Nutrition and Gastroenterology for MECP2 Duplication Syndrome

Kathleen J. Motil, M.D., Ph.D.
USDA Children’s Nutrition Research Center
Baylor College of Medicine
Houston, TX 77030
Growth and Nutritional Status

- Patterns of growth vary
  - Normal height, short stature
  - Genetic downsizing at puberty?
  - Normal, poor, excess weight gain
  - Less muscle mass than body fat?
- Poor growth caused by inadequate dietary intake relative to growth needs
  - Poor chewing/swallowing skills
  - GE reflux, constipation, seizures
  - Unlikely intestinal malabsorption
Chewing and Swallowing Problems

Features

- Poor chewing skills
- Poor tongue lateralization, food bolus formation
- Pooling of liquids/solids in back of throat
- Laryngeal penetration of thin liquids
- Aspiration of liquids
- OT/speech therapy for oral motor feeding skills
- “Don’t use it, lose it!”
Body Mass Index

- BMI “gold standard” for nutritional status
  - Ratio of body weight to height
  - Normal: 25-75th %ile
  - Trigger: <5th %ile, >85th %ile
- Alternative feeding methods if BMI <5th %ile
  - Oral supplements, nasogastric or gastrostomy tube feeding
- Dietary energy reduction if BMI >85th %ile
Low Bone Mass

- Low bone mineral content, low bone mineral density
- Minimal information in MECP2 duplication
  - Increased fracture risk
- Risk factors include poor diet, anticonvulsants, immobility
- Test by x-ray, DXA scan
- Treatment strategies
  - Dietary calcium, vitamin D important
Recommendations: Calcium

- **DRI-for-age Ca**
  - <4 y = 700 mg/d
  - 4-8 y = 1000 mg/d
  - 9-18 y = 1300 mg/d
  - >18 y = 1000 mg/d

- **Milk/yogurt good Ca source**
  - 8 oz milk = 300 mg
  - 1 c low-fat, plain yogurt = 415 mg
  - 1 oz American cheese = 175 mg

- **Ca supplement**
  - 600 mg elemental Ca per tablet
Recommendations: Vitamin D

- Vitamin D promotes Ca absorption
- Risk factors for deficiency
  - Dark skin, ↓ sun exposure, anticonvulsants
- Sunlight, milk sources of vitamin D
- DRI-for-age Vitamin D
  - 1-70 y = 600 IU/d
- AAP recommends four 8-oz glasses of milk daily (vitamin D = 400 IU/d)
- Supplement if Vitamin D <20 ng/mL
Gastroesophageal Reflux

- Definition - passage of stomach contents back into esophagus
- Caused by poor motility of esophagus, LES, stomach
- Symptoms: Irritability, nighttime awakening, vomiting, wet burps, regurgitation, feeding refusal, coughing, wheezing
- Tests: UGI series, pH probe, gastric emptying scan
Treatment

- **Diet**
  - ↓ Spicy food, caffeine, chocolate, citrus (OJ, tomato)
- **Position**
  - Upright 30 min after eating
  - Elevate head of bed 45°
- **Medications**
  - Acid blockers (antacids, H$_2$-receptor inhibitors, proton pump inhibitors)
  - Prokinetics
- **Surgery** (fundoplication)
Gas Bloating

- **Definition**: gas trapping in stomach, intestines; tummy distention worse end of day
- **Caused by**: air swallowing, aggravated by constipation (bacterial overgrowth)
- **Symptoms**: air swallowing, burping, distension, abdominal pain, flatus, diarrhea, poor appetite
- **Tests**: abdominal x-ray, lactulose breath test, celiac
Treatment

- **Diet**
  - Modify CHO beverage, sorbitol, legume consumption
  - Modify dairy with lactase enzyme
  - Gluten-free diet for celiac disease

- **Medications**
  - Anti-gas
  - Antibiotics
  - Probiotics (Lactobacillus, Bifidobacterium)
  - Laxatives
Constipation

- **Definition** – difficulty having bowel movements
- **Causes** - functional, neuromuscular (Hirschsprung), endocrine (hypothyroid), drugs (phenytoin)
- **Symptoms**: infrequent motions < 2/week, hard stools, ± blood, straining with evacuation, abdominal distention, pain, flatus, urinary tract infection, feeding refusal, vomiting
- **Tests**: Rectal exam, barium enema
Treatment

- **Diet**
  - Fiber (fruits, veggies, cereal)
  - Sorbitol-containing fruits
  - Probiotics (Lactobacillus)

- **Medications**
  - Softeners: Polyethylene glycol
  - Pushers: Milk of Magnesia
  - Pullers: suppositories
  - Disimpact: enemas, GT lavage

- **Physical activity (physical therapy)**
Summary

- Nutritional, gastrointestinal problems common in MECP2 mutations
- Goal: be proactive in diagnosis and treatment to maintain quality of life
- Any symptom that causes parental concern should be evaluated by a physician
- Have your physicians consult MECP2 duplication team with questions