Objectives

- The learner should be able to identify the causes of encopresis in children.
- The learner should be able to discuss the strategies for treating encopresis in children.
- The learner should be able to define celiac disease.
- The learner should be able to discuss the prevalence of celiac disease in the population and amongst relatives.

Definition and Prevalence of Constipation

- Defined as a delay or difficulty in defecation that is present for 2 weeks or more and sufficient to cause significant distress to the patient
- Relatively common condition
- 3% of general pediatric office visits in the US
- 25% of pediatric gastroenterology clinic visits in the US
- 95% of cases are functional (no underlying cause).

Definition and Classification Encopresis

- Passage of stool in the underpants typically in a child that has already been toilet trained (>4yrs)
- May be voluntary (most often behavioral) or involuntary
- Introduced as a term in 1926 by Weissenberg
- 2 broad classifications:
  - Functional Encopresis (90% of cases)
  - Organic Encopresis - due to a defined disease process. (Anatomical, neurologic, metabolic)

Prevalence of Encopresis

- Affects 3% of 4 year olds and 1.6% of 10 year olds
- Most commonly affects 5-10 year olds
- Median age based on two large studies was 7-9 years
- Rarely affects adolescents
- Affects more boys than girls (M:F = 3-6:1)
Physiology of Normal Defecation

- Stool enters the rectum
- Internal anal sphincter relaxes
- Stool enters the anal canal
- External anal sphincter (under voluntary control)

Causes of Encopresis

- Functional Encopresis
  - Occurs in 90% of cases
  - No identifiable disease process
- Organic
  - Uncommon reason for encopresis
  - There is often a clearly recognized underlying condition
  - The causes may be anatomical, neurologic, or metabolic

Functional Encopresis

- Seen in 90% of all cases of chronic encopresis
- Most children with this withhold stool and this leads to chronic stretching of the rectal walls
- May be triggered by an event:
  - Passage of painful stool
  - Unsubstantiated fears
  - Difficulties with toilet training
  - Issues with the use of public toilets (e.g. school)

Organic Encopresis

- 5-10% of all cases of chronic encopresis
- Anatomic
  - Imperforate anus, ectopic anus or anal stenosis
  - Prior bowel surgery
- Neurologic
  - Hirschsprung’s disease, Spinal cord damage
- Metabolic
  - Hypothyroidism

Evaluation of Patients with Encopresis

- Good clinical history and physical examination
- Plain abdominal radiograph
  - Determine the degree of stool impaction in the colon
- Anorectal manometry
  - Screen for Hirschprung’s disease and voiding abnormalities like dyssynergic defecation
- Rectal mucosal biopsy
  - Confirm Hirschprung’s disease or anal achalasia
- Blood tests to evaluate for anemia, underlying thyroid abnormalities and celiac disease
3. Review of Celiac Disease and Encopresis in Children

**Treatment of Encopresis**
- Fecal disimpaction
  - Oral laxatives (stimulants and osmotic laxatives)
  - Enemas
- Maintenance Therapy
  - Daily laxatives
  - High fiber diet (age in years + 5 = daily requirement)
- Behavioral modification
  - Avoid withholding stool
  - Scheduled bowel habits: Sit on the toilet for 10 minutes or 1 minute per year of life after meals (2-3 times a day).

**Prognosis of Encopresis**
- Prolonged and consistent treatment is often required
- Frequent relapses occur
- 10 year follow up study of patients with functional constipation revealed:
  - 46% remained constipated
  - 25% continued to have encopresis
  - 56% still had recurrent abdominal pain

**Summary of Encopresis**
- Functional constipation is the most common cause for encopresis
- More common in boys than girls
- Often initiated by a precipitating event such as painful stools
- Enuresis (soiling with urine) and Urinary tract infections (UTI) are frequent associations
- Children and families often feel isolated and ostracized
- Treatment involves: laxatives, behavioral modification and counseling when deemed appropriate
- Treatment is a long term process with frequent relapses
Definition of Celiac Disease

- An immune mediated enteropathy (disorder of the intestine).
- There is a permanent sensitivity to gluten
- Affects genetically susceptible individuals
- Patients may be fall into one of the following groups:
  - Symptomatic
    - Gastrointestinal
    - Non gastrointestinal
  - Asymptomatic

Epidemiology of Celiac Disease

- Females more affected than males
- Affects ~1% in North America and Europe
- Affects other ethnic populations including:
  - Middle East
    - Iran: prevalence amongst 2,000 blood donors was 1:166
  - North Africa
    - Saharawis (1 in 18 children affected)
  - Asia
    - common cause of chronic diarrhea in children and adults in India
  - South America

Pathogenesis of Celiac Disease

- Genetic
- Environmental
- Dietary
  - Gluten (present in wheat, rye and barley)
  - α-gliadin (main toxic component of Gluten)
- Immune responses
  - Humoral
  - T cell
  - Mucosal

Clinical Presentation
Clinical Presentation of Celiac Disease

- Gastrointestinal ("classical")
- Non-gastrointestinal ("atypical")
- Asymptomatic

Gastrointestinal Manifestations of Celiac Disease

- Most common age of presentation: 6-24 months
  - Chronic or recurrent diarrhea
  - Abdominal pain and distension
  - Anorexia
  - Failure to thrive or weight loss
  - Vomiting
  - Constipation
  - Irritability

Malnourished patients with Celiac Disease

Image courtesy of CDHNF/NASPGHAN

Dermatitis Herpetiformis in Celiac Disease

- Erythematous macule > urticarial papule > tense vesicles
- Severe pruritus
- Symmetric distribution
- 90% no GI symptoms
- 75% villous atrophy
- Gluten sensitive

Image courtesy of CDHNF/NASPGHAN

Non Gastrointestinal Manifestations

- Dermatitis Herpetiformis
- Dental enamel hypoplasia of permanent teeth
- Osteopenia/Osteoporosis
- Short Stature
- Delayed Puberty/Menarche
- Iron-deficient anemia resistant to oral Fe
- Hepatitis
- Arthritis
- Epilepsy with occipital calcifications

Dental Enamel Defects in Celiac Disease

Image courtesy of CDHNF/NASPGHAN
Osteoporosis in Celiac Disease

Low bone mineral density improves in children on a gluten-free diet

Image courtesy of CDHNF/NASPGHAN

Iron Deficiency Anemia in Celiac Disease

• The most common non-gastrointestinal finding in some adult studies of patients with celiac disease
• Most patients would not respond to oral iron alone without treating the celiac disease.
• 5-8% of adults with unexplained iron deficiency anemia have celiac disease
• In children with newly diagnosed Celiac Disease:
  - Anemia is common
  - Little evidence that celiac disease is common in children presenting with anemia

Asymptomatic Celiac Disease

Silent Latent

• Silent: Patient has no or minimal symptoms, “damaged” mucosa and positive serology
• Identified by screening asymptomatic individuals from groups at risk such:
  • First degree relatives
  • Down syndrome patients
  • Type 1 diabetes patients, etc.

Disorders Associated with Celiac Disease

• The prevalence of Celiac Disease is higher in patients who have the following:
• Certain genetic disorders or syndromes
  - Downs (4-19%)
  - Turners (4-8%)
• Other autoimmune conditions
  - Type I Diabetes (3.5 - 10%)
  - Auto immune thyroiditis (4-8%)
• 1st degree relatives (~5%)

Short Stature and Delayed Puberty in Celiac Disease

• Short stature occurs in children and teens
  - ~10% of short children and teens have evidence of celiac disease
• Delayed menarche
  - High prevalence in teens with untreated Celiac Disease
Diagnosis

- Diagnosis of Celiac Disease requires:
  - Characteristic small intestinal histology in a symptomatic child
  - Complete symptom resolution on gluten-free diet
- Serological (blood tests) may support diagnosis

Serological Tests

- Antigliadin antibodies (AGA)
- Antiendomysial antibodies (EMA)
- Anti tissue transglutaminase antibodies (TTG)

Serological Test Comparison

<table>
<thead>
<tr>
<th>Test</th>
<th>Sensitivity %</th>
<th>Specificity %</th>
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<tbody>
<tr>
<td>AGA-IgG</td>
<td>69 – 85</td>
<td>73 – 90</td>
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<tr>
<td>AGA-IgA</td>
<td>75 – 90</td>
<td>82 – 95</td>
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<tr>
<td>EMA (IgA)</td>
<td>85 – 98</td>
<td>97 – 100</td>
</tr>
<tr>
<td>*TTG (IgA)</td>
<td>90 – 98</td>
<td>94 – 97</td>
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</tbody>
</table>

Endoscopic Findings

- Normal Appearing
- Scalloping
- Nodularity

Histology in Celiac Disease

- Normal duodenal mucosa
- Duodenal mucosa in celiac disease
Treatment of Celiac Disease

- Only treatment for celiac disease is a gluten-free diet (GFD)
- Advisable to see a dietitian knowledgeable in Celiac Disease
  - Strict, lifelong diet
  - Avoid:
    - Wheat
    - Rye
    - Barley

Gluten-Containing Grains to Avoid

<table>
<thead>
<tr>
<th>Gluten-Containing Grains to Avoid</th>
<th>Sources of Gluten</th>
<th>Ingredients to Question (may contain gluten)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Wheat</td>
<td>OBVIOUS SOURCES</td>
<td>- Seasonings and spice blends or mixes</td>
</tr>
<tr>
<td>Wheat Bran</td>
<td>- Bread</td>
<td>- Modified food starch</td>
</tr>
<tr>
<td>Wheat Starch</td>
<td>- Bagels</td>
<td>- Malt/ malt extract/ flavoring</td>
</tr>
<tr>
<td>Wheat Germ</td>
<td>- Cakes</td>
<td>- Modified hop extract and yeast-malt sprout extract</td>
</tr>
<tr>
<td>Flour/Meal</td>
<td>- Cereal</td>
<td>- Dextrin</td>
</tr>
<tr>
<td>Semolina</td>
<td>- Cookies</td>
<td>- Caramel color</td>
</tr>
<tr>
<td>Spelt</td>
<td>- Pasta / noodles</td>
<td></td>
</tr>
<tr>
<td></td>
<td>- Pastries / pies</td>
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</tr>
<tr>
<td></td>
<td>- Rolls</td>
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<td></td>
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<tr>
<td>Wheat</td>
<td>POTENTIAL SOURCES</td>
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<tr>
<td>Wheat Bran</td>
<td>- Candy</td>
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<td>Wheat Starch</td>
<td>- Communion wafers</td>
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<td>Wheat Germ</td>
<td>- Cured Pork Products</td>
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<td>- Drink mixes</td>
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<td>Semolina</td>
<td>- Gravy</td>
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<td>- Imitation meat / seafood</td>
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<tr>
<td></td>
<td>- Sauce</td>
<td></td>
</tr>
<tr>
<td></td>
<td>- Self-basting turkeys</td>
<td></td>
</tr>
<tr>
<td></td>
<td>- Soy sauce</td>
<td></td>
</tr>
</tbody>
</table>
Gluten-Free Grains and Starches

- Amaranth
- Arrowroot
- Buckwheat
- Corn
- Flax
- Millet
- Montina
- Oats*
- Potato
- Quinoa
- Rice
- Sorghum
- Tapioca
- Teff
- Flours made from nuts, beans and seeds

*for possible cross-contamination with gluten containing grains

Other Potential Sources of Gluten Contamination

- Lipstick/Gloss/Balms
- Mouthwash/Toothpaste
- Play Dough
- Stamp and Envelope Glues
- Vitamin, Herbal, and Mineral preparations
- Prescription or OTC Medications

Some Complications of Celiac Disease

- Short stature
- Nutritional Deficiencies
- Osteoporosis and bone fractures
- Infertility
- Gluten ataxia and other neurological disturbances
- Intestinal lymphoma

Celiac Disease Complicated by Enteropathy-Associated T-cell Lymphoma (EATL)

CT Scan Showing Occipital Calcifications in Celiac Disease

By permission of G. Holmes, Derby (UK)
Non Celiac Gluten Sensitivity

- Presence of clinical symptoms that overlap with celiac disease and wheat allergy
- Negative immune allergen test to wheat
- Negative celiac disease serology and normal duodenal histology
- Resolution of symptoms on gluten free diet (double blind)

Summary of Celiac Disease

- Celiac disease is an immune disorder that occurs in genetically predisposed individuals
- Patients have a permanent and life long sensitivity to gluten
- Symptoms may be gastrointestinal or non gastrointestinal or asymptomatic (silent)
- Diagnosis is made by a combination of endoscopy, serology and resolution of symptoms on gluten free diet
- Major complications occur if untreated
- 1st degree relatives and other high risk groups should be screened for the disease
- Treatment is a strict gluten free diet

References

- Sapone et al BMC Medicine 2012 10:13

Thank you