Gastrointestinal Tract

Symptoms

Gastrointestinal (GI) symptoms that result from dysfunction of alimentary tract skeletal or smooth muscles are common. They can be a disabling and potentially serious feature of myotonic dystrophy (DM). Common GI symptoms include:

- Chewing and swallowing difficulties due to mouth, tongue or throat weakness or myotonia
- Gastroesophageal reflux caused by esophageal sphincter laxity
- Abdominal or chest pain (dyspepsia), nausea, vomiting, bloating or bowel pseudo-obstruction due to ineffective peristalsis
- Cholestasis (gallstones) due to ineffective gallbladder or bile duct musculature
- Constipation, diarrhea or malabsorption caused by bowel dysmotility (with secondary bacterial overgrowth), creating risk for fecal impaction, megacolon, bowel perforation and sepsis
- Impaired or painful bowel movements (dyschezia)
- Fecal incontinence due to anal sphincter and pelvic floor muscle weakness

Patterns of Gastrointestinal System Problems

Congenital DM1
Prenatal • Accumulation of amniotic fluid in the mother caused by reduced ingestion of amniotic fluid by the fetus (polyhydramnios)
Newborn • Ineffective nursing and failure to thrive due to weak suck • Ineffective swallow caused by craniofacial skeletal abnormalities and weakness of face, tongue, and jaw muscles • Inhalation of ingested liquid or secretions due to pharyngeal weakness and incoordination, potentially causing aspiration pneumonia (aspiration)
Childhood/Adolescence/Adulthood • Ineffective swallow (dysphagia) caused by craniofacial skeletal anomalies, or weakness, incoordination and myotonia of face, tongue, jaw, esophagus, and throat muscles • Aspiration due to pharyngeal weakness, potentially causing pneumonia • Recurrent post-prandial abdominal pain and bloating due to ineffective peristalsis or bowel pseudo-obstruction • Constipation, diarrhea, irritable bowel syndrome, caused by ineffective peristalsis or secondary intestinal bacterial overgrowth • Gallstones, due to abnormal gallbladder, bile duct, or sphincter musculature • Dilated colon, potentially leading to stool impaction, bowel perforation or megacolon
Childhood Onset DM1 and Adult Onset DM1

• Difficulty swallowing (dysphagia) caused by weakness or myotonia of the face, tongue, jaw, esophagus, and throat muscles • Aspiration due to pharyngeal weakness, potentially causing pneumonia • Recurrent abdominal pain and bloating, especially post-prandially • Constipation, diarrhea and irritable bowel symptoms • Gallstones due to abnormal muscle function of the gallbladder, bile duct and sphincter • Dilated colon, which can result in fecal impaction, possibly associated with megacolon or bowel perforation and sepsis
DM2
• Common symptoms include constipation, diarrhea, irritable bowel complaints, post-prandial bloating and abdominal pain, or gastroesophageal reflux • Additional investigations are required to determine whether these features and their molecular and cellular causes are similar.

Diagnosis

Careful assessment of the digestive tract is essential to relieve symptoms and to avoid secondary effects and complications. Gastrointestinal symptoms often develop gradually so that patients adopt compensatory mechanisms and consequently avoid necessary examinations. Patients and physicians can thus be unaware of gastrointestinal dysfunction until it comes to clinical attention due to acute exacerbation. For example, mild bowel dysmotility can be overlooked until a patient presents with symptoms of advanced pseudo-obstruction, at which time misdiagnosis of the severe abdominal pain and bloating as a complete mechanical bowel obstruction can lead to the potentially disastrous consequences of inappropriate abdominal surgery. This situation can be avoided only by conscientious and detailed inquiry about gastrointestinal problems at the time of routine clinical care, investigating, treating and educating patients at an early stage rather than when symptoms climax in an acute abdomen.

Routine gastrointestinal assessment
History and review of symptoms should cover chewing problems (myotonia or fatigue); difficulty swallowing (dysphagia for solids; aspiration of liquids, or frequent dry cough suggesting aspiration of secretions) gastroesophageal reflux; eating patterns; post-prandial bloating or pain and characteristics of any abdominal pain; frequency and character of bowel movements; fecal or urinary incontinence.

Routine physical examination
Special attention should be paid to evidence of involuntary weight loss, dysphonia indicative of pharyngeal weakness, frequent cough indicative of aspiration, abdominal pain on palpation, either generally or at gallbladder, and abdominal bloating.

Evaluation of asymptomatic individuals
Additional evaluation may include:

- Abdominal X-ray to evaluate abnormal bowel gas or stool, or free abdominal air
- A swallow study to characterize dysynergic movements, pharyngeal weakness, pharyngeal or esophageal constriction, or aspiration
- Abdominal ultrasound or MRI scans can detail stomach, small bowel, large bowel or gallbladder anatomy

- Barium upper GI radiographic evaluation to assess lower esophageal function and reflux, gastric emptying, and small bowel anatomy and function. If acute bowel obstruction is considered, a barium radiographic investigation with small-bowel follow-through distinguish pseudo-obstruction from the surgical emergency of true bowel obstruction.
- Manometry to demonstrate weakness or disordered contraction of esophagus, gastroesophageal sphincter, stomach, small bowel, rectum and anal sphincter
- Endoscopy to define abnormal structure or function of pharynx, esophagus, stomach, small intestine, or large intestines
- Blood tests to investigate cholestasis or hepatic involvement. Results should be interpreted cautiously since elevated AST and ALT in myotonic dystrophy can be evidence of muscle damage rather than liver dysfunction. Similarly, gamma-glutamyltransferase (GGT) blood level does not correlate with liver damage in myotonic dystrophy because it too is often elevated in all DM1 and DM2 subjects. Alternatively, serum alkaline phosphatase and bilirubin elevation do correlate with cholestasis in myotonic dystrophy.

Treatment

Accurate diagnosis is critical when treating GI symptoms in people with myotonic dystrophy. For example, painful bowel dilitation caused by myotonic dystrophy pseudo-obstruction may be mistakenly diagnosed as an acute bowel obstruction, which could expose the patient needlessly to the risks of anesthesia and surgery and post-surgical complications. Alternatively, clinical or radiographic verification of pseudo-obstruction allows conservative management with medication and other measures.

Pharmacologic approaches to GI symptoms

- Mexiletine to reduce myotonia in muscles of mastication that interfere with chewing, or in pharyngeal and proximal esophageal muscles responsible for dysphagia
- Prokinetic drugs (such as metoclopramide, and erythromycin) used intermittently to reduce symptoms of bowel hypomotility (bloating, abdominal pain, constipation), although diminished response prevents the utility of chronic treatment with these medications (Prokinetic agents can sometimes help control diarrhea that results from the bacterial overgrowth that is caused by hypomotility and malabsorption.)
- Cholestyramine to treat diarrhea, incontinence, and pain

Treatments for dysphagia

- Dietary modification (mechanically soft foods are easiest to swallow)
- Involvement of a speech therapist to teach behavioral and postural modification (e.g., neck flexed when swallowing, reduction of mouthful volume, alternation of solids and liquids, use of a particular implement, such as a cup, straw or spoon that improves swallowing)
- Gastrostomy feeding to maintain nutrition and protect the airway. Nasogastric tubes are typically contraindicated in myotonic dystrophy patients because they increase risk of aspiration.