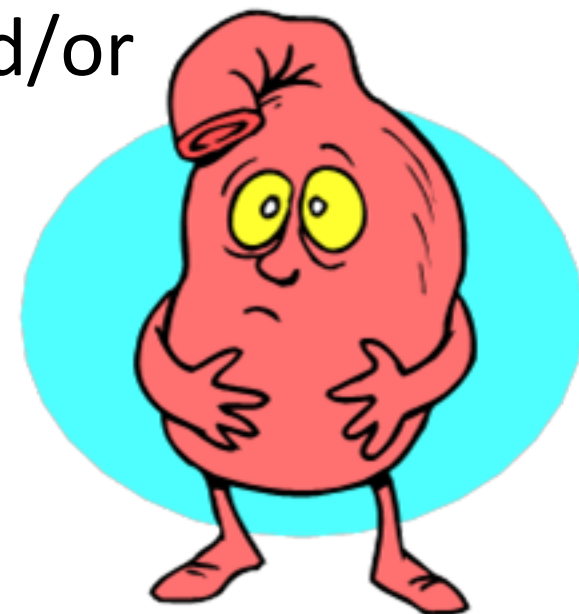
**PEG Procedure**

# Treatment of GERD

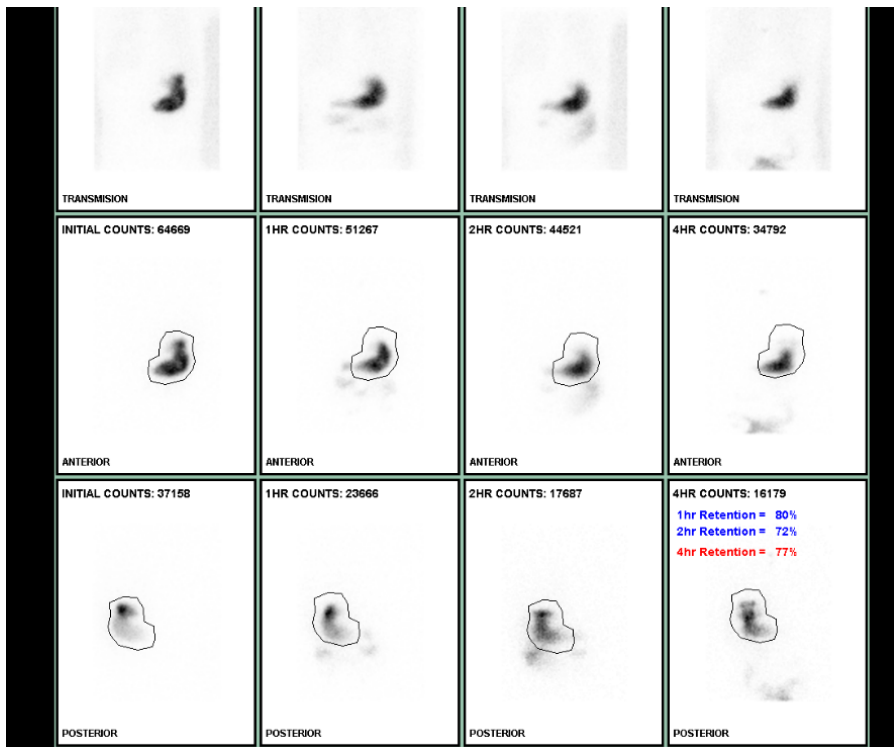
- Dietary changes
  - Avoid: acidic foods, spicy foods, fatty foods, caffeine, alcohol
  - Remain upright 3 hours after eating
- Elevate the head of the bed
- Acid suppression therapy
- Reglan

# Gastroparesis

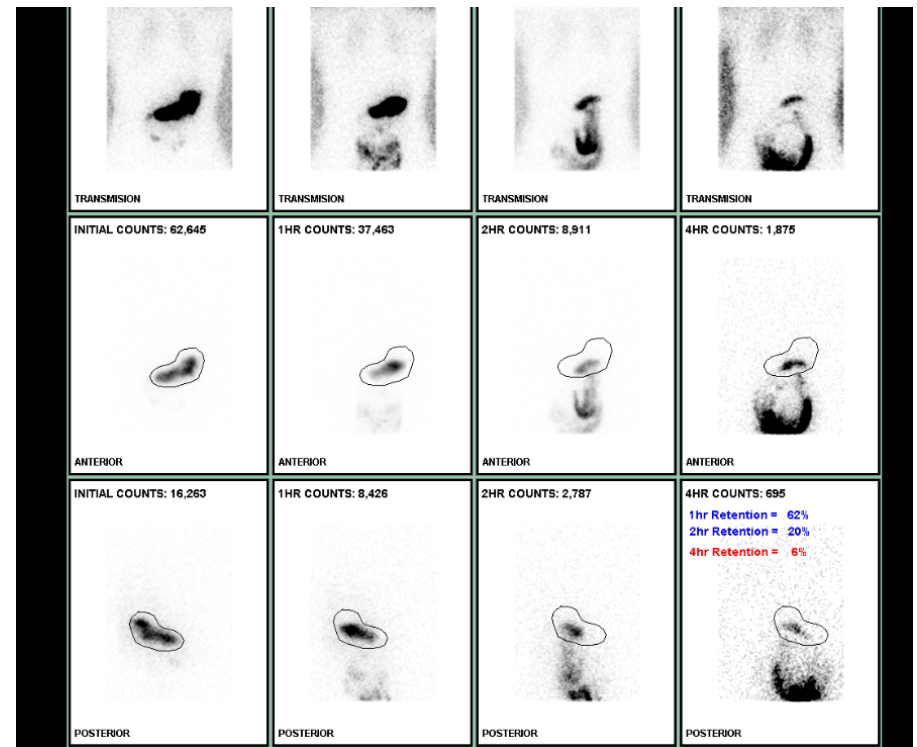
- Slow stomach emptying
- DM patient have slower gastric emptying compared to healthy controls
  - Even without symptoms
- Symptoms: Nausea, vomiting and/or abdominal pain (after eating)
- May be worsening acid reflux



# Diagnosing Gastroparesis: Gastric Emptying Study



Delayed Gastric Emptying



Normal Gastric Emptying

# Treatment of Gastroparesis

- Dietary changes
  - Low fat diet (fat slower to digest)
  - Low fiber (avoid “roughage”)
  - Small frequent meals
- Stay hydrated with electrolytes
  - Gatorade
  - Pedialyte

# Available Treatment Options for Gastroparesis

- Dopamine antagonists (D<sub>2</sub>-receptor): reglan, domperidone
- Serotonin agonist 5-HT<sub>4</sub> (i.e. tegaserod, cisapride)
- Cholinergic agonists (i.e. Neostigmine, bethanechol)
- Macrolides-motilin agonist: erythromycin, azithromycin
  - Improves gastric emptying with minimal affect on symptoms [Meganty et. al. Am J Gastroenterol 2003](#)
- Intrapyloric Botulinum Toxin
- Jejunal feeding tube
- Gastric electrical stimulation

# Treatment of Gastroparesis

- Therapies reported/studied in DM
  - Metoclopramide (N=16): increases gastric emptying
  - Erythromycin (N=10): did not improve gastric emptying or symptoms except diarrhea
  - Cisapride (no longer available)

# Intestinal Pseudoobstruction



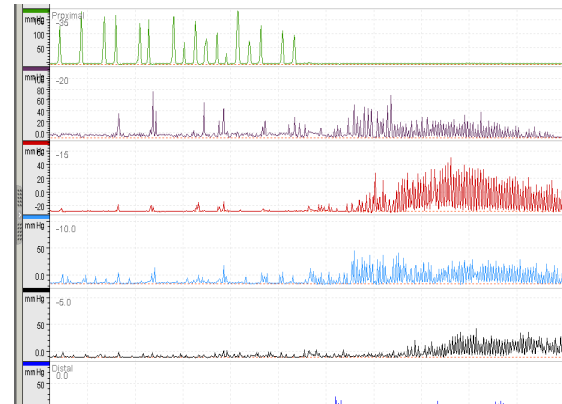
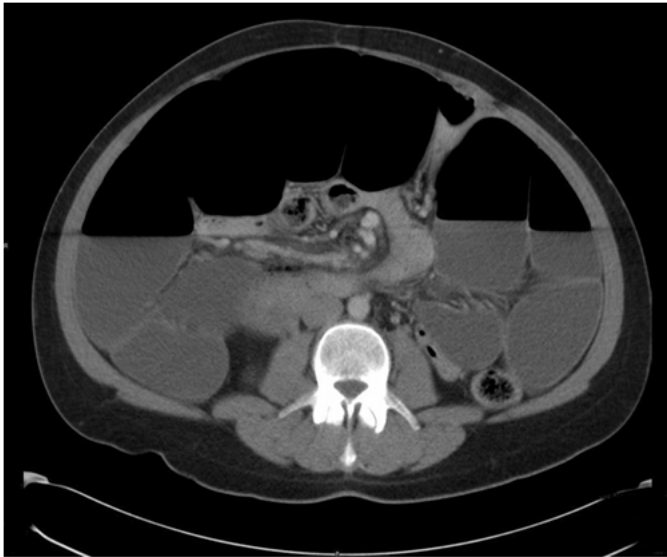


# Chronic Intestinal Pseudoobstruction

- Disordered small bowel motility (neuropathic or myopathic) leading to obstructive-like symptoms and dilated bowel
  - Distension – 75%
  - Abdominal pain – 58%
  - Nausea - 49%
  - Constipation - 48%
  - Heartburn/regurgitation – 46%
  - Fullness – 44%
  - Epigastric pain/burning – 34%
  - Early satiety – 37%
  - Vomiting – 36%

# Diagnosing CIP

- Imaging (Xray, CT)
  - Avoid barium studies
- Small bowel manometry



# Treatment of CIP

- **AVOID UNNECESSARY SURGERY**
- Nutritional support, IV hydration, decompression
- Evaluate and treat small intestinal bacterial overgrowth (if present)
- Promotility agents
  - Erythromycin/Azithromycin
  - Domperidone or metoclopramide
  - Octreotide
  - Cholinergic agonist: Neostigmine, pyridostigmine, bethanechol

# Constipation



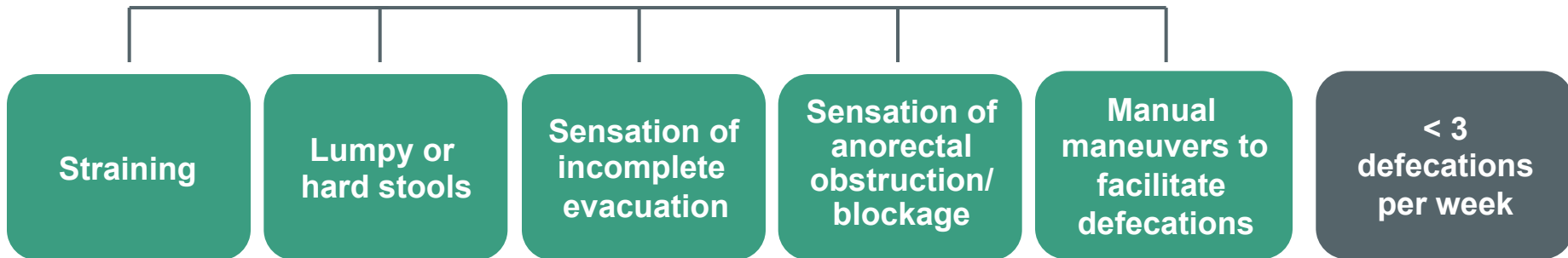
# Constipation Impairs Quality of Life

- HRQoL is impaired in patients with DM
- GI Factors associated with decreased QOL
  - Constipation
  - Gallstones

# Defining Constipation

Chronic constipation must include 2 or more of the following:

During at least 25% of defecations










- Loose stools are rarely present without the use of laxatives
- Insufficient criteria for irritable bowel syndrome

\*Criteria fulfilled for the last 3 months with symptom onset at least 6 months prior to diagnosis

Longstreth GF et al. *Gastroenterology*. 2006;130:1480-1491.

# Bristol Stool Chart

Type 1		Separate hard lumps, like nuts (hard to pass)
Type 2		Sausage-shaped but lumpy
Type 3		Like a sausage but with cracks on its surface
Type 4		Like a sausage or snake, smooth and soft
Type 5		Soft blobs with clear-cut edges (passed easily)
Type 6		Fluffy pieces with ragged edges, a mushy stool
Type 7		Watery, no solid pieces. <b>Entirely Liquid</b>

# Causes of Constipation in DM

- Slow colon transit
  - Altered colonic smooth muscle activity
  - Abnormal enteric nervous system function
  - Autonomic dysfunction
  - Decreased mobility
- Anal sphincter dysfunction (up to 90%)
  - Inability to relax anal sphincter with straining
  - Difficulty with defecation/excessive straining

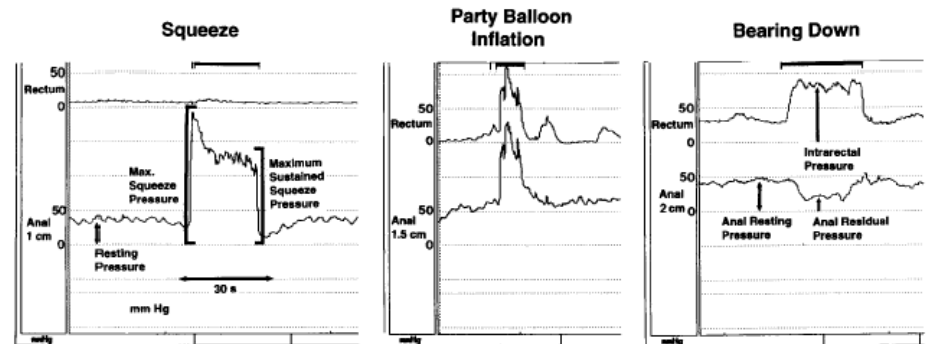
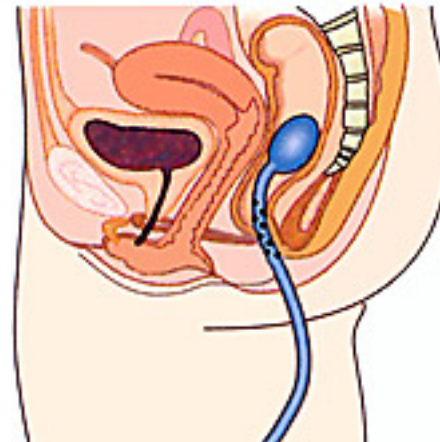


# Diagnostic Testing

- Sitz marker study

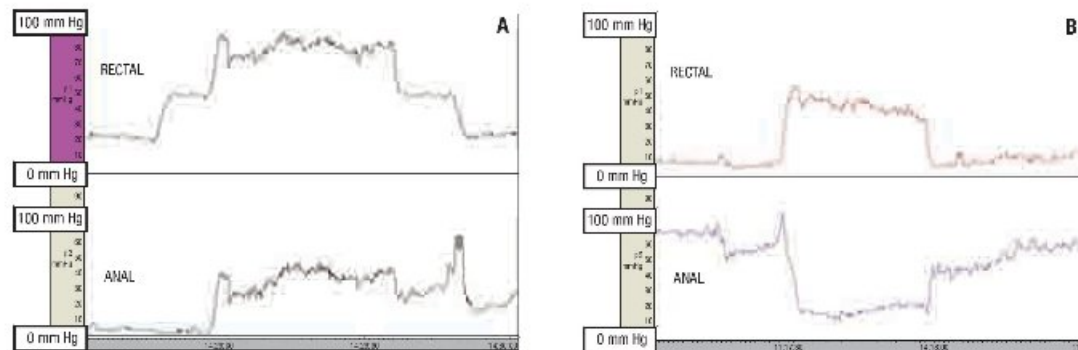


- Anorectal manometry



# Pelvic floor function in DM

- Low to Normal resting sphincter pressure
- Weaker squeeze pressure
- Myotonic contraction of the anal sphincter following the rectoanal inhibitory reflex (RAIR)
- Pelvic dyssynergia (Anismus)



# Treatment of Constipation

- Non-medical Therapy
  - Exercise
  - Diet
- Medical Therapy
  - Fiber
  - Laxatives/Stool softeners
  - Promotility or prosecretory agents
- Surgery

# Soluble vs. Insoluble Fiber

- Total Fiber intake 20-30 grams per day
  - Too much fiber can cause excessive bloating and gas
- Soluble Fiber = attracts water and forms a gel slowing gastric emptying
  - Dried beans, oats, oat bran, rice bran, barley, citrus fruits, apples, strawberries, peas, potatoes
- Insoluble Fiber = adds bulk to stool increasing colonic transit
  - Wheat bran, whole grains, cereals, seeds, skins on fruits and vegetables

# Medical Therapies

- Fiber (if diet insufficient)
- Osmotic laxatives (lactulose, magnesium citrate, Miralax)
- Stimulant laxative (bisacodyl, senna, glycerin)
- Prosecretory agents (lubiprostone, linaclotide)
- Suppositories/Enema- help with rectal evacuation

# Treatment of Defecatory Disorders

- Pelvic floor dysfunction
  - Biofeedback therapy
    - Teach relaxation of pelvic floor
    - Colostomy
- Rectocele or Rectal Prolapse
  - Surgery

# Principles of Biofeedback

- Push with <50% of maximal force
- Kegel exercises
  - Helps develop awareness of pelvic floor muscles
- Abdominal exercises
- Timing BMs after meals and when urge present
- Forward leaning or Squatting position
  - Facilitates whole body relaxation
- Stop trying after 10-15 minutes

# Diarrhea

- Malabsorption
  - Bacterial overgrowth
    - Treatment: Antibiotics and probiotics
  - Bile salt malabsorption
    - Treatment: cholestyramine
- Fecal impaction with overflow
  - Treatment: fiber, laxatives
- Medications



# Gallstones

- Present in 25-50% of DM patients
- Results from poor gallbladder function
- Causes abdominal pain after eating
- Treatment
  - Surgery (cholecystectomy)
  - Ursodeoxycholic acid (Ursodiol): 8-10 mg/kg/d
    - Dissolves small gallstones at a rate of 1 mm/month
    - Prevents complications i.e. cholecystitis

# Summary

- GI symptoms are common in patients with DM
- GI symptoms can precede the diagnosis of DM
- Symptoms can present gradually
- DM can affect the GI tract from the mouth to the anus
- Treatment options are limited but available

# Take Home Points

- GI symptoms affect quality of life (QOL)
- Symptomatic treatment can improve symptoms and QOL
- Targeted testing can help guide therapy
- Routine GI questionnaires/assessments should be a part of regular DM care