

# Management of GI Issues in Duchenne

Kent Williams, MD  
Assistant Professor  
Nationwide Children's Hospital  
Columbus Ohio

# Objectives

- Current GI recommendations
- What is known and not known
- Case Presentation: Application

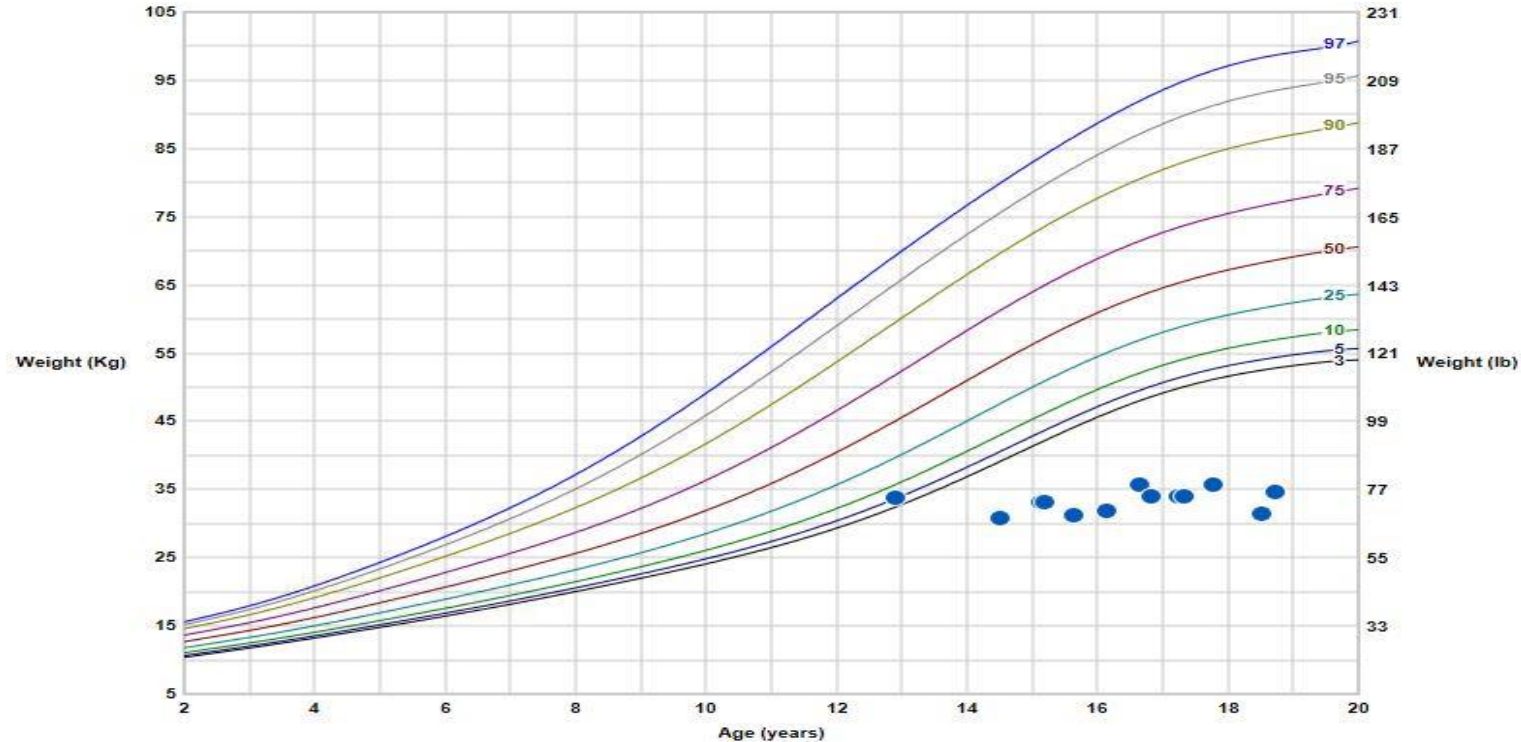
# Current Recommendations

- 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)
  - Archer SK, Garrod R, Hart N, Miller S. Dysphagia in Duchenne muscular dystrophy assessed by validated questionnaire. *Int J Lang Commun Disord*. 2013;**48**(2):240–6.
- 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
  - Bensen ES et al. Acute gastric dilatation in Duchenne muscular dystrophy: a case report and review of the literature. Archives of physical medicine and rehabilitation. 1996;77:512-4
- Progressive Changes
  - Borrelli O, et al. Evolution of gastric electrical features and gastric emptying in children with Duchenne and Becker muscular dystrophy. Am J Gastroenterol. 2005;100:695-702.

# Impaired Gastric Function?

- Range of symptoms
  - Vomiting
  - Feel full quickly
  - Bloating/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

- Leon SH, et al. Chronic intestinal pseudoobstruction as a complication of Duchenne's muscular dystrophy. *Gastroenterology*. 1986;90:455-9.
- Gottrand F, et al. Segmental colonic transit time in Duchenne muscular dystrophy. *Arch Dis Child*. 1991;66:1262.
- Lo Cascio CM, et al. Gastrointestinal Dysfunction in Patients with Duchenne Muscular Dystrophy. *PLoS One*. 2016;11:e016377

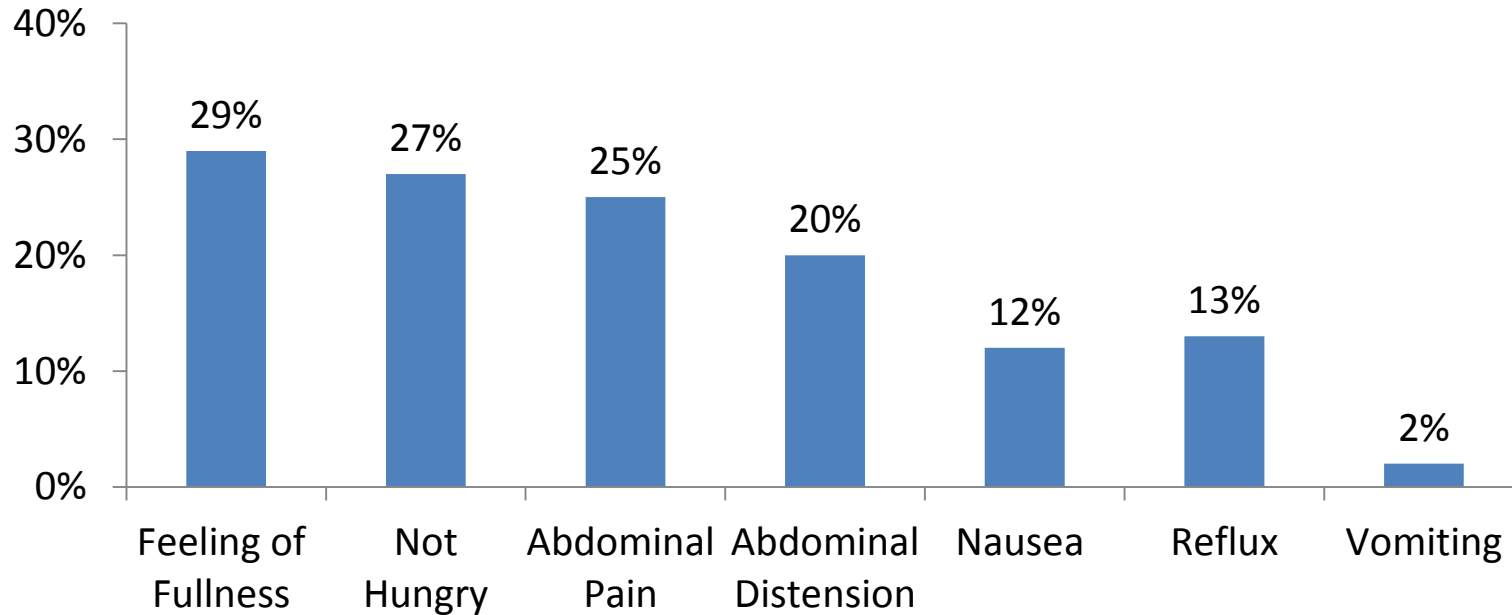
- Evidence against Colonic Dysmotility

- Kraus D, Wong B, Hu S, Kaul A. Gut Transit in Duchenne Muscular Dystrophy Is Not Impaired: A Study Utilizing Wireless Motility Capsules. *J Pediatr*. 2017

# Constipation

- Overlooked Cause for Reducing Appetite
- Cause of Other GI Symptoms

# PPMD Online GI Survey (N=338)



# Constipation








- Under Reported and Under Treated
  - Kraus et al. Constipation in Duchenne Muscular Dystrophy: Prevalence, Diagnosis, and Treatment. J Pediatr 2016;171:183-8
  - Almost half of 120 surveyed met criteria
- Unrecognized



# Criteria for Diagnosing Constipation

- Consistency
  - “Hard Stool or Little Balls or Lumpy Bumpy?”

**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>

↕ **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 ( $\geq 4$  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





R<sub>M</sub>C

STRETCH

# 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

G1.2e#0.60+0.45,MCQ2.0AF0.4



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JPINE

L SET

# 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