Gastrointestinal Manifestations

- Feeding
- Nutrition
- Esophagus - GERD
- Stomach
- Small intestine
- Colon – Constipation
- Hepatobiliary
Gastrointestinal Manifestations

- Rett Syndrome Center Data
- Literature
Rett Syndrome Center CHAM
GI manifestations

• 29 patients out of 120 referred to GI (25%)
• Parental reported common problems:
  – Feeding
  – Suboptimal weight gain
  – GERD
  – Gastrostomy tube placement/management
  – Abdominal pain
  – Aerophagia/Abdominal Distension
  – Constipation
Rett Syndrome Center CHAM
GI manifestations

- Feeding: 27
- Nutrition: 22
- GERD: 18
- PEG: 10
- Ab Pain: 7
- Ab Distension: 5
- Constipation: 25
Rett Syndrome Center CHAM
GI manifestations

Neuron. 2007 Nov 8;56(3):422-37.
Rett Syndrome Center CHAM
GI manifestations

Age vs. Complications

$\text{# of Complications}$

$\text{Age}$

$r^2 = 0.17826$
Rett Syndrome Center CHAM

Feeding

- Appetite issues – decreased intake
- Feeding selectivity or necessity (depending on swallow evaluation), ex. pureed foods only
- Unclear hunger and satiety cues
- Inability to self feed
- Increased time to complete meals
Rett Syndrome Feeding

- Isaacs et. al 2003, Rett syndrome group was significantly different compared with the developmental disability group.
  - Lower body weights
  - More swallowing problems
  - Less self-feeding
  - Lower texture tolerance for chewy and crunchy foods, with $p = 0.008$

Rett Syndrome
Feeding and Nutrition

20% GT*

80% No GT

*did not state why tubes placed
Feeding Issues

- Difficulty eating or unable to feed: 62
- Needed specific texture: 60
- Liquids thickened: 9.3
- Daily choking or gagging: 6.3
- No issues: 7

Percentiles
Coughing With Feeds

<table>
<thead>
<tr>
<th>Texture</th>
<th>Percentiles</th>
</tr>
</thead>
<tbody>
<tr>
<td>None</td>
<td>42</td>
</tr>
<tr>
<td>Liquids</td>
<td>22</td>
</tr>
<tr>
<td>Purees</td>
<td>8</td>
</tr>
<tr>
<td>Solids</td>
<td>11</td>
</tr>
<tr>
<td>&gt;1 texture</td>
<td>10</td>
</tr>
</tbody>
</table>

Rett Syndrome
Feeding and Nutrition

- Schwartzman et. al (2008):
  - 41% were undergoing feeding therapy at the time of the study.
  - 32% had never gone through any evaluation with a speech language pathologist.

Rett Syndrome
Feeding and Nutrition

^ compared to general population

# Rett Syndrome Nutrition

## Table 1
Clinical features of Rett syndrome and nutritional implications

<table>
<thead>
<tr>
<th>Seizures</th>
<th>Medication side effects such as sedation or nausea may change appetite or meal pattern. Feeding or eating during a postictal state (if present) is not recommended</th>
</tr>
</thead>
<tbody>
<tr>
<td>Constipation</td>
<td>May decrease appetite if unmanaged. Food sources of dietary fiber are not as effective as prescribed medications that affect colonic motility.</td>
</tr>
</tbody>
</table>
Table 1. Characteristics and growth and body composition measurements of girls with Rett syndrome and healthy girls

<table>
<thead>
<tr>
<th>Metric</th>
<th>Girls with RS (mean ± SD)</th>
<th>Healthy girls (mean ± SD)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Head circumference (cm)</td>
<td>47.2 ± 1.9*</td>
<td>50.8 ± 1.3</td>
</tr>
<tr>
<td>Height (cm)</td>
<td>104.6 ± 11.7</td>
<td>116.0 ± 14.4</td>
</tr>
<tr>
<td>Height-for-age (z score)</td>
<td>−1.7 ± 1.0*</td>
<td>−0.2 ± 0.7</td>
</tr>
<tr>
<td>Weight (kg)</td>
<td>14.8 ± 3.4*</td>
<td>21.1 ± 5.6</td>
</tr>
<tr>
<td>Weight-for-age (z score)</td>
<td>−2.3 ± 1.0*</td>
<td>−0.2 ± 1.0</td>
</tr>
<tr>
<td>Weight-for-height (z score)</td>
<td>−1.9 ± 0.8*</td>
<td>0 ± 1.3</td>
</tr>
</tbody>
</table>

Data presented as mean ± SD.

*p < 0.01, girls with RS versus healthy girls.
Rett Syndrome Center CHAM Nutrition

Range of BMI Percentile

Percentile on Growth Chart

Patients

- 0
- 20
- 40
- 60
- 80
- 100

Patients: 1 3 5 7 9 11 13 15 17 19 21 23 25 27 29

Percentile: 0 20 40 60 80 100

66%
38%
Rett Syndrome Center CHAM Nutrition

![Graph showing Age vs. BMI% with r^2 = 0.2548](image-url)
Rett Syndrome Center CHAM Nutrition

Z-scores

Patient yrs

7 - 20

2 - 6.2
Rett Syndrome Center CHAM Nutrition

Nutritional Intake

<table>
<thead>
<tr>
<th>Number of patients</th>
<th>Regular Diet</th>
<th>High Calorie Diet (HCD)</th>
<th>HCD and Enteral Nutrition</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>6</td>
<td>8</td>
<td>12</td>
</tr>
</tbody>
</table>
Rett Syndrome Center CHAM Nutrition

**Gastrostomy Tube**

<table>
<thead>
<tr>
<th>Presence of Gastrostomy Tube</th>
<th>Number of patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>No</td>
<td>19</td>
</tr>
<tr>
<td>Yes</td>
<td>10</td>
</tr>
</tbody>
</table>

The chart above shows the number of patients with and without a gastrostomy tube. It indicates that 19 patients do not have a gastrostomy tube, while 10 patients have one.
Rett Syndrome
Gastrostomy tube

• PEG in Rett syndrome indications:
  – Need to administer supplemental nutrition and/or hydration
  – Severe gastrooesophageal reflux (usually combined with fundoplication) causing retrograde aspiration
  – Inordinately long (>3 h per day) oral feeding time
  – Dependant on nasogastric tube feeding

Brain Dev 1992;14:S11–20
Rett Syndrome
Gastrostomy tube

• PEG in Rett syndrome indications:
  – Growth failure
  – Inability to close the mouth during swallowing
  – Need to administer medication
Rett Syndrome

Gastrostomy tube

• Motil et al 2009: Gastrostomy Placement Improves Height and Weight Gain in Girls With Rett Syndrome

• 92 girls with Rett
  – Height
  – Weight
  – Body mass index (BMI) z score

FIG. 3. Body mass index (BMI) z score slopes before and after gastrostomy placement in girls with Rett syndrome ($n = 81$, $P < 0.001$) (—, mean; ..., ±SD).
**GERD**: Symptoms or complications that may occur when gastric contents reflux into esophagus or oropharynx.

**CHAM**: Diagnosis of GERD: 1 pH probe study, congestion, vomiting, feeding intolerance, abdominal pain, irritability.
Rett Syndrome Center CHAM
GERD

Number of Patients

Treatment of GERD

No

Yes

PPI

H₂

10

19
Rett Syndrome

GERD

- “High incidence of GERD” reported in Rett patients, but no studies specifically looking at incidence, prevalence or results of pH probes/impedence studies

- ASD and Neurodevelopmental Disabled (ND) Population
Rett Syndrome – GERD ASD populations

• Gastrointestinal conditions that are reported to be common in individuals without ASDs are also encountered in individuals with ASDs.

• May manifest as:
  – Sleep disturbance
  – Self-injurious behavior
  – Tantrums
  – Aggression
  – Oppositional behavior

Pediatrics 2010;125:S1–S18
Rett Syndrome – GERD ND populations

- The high incidence of GER (15–75%) in neurologically impaired children is well recognized.
- Objective hallmark of GERD is recurrent vomiting which occurs in over 80% of cases.

Rett Syndrome – GERD ND populations

- Individuals with neurological impairment who are suspected of having GERD:
  - Vomiting
  - Hematemesis
  - Anemia
  - Rumination
  - Regurgitation

Rett Syndrome – GERD ND populations

- Dental problems are an under recognized consequence of GER
  - Increased prevalence of tooth erosion

www.annals.org/content/122/11/809.full
Rett Syndrome – GERD ND populations

• GERD and respiratory complications have been recognized for decades
  – Apnea
  – Asthma/wheezing
  – Chronic cough
  – Chronic pulmonary aspiration
  – Recurrent pneumonia
  – Progressive lung injury

Rett Syndrome – GERD
ND populations

• Otolaryngologic Manifestations:
  – Laryngitis
  – Dysphagia
  – Edema of the posterior larynx

Rett Syndrome – GERD
ND populations

Normal
Reflux laryngitis

NASPghan Slide set GERD

There are still many unsettled questions before one accepts the prevalent cult of “reflux disease.” The response to therapeutic intervention still lacks serious well-controlled studies to allow drawing reasonable conclusions.
Rett Syndrome Center CHAM
Esophagus: Dysmotility
Rett Syndrome
Esophagus: Dysmotility

• Esophageal dysmotility via UGI (Motil et. al 1999)
  – Absent primary or secondary waves
  – The presence of tertiary waves
  – Atony
  – Spasm
  – Gastroesophageal reflux

• Symptomatic patients had abnormal peristalsis on manometric evaluation in a study from 2008.

Rett Syndrome
Stomach: Dysmotility

• Gastric dysmotility via UGI
  – Diminished peristalsis
  – Atony

Aerophagia
Aerophagia

- 5 patients with aerophagia
- Caused by repeated swallowing of air
- Can manifest with:
  - Burping
  - Abdominal pain and/or distension
  - Flatulence
  - Diarrhea
  - Poor appetite
Aerophagia

- Can occur in individuals who have ASD or ND and in those without: psychological stressors
- Consider celiac disease, lactose intolerance or parasitic infection on differential.
Rett Syndrome Aerophagia

• Difficult to treat
  – Behavioral modification
  – Anxiolytics
  – Eat slowly
  – Don’t use straws or sippy cups for drinking
  – Don’t drink carbonated beverages or chew chewing gum
  – Trial of PPI
  – Trial of Simethicone
  – Trial of probiotic or antibiotic for bacterial overgrowth
  – Treat constipation
Rett Syndrome Center CHAM Constipation

- Yes: 24 patients
- No: 5 patients

Number of Patients

Patients
Rett Syndrome Center CHAM
Constipation

### Treatment of Constipation

<table>
<thead>
<tr>
<th>Treatment</th>
<th>Number of Patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>MiraLax and diet</td>
<td>18</td>
</tr>
<tr>
<td>Diet Alone</td>
<td>4</td>
</tr>
<tr>
<td>Other medication and diet</td>
<td>1</td>
</tr>
</tbody>
</table>
Rett Syndrome
Constipation

• Common GI Complaint
• Schwartzman et. al (2008):
  – 74% presented with constipation

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

http://upload.wikimedia.org/wikipedia/commons/1/1b/Hepato-biliary.jpg
Rett Syndrome
Hepatobiliary

• International Rett Syndrome Association

• 34 patients had evidence of gallbladder dysfunction.

J Child Neurol. 2005 Sep;20(9):718-21
Rett Syndrome
Hepatobiliary

3 to 43 years

Am J Gastroenterol 1989; 84:1378-82.

J Child Neurol. 2005 Sep;20(9):718-21
Rett Syndrome
Hepatobiliary

Table 5. Gallbladder Disease Clinical Status

<table>
<thead>
<tr>
<th>Feature</th>
<th>Number</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gallstones</td>
<td>19</td>
<td>66</td>
</tr>
<tr>
<td>Sludge</td>
<td>3</td>
<td>10</td>
</tr>
<tr>
<td>Cholecystomy</td>
<td>25</td>
<td>86</td>
</tr>
<tr>
<td>Death*</td>
<td>2</td>
<td>7</td>
</tr>
</tbody>
</table>

* One gallbladder dysfunction and one postoperatively
GI manifestations
Conclusions

• GI manifestations are common in Rett
• Feeding/nutrition and constipation most are the most frequent GI symptoms, but children also have issues with
  – GERD
  – Dysmotility
  – Needing a gastrostomy tube
  – Aerophagia
  – Abdominal pain and distention
  – Hepatobiliary issues
GI manifestations

Conclusions

• Increased awareness to identify individuals which are affected and at risk are important to improve desired outcomes
Rett Syndrome
Gastrostomy tube

• Complications:
• 4–26% of cases
• Major complications occur during or shortly after PEG insertion
  – Anesthesia risk
  – Laceration of the esophagus
  – Pneumoperitonium
  – Peritonitis
  – Colonic perforation with the risk of colo-gastric fistula formation.
Rett Syndrome
Gastrostomy tube

• Complications:
  Minor complications
  – Stoma leakage
  – Cellulitis
  – Granulation tissue formation around the site of PEG insertion
  – Loss of GT and closure of stoma
  – “Buried bumper”
### TABLE 3. Features of intestinal habit and characterization of constipation in Rett syndrome

<table>
<thead>
<tr>
<th>Evacuation with</th>
<th>Stool</th>
<th>Evacuations per week</th>
<th>Diagnostic of constipation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pain</td>
<td>Difficulty</td>
<td>Consistency</td>
<td>Form</td>
</tr>
<tr>
<td>1</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>2</td>
<td>No</td>
<td>Yes</td>
<td>Pasty</td>
</tr>
<tr>
<td>3</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>4</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>5</td>
<td>No</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>6</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>7</td>
<td>Yes</td>
<td>Yes</td>
<td>Pasty</td>
</tr>
<tr>
<td>8</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>9</td>
<td>No</td>
<td>Yes</td>
<td>Hard to pasty</td>
</tr>
<tr>
<td>10</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>11</td>
<td>Does not know</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>12</td>
<td>No</td>
<td>No</td>
<td>Pasty</td>
</tr>
<tr>
<td>13</td>
<td>No</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>14</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>15</td>
<td>No</td>
<td>No</td>
<td>Pasty</td>
</tr>
<tr>
<td>16</td>
<td>No</td>
<td>No</td>
<td>Pasty</td>
</tr>
<tr>
<td>17</td>
<td>No</td>
<td>Yes</td>
<td>Pasty</td>
</tr>
<tr>
<td>18</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>19</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>20</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>21</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>22</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>23</td>
<td>Yes</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>24</td>
<td>No</td>
<td>Yes</td>
<td>Pasty</td>
</tr>
<tr>
<td>25</td>
<td>No</td>
<td>No</td>
<td>Hard</td>
</tr>
<tr>
<td>26</td>
<td>No</td>
<td>Yes</td>
<td>Hard</td>
</tr>
<tr>
<td>27</td>
<td>No</td>
<td>Yes</td>
<td>Hard</td>
</tr>
</tbody>
</table>
• 5 parents state the use of CAM in their children
• CDC estimated that 11.8% of U.S. children used CAM in 2007
• Prevalence of CAM use in children with ASD 52% and 95%.
Rett Syndrome
Complementary and Alternative Medicine

**Alternative medical systems**
- Aromatherapy
- Homeopathic remedies
- Acupuncture
- Naturopathic remedies

**Biological based therapies**
- Diets
  - Casein free
  - Gluten free
  - Low glycemic diet
  - No additive and preservatives
  - Sugar free
  - Yeast free
  - Caffeine free
  - Digestive enzymes
  - Essential fatty acids liquid
  - Melatonin
- Mineral supplements
  - Magnesium
  - Calcium
  - Selenium

**Mineral and Vitamin supplement**
- Magnesium/B6
- Magnesium/B12
- Nutritional supplements
- Omega 3 oil

**Vitamin supplements**
- Vitamin B
- Vitamin C
- Echinacea
- Garlic oil
- Strawberry extract
- Vitamin D

**Celation Therapy**

**Manipulative and body-based therapies**
- Body based relaxation therapies
- Chiropractic
- Massage
- Sensory integration
- Therapeutic horseback riding

**Mind-body and psychological therapies**
- Music therapy
- Spiritual healing
Rett Syndrome
Complementary and Alternative Medicine

<table>
<thead>
<tr>
<th>Alternative medical systems</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aromatherapy</td>
</tr>
<tr>
<td>Homeopathic remedies</td>
</tr>
<tr>
<td>Acupuncture</td>
</tr>
<tr>
<td>Naturopathic remedies</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Biological based therapies</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diets</td>
</tr>
<tr>
<td>Casein free</td>
</tr>
<tr>
<td>Gluten free</td>
</tr>
<tr>
<td>Low glycemic diet</td>
</tr>
<tr>
<td>No additive and preservatives</td>
</tr>
<tr>
<td>Sugar free</td>
</tr>
<tr>
<td>Yeast free</td>
</tr>
<tr>
<td>Caffeine free</td>
</tr>
<tr>
<td>Digestive enzymes</td>
</tr>
<tr>
<td>Essential fatty acids liquid</td>
</tr>
<tr>
<td>Melatonin</td>
</tr>
<tr>
<td>Safe, effective</td>
</tr>
<tr>
<td>Mineral supplements</td>
</tr>
<tr>
<td>Magnesium</td>
</tr>
<tr>
<td>Magnesium/B6</td>
</tr>
<tr>
<td>Magnesium/B12</td>
</tr>
<tr>
<td>Nutritional supplements</td>
</tr>
<tr>
<td>Omega 3 oil</td>
</tr>
<tr>
<td>Vitamin supplements</td>
</tr>
<tr>
<td>Vitamin B</td>
</tr>
<tr>
<td>Vitamin C</td>
</tr>
<tr>
<td>Echinacea</td>
</tr>
<tr>
<td>Garlic oil</td>
</tr>
<tr>
<td>Strawberry extract</td>
</tr>
<tr>
<td>Vitamin D</td>
</tr>
<tr>
<td>Celation Therapy: Definitely unsafe: Discourage</td>
</tr>
</tbody>
</table>

| Mineral and Vitamin supplement           |
|                                         |

| Manipulative and body-based therapies   |
|                                         |
| Body based relaxation therapies         |
| Chiropractic                             |
| Massage                                  |
| Sensory integration                     |
| Therapeutic horseback riding            |

| Mind-body and psychological therapies   |
|                                         |
| Music therapy                           |
| Safe, unknown efficacy: encourage objective monitoring. |
| Spiritual healing                        |

Rett Syndrome
Abdominal Pain in Rett

• Difficult to ascertain - 8 patients at CHAM

<table>
<thead>
<tr>
<th>Vocal Behaviors</th>
<th>Motor Behaviors</th>
<th>Changes in Overall State</th>
</tr>
</thead>
<tbody>
<tr>
<td>Frequent clearing of throat, swallowing, tics, etc</td>
<td>Facial grimacing</td>
<td>Sleep disturbances: difficulty getting to sleep, difficulty staying asleep</td>
</tr>
<tr>
<td>Screaming</td>
<td>Gritting teeth</td>
<td>Increased irritability (exaggerated responses to stimulation)</td>
</tr>
<tr>
<td>Sobbing “for no reason at all”</td>
<td>Wincing</td>
<td>Noncompliance with demands that typically elicit an appropriate response (oppositional behavior)</td>
</tr>
<tr>
<td>Sighing, whining</td>
<td>Constant eating/drinking/swallowing (“grazing” behavior)</td>
<td></td>
</tr>
<tr>
<td>Moaning, groaning</td>
<td>Mouthing behaviors: chewing on clothes (shirt sleeve cuff, neck of shirt, etc), pica</td>
<td></td>
</tr>
<tr>
<td>Delayed echolalia that includes reference to pain or stomach (eg, child says, “Does your tummy hurt?” echoing what mother may have said to child in the past)</td>
<td>Application of pressure to abdomen: leaning abdomen against or over furniture or kitchen sink, pressing hands into abdomen, rubbing abdomen</td>
<td></td>
</tr>
<tr>
<td>Direct verbalizations (eg, child says “tummy hurts” or says “ouch,” “ow,” “hurts,” or “bad” while pointing to abdomen)</td>
<td>Tapping behavior: finger tapping on throat</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Any unusual posturing, which may appear as individual postures or in various combinations: jaw thrust, neck torsion, arching of back, odd arm positioning, rotational distortions of torso/trunk, sensitivity to being touched in abdominal area/flinching</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Agitation: pacing, jumping up and down</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Unexplained increase in repetitive behaviors</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Self-injurious behaviors: biting, hits/slaps face, head-banging, unexplained increase in self-injury</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Aggression: onset of, or increase in, aggressive behavior</td>
<td></td>
</tr>
</tbody>
</table>

A functional behavioral assessment would be useful in interpreting these behaviors.

* Motor behaviors also may be markers of pain or discomfort arising in other parts of the body.
# Rett Syndrome

## Abdominal Pain in Rett

### TABLE 3 Diagnostic Evaluation of Gastrointestinal Symptoms and Disorders in Individuals With ASDs

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Possible Associated Gastrointestinal Disorder</th>
<th>Definition</th>
<th>Diagnostic Evaluations to Be Considered</th>
</tr>
</thead>
<tbody>
<tr>
<td>Perceived abdominal discomfort: pressing abdomen, holding abdomen and crying, problem behaviors related to meals</td>
<td>Constipation, GERD, intestinal inflammation, malabsorption, maldigestion</td>
<td></td>
<td>(1) Diagnostic trial of proton-pump inhibitor or PEG 3350; (2) abdominal radiograph; (3) lactose breath test (or measure lactase-specific activity); (4) pH probe, EGD, colonoscopy</td>
</tr>
</tbody>
</table>
Evidence-based algorithms for the assessment of abdominal pain, constipation, chronic diarrhea, and gastroesophageal reflux disease (GERD) should be developed.
Introduction

- Rett syndrome – a postnatal neurodevelopmental disorder caused by mutations in the gene encoding methyl-CpG binding protein 2 (MeCP2).
Introduction
Introduction

Rett Syndrome

Autistic Spectrum Disorder (ASD)

Neurodevelopmental Disability (NPD)

Time
Introduction

• Clinical of Rett features varied can influence:
  – Neurologic system
  – Endocrinologic
  – Pulmonary
  – Cardiologic
  – Gastrointestinal
Rett Syndrome Center CHAM Nutrition

Z-Scores

Number of patients

Z-score

Series 1
Rett Syndrome Center CHAM Nutrition

Z-scores

Patients

z-scores