Management of GI Issues in Duchenne

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Objectives

• Current GI recommendations

• What is known and not known

• Case Presentation: Application
Current Recommendations

• Experts from a wide range of disciplines
  – U.S. Centers for Disease Control and Prevention (CDC)
  – TREAT-NMD
  – Patient Advocacy Organizations

• Schedule for Screening GI issues
  – 6 months
  – Annual
Recommendations (6 Months)

• Weight and height

• Meet with Registered Dietician
Growth Chart Teen/Adult
Recommendations (6 Months)

- Screen for Swallowing/Dysphagia
- Screen for Reflux
- Screen for Gastroparesis / Stomach Issues
- Screen for Constipation
Recommendations (Annual)

- Vitamin D levels
- Dietary intake of Calcium
Dystrophin and GI Muscle?

• Chewing/swallowing: skeletal muscle
• Distal Esophagus: smooth muscle
• Stomach and Colon: smooth muscle
Routine Screens of Swallowing

- Specific questions on eating and swallowing

- Validated Survey: Sydney Swallow Questionnaire (SSQ)

- Speech Therapy Evaluation

- Video Swallow Study (V.S.S)

- Fiberoptic Endoscopic Evaluation of Swallowing (F.E.E.S.)
Problem Swallowing / Dysphagia

• Oral Pharyngeal Phase
  – Tired with chewing / reduced endurance
  – Started eating softer foods
  – Choking when swallowing
  – Any weight loss

• Esophageal Phase
  – Feel Food Getting Stuck
  – Chest Pain
  – More likely reflux or allergy
Impaired Gastric Function

• Reports of Gastroparesis

• Progressive Changes
Impaired Gastric Function?

- **Range of symptoms**
  - Vomiting
  - Feel full quickly
  - Bloated/Nausea
  - Decreased appetite
  - Unable to eat full meals

- **Gastric Emptying studies**
  - Standard Nuclear Medicine Scans
  - Breath tests study

- **Would get Emptying Study prior to getting G-tube**
Colonic Motility in DMD?

• Evidence for Colonic Dysmotility

• Evidence against Colonic Dysmotility
Constipation

- Overlooked Cause for Reducing Appetite
- Cause of Other GI Symptoms
Feeling of Fullness: 29%
Not Hungry: 27%
Abdominal Pain: 25%
Abdominal Distension: 20%
Nausea: 12%
Reflux: 13%
Vomiting: 2%
Constipation

• Under Reported and Under Treated

  – Almost half of 120 surveyed met criteria

• Unrecognized
Criteria for Diagnosing Constipation

• Consistency
  – “Hard Stool or Little Balls or Lumpy Bumpy?”

<table>
<thead>
<tr>
<th>Bristol stool chart</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>

Constipated
Criteria for Diagnosing Constipation

• Consistency

• Size
  – “Ever pass anything that amazes you?”
  – “How often clog toilet?”
Criteria for Diagnosing Constipation

• Consistency

• Size

• Effort/Pain
  – “Hurt when stool comes out?’
  – “Straining, holding breath, face change color?”
  – “How long in bathroom?”
Criteria for Diagnosing Constipation

- Consistency
- Size
- Effort/Pain
- Continence
  - “Stool Accidents?”
  - “Smears in Underwear?”
Criteria for Diagnosing Constipation

• Consistency
• Size
• Effort/Pain
• Continence
• Frequency \((\geq 4 \text{ days})\)
Clinical Case (14 yo with DMD)

- Wheel chair dependent. No significant cardiac or respiratory issues. Eats well by mouth.
- Issues with constipation since first year of life.
- Diagnosed Autism age 4.
- Miralax as needed. Miralax every day had loose stools. Never used stimulants.
- **Upper GI Symptoms:** none
- **Bowel Habits/Patterns:**
  - stool frequency: every 10-14 days
  - stool consistency: firm and large volume
  - blood/mucus: blood streaks
  - stooling associated with: straining
  - encopresis: Yes daily for the past 2 weeks
Clinical Case (cont.)

• Xray showed large amount of stool

• Did bowel clean out and started on aggressive daily bowel regimen

• Patient never followed up
Clinical Case (cont.)

• Called to see patient in Neuromuscular Clinic

• 16 yo malnourished / losing weight

• Had not liked result of bowel regimen
  – Increased stool output
  – Difficult to regulate with miralax
Clinical Case (cont.)

Two Issues

• Nutrition

• Function of Colon
Work-Up for G-tube

• Evaluate Swallow Function

• Gastric Emptying Study

• UGI (evaluate anatomy)

• Trial of NG tube feeds
Work-Up for Colon Function

- Aggressive Medical Therapy
- Actually could use NG tube for medications
- Colonic Manometry
Results of Studies

• Decreased endurance of chewing

• No aspiration

• Stomach anatomy and function normal

• Proximal Colon normal function

• Distal Colon segment not functioning
Recommendations

• G-tube

• Resect non-functioning segment

• Appendicostomy
  – Antegrade enema regimen
Appendicostomy

• Used frequently in wheelchair dependent patients

• Flush colon on daily basis

• “Take over” colonic function
Outcome

• Mother “Best thing we ever did”

• Patient “Feel much better. I have more energy.”

Actually decreasing amount of tube feeds
Importance of GI Screen

• Identify at nutritional risk
  – Obesity
  – Malnutrition

• Prevent infection
  – Aspiration
  – Poor immune system from malnutrition

• Maintain function
  – Loss of muscle function
  – Malnutrition causes muscle loss