Pediatric Celiac Disease

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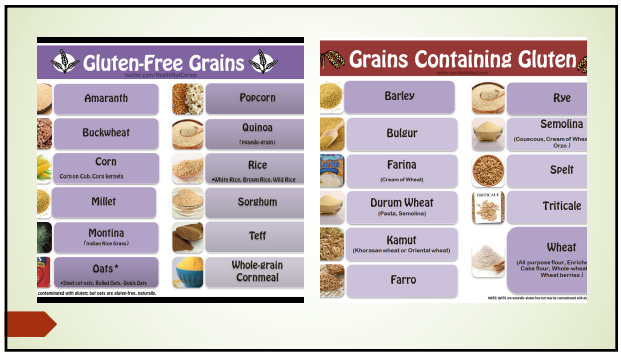
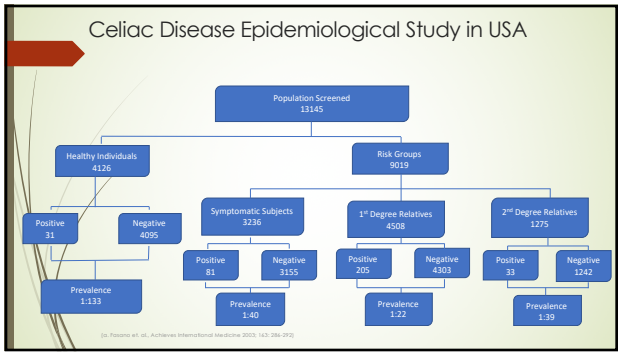
Disclosures

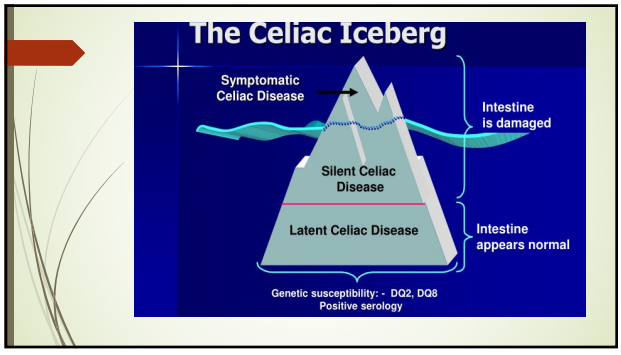
- I have nothing to disclose
- I will not discuss any off brand use of medications

Celiac Disease Is: (Objective 1)

- An inherited, lifelong intolerance to gluten
- Chronic inflammatory disorder of the small intestine
- A unique autoimmune disorder


Gluten-Free Grains
Amaranth
Buckwheat
Corn
Millet
Montina
Oats*
Popcorn
Quinoa
Rice
Sorghum
Teff
Whole-grain Cornmeal
Grains Containing Gluten
Barley
Bulgur
Farina
Durum Wheat
Kamut
Farro
Rye
Semolina
Spelt
Triticale
Wheat


Celiac Disease Epidemiological Study in USA
Population Screened 13145
Healthy Individuals 4126
Risk Groups 9019
Positive 31
Negative 4095
Prevalence 1:133
Symptomatic Subjects 3236
1st Degree Relatives 4508
2nd Degree Relatives 1275
Positive 81
Negative 3155
Prevalence 1:40
Positive 205
Negative 4303
Prevalence 1:22
Positive 33
Negative 1242
Prevalence 1:39


The Celiac Iceberg
Symptomatic Celiac Disease
Intestine is damaged
Silent Celiac Disease
Latent Celiac Disease
Intestine appears normal
Genetic susceptibility: - DQ2, DQ8
Positive serology

Why Do People Get Celiac Disease?
(Objective 2)

- Genetic predisposition
  - HLA-DQ2 & HLA-DQ8
  - Non-HLA (one may be chromosome 19, in the myosin IXB i.e. MYO98)
- Exposure to gluten (environmental trigger)
- Another trigger, such as illness, stress, other autoimmune disease
- Occurs in people of all ages
- Most common genetic disorder in North America & Europe

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)

Environmental Factors

- Feeding patterns the 1st year of life
- Potential infections
- Debated factors
  - Heavy metals

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)

Clinical Presentation

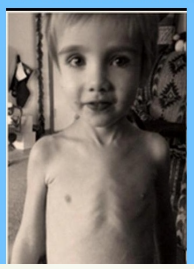
- Greatly heterogeneous depending in part on
  - Patient's age
  - Duration and extent of the disease
  - Presence of extra-intestinal comorbidities
- Various subtypes
  - Classical or Typical form
  - Atypical form
  - Silent or Asymptomatic form
  - Latent form
  - Potential form
  - Refractory form

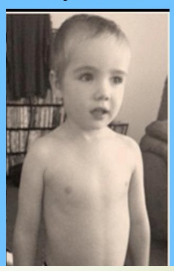
"Classic or Typical" Form
(Objective 3)

- Gastrointestinal Manifestations
  - Chronic or recurrent diarrhea
  - Abdominal distension
  - Anorexia
  - Failure to thrive or weight loss
  - Abdominal pain
  - Vomiting
  - Constipation
  - Irritability

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)

This is Anthony. He spent his first 3 1/2 years as a very sick baby and toddler. Here he is just before finally receiving a proper diagnosis of celiac disease and then just three months later after being put on a strict gluten free diet.





Atypical or Silent Form
(Objective 3)

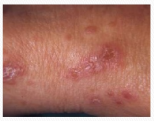
- Dermatitis Herpetiformis
- Dental enamel hypoplasia
- Osteopenia/Osteoporosis
- Short Stature
- Delayed Puberty/Infertility
- Peripheral Neuropathy/Ataxia
- Chronic Fatigue
- Normal weight or overweight
- Iron-deficient anemia resistant to oral iron
- Hepatitis
- Arthritis
- Epilepsy with occipital calcifications
- Behavioral
  - Depression
  - Poor school performance
  - Irritability

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)

Another Form of Celiac

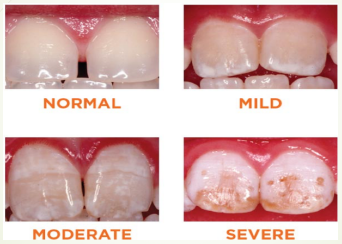
Dermatitis Herpetaformis

- Severe, itchy, blistering skin condition
- "Sister" to Celiac
- Everyone with DH has Celiac, but not everyone with Celiac has DH!!!
- Found on elbows, knees, buttock , scalp, back of neck



Blistering Dermatitis Herpetaformis

Dental Enamel Hypoplasia



Asymptomatic or Silent Form

(Objective 3)

- Most commonly diagnosed in those who also have
  - Type 1 diabetes
  - Selective IgA deficiency
  - Down syndrome
  - Turner syndrome
  - Williams syndrome
  - Autoimmune thyroiditis
  - A 1st degree relative with CD

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)

Latent Form

- Characteristic of subjects with previous asymptomatic celiac disease not on a GF diet
- Positive serology but no villous atrophy or other tissue abnormalities are recognized

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)

Potential Form

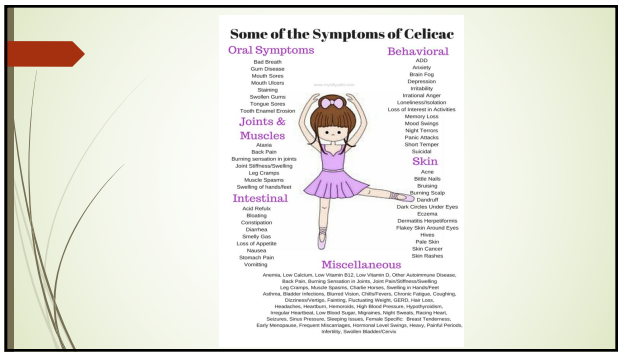
- Used in individuals who have never had a diagnosis of CD
- Show presence of appropriate genetic background
- Positive serology
- Normal or mildly abnormal histology

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)

Refractory Form

- Presence of malabsorptive symptoms and villous atrophy that persist 1 year after a strict GF diet
- Roughly 5-30% never respond to a GF diet
- Subtypes
  - Type 1
  - Type 2

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)



Diagnostic Tests
(Objective 4)

- Serological tests
  - Transglutaminase-IgA (tTG-IgA) AND deamidated gliadin peptide-IgG (DGP-IgG) antibody tests

  OR
  - Transglutaminase-IgA (tTG-IgA) antibody test AND the total IgA level

(Parzanese, I., et al. (2017). Celiac disease: From pathophysiology to treatment. World Journal of Gastrointestinal Pathophysiology, 8(2): 27-38)

Diagnostic Tests

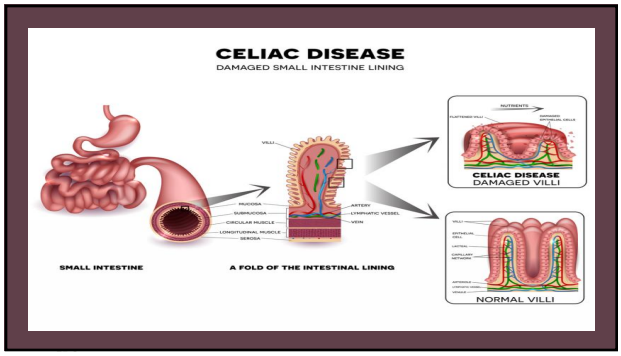
- Gene testing (HLA-DQ2/8)
  - Tests for the main genes: HLA-DQ2 and HLA- DQ8. This test is not always required but may be useful in certain situations, mainly as a way of excluding a diagnosis of coeliac disease. Some guidelines recommend it be used in conjunction with coeliac serology when assessing relatives of a person with coeliac disease to determine their potential risk for developing coeliac disease.
  - Genotyping is usually performed on a blood sample, although some pathology collection centers can use a buccal scrape (sample from inside the cheek), which avoids the need for a needle.

(Journal of Pediatric Gastroenterology and Nutrition (2005), 40: 1-19)

Diagnostic Tests

- Small Bowel Intestinal biopsy
  - Gold standard for confirming a diagnosis
  - Samples are analyzed under the microscope and the presence of typical changes to the lining of the small intestine help confirm the diagnosis of coeliac disease.

(Journal of Pediatric Gastroenterology and Nutrition (2005), 40: 1-19)



Can coeliac disease be diagnosed without small intestinal biopsies?

- The child is unwell with symptoms suggestive of CD
- The tTG antibody level is more than 10 times the upper limit of normal
- On a blood sample collected at different points in time, there is a POSITIVE endomysial antibody (EMA) result (another highly specific coeliac antibody test) AND ALSO a POSITIVE HLA-DQ2 and/or DQ8 coeliac gene test result
- The diagnosis is made by a pediatric specialist/gastroenterologist (not a general practitioner) after detailed consideration of all the circumstances
- If these criteria are not all met then the "gold standard" approach based on gastroscopy and small intestinal biopsies is necessary

Treatment
(Objective 4)

- Lifelong GF diet is the only current treatment
- Avoid milk and dairy products
- GF multivitamin

(Journal of Pediatric Gastroenterology and Nutrition (2005), 40: 1-19)

Management
(Objective 4)

- May reduce risk of developing CD if:
  - Prolong breast-feeding
  - Delaying and gradually introducing gluten in the first year of life
- Monitor
  - Assessment of symptoms, growth, and adherence to GFD
  - Measure TTG after 6 months of GFD to demonstrate a decreased antibody titer
    - Also recommended with persistent or recurrent symptoms at any time after starting GFD
    - In asymptomatic patients at intervals of 1 year or longer may help monitor adherence to GFD
- Additional biopsies are not recommended in children with symptoms and Marsh type 3 biopsy characteristics

(Journal of Pediatric Gastroenterology and Nutrition (2005), 40: 1-19)