Digestive Woes in Myotonic Dystrophy

Linda Nguyen, MD
Director, Neurogastroenterology and Motility
Clinical Assistant Professor
Stanford University
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

<table>
<thead>
<tr>
<th>Symptoms</th>
<th>Clinical Conditions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Difficulty Chewing, Swallowing or Coughing while eating (52-62%)</td>
<td>- Oropharyngeal dysphagia</td>
</tr>
<tr>
<td></td>
<td>- Esophageal dysmotility</td>
</tr>
<tr>
<td></td>
<td>- Acid reflux</td>
</tr>
<tr>
<td>Nausea and Vomiting</td>
<td>- Gastroparesis</td>
</tr>
<tr>
<td></td>
<td>- Acid reflux</td>
</tr>
<tr>
<td>Abdominal pain (45-62%)</td>
<td>- Gastroparesis/Pseudoobstruction</td>
</tr>
<tr>
<td></td>
<td>- Gallstones</td>
</tr>
<tr>
<td></td>
<td>- Sphincter of Oddi dysfunction</td>
</tr>
<tr>
<td>Constipation (55-62%)</td>
<td>- Slow transit constipation</td>
</tr>
<tr>
<td></td>
<td>- Anal spasm</td>
</tr>
<tr>
<td></td>
<td>- Megacolon</td>
</tr>
<tr>
<td>Diarrhea</td>
<td>- Bacterial overgrowth</td>
</tr>
<tr>
<td></td>
<td>- Bile salt malabsorption</td>
</tr>
<tr>
<td>Fecal incontinence</td>
<td>- Weak anal sphincter</td>
</tr>
</tbody>
</table>
Dysphagia

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

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)
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$_2$-receptor): reglan, domperidone
- Serotonin agonist 5-HT$_4$ (i.e. tegaserod, cisapride)
- Cholinergic agonists (i.e. Neostigmine, bethanechol)
- Macrolides-motilin agonist: erythromycin, azithromycin
  - Improves gastric emptying with minimal affect on symptoms \cite{Meganty2003}
- 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%

Stanghellini V et al. Gut 1987
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:

- Straining
- Lumpy or hard stools
- Sensation of incomplete evacuation
- Sensation of anorectal obstruction/blockage
- Manual maneuvers to facilitate defecations
- < 3 defecations per week

- 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

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