Gastroenterology:
Congenital Abnormalities and Acquired Disorders of the GI Tract

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Outline

• Esophageal Disorders
• Vomiting
• Abdominal Pain
• Diarrhea
• Constipation
• Upper and Lower GI Bleeding
Esophageal Disorders
Esophageal Disorders

- Caustic Ingestions
- Foreign Bodies
- GERD
- Eosinophilic Esophagitis
Esophageal Disorders: Caustic Ingestions

• Include alkalis, acids, bleach, detergent, button batteries
  • **Alkalis** cause most profound injury: deep liquefaction necrosis potentially through all esophageal layers
• Most common caustic ingestions: household cleaners
• Peak age is < 5 years old
Esophageal Disorders: Caustic Ingestions

- **Presentation: signs and symptoms:**
  - Drooling, refusal to drink, nausea & vomiting, epigastric pain, oral ulcerations (not always present), fever and leukocytosis
  - Beware of esophageal perforation (especially with button batteries!)
  - Esophageal strictures develop over weeks, and cause dysphagia and weight loss
Esophageal Disorders: Caustic Ingestions

**Treatment:**

- Upper airway edema may require intubation
- Hospitalization, NPO, IVFs
- Broad spectrum antibiotics for suspected perforation, mediastinitis
- EGD w/in 48 hours: preferred method of staging injury
  - Early detection and dilation important
  - Nasogastric tube placement to establish feeding route
  - Strictures more common in circumferential burns
Esophageal Disorders: Foreign Bodies

• Typically lodge below cricopharyngeal muscle, at the aortic arch, or just above diaphragm
• Coins most common (< 5 yo)
• Symptoms: Coughing, drooling, choking, refusal to take POs, pain, dysphagia
Esophageal Disorders: Foreign Bodies

• **Diagnosis:**
  - PA & lateral films (radiopaque)
  - Radiolucent bodies must be evaluated by endoscopy (some plastic toys, fishbones)

• **Treatment:**
  - Endoscopy: therapeutic removal and assess mucosa for injury
  - Sharp objects (safety pins), and button batteries must be removed emergently
    - Can cause significant injury and perforation in < 4 hours
  - Gastric foreign bodies usually only require follow-up for passage into stool
Esophageal Disorders: GE Reflux

• Physiologic reflux:
  • 50% of all infants have physiologic reflux (“happy spitters”, worse when lying down)
  • Peaks at 4-6 months of age

• Reflux associated with symptoms is termed GERD
  • Abnormal beyond 1 year of life
  • More common in Down syndrome, MRCP, prematurity, family history of severe GERD, h/o TEF
Esophageal Disorders: GERD Presenting Symptoms

**Infants**
- Recurrent vomiting
- Poor weight gain
- Irritability
- Apnea/ ALTE
- Upper airway symptoms/ Reactive airway disease
- Sandifer syndrome

**Older Children**
- Recurrent vomiting
- Poor weight gain
- Heartburn/ Chest pain
- Dysphagia/ odynophagia
- Asthma/ RAD
- Recurrent pneumonia
Esophageal Disorders: GERD Pathophysiology

- Transient lower esophageal sphincter relaxations
- Delayed gastric emptying
- Hiatal hernia
Esophageal Disorders: GERD

• Diagnosis:
  • **Esophageal pH monitoring** gold standard
  • **Upper GI** defines anatomy
    • GER is episodic, may be seen on UGI, but not diagnostic
  • Nuclear medicine **gastric emptying study**
  • **Endoscopy** may be used to detect esophagitis (chronic / severe), but is rarely indicated
Esophageal Disorders: GERD

• Treatment:
  • GER self resolves in 95% of all healthy infants by 1 year of life ("reassurance", reduce overfeeding)
  • Upright position, thickened formula
  • Promotility agents
    • Erythromycin: motilin agonist
    • Metoclopramide: 5HT4 agonist, EPS: dose related side effect
  • H2 blockade, PPIs for symptomatic relief, decrease gastric fluid volume and neutralize acid
  • Anti-reflux surgery: Nissen fundoplication successful in 90% of patients
Esophageal Disorders: Eosinophilic Esophagitis

- Chronic immune mediated esophageal disease
- Male, caucasian predominance
- Symptoms related to esophageal dysfunction:
  - Infants/ toddlers: feeding difficulties, FTT
  - School age: nausea, vomiting, pain
  - Teenagers: solid food dysphagia/ food impaction (meat, dry bread)
- Personal/FH atopy: eczema, allergic rhinitis, food allergy, asthma
Esophageal Disorders: Eosinophilic Esophagitis

• Diagnosis

  • EGD gross appearance - white specks, mucosal edema, linear furrows
  • Microscopic /biopsy appearance- ≥ 15 eosinophils/ high power field throughout the esophagus
    • Note: GERD tends to involve distal 1/3 of the esophagus

![Endoscopic pictures showing rings (crinoidization), white exudates, linear furrowing](image)

![Microscopic image of eosinophilic esophagitis](image)
Esophageal Disorders: Eosinophilic Esophagitis

**Treatment:**

- Often refractory to PPI trial
- Elimination diet
  - Identify & avoid trigger foods (milk, wheat, egg, soy, nuts)
- Topical steroids (fluticasone, viscous budesonide)
- Leukotriene inhibitors
- Esophageal dilation
Case #1: Esophageal Disorders

• A 6 month old baby boy is brought into the office with a chief complaint of irritability, and frequent vomiting. Emesis is non-bloody, non-bilious and occurs after shortly after most feeds. Child is formula fed (cow’s milk protein formula) since 2 months of age. 95% wt & 90% ht for age. The baby stools regularly BID. No signs of eczema on physical exam.
Case #1: Esophageal Disorders

Which of the following is the most appropriate workup?

1. Upper GI
2. Trial of soy-protein formula
3. 24 intra-esophageal pH probe
4. Reassurance
5. Nuclear medicine gastric emptying study
Vomiting
Vomiting

- Nonbilious
- Biliary: Malrotation, Volvulus, Intestinal Atresia
- Projectile
- Cyclic Vomiting
Nonbilious Vomiting: Potential Etiologies

• Infectious: AGE, PNA, UTI, AOM
• Inflammatory: PUD/gastritis, pancreatitis, hepatitis
• Motility disorders: gastroparesis, pseudo-obstruction, CHF
• CNS: tumor, meningitis
• Endocrine: DKA, inborn errors of metabolism, thyroid, adrenal
• Renal: uremia
• Medications/ Ingestion
Nonbilious Vomiting: Clues to Etiology

- Fever suggests infection/inflammation
- Tenderness/guarding: inflammation
- Skin: sclerodactyly in scleroderma
- Loss of dental enamel: severe GERD, bulimia
- Adenopathy: neoplasm
- High pitched bowel sounds: obstruction
- Absent bowel sounds: Ileus
- Papilledema: CNS
Bilious Vomiting: Potential Etiologies

- Malrotation
- Volvulus
- Intestinal Atresia
- Obstruction: Adhesions, SMA syndrome
Bilious Vomiting: Malrotation

- 3rd portion of duodenum lies to R of vertebral column; Cecum in upper abdomen
- Mesentery not attached to the posterior abdominal wall
- Midgut is prone to twist around SMA
- Risk of volvulus: highest in neonates, may present at any age with severe/recurrent abd pain + bilious emesis

- Workup: UGI
- Treatment: Surgical emergency

- R sided jejunum, duodenal obstruction w/ beak shape, corkscrew duodenum midline cecum.
Vomiting: Diagnostic Workup

- KUB and upright
  - proximal air-fluid levels with paucity of air distally (obstruction) vs. diffuse air-filled distended loops (ileus, pseudo-obstruction)
- Upper GI+ SBFT: focal obstruction
- Abdominal Ultrasound or CT: Inflammatory causes, mass
- MRI brain: increased intracranial pressure, CNS lesion
Projectile Vomiting: Pyloric Stenosis

• Progressively worse, postprandial, non-bilious, first-born males, 4 weeks old

• Hypochloremic, hypokalemic alkalosis

• Diagnosis:
  • Palpable epigastric mass
  • UTZ (thickness > 3 mm, length >14 mm)
  • UGI: String sign

• Treatment: rehydration/electrolyte stabilization + pyloromyotomy
Recurrent Vomiting: Cyclic Vomiting Syndrome

• Triad:
  • ≥ 3 recurrent episodes of intense nausea & NBNB vomiting
  • Intervals of completely normal health between episodes
  • Episodes are stereotypical in timing of onset, symptoms, & duration

• Typically school-aged
• Episodes last hours - a few days
• At risk of developing migraines & IBS
• Treatment: if severe → IV hydration, ondansetron, lorazepam
Cyclic vomiting syndrome

• Differential: non-GI causes
  - Medications, toxins
  - CNS (↑ICP, tumor, hemorrhage, abscess, meningitis, congenital malformation)
  - Psychiatric (anorexia, bulimia, depression, anxiety)
  - Pancreatitis, metabolic defects

• Treatment: on occasion, IV hydration, ondansetron, lorazepam
Case #2: Vomiting

You are called by the newborn nursery to evaluate an infant born 12-hours ago who has had bilious emesis x 2. The prenatal and birth history are unremarkable.

Which of the following radiographic findings would likely be seen in this child?

1. Hypertrophic pylorus on ultrasound
2. GE Reflux on UGI
3. Choledochal cyst on CT scan
4. A “double bubble” sign on KUB
Abdominal Pain
Abdominal Pain

• Acute Abdominal Pain
  • Appendicitis
  • Intussusception
  • Pancreatitis
• Chronic Abdominal Pain
  • Functional abdominal pain (recurrent)
  • Irritable Bowel Syndrome (IBS)
  • Dyspepsia
  • Abdominal Migraines
Acute Abdominal Pain: Appendicitis

• Lifetime risk 7%
  • Peak incidence males 10-14 y/o, females= 15-19 y/o
• Natural course: 20% perforation rate within 24 hrs of start symptoms; 80% perforation within 48 hrs
  • Observation of early suspected appy in the ER/clinic is key
  • Timely involvement of pediatrics surgery is as important
  • Must be in DDx of any child with severe abdominal pain
Appendicitis: History

• AGE: emesis often prior to pain vs. Appy: pain is often first
• Immediately after perforation:
  • The patient may feel better temporarily, followed by desire to lie still, low grade F, ↑ vomiting (which can become bilious) due to ileus
Appendicitis- Physical Exam

- Appearance: Listless, low grade fever
- ↓ Bowel Sounds (vs. AGE= hyperactive)
- Periumbilical pain/tenderness migrating to RLQ over time
- As peritoneal irritation worsens → guarding/rebound.
Appendicitis: Workup

- RUA: sterile pyuria
- ↑ WBC w/ shift to left.
- Plain films may be helpful to r/o obstruction or perforation
- UTZ: normal appendix is not well seen
  - Operator/Interpreter dependent
- Thin cut CT with oral/IV/rectal contrast
  - Wall thickening, fat stranding, appendicolith
  - Sensitivity & specificity as high as 94%
Appendicitis: Differential Diagnosis

• AGE
• Constipation
• UTI
• IBD
• Ovarian Cyst
• PID
• Pneumonia
• Mesenteric adenitis
Appendicitis: Treatment

- Broad spectrum antibiotics (gram neg & anaerobes)
- Pain control once diagnosis is made/ seen by surgery
- Generous IVFs
- Appendectomy
Acute Abdominal Pain: Intussusception

• Previously healthy, infant/toddler with triad of:
  • episodes of colicky abd pain
  • bilious emesis
  • “currant jelly stool” = mixture blood, exudate & stool.

  • Exam may reveal palpable sausage mass.
Acute Abdominal Pain: Intussusception

- Abdominal UTZ vs. air or contrast enema (diagnose & reduce)
- Peds surgery on-call/available in case of perforation or inability to reduce
Acute Abdominal Pain: Pancreatitis

- Epigastric or RUQ abdominal pain
- Anorexia/ Nausea/ Vomiting
- Rebound tenderness or rigidity
- Abdominal distention & diminished bowel sounds
- Fever

- Severe cases: Hypotension / shock
Common Causes of Pancreatitis in Children

- Idiopathic (20-25%)
- Mechanical/Structural
  - **Trauma, Gallstones**, Post-ERCP, Post-surgical, Tumor obstruction, Pancreatic divisum, Choledochal cyst
- Infection:
  - Sepsis/shock, bacterial, viral & parasitic
- Hereditary
  - Mutations in gene encoding for trypsin (SPINK, PRSS)
  - 1% of chronic/recurrent cases
- Inflammatory disorders
  - SLE, Dermatomyositis, HSP, Crohn’s
- Metabolic
  - CF, Hypertriglyceridemia, Hypercalcemia, DKA, Malnutrition, Organic acidemias
- Drugs/Toxins
  - Chlorothiazides, Furosemide, Tetracyclines, Sulfonamides, Estrogens/ OCPs, 6-MP, L-Asparaginase, Valproic acid
  - EtOH, Heroin, Amphetamines,
  - Acetaminophen
Pancreatitis: Pathogenesis

1. Blockage/damage to collecting ducts and acinar cells
2. Activation and release of digestive enzymes
3. Auto-digestion of parenchyma with inflammation
4. Interstitial edema, pancreatic necrosis, blood vessel occlusion/disruption, SIRS
Pancreatitis: Laboratories & Imaging

• Amylase: ↑ within hours, peaks at 48 hrs, remains ↑ for 4 days in uncomplicated cases
  • False elevation: intestinal, salivary, ovarian, CKD
• Lipase: more specific for pancreatitis. Remains ↑ longer than amylase levels (8-14 days)
• Radiographic evidence of pancreatitis, abnormal ducts, stones are important to look for
  • Ultrasound, CT, MRCP, ERCP
## Pancreatitis: Management

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<tr>
<td>NPO (&quot;pancreatic rest&quot;), followed by early feeds once stable</td>
<td>IV hydration</td>
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<td>Nasogastric decompression</td>
<td>Pain management</td>
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<td>$H_2$-blockers, PPI’s</td>
<td>Monitoring for complications</td>
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<td></td>
<td>• Antibiotics</td>
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<td>• Surgical consult</td>
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Chronic abdominal pain

- Functional abdominal pain (recurrent)
- Irritable Bowel Syndrome (IBS)
- Dyspepsia
- Abdominal migraine
Functional abdominal pain

• Definition: Recurrent abdominal pain without organic cause
  • Occurring over weeks - months (present at least once/week x past 2 months)
  • Some loss of daily activity
  • Most common cause of abdominal pain in children

• Etiology
  • Visceral hyperalgesia
Functional abdominal pain

• Presentation: Signs and symptoms
  • Non-specific, generalized or periumbilical pain
  • No associated “red flags”
    • No vomiting or diarrhea, no hematochezia, no fever, rash, joint pain, weight loss, or growth failure
    • School absence, missed activities

• Diagnosis:
  • Careful history and physical
Functional abdominal pain

**Treatment:**

- Reassurance
- Supportive: distraction, guided imagery, biofeedback, counseling, close follow up
- Anticholinergic medications
- If disruptive (school absences, missed activities) or superimposed anxiety: TCA, SSRI
Irritable bowel syndrome (IBS)

• Definition:

  • The presence of abdominal pain which is either relieved by defecation, or the onset of which is associated with a change in form or frequency of bowel movements
  • At least once per week over the course of 2 months
  • No organic cause suspected or identified (NO RED FLAGS!!)
  • Tends to occur in families

• Treatment

  • Supportive: Dietary changes (reducing sorbitol, fructose; increasing soluble fiber); TCAs/SSRIs?
Non-Ulcer Dyspepsia

- Symptoms suggest peptic ulcer disease
  - Pain centered in upper abdomen
- But no signs of gastritis on endoscopy, esophageal pH testing negative
- NOT characterized by: pain radiating to back, bilious emesis/ hematemesis, melena, weight loss, anemia, other constitutional symptoms…
Abdominal migraine

• Acute, incapacitating, periumbilical pain
• May include anorexia, nausea, vomiting, headache, pallor, photophobia
• Symptom-free periods (weeks to months)
• Often family history of migraines
• Treatment: remove triggers (caffeine, nitrates, stress), pharmacologic: propanolol, cyproheptadine, amitriptyline
• DO NOT order labs/imaging for functional pain
Diarrhea
Diarrhea

- Stool volume > 10 mL/kg/day (up to 20 kg), or > 200 mL/day
- Decrease in consistency & increase in frequency of stool
- Acute vs. chronic (2 weeks)
Diarrhea: Pathophysiology

• 4 processes that may contribute to diarrhea

  • **Secretory**: enterotoxin stimulates secretion of fluid and electrolytes from mucosal crypt cells
  
  • **Cytotoxic**: destruction of small bowel villi by infectious agent → decrease small bowel surface area & absorptive capacity
  
  • **Osmotic**: malabsorbed substance causes a net flux of water into the intestinal lumen → loose stools, gas, bloating, abd pain
  
  • **Inflammatory**: inflammation of mucosa and submucosa of small & large bowel; may be accompanied by tenesmus, GI bleeding & urgency
Infectious Causes of Acute Diarrhea

**Secretory**
- E. coli
- Vibrio cholerae
- Clostridium difficle
- Clostridium perfringes
- Aeromonas hydrophila
- Staphylococcus aureus
- Vibrio parahemolyticus
- Bacillus cereus
- Giardia lamblia

**Cytotoxic**
- Rotavirus
- Norwalk agent
- Cryptosporidium
- Escherichia coli

**Inflammatory**
- Campylobacter fetus
- Clostridium difficle
- Salmonella
- Shigella
- Yersinia enterocolitica
- Entamoeba histolytica
Acute Diarrhea

• **Associations:**
  • *Reactive arthritis*: Salmonella, Shigella, Yersinia, Campylobacter
  • *Guillain-Barre*: Campylobacter
  • *Bloody diarrhea, HUS*: Enterohemorrhagic E.coli
  • *Short incubation*: C. perfringens, B. cereus
  • *Seizures*: Shigella
Acute Diarrhea

• Viral
  • Rota, adeno, astro, norovirus: fever, 1-2 days vomiting followed by 5-7 days diarrhea, no stool WBCs

• Parasitic
  • Giardia, cyclospora, isospora
  • Routine O+P not indicated unless endemic area, outbreak, stool cultures negative, diarrhea persists >1 week
Acute Diarrhea: Treatment

• Treatment: Antidiarrheals are NOT to be used in children
• BRAT diet may be too restrictive (AAP: continue to feed age-appropriate diet)
• Oral rehydration solution
• Avoid juices, soft drinks (dairy only if prolonged)
Acute Diarrhea – Age of Onset

- Neonatal diarrhea beginning in first month of life
  - Allergic causes: cow’s milk protein allergy (formula or breast milk)
  - Dissacharidase deficiency: sucrase-isomaltase, glucose-galactose malabsorption
  - Pancreatic insufficiency: Schwachman-Diamond syndrome (skeletal abnormalities)
  - NEVER think lactase deficiency in infancy: congenital LD is 1:1,000,000
Diarrhea in Infants: Sucrase-isomaltase deficiency

- Rare, autosomal recessive disorder
- Absent sucrase, diminished maltase activity
- Symptoms depend on time of introduction of pureed fruits (or juices)
  - Diarrhea, abdominal pain, and/or bloating when sucrose is ingested
- Starch and cornstarch (isomaltose) also cause symptoms
Diarrhea in Infants: Sucrase-isomaltase deficiency

- Reducing substances in stool will not detect this deficiency (sucrase non-reducing)
- Can detect with hydrogen breath testing or enzyme activity in of small bowel biopsy
- Treatment: restriction of sucrose containing foods, enzyme replacement (Sucraid)
Diarrhea in Infants: Glucose-galactose malabsorption

- Rare, autosomal recessive
- Presents at birth with diarrhea, dehydration and metabolic acidosis in breast fed and lactose containing formula fed infants
- Fructose only carbohydrate is tolerated
Diarrhea in Infants: Pancreatic insufficiency

• Cystic fibrosis
  • Most common cause of malabsorption (steatorrhea) among white American children of European descent
  • Pancreatic insufficiency (PI) with malabsorption is seen in 90% of CF patients by one year of age
  • Dysfunction of CFTR
  • Treatment of pancreatic insufficiency with enzyme replacement leads to better absorption, growth /weight gain & normal stools
  • Test for PI with fecal elastase and fecal fat
Diarrhea in Infants: Pancreatic insufficiency

- Schwachman-Diamond Syndrome
  - Pancreatic insufficiency, bone marrow dysfunction, cyclic neutropenia/defect in neutrophil chemotaxis, skeletal abnormalities (metaphyseal dysostosis), FTT/short stature
  - 2nd most common cause of PI in children (behind CF)
  - Autosomal recessive, defect in SBDS gene on Chr. 7 (ribosome dysfunction)
  - Present with oily, sticky, foul smelling stools
Diarrhea in Infants: Schwachman-Diamond

- Differentiated from CF by normal sweat chloride testing/negative CF genetic mutation + hematologic & ortho manifestations
  - Thrombocytopenia (70%), Anemia (50%)
  - Genetic testing can be performed
- Often have poor growth with treatment of PI
- Pyogenic infections major cause of morbidity and mortality (neutropenia)
Acute Diarrhea – Age of Onset

• One month to two years of age

  • Toddler’s Diarrhea: excess fructose/sorbitol
  • Rotavirus-leading cause of diarrhea in infants/ toddlers worldwide, (but with vaccination, being replaced by norovirus and adenovirus)
  • Parasitic - Giardia lamblia (daycare, stool antigen, metronidazole), cryptosporidiosis
  • Less common: CF, lymphangiectasia (PLE)
Chronic Diarrhea – Age of Onset

- Age two to eighteen years of age
  - Post-infectious – parasitic, bacterial
  - Chronic nonspecific diarrhea (IBS)
  - Lactase Deficiency
  - Excessive juice (containing sorbitol) intake
  - Gluten sensitive enteropathy (Celiac disease)
  - Inflammatory bowel disease (IBD)
Chronic Diarrhea Older Children: Lactase deficiency

- Congenital presentation (extremely rare!)
- Late onset
  - Most common cause of genetically determined carbohydrate malabsorption
  - Results from decline in intestinal lactase specific activity with age, typically begins after age 3 years
  - Decline in lactase levels varies among ethnicities: 40% of Asian American, 85% of African American adults
  - Diagnosed by lactose breath hydrogen testing, lactase activity on duodenal biopsy taken by upper endoscopy, or by simple challenge/withdrawal
  - Treatment: lactose-free diet; lactase supplementation
Chronic Diarrhea: Celiac Disease

• Immune-mediated enteropathy caused by permanent sensitivity to gluten in a genetically predisposed host
  • Gluten is found in wheat, rye & barley
  • Suspect celiac disease in other immune-mediated diseases (IgA deficiency, Type I DM, dermatitis herpetiformis, autoimmune thyroiditis), 1st degree relatives, Trisomy 21

• U.S. incidence = 1:133
Chronic Diarrhea: Celiac Disease

- Most commonly presents 6-24 months of age
- Presenting symptoms:
  - Chronic diarrhea (bulky, frothy, foul smelling stools)
  - Abdominal pain & distension
  - Proximal muscle wasting
  - Weight loss/ FTT
  - Anorexia
Celiac: Non Gastrointestinal Manifestations

Often in older children/adolescents

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

- Circulating autoantibodies
  - IgA, IgG
    - Serologies must be checked with total IgA, as IgA deficiency will result in false negative test
  - Tissue transglutaminase: 87% sensitivity, 95% specificity
  - Antiendomysial antibodies increase sensitivity of testing
  - Anti-gliadin antibody screening not recommended: poor sensitivity & specificity
- HLA typing:
  - Useful in ruling out celiac, helps diagnose patients already on gluten-free diet
Celiac Disease: Diagnosis/ Treatment

- Endoscopy with biopsies
  - Gold standard (villous atrophy)
    - Characteristic features on small intestinal biopsy
- Clinical response to withdrawal of all gluten from the diet
  - Significant weight gain/relief of symptoms
Case #3: Celiac Disease

• A 10 year old boy with Type I Diabetes Mellitus has always been shorter than his siblings and peers. Parents are of European descent. Dad is 6’0”, Mom is 5’10”. The boy has had an itchy vesicular rash on the back of his neck and on his elbows for the past 6 months. His paternal grandfather died of non-Hodgkin lymphoma. Mom has had the whole family on a gluten-free diet for years because she feels it provides certain health benefits. You have read about the importance of screening children with Type I DM for celiac disease.
You tell the child’s parents:

1. Restart a gluten containing diet, as the risk of celiac disease is likely low
2. The child needs an upper endoscopy as celiac serologies will likely be negative
3. Performing HLA testing (DQ2, DQ8) will provide valuable information regarding the risk of having celiac disease
4. Stool studies can be performed to make the diagnosis of celiac disease
Chronic Diarrhea: IBD- Crohn’s Disease

- Chronic idiopathic inflammatory bowel disease, transmural, affects GI tract anywhere from mouth to anus, skip areas with intervening normal bowel
  - >50% of patients present with ileocolitis
  - Bimodal age distribution, first peak during adolescence
  - Incidence: increasing, 1/30,000
  - Small bowel disease more likely obstructive, RLQ pain, fibro-stenotic
  - Colonic disease more likely to cause cramping, diarrhea, bleeding
  - Evolves over time: inflammatory becomes stricturing, can perforate
Inflammatory bowel disease: Crohn’s Disease

- **Systemic signs** more prevalent than in UC: malaise, anorexia, growth failure/weight loss (may be presenting sign), pubertal delay
- Perianal disease common (skin tags, fistulae)
- Penetrating disease may cause fistulae
  - Between loops of bowel, from bowel to vagina or bladder, or from bowel to skin
- **Extraintestinal manifestations** more common than in UC: arthritis, aphthous ulcers, erythema nodosum, clubbing, episcleritis/uveitis, renal stones and gallstones
Inflammatory bowel disease: Crohn’s Disease

**Diagnosis:**

- CBC often shows anemia, thrombocytosis
- Hypoalbuminemia common
- Serologies: Anti-saccharomyces antibodies
- Fecal calprotectin and lactoferrin (neutrophil proteins shed in stool)
- Radiology studies:
  - Upper GI/SBFT: thumbprinting, cobblestoning
  - MR or CT enterography
- Upper endoscopy and colonoscopy: non-caseating granulomas = hallmark
Inflammatory bowel disease: Crohn’s disease

**Treatment:**

- Induction and maintenance of remission
- Corticosteroids, aminosalicylates (mesalamine), antibiotics (metronidazole), immunomodulators, anti-TNF biologics (infliximab)
- Nutritional support
- Medications are NOT curative, they decrease morbidity (induce, maintain remission)
- Steroids induce remission in 70% of patients, but high relapse rate after weaning
Inflammatory bowel disease: ulcerative colitis

- Limited to mucosal layer of colon; spares upper GI tract
- Children much more likely to have pancolitis (80%) where adults more likely to have limited distal colitis
- Incidence: stable, 1/6,500
- Presentation: bloody (mucoid, purulent) diarrhea, tenesmus, urgency, frequency, cramping, nocturnal stools
- Chronic: symptoms persist beyond 2 weeks
- Extraintestinal symptoms: more likely to have pyoderma gangrenosum, PSC, chronic active hepatitis, ankylosing spondylitis (HLA B27)
Inflammatory bowel disease: ulcerative colitis

• Diagnosis: colonoscopy with biopsies shows histologic chronic change (crypt abscesses, crypt branching)
• Pseudopolyps, continuous erythema, loss of vascularity
• No perianal disease
• Severe disease: toxic megacolon potential complication, fevers, hypoalbuminemia
Inflammatory bowel disease: ulcerative colitis

- 5% of UC patients enter prolonged remission, 25% of those with severe UC at diagnosis will require colectomy within 5 yrs of diagnosis
- Risk of colon CA increases after 8 yrs of diagnosis by ~ 0.5% per year, requiring surveillance colonoscopies every 1-2 years for dysplasia (colectomy then recommended)
IBD: UC treatment

• 5-ASA (mesalamine) prevents relapses

• Surgery
  • For intractable or fulminant colitis, complications of therapy, dysplasia
  • Total colectomy with endorectal pull-through (ileal pouch anal anastomosis, J pouch), temporary ileostomy
  • Pouchitis possible complication, requiring antibiotic therapy
  • Probiotics shown to prevent pouchitis
Chronic Diarrhea: Crohn’s vs UC

- UC: Continuous colitis, mucosal inflammation, cured by surgical excision
- CD: Skip lesions from mouth to anus, cobblestoning, non-caseating granulomas, (string or thumb-print sign on SBFT), excision NOT curative, growth retardation/pubertal delay more common, transmural inflammation
Constipation
Constipation

• Functional Constipation
• Hirschsprung Disease (HD)
## Functional Constipation vs. Hirschsprung

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<th>Functional Constipation</th>
<th>Hirschsprung</th>
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<tr>
<td>Onset</td>
<td>After 2 y/o (toilet training)</td>
<td>Birth</td>
</tr>
<tr>
<td>Encopresis</td>
<td>Common</td>
<td>Very rare</td>
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<tr>
<td>Stool size</td>
<td>Very large</td>
<td>Small, ribbon-like</td>
</tr>
<tr>
<td>Enterocolitis</td>
<td>None</td>
<td>Possible</td>
</tr>
<tr>
<td>FTT</td>
<td>Uncommon</td>
<td>Common</td>
</tr>
<tr>
<td>Anal tone</td>
<td>Normal</td>
<td>Tight</td>
</tr>
<tr>
<td>Rectal exam</td>
<td>Large rectum full of stool</td>
<td>small, empty rectum</td>
</tr>
<tr>
<td>Treatment</td>
<td>Dietary, laxatives</td>
<td>Surgery</td>
</tr>
</tbody>
</table>
Constipation: HD Presentation

- Infants
  - 95% fail to pass meconium in first 24 hrs
  - 2/3 abdominal distension, vomiting, constipation

- Older children
  - Chronic constipation (from birth)
  - Abdominal mass/distention
  - Ribbon-like stools
  - Enterocolitis
Constipation: HD- Pathogenesis

• The enteric nervous system is formed by cells that migrate to the bowel from the neural crest

• Failure of craniocaudal migration of neural crest ganglion cells during the 5th-12th week of gestation.
  
  • The earlier it occurs the longer the segment (85% rectum and sigmoid (short segment), 10% total colon (long segment)
  
  • Absence of ganglion cells interrupts expression of parasympathetic nerves in the myenteric plexus
    • Therefore there is a failure of relaxation
Constipation: Hirschsprung Disease

• **Diagnostic modalities**
  
  • Barium enema – transition zone
  
  • Anal manometry – abnormal response
  
  • Rectal suction biopsy – absence of ganglion cells
Constipation: HD Diagnosis - Barium Enema

- Bowel proximal to the aganglionic segment becomes dilated due to the distal obstruction. This produces the appearance of a transition zone.
  - 80% show transition zone
- B.E. should be unprepped; no rectal manipulation 48 hrs prior.
Constipation: HD Diagnosis- Anorectal Manometry

![Diagram of anorectal manometry]

**Normal Response to Rectal Distension**

- **internal sphincter**
- **external sphincter**
- **intrarectal**

![Graph of normal response to rectal distension]

Mattel Children's Hospital UCLA
Constipation: HD Diagnosis - Rectal Biopsy

• Gold standard in diagnosing HD
• Absence of ganglion cells in the myenteric (Auerbach’s) plexus and submucosal (Meissner’s) plexus defines HD
• Biopsies are typically taken at 3, 4, and 5cm
• Occasionally not diagnostic and full thickness surgical biopsies are required
Constipation: HD- Enterocolitis

• 20% of HD patients may present with enterocolitis
  • Fever, abdominal distension, loose/bloody stools, sepsis

• Pathophysiology
  • Mucosa compromised by distention, → stasis of fecal contents/bacterial overgrowth → translocation → foul-smelling, bloody diarrhea & sepsis-like picture

• Treatment
  • Broad spectrum antibiotics, NG tube decompression, surgery
Case #4: Constipation

- A 4 y/o boy with Trisomy 21, who recently immigrated from central America is accompanied by his aunt for his first well-child check up in the U.S. He is noted to have significant abdominal distension. It is unclear if he passed stool in the first 48 hours of life. His aunt states he passes thin stools once per week and has required enemas for some time.
Case #4: Constipation

Which of the following is true regarding this child?

1. Encopresis would be quite rare in this child
2. He likely developed issues with constipation around the time of toilet training
3. Dietary education, increased fluid intake and polyethylene glycol will help this child a great deal
4. Rectal exam will reveal an enlarged rectum full of stool
GI Bleeding
GI Bleed: Localizing the Source

• Upper GI Bleed
  • Proximal to the ligament of Treitz
  • Hematemesis/coffee-ground emesis, melena (black, tarry stool)

• Lower GI Bleed
  • Tend to present with hematochezia (BRB per rectum or maroon colored stools)
Differential Diagnosis LGI Bleeding: Newborn- 2 month

- Sepsis/ DIC
- Necrotizing Enterocolitis
- Malrotation w/ Volvulus
  - (Bilious emesis, abd pain/distension, melena/BRBPR late)
- Hirschsprung Disease Enterocolitis
- Hemorrhagic Disease of Newborn
  - (Vit K refusal, begin 2-7 days, other organ systems)
- Swallowed Maternal Blood
  - Apt-Downey Test: maternal vs fetal Hgb
- Allergic Proctocolitis (Cow’s milk protein allergy)
Lower GI Bleed: Cow’s Milk Protein Allergy

• Typically occurs in first few weeks - months of life
• Prevalence: 2-3% of infants
• Often well appearing, normal wt gain, occult or gross blood in stool ± eczema, + FH, may occur in breast fed infants
• Diagnosis: Breast fed infants first eliminate milk/ dairy products from mom’s diet; vs. formula fed: → soy protein*, hydrolyzed or free amino acid based formula.
• If diagnosis in doubt → sigmoidoscopy (grossly focal erythema; histology= eosinophils in mucosa & lamina propria)
• 1 y/o re-challenge; excellent long-term prognosis

*40 also sensitized to soy protein; may have honeymoon period of 2 weeks
Case#5: LGI Bleeding in a Toddler

• You are working in the pediatric ER when a 2 y/o previously healthy female presents w/ a large amount of BRB per rectum.
• No recent illnesses. No associated diarrhea, abdominal pain, nausea or vomiting
• In the ER the child is pale, HR 170, BP: 95/ 40, cap refill 3 sec
• Hemacue= 4.5 g/dL
• Which is the most appropriate diagnostic test to order?
Case#5: LGI Bleeding in a Toddler

Which of the following is the most appropriate diagnostic test to order?

1. CT scan with oral and IV contrast
2. Nuclear medicine Technetium-99m scan
3. Stool studies for C Dificile, bacterial culture, O&P
4. Angiogram
LGI Bleeding: Meckel’s Diverticulum

- Ectopic gastric mucosa is present in 90% of children
  - Bleeding: due to peptic ulceration of the adjacent ileum

- Most common congenital anomaly of GI tract; Remnant of the omphalomesenteric duct (vitelline duct).

- Most common cause of painless & significant LGI bleeding in toddlers

- Rule of 2s: 2% of the population, 2:1 M:F prevalence, majority under 2 y/o, most within 2 feet from ileocecal valve, 2 cm in length
LGI Bleeding: Meckel’s- Diagnosis

- Meckel’s scan = Nuclear medicine test which identifies ectopic gastric mucosa
- Sensitivity & specificity for identifying Meckel's are 85% & 95% respectively
## Additional Tests to Localize GI Bleeding

<table>
<thead>
<tr>
<th>Test</th>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
<tbody>
<tr>
<td>Endoscopy</td>
<td>Diagnostic &amp; Therapeutic</td>
<td>Areas of small bowel unreachable</td>
</tr>
<tr>
<td>Capsule Endoscopy</td>
<td>Noninvasive</td>
<td>Must be able to swallow capsule</td>
</tr>
<tr>
<td></td>
<td>Screens entire GI tract</td>
<td>No biopsies</td>
</tr>
<tr>
<td>Tagged RBC Scan</td>
<td>Localize Upper vs. Lower Bleed</td>
<td>Imprecise</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Requires Brisk Bleed</td>
</tr>
<tr>
<td>Angiography</td>
<td>Diagnostic &amp; Therapeutic</td>
<td>Invasive</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Requires Brisk Bleed</td>
</tr>
</tbody>
</table>
GI Bleeding: Initial Assessment/ Management

• ABCs
  • Tachycardia = sensitive indicator of blood loss vs. cap refill, hypotension & widened pulse pressure = later signs

• Access
  • 2 large-bore IVs
  • Labs: CBC c plts, PT, PTT, Chem 7 (BUN>>Cr in UGI bleed), Type and Cross.
Differential Diagnosis LGI Bleeding in Toddlers

- Meckel’s Diverticulum
- Anal fissure
- Infectious Colitis
- Allergic Proctocolitis
- Intussusception
- Lymphonodular Hyperplasia
- Malrotation w/ Volvulus
- Hirschsprung Disease Enterocolitis
Differential Diagnosis LGI Bleeding: School Age

- Anal Fissure
- Infectious Colitis
- Lymphonodular Hyperplasia
- Henoch-Schonlein Purpura
  - Classic Triad: LE/buttocks purpuric rash, arthritis, nephritis (proteinuria)
- Hemolytic Uremic Syndrome
  - Bloody D → 3-4 days later = Hemolytic anemia, ↓ plts, ARF; Ecoli O157:H7.
- Inflammatory Bowel Disease
- Polyp
LGI Bleeding: Polyps

- Majority of polyps in children hamartomatous (benign juvenile polyps)
  - Affects 1-2% of children, between ages 2-5 yrs, 50% solitary, 80% of pts have <5 polyps
  - Bleed as outgrow blood supply, autoamputate
  - Therapy: colonoscopy with polypectomy
Polyposis Syndromes

- Familial adenomatous polyposis (FAP), colonic adenomas, peak age 16 years, **APC** gene mutation, 100% risk of colon CA
- Gardner’s Syndrome, small bowel and colonic adenomas, APC mutation, 100% risk of colon CA (*supranumerary teeth*)
- Peutz-Jegher’s Syndrome, hamartomatous polyps, lip/gum freckling, slight increased risk of colon CA (*polypectomy*)
- Juvenile Polyposis Syndrome, hamartomatous polyps in small bowel and colon, PTEN, SMAD4, or BMPR1 gene mutations, ~10% risk of colon CA
Upper GI Bleeding

• Esophagitis
• Mallory-Weiss tear
• Peptic Ulcer Disease
  • Gastritis (NSAID or steroid-induced)
  • H Pylori
• Portal Hypertension/ Esophageal Varices
• Trauma
• Other: oropharynx, epistaxis
Peptic ulcer disease

- Epigastric pain, guaiac positive stools, hematemesis, melena
- Often diagnosed by endoscopy
- NSAIDS- disrupt cyclooxygenase-prostaglandin synthesis
  - Patient with JRA and epigastric pain
- H pylori (up to 80% of PUD)
  - Serologic testing may represent old infection or exposure
  - Stool H pylori Ag, urea breath test preferred
  - Triple therapy: PPI plus two antibiotics (amoxicillin and clarithromycin) for 14 days
UGI Bleed Work-up

- ABCs, Vitals (screen for tachycardia, postural hypotension)
- History: Ask about medications, umbilical venous catheter
- Physical exam: pallor, CR> 2secs, Examine naso/oropharynx, signs of portal HTN (HSM, caput medusa)
- Stat hemoglobin, coags and type and cross
- IV access, NG lavage
- H pylori screening
- Endoscopy- diagnostic and therapeutic
Thank you and best wishes for a successful exam!