Diagnosis and management of emergent GI issues, treatments plans and connecting PCPs with GI subspecialists
Speakers

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Primary Care Pediatrician

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Pediatric Gastroenterology

The speakers have no disclosures and no off label products will be discussed.
Objectives

• Differentiate the diagnosis and management of functional abdominal pain from other causes of abdominal pain including inflammatory bowel disease
• Implement a plan for the treatment of constipation
• Identify when to refer to a specialist
Common GI office Visits

- Abdominal Pain
- Constipation
- Gastroesophageal Reflux
- When to Refer to Gastroenterologist
- Cases
**Common Causes of Abdominal Pain**

<table>
<thead>
<tr>
<th>Cause</th>
</tr>
</thead>
<tbody>
<tr>
<td>Constipation</td>
</tr>
<tr>
<td>Gastrointestinal Infection</td>
</tr>
<tr>
<td>Other Infections: UTI, Strep, Pneumonia, Viral Illness, PID</td>
</tr>
<tr>
<td>Mesenteric Lymphadenitis</td>
</tr>
<tr>
<td>Ruptured Ovarian Cyst</td>
</tr>
<tr>
<td>Foreign Body Ingestion</td>
</tr>
<tr>
<td>Colic</td>
</tr>
</tbody>
</table>
### Other Causes

#### Gastrointestinal
- Inflammatory Bowel Disease
- Pancreatitis
- Acute Cholecystitis
- Intra-abdominal Abscess
- Food Allergy
- Malabsorption
- Meckles Diverticulum
- Abdominal Migraine
- Wandering Spleen

#### Non Gastrointestinal
- IgA Vasculitis
- Hepatitis
- Sickle Cell crisis
- Neoplasm
- Urolithiasis
- Testicular Torsion
- Ovarian Torsion
- Poisoning
- Acute porphyrias
- Familial Mediterranean fever
Causes of Acute Abdominal pain in children by age

**Neonate**
- Adhesions
- NEC
- Volvulus
- Colic
- Dietary protein allergy
- Testicular torsion

**1 month to 2 years**
- Adhesion
- FB ingestion
- HUS
- Hirschsprung disease
- Incarcerated hernia
- Intussusception
- Trauma
- Gastroenteritis
- Viral Illness
- Dietary protein allergy
- Hepatitis
- IB
- Meckel's diverticulum
- Sickle cell syndrome vasoocclusive crisis
- Toxin
- Tumor
- UTI
Causes of Acute Abdominal pain in children by age

2 to 5 years
- Adhesions
- Appendicitis
- FB ingestion
- HUS
- Intussusception
- Primary bacterial peritonitis
- Trauma
- Gastroenteritis
- Viral illness
- Pharyngitis
- Constipation
- HUS
- Hepatitis
- IBS
- Intraabdominal abscess
- Meckel's diverticulum
- UTI
- Ovarian torsion
- Pancreatitis
- Pneumonia
- Sick cell syndrome vasoocclusive crisis
- Toxin
- Tumor

>5 years
- Adhesions
- Appendicitis
- DKA
- Myocarditis
- Pericarditis
- Perforated ulcer
- Primary bacterial peritonitis
- Trauma
- Constipation
- Gastroenteritis
- Pharyngitis
- Viral illness
- Abdominal migraine
- Cholecystitis or cholelithiasis
- Familial Mediterranean fever
- Gastrointestinal dysmotility
- HUS
- Hepatitis
- IBS
- Intraabdominal abscess
- Meckel's diverticulum
- Ovarian torsion
- Pancreatitis
- Pneumonia
- Acute porphyria
- Ruptured ovarian cyst
- Sick cell syndrome vasoocclusive crisis
- Testicular torsion
- UTI
- Urolithiasis
Constipation Symptoms

• Infrequent bowel evacuation
• Hard, small feces
• Difficult or painful evacuation of large-diameter stool
• Fecal incontinence (voluntary or involuntary evacuation of feces into the underwear)
Constipation

• 3 – 5 % of Office Visits
• 95% are caused by Functional Constipation, healthy children 1 year or older
• Important to identify Organic causes of Constipation
Normal Stooling Pattern

- Infants
- Toddlers
- Children
Causes of Constipation

**Functional**
- 95% of Constipation
- Common in Preschool
- Presence of at least 2/6 criteria describing stool frequency, hardness, and size
- Fecal Incontinence
- Stool Retention
- Exclusion of organic causes

**Organic**
- 5% of Constipation
- Young Infants
- Main causes in table for Infants and older children
Rome IV criteria for the diagnosis of functional constipation in children

<table>
<thead>
<tr>
<th>Infants and toddlers up to 4 years old</th>
<th>Children and adolescents (developmental age ≥ 4 years)</th>
</tr>
</thead>
<tbody>
<tr>
<td>At least 2 of the following present for at least 1 month:</td>
<td>At least 2 of the following present at least once per week for at least 1 month:*</td>
</tr>
<tr>
<td>2 or fewer defecations per week</td>
<td>2 or fewer defecations in the toilet per week</td>
</tr>
<tr>
<td>History of excessive stool retention</td>
<td>At least 1 episode of fecal incontinence per week</td>
</tr>
<tr>
<td>History of painful or hard bowel movements</td>
<td>History of retentive posturing or excessive volitional stool retention</td>
</tr>
<tr>
<td>History of large-diameter stools</td>
<td>History of painful or hard bowel movements</td>
</tr>
<tr>
<td>Presence of a large fecal mass in the rectum</td>
<td>Presence of a large fecal mass in the rectum</td>
</tr>
<tr>
<td>In toilet-trained children, the following additional criteria may be used:</td>
<td>History of large-diameter stools that may obstruct the toilet</td>
</tr>
<tr>
<td>At least 1 episode/week of incontinence after the acquisition of toileting skills</td>
<td>The symptoms cannot be fully explained by another medical condition</td>
</tr>
<tr>
<td>History of large-diameter stools that may obstruct the toilet</td>
<td></td>
</tr>
</tbody>
</table>

* In addition, the symptoms are insufficient to fulfill the diagnostic criteria of irritable bowel syndrome.

Data from:
## Causes of constipation in children

**Physiologic causes of constipation**
- Functional constipation (only have dietary or behavioral triggers, exacerbated by painful defecation and stool withholding)
- Cow’s milk or other dietary protein intolerance
- Low dietary fiber
- Slow-transit constipation
- Inadequate fluid intake (drying fever or hot weather)
- Immobility
- Anorexia nervosa
- Starvation

**Neurogenic causes**
- Hirschsprung disease
  - Cerebral palsy
  - Myelomeningocele
  - Spinal cord injury
  - Closed spinal dysraphism (e.g., tethered cord, sacral agenesis, split spinal cord malformation, dural ectasia)
  - Generalized laxity
  - Neurofibromatoses
  - Muscular weakness (may be generalized, as in Ossowsky syndrome, or due to abnormal abdominal musculature, as in prune belly syndrome or paralytic ileus)
  - Infantile botulism (constipation is an early complaint; other features are facial and oracular paresis, poor suck and hyporeflexia)
  - Prune-belly syndrome (e.g., vesicoureteral reflux, nephropathies, microcephaly, neuropathies)
  - Intestinal neural dysplasia
  - Familial or acquired distal enterocolitis
  - Duchenne muscular dystrophy

**Endocrine and metabolic causes**
- Cystic fibrosis
- With meconium ileus in neonates or distal intestinal obstruction syndrome in older children
- Hypokalemia
- Lead poisoning
- Vitamin D intoxication
- Hypo- or hypercalcemia
- Hypothyroidism
- Diabetes mellitus
- Phenylketonuria
- Multiple endocrine neoplasia type 2 (Menkes)
- Polycystic kidney (leading to dehydration)
- Juvenile systemic adenohypophyseal (adrenocortical) dysplasia
- Acute intermittent porphyria

**Anatomic causes**
- Anorectal anomalies (impaired anus, anteriorly displaced anus)
- Intestinal obstruction (in neonates, consider atrumatic, volvulus, or volvulus in small left colon syndrome)
- Other causes
  - Inborn errors of metabolism
    - Medications (corticosteroids, antihistamines, anticholinergics, chemotherapy, salicylates, containing antidepressants)
    - Generalized joint hypermobility

*Relative or common cause.
† Prompt diagnosis is important for outcome.
‡ Generalized laxity during infancy.
§ Similar findings may occur in infants with Hirschsprung disease.

Data from:
Evaluation of Constipation - History

- Parent and child if age appropriate
- Acute signs or symptoms – delayed passage of meconium, fever, V/D, rectal bleeding, abdominal distention
- Chronic signs or symptoms – since infancy, ribbon stools, weight/growth, extraintestinal, anorectal, congenital anomalies, FMHX Hirschsprungs
- Young Infants – straining with soft stool? "Infant dyschezia", hard stools, Hirschprungs or CF
- Psychosocial or environmental factors – onset, stool withholding, good response to treatment
- Risk of lead intoxication – pica
- Developmental history – hypothyroidism, mitochondrial or neurologic - motility
- Medical history – celiac, hypothyroidism, neuromuscular d/o
### Differential diagnosis of straining in infants

<table>
<thead>
<tr>
<th>Cause of straining</th>
<th>Clinical characteristics</th>
</tr>
</thead>
</table>
| Infant dyschezia          | • Healthy infant 0 to 9 months of age  
• Soft stool passed after straining                                                   |
| Constipation              | • Healthy infant  
• Stools are hard, large or pellet-like                                                  |
| Anal fissure              | • Healthy infant  
• May or may not have history of constipation  
• Fissure identified on inspection of anus  
• Straining may be caused by voluntary stool withholding                                   |
| Cow’s milk intolerance    | • Healthy infant  
• Diet contains cow’s milk protein (breast- or formula-fed)  
• Normal or loose stools with gross or occult blood and/or mucus                          |
| Hirschsprung disease      | • Newborn or infant  
• History of delayed passage of meconium (after 48 hours of life)  
• Well- or ill-appearing  
• Constipation or abdominal distension, occasionally diarrhea  
• Rectal examination may reveal tight sphincter, empty or narrow ampulla, and/or explosive squirt of stool on withdrawal of finger  
• Anorectal manometry demonstrates absence of rectosphincteric relaxation reflex  
• Ganglion cells absent on rectal biopsy                                                |
| Internal anal sphincter achalasia | • Presentation and anorectal manometry similar to Hirschsprung disease, as described above  
• Ganglion cells present on rectal biopsy                                                  |

Courtesy of Dr. Manu Seed.
Evaluation of Constipation – Physical Exam

- General
- Neurologic
- Perineum
- Digital rectal examination – not routine
- Findings suggestive of functional constipation are a distended rectum that is full of stool - lack of stool does not exclude the possibility of functional constipation
- Testing for Occult blood
Testing

• Labs – celiac screening, UA and urine culture, electrolytes and calcium, TSH, blood lead level
• Imaging – Abdominal radiograph, Barium Enema, Spine radiograph
When to Refer to Gastroenterology

• Constipation resistant to common treatments like laxatives (particularly, stool softeners like PEG), diet, and lifestyle management
• Significant abdominal distension
• Signs of intestinal obstruction
• Suspect Hirschsprung's Disease (short-segment)
• Persistent hematochezia
• Poor growth
• Failure to thrive
• Abnormal physical exam- local or systemic
Treatment of Functional Constipation

**Infants**
- Infants with functional constipation frequently respond to treatment with nondigestible osmotically active carbohydrates, such as sorbitol-containing juices (apple, prune, or pear)
- Pharmacotherapy – Lactulose, PEG, Glycerin suppository

**Children**
- Dis-impaction (for children with a large rectal stool mass or fecal incontinence)
- Prolonged laxative treatment and behavioral therapy to achieve regular evacuation and avoid recurrent constipation
- Dietary changes (primarily increasing fiber and fluid content) to maintain soft stools
- Gradual tapering and withdrawal of laxatives as tolerated
Typical regimen for a child with recurrent or chronic constipation without fecal impaction or incontinence

<table>
<thead>
<tr>
<th>Step</th>
<th>Instruction</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Start 2 to 4 teaspoons (3.5 teaspoons = 17 g) of PEG 3350 (e.g., MiraLax, GlycoLax) once daily, in 4 to 8 ounces (120 to 240 mL) of noncarbonated beverage (or appropriate dose of another laxative). Dietary counseling to add dietary fiber and extra liquids to the diet each day.</td>
</tr>
<tr>
<td>2</td>
<td>Increase or decrease PEG 3350 by 1 to 2 teaspoons every 2 to 3 days, until the desired result of daily soft stools is achieved. Maximum dose is 1 heaping tablespoon (17 g) twice daily.</td>
</tr>
<tr>
<td>3</td>
<td>Follow-up by phone or a return visit within 1 month to be sure the laxative is effective.</td>
</tr>
<tr>
<td>4</td>
<td>Continue to add dietary fiber and extra liquids to the diet each day.</td>
</tr>
<tr>
<td>5</td>
<td>After 6 to 8 weeks of soft daily bowel movements, begin to taper the dose of PEG 3350 by 0.5 to 1 teaspoon every 2 weeks, until daily movements continue without the need for a laxative.</td>
</tr>
<tr>
<td>6</td>
<td>If stools become hard again, increase the dose slightly and retry weaning off the laxative in another 6 to 8 weeks.</td>
</tr>
<tr>
<td>7</td>
<td>This process may take from 2 to 4 weeks to 6 months, but the end result should be resolution of the constipation.</td>
</tr>
</tbody>
</table>

This table describes a typical regimen for children older than 3 years with mild or moderate constipation without fecal impaction. Children with mild chronic constipation may be treated with dietary changes alone rather than medication, if desired. Children with fecal impaction, with or without overflow incontinence, should first be disimpacted with a regimen of oral and/or rectal medications. In most cases, these steps should be combined with family education and behavior modification (toilet training) to enhance efficacy and prevent relapse. For details, refer to the UpToDate topic on chronic constipation in children.

PEG 3350: polyethylene glycol without electrolytes, also known as macrogol.
Behavioral Modification

• Toilet Sitting: sit on the toilet shortly after a meal for 5 to 10 minutes, two to three times per day
• Toilet sitting episodes should occur at the same time each day and be timed with a timer or stopwatch
• The routine should be followed every day
• The child's adherence to the program should be encouraged with positive reinforcers, rewards, rather than negative reinforcers (criticism or punishment)
• Use a stool - for Child whose feet do not touch the floor sitting on a regular toilet seat
GER in Infants

• Uncomplicated GER vs GERD
• GERD - esophagitis, nutritional compromise with weight loss, or respiratory complications
• Warning signs and symptoms
• Happy spitters
• Lifestyle measures - feeding breast milk as much as possible to infants who are fed both breast milk and formula, avoiding tobacco smoke, and avoiding overfeeding
• Trial of thickened feeds, upright positioning after feeds, or trial of a hypoallergenic diet
• Usually resolve by 1 year of age, if not improving by 18-24 months further evaluation
Reflux and poor weight gain

- Caloric intake, dysphagia
- UGI – rule out anatomic abnormalities
- Pyloric US
- Labs – occult blood, CBC, electrolytes, review of Newborn screen
- GERD – thickening formula or expressed breast milk, increase caloric density, trial to reduce gastric acidity
- Reflux and feeding refusal
- Reflux and irritability
- Reflux and rectal bleeding
GER in Children and Adolescents

• Higher rate of GERD – hx of prematurity, pulmonary disease, developmental and neurologic disorders – CP, MD, Down syndrome; other – CF, Obesity
• Pre-schoolers – Presents with Sandifer Syndrome
• Children/Adolescents - Present with Heart burn or regurgitation
• History - Presence of heartburn/chest pain or abdominal pain, regurgitation or vomiting, water brash, onset of symptoms and relation to meals, dysphagia or odynophagia, underlying disorders including neurologic dysfunction or congenital anomalies, Asthma, pneumonia, or chronic cough, Functional constipation, Medication history
General approach to GER

- Recurrent vomiting or regurgitation – hx and physical, DDX is wide, warning signs such as fever, weight loss, abdominal distention, bilious vomiting, hematemesis, Hepatosplenomegaly, headache or new neuro signs
- Gastroparesis, rumination or psychogenic vomiting, pregnancy, cannabis hyperemesis
- Heartburn – trial of lifestyle changes and/or acid suppressing medication
- Dysphagia or Odonophagia – esophageal inflammation, pill esophagitis
- Asthma – acid suppressing medication for 3 months
- Recurrent pneumonia or chronic cough – imaging to look for aspiration
When to Refer to Gastroenterology

- Infantile GER symptoms persisting beyond 15 months of age
- Feeding difficulties (infants, toddlers)
- Treatment resistant GERD (acid reducing medication +/- lifestyle management)
- Dysphagia
- Hematemesis
- GER symptoms + Failure to thrive
- Concern for Eosinophilic esophagitis
Connecting PCP with GI Specialist
Case Discussions

• Understand the basic principles of these scenarios
• “Key words, phrases”
• Specific disorders
• Open discussion
Abdominal pain

- Common symptom
- Organic vs. functional
- Acute vs. chronic
- “Red flags” suggest a possible organic etiology
Red flag symptoms

• Age < 5 yrs.
• Fever
• Bile or blood stained emesis
• Unexplained weight loss
• Focal, non-periumbilical abdominal pain
• Referred pain to shoulder, groin or back
Red flag symptoms

• Night time awakening (pain, or a distinct change in BM pattern)
• Mass per abdomen
• Laboratory tests: elevated ESR, anemia, elevated liver enzymes etc.
• Family h/o IBD, Celiac,
Focal abdominal pain

<table>
<thead>
<tr>
<th>Right</th>
<th>Left</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gallstones</td>
<td>Stomach Ulcer</td>
</tr>
<tr>
<td>Stomach Ulcer</td>
<td>Heartburn/Indigestion</td>
</tr>
<tr>
<td>Pancreatitis</td>
<td>Pancreatitis, Gallstones</td>
</tr>
<tr>
<td>Epigastric hernia</td>
<td></td>
</tr>
<tr>
<td>Kidney stones</td>
<td>Stomach Ulcer</td>
</tr>
<tr>
<td>Urine Infection</td>
<td>Duodenal Ulcer</td>
</tr>
<tr>
<td>Constipation</td>
<td>Ulcer</td>
</tr>
<tr>
<td>Lumbar hernia</td>
<td>Biliary Colic Pancreatitvs</td>
</tr>
<tr>
<td>Appendicitis</td>
<td>Kidney Stones</td>
</tr>
<tr>
<td>Constipation</td>
<td>Diverticular Disease</td>
</tr>
<tr>
<td>Pelvic Pain (Gynae)</td>
<td>Constipation</td>
</tr>
<tr>
<td>Groin Pain (Inguinal)</td>
<td>Inflammatory Bowel</td>
</tr>
<tr>
<td>Groin Pain (Inguinal)</td>
<td>Small bowel</td>
</tr>
<tr>
<td></td>
<td>Umbilical hernia</td>
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<td>Appendicitis</td>
<td>Diverticular Disease</td>
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<td></td>
</tr>
</tbody>
</table>
Corkscrew sign
Acute abdomen: Infants

• Infants
• Bilious vomiting, abdominal distension -> think malrotation and volvulus -> UGI
• Relationship of the ligament of Trietz to the spine
• Volvulus on plain radiograph: “corkscrew appearance”
Malrotation
Donut sign
Intussusception

- Colicky abdominal pain, **currant jelly stools** -> intussusception
- 6 months - 2 years
- Most common abdominal emergency < 2 yrs
- **Ileocolic**
- Recognizable lead points found in 2-8% e.g. Meckels, polyp, duplication cysts
Intussusception

- Idiopathic
- Cystic Fibrosis
- HSP
- PE: sausage-shaped mass, tender
- US: target/doughnut sign
- Rx: Pneumatic reduction/ air-contrast enema
• Bilious vomiting, no abdominal distension in a baby w/ Downs syndrome -> ?, “double bubble sign” on plain radiograph
• ½ are premature
• ½ have Downs syndrome
• h/o polyhydramnios
• Rx: NG to decompress->evaluate for other congenital abnormalities (1/3 have another life threatening congenital anomaly)

• CHD (30%), malrotation (30%), anorectal (5%), annular pancreas (30%), EA (5-10%)

• Duodenoduodenostomy
Double bubble sign
Acute abdomen: Children

• PU ➔ RLQ pain, anorexia, fever ➔
• Blunt trauma to epigastrium, abdominal pain, vomiting, “Sentinel loop” ➔
• “Bike handle bar injury” ➔ non-bilious emesis, abdominal pain ➔
Appendicitis

• Most common acute abdominal emergency in children
• 4/1000 children; 12-18 yrs.
• < 50% present with classic symptoms
  • Acute onset RLQ pain, low grade fever, nausea, vomiting, anorexia
• Starts PU → RLQ, rapid progression 24-48 hour
Appendicitis

• > 36 – 48 hrs ➔ perforation (65%)
• “bumpy road” pain, guarding
• Diagnosis: single most reliable exam finding: Localized tenderness at Mc Burney’s point
• Clinical scoring systems exist (not superior to clinical judgment)
Appendicitis

• Equivocal cases: add imaging
• First imaging of choice: USG (≈ 90%)
• If inconclusive and index of suspicion high→ CT scan (gold standard ≈ 95%)
• Acute appendicitis- Rx is appendectomy
• If perforation/ peritonitis has occurred→ Abx, bowel rest, drainage etc. -> Interval appendectomy
Pancreatitis

- Acute severe epigastric pain, radiating to back + N/V
- Most common etiology: Idiopathic (? Viral)
- Abdominal trauma
- Gallstones
- Drug toxicity
Drugs associated w/ pancreatitis

- Valproate,
- Carbamazepine,
- Azathioprine,
- Steroids,
- L-asparaginase,
- Cimetidine
Diagnosis of pancreatitis

- Acute severe pancreatitis
- *Cullen* sign (dyscoloration around umbilicus)
- *Grey Turner* sign (flanks)
- Diagnosis
- Elevated Lipase (x 3)
- X-ray: Sentinel loop, “cut off sign”
- CT scan
Cholecystitis

- RUQ pain, N/V
- Increasing incidence in children
- Gallstones in hemolytic anemia(s) (HS, Thalassemia etc.)
- Obesity
- Rare: Bile acid transport protein defects
Foreign body in the GI tract

- “Sites of FB lodging” in the GI tract
- Cricopharyngeus
- mid-esophagus
- LES
- Pylorus
- Ileocecal valve
Esophageal constrictions

The esophagus has 3 areas of narrowing:

- Superiorly: level of cricoid cartilage, juncture with pharynx
- Middle: crossed by aorta and left main bronchus
- Inferiorly: diaphragmatic sphincter
Foreign body in the GI tract

• Coins, most common
• Lodge in upper esophagus
• No FB should be left in esophagus >24 hours
Coins

- Esophagus: 25% will pass spontaneously, so if no respiratory distress, no other known esophageal pathology, can observe for 4-6 hours. Repeat x-ray➡️ not moved➡️ EGD
- Stomach: Most will pass spontaneously, unless evidence of pyloric obstruction (unlikely with an object < 5cm long).
- 4 weeks
Button battery

• Esophagus: Emergent EGD and removal!
• Stomach or beyond:
• If a magnet co-ingested, symptomatic, >15 mm battery in child < 6 years, not passed in 4 days -> Remove
• Battery < 12 mm, child > 6 yrs and asymptomatic -> can wait for passage
• http://pediatrics.aappublications.org/cgi/content/abstract/peds.2009-3037v1
Other scenarios

• Sharp Objects: Should be removed immediately
• Rod shaped: Nails, pins, pencils, if \(< 5\, \text{cm}\) will pass spontaneously.
• Magnet > 1 or with metal: Remove immediately
• Single: Asymptomatic, removal is preferable but can follow w/ careful considerations.
• www.naspghan.org
Caustic ingestion

- Alkaline: Liquefactive necrosis -> deep ulceration
  - e.g. drain cleaners, oven/grill cleaners, dishwasher detergent
- Acids: Coagulative necrosis -> eschar formation
  - E.g. lead battery acid, con. vinegar, toilet cleaner
- Household bleach
- Hair relaxer
Caustic ingestion

- H and P: find out what was ingested, e.g. hair relaxers have shown to not cause much damage
- **No proven role for common neutralizers like milk etc.**
- Symptomatic child: After stabilization, **early EGD to assess extent of injury**, then place an NG, ND tube.*
Q 1

• Healthy, thriving young infant (usually first few months) with blood streaked stools, +/- mucus:
Hematochezia: Scenarios and differentials

• Cow’s milk protein intolerance
• Prevalence up to 7.5%
• Anal fissure may be an associated finding
• Cross reactivity w/ soy protein is so switch to a protein hydrolysate formula, not soy formula*
Hematochezia

- Milk and soy exclusion is recommended for breastfeeding mothers.
- Conventional allergy tests like skin prick or RAST are not helpful in diagnosis,* as these are IgE based Vs the non IgE mechanisms in CMP allergy
- Most infants develop tolerance and “grow out of milk allergy” by 12 months of age.
Q 2

- 3 y/o otherwise healthy child with bright red blood in stools and a “mass protruding from the bottom”*
Hematochezia

- First and foremost ask about the consistency of the stools, and whether there’s associated rectal pain as this is very crucial in guiding further work-up and management
- Constipation w/ Rectal prolapse
- Requires stool softening
- Other consideration in a child w/ Rectal Prolapse? Cystic Fibrosis (20%)
Hematochezia

- Juvenile Polyp
- Sporadic or familial (JPS)
- Sporadic is usually a single, hamartomatous polyp found in the rectum of a young child (3-7 years), prone to auto-amputation (small pedicle) and has no malignant potential.
- Rx: Polypectomy
Hematochezia

- JPS is mostly familial, AD w/ variable penetrance.
- > 5 JP in colon or rectum or any no. with + family history.
- Prophylactic colectomy due to high risk of Colorectal carcinoma.
Q 3

• 2 y/o with profuse, painless brick red colored or maroon stools who is tachycardic on exam and has anemia (Hb 6.8 g/dl)
Hematochezia

- Meckel’s Diverticulum:
- Rule of 2s, In a 2 year old, 2 cm long, 2 ft from the IC valve, on the antimesentric border. Symptomatic MD usually has ectopic gastric mucosa.
Hematochezia

- Meckel’s scan (uptake of i.v. 99mTc by gastric mucosa) is the test of choice*
- Sensitivity and specificity: 85% and 95% respectively.
- Rx: First is always hemodynamic stabilization* of an unstable patient, fluid resuscitation, blood transfusion etc.
- Then surgery
Q 4

• 13 y/o with progressively worsening diarrhea with blood, abdominal pain, fecal urgency, reduced appetite and weight loss.
Hematochezia

• IBD*
• Crohn’s Disease Vs Ulcerative Colitis
• Chronic enteric infections are possible, Yersinia, but usually there will be some clues like ingestion of eggs, meat, day-care outbreak etc.
Select associated Extra-intestinal manifestations

- Erythema nodosum (10% CD): painful nodules usually on shins*
- Pyoderma gangrenosum (1-2%): painful, violaceous ulcers*
- Oxalate urinary crystals/renal stones in CD
Hematochezia

• A child with sudden onset of fever, crampy abdominal pain, bloody diarrhea with mucus a.k.a. dysentery and usually there will be some clue pointing to an infectious exposure, suspect one of the following:
  • Shigellosis: h/o day care outbreak*
  • EI manifestation: non-suppurative arthritis, seizures in young children*
Hematochezia

- Salmonellosis (non-typhoidal)
- Poultry/meat, raw milk, insanitary water supplies, eggs, exotic pets like turtles, lizards etc.
- Usually self-limited course, *Abx not required routinely*
- Infants< 3 months, immunosuppressed, cancer, chronic debilitating inflammatory diseases require Rx.
Hematochezia

• Yersinia
• Campylobacter
• C. diff
• Remember, it can also occur w/ viral gastroenteritis!
• Reiter's syndrome: Reactive RF (-), HLA B27 (+) arthritis. *(Salmonella, Shigella, Campylobacter)*
• 4 year old boy develops colicky abdominal pain, vomiting, hematochezia, and shortly afterwards petechia, macular rash (non-blanching) on the buttocks and extensor surfaces of arms and legs
Hematochezia

- Henoch-Schonlein Purpura; vasculitis*
- Rash, arthritis, renal disease, GI (75%)
- Hemorrhage in bowel wall, colicky abdominal pain, intussusception (SB), perforation, pancreatitis, hydrops of GB
- Rx: Steroids*
• 3 y/o presents to the ED with mild jaundice, pallor, lethargy and dehydration. Labs reveal: Hb 12* g/dl, Bil 3.5 g/dl, WBC 15K, Platelets 80K, BUN 42, Cr 2.4. A few days ago he had developed acute onset febrile illness, vomiting, bloody diarrhea
Hematochezia

- HUS: Microangiopathic hemolytic anemia, thrombocytopenia and uremia
- ETEC, *E. coli* O157:H7 (meat)
- Toxin A, B: B binds to GB3 receptors in intestine, glomerular and glial cells; A subunit is internalized and disrupts protein synthesis, cell death
- Supportive management
- Antibiotics are contraindicated
• 2 y/o, otherwise healthy child with normal growth parameters with multiple, loose stools/day, with noticeable undigested food materials.
Chronic Diarrhea

• Toddler’s diarrhea, benign diarrhea of Infancy.
• Due to a combo of “colonic fluid-overload” + osmotic effects of sorbitol and fructose in juices
• Rx: Limiting juices, fluids; increasing fiber and fat in the diet
Chronic Diarrhea Scenarios

- Chronic giardiasis: FTT, steatorrhea
- Immunocompromised hosts: Cryptosporidium can cause watery, “cholera like” diarrhea.
- **Terminal ileitis, pseudoappendicular syndrome**: Yersinia. Rx only in special populations
- **a/w GB syndrome, Reiter’s syndrome, Miller-Fisher syndrome**: Campylobacter
Chronic Diarrhea Scenarios

- Bloody diarrhea in the context of recent Abx use: C. diff induced pseudomembranous colitis
  
  Rx Flagyl

- Protracted diarrhea, flatulence, weight loss in the setting of immunosuppression or exposure to contaminated water sources: think Giardiasis (they also have lactose intolerance due to sub-total villous atrophy)
• 7 m/o with poor growth, irritability, weeping rashes in the perioral area and perineum, chronic diarrhea and alopecia.
Chronic Diarrhea

- Acrodermatitis Enteropathica
- Primary
- Secondary: Crohn's disease, Celiac, CF, Chronic liver disease, sickle cell etc.
- Zn deficiency; low alkaline phosphatase
• 15 m/o Caucasian (Irish, Italian) infant with poor weight gain, loose stools, abdominal distension, irritability, thin extremities with loss of subcutaneous fat.
Chronic Diarrhea

• Celiac Disease
• Autoimmune gluten sensitive enteropathy
• HLA DQ2, DQ8
• Screening: tTG IgA (>95% sensitivity and specificity)
• Ig A deficiency, so also check Ig A
• Rx: Lifelong strict Gluten free diet.
Extra-intestinal manifestations

- Dermatitis Herpetiformis (90% have mucosal abnormalities)
- Refractory iron deficiency anemia
- Short stature (10%)
- Arthritis
- Peripheral neuropathy
- Dental enamel hypoplasia (30%)
Celiac Disease Associations

- Down
- Turner’s
- Williams
- Selective IgA deficiency
- Other autoimmune diseases: DM type 1, Thyroid, Addison’s etc.
Hematemesis

- UGI Bleed
- May also lead to melena
- First and foremost step and answer to any question related to this, would be hemodynamic evaluation and stabilization of the patient as needed.
- Second would be to insert an NG tube for a gastric lavage to determine whether active bleeding is ongoing or has ceased.
Hematemesis

- Further steps will be based on the color of the lavage fluid and other factors,
- Bright red ➔ ongoing bleeding ➔ NPO, IVF, IV PPI, Octreotide and prepare for emergent Endoscopy.
- Coffee-grounds with clearing fluid, usually implies that active bleeding has stopped. No need for emergent endoscopy.
Scenarios

• Cirrhosis, shrunken liver, splenomegaly, thrombocytopenia, +/- jaundice, spider angioma etc -> signify portal hypertension and esophageal varices.*
• Teenager with acne on tetracyclines, odynophagia -> pill esophagitis.*
• Other corrosive medications: NSAIDs (chronic pain condition), Alendronate.*
NSAID induced GI injury*

- Inhibition of COX catalyzed conversion of AA to protective Prostaglandins
- Mucus and HCO3
- Epithelial integrity
- Microvascular supply
- Aspirin also reduces Thromboxane production from platelets
- Rx: Judicious use, PPI, Misoprostol
Hematemesis

- Violent retching, purging-> Mallory-Weiss tear
- Poor sanitation, epigastric abdominal pain-> think Helicobacter Pylori gastritis/PUD
- Helicobacter Pylori
- Causes dyspeptic symptoms: Chronic active gastritis, PUD, Duodenal ulcers
- Diagnosis: EGD w/ rapid urease or culture
Helicobacter Pylori

• Serological screening is NOT RECOMMENDED
• Rx: Triple therapy: 2 antibiotics + PPI x 14 days. Continue PPI x 2-3 months.
• Test of eradication: Stool antigen test in 4-6 weeks after completion of Abx Rx or breath test, both of which are equally reliable
2 y/o male infant with poor weight gain, loose, greasy, bulky, foul smelling stools.
Steatorrhea

- Steatorrhea: **Pancreatic insufficiency** >> inflammatory mucosal disease
- Causes?
- Cystic fibrosis: Diagnostic test: **Sweat Chloride**
- Shwachman-Diamond syndrome
SDS

- Pancreatic insufficiency, bone marrow dysfunction and skeletal abnormalities
- Acinar tissue replaced by fat -> Pancreatic lipomatosis
- Increase in normal tissue volume occurs in may SDS ➔ restoring pancreatic sufficiency
- Neutropenia and predisposition to myelodysplasia and leukemias
SDS

- Thoracic cage abnormalities: **metaphyseal chondrodysplasia**, 
- Long bone dysostosis 
- Diagnosis: Clinical + **SBDS** gene testing
Steatorrhea Syndrome

- Johanson-Blizzard syndrome
- Pancreatic lipomatosis
- Deafness
- Dental abnormalities
- Imperforate anus
- Urogenital abnormalities
- Hormonal: hypothyroid, panhypopituitarism
Steatorrhea Syndrome

- Pearson’s marrow-pancreas syndrome
- Deletions in mitochondrial DNA
- Pancreatic fibrosis
- Bone marrow: ringed sideroblasts
- Lactic acidosis
- Sepsis, multiorgan failure and metabolic derangements lead to early death
Steatorrhea syndrome

• Juene Syndrome: Asphyxiating thoracic dystrophy + Pancreatic insufficiency (fibrosis)
• Gold standard indirect test for exocrine pancreatic insufficiency: 72 hour fecal fat test
• Gold standard direct test: CCK/Secretin stimulation test
Steatorrhea

- Rx: Pancreatic enzyme replacement
- Have protease and Lipase (500-2,500 IU/kg/meal); max 10,000 U/kg/day.
- Excess can cause Fibrosing colonopathy
- Gastric acid suppression may help decrease inactivation of Lipase and improve steatorrhea
Recurrent Pancreatitis

- Hereditary pancreatitis; *SPINK 1* (mutations in gene coding for trypsin inhibitor), *PRSS1* (cationic trypsinogen gene)
- Autoimmune pancreatitis
- Anatomical malformations of the Pancreatic duct: Pancreas Divisum, choledochal cyst
Recurrent Pancreatitis

• Hypertriglyceridemia
• Hypercalcemia
• Gall stones
• Tropical chronic pancreatitis
• Organic acidemias
Jaundice and Neonatal Cholestasis

- Newborns
- Physiological (unconjugated)
- Pathological
  - Direct hyper-bilirubinemia is always pathological
  - Persistent Jaundice beyond 14 days, warrants checking a direct bilirubin. This may be the first indication of Biliary Atresia
Biliary atresia

• Progressive fibro-obliterative disease of the hepato-biliary system
• Perinatal form: BA w/o associated malformations
• BASM: embryonal form: BA+ splenic malformations, situs inversus, malrotation, annular pancreas, cardiac anomalies
Biliary Atresia

• Progressive jaundice, acholic stools, poor weight gain, fat-soluble vitamin deficiencies, fibrosis of the liver-> cirrhosis, ESLD with PHT, Esophageal varices can occur very rapidly, thus establishing an accurate diagnosis in a timely manner is very important!

• HIDA scan is the initial test of choice.

• US: neither sensitive nor specific. Triangular cord sign : hyperechogenecity at porta hepatis
Biliary Atresia

- If suspicion still persists, an IOC (Intra-operative cholangiogram) is the most definitive test. Usually is also combined with a liver Bx.
- Rx: Kasai Portoenterostomy to re-establish biliary drainage
- 10 yr survival 75% if Rx before 60 days Vs 11% in those where diagnosis is delayed for 3 months
Biliary Atresia

• 50% will end up requiring a liver transplant by age 2 years
• At 3 months Bili > 6 mg/dl is a/w high likelihood of failure of Kasai and need for liver transplant under 2 years of age.
• About 20% may remain transplant free for whole life
“Key points” for other neonatal cholestatic diseases

- Alpha 1 antitrypsin deficiency: Phenotype normal MM, abnormal: ZZ, SZ, SS; heterozygote: MZ.
- A1AT level in blood and phenotype
- A1AT granules in periportal hepatocytes are PAS positive, Diastase resistant
- Lung disease is very rare in children
Galactosemia

- NBS. Galactose-1-phosphate uridyl transferase level (GALT) is diagnostic
- Presentation can mimic neonatal sepsis
- Jaundice, vomiting, diarrhea after initiation of feeds (lactose), FTT, hypoglycemia, encephalopathy
- E. Coli sepsis, cataracts
- Rx: Elimination of Lactose
Tyrosenemia

- Tyrosinemia type 1, deficient function of FAH, leading to accumulation of FAA, MAA (hepato and reno-toxic- renal rickets) -> SAA succinylacetone -> increased ALA -> neurotoxic.
- Fulminant hepatic failure or a chronic hepato-renal presentation with neurological crisis (porphyria like)
- Fanconi syndrome with renal rickets
- Rx: Dietary restriction of Phenylalanine and Tyrosine
- 2 NTBC
Neonatal Cholestasis

TORCH
Acquired sepsis
UTI
Hormonal: Thyroid
Pan-hypopituitarism: Hypoglycemia, a/w SOD, ON hypoplasia, microphallus
Chronic Hepatitis

- Infectious: Viral: Hepatitis B, C
- Interpret Hepatitis B serology:
  - HBsAg +: Ongoing HBV infection
  - Anti-HBS: Resolving or past infection
  - Protective immunity
  - Vaccinated status
- HBeAg: Active viral replication; viremia
Chronic Hepatitis

- Anti-HBe: Cessation of active viral replication
- HBV DNA: Active viremia, used to follow response to Rx
- HBcAg: Detected in Liver, active replication
- Anti-HBc IgM: Recent infection
- Chronic Hepatitis B: HBs Ag+ for 6 months and other evidence of ongoing hepatitis.
Infectious Hepatitis

- Amoebic Liver abscess: E. Histolytica, developing countries, amoebic colitis, usually right lobe, margin with peripheral halo on US, “Anchovy paste” liquefactive drainage from the abscess
- Rx: Metronidazole + Diloxanide furoate, Paromomycin
Other Chronic Hepatitis

- Autoimmune Liver Disease: ANA+, SMA+ (type 1), anti LKM + (Type 2)
- Primary Sclerosing Cholangitis a/w IBD
- Wilson’s Disease/Hepato-lenticular degeneration, AR, 1:30,000, ATP7B-> decreased excretion of Cu-> accumulation in liver and brain, cornea, kidneys
Wilson’s Disease

• Chronic active hepatitis in adolescents and as a neuropsychiatric illness in adults
• Low ceruloplasmin, may be normal
• Liver disease: quantitative Cu testing in liver is the most definitive test: >250 mcg Cu/g dry liver
• KF rings (Liver disease: 50%; neurological disease: 90%)
Wilson’s Disease

• s/s of Liver disease
• Coomb’s negative hemolytic anemia
• Coagulopathy unresponsive to Vit K
• Neurological symptoms: Behavioral and motor, coordination tasks, changes in school performance, bulbar symptoms: dysarthria, dysphagia, drooling etc.
Wilson’s Disease

- Rx: Cu chelation:  Pencillamine
  Trientene
  Tetrathiomolybdate
Constipation

- 4 y/o with history of increasingly difficult passage of large, hard BMs, intermittent hematochezia, “stool holding and tiptoeing”.
- Chronic functional constipation!
- Increased rectal caliber with secondary increase in rectal sensory thresholds -> decreased frequency -> hardening of stools -> further holding
Constipation

• RAIR is preserved in functional constipation, but the sensory thresholds are elevated during a manometry
• Rx: positive reinforcement, adequate stool softening, regular bowel habits, diet etc.
Hirschsprung’s Disease

- Congenital absence of Ganglion cells in Auerbech and Meissner plexus of colon.
- 1:5000 live births
- M:F 4:1 for classic, short segment affects the recto-sigmoid colon
- 30% are syndromic (other neural crest derivatives): cardiac anomalies, melanocyte abn, cranio-facial, skeleton, iris
HD

- Down syndrome
- Waardenburg
- Smith-Lemli-Opitz
- Congenital hypoventilation syndrome
- MEN 2
- NF
- X-linked hydrocephalus
HD

- Severe, refractory constipation in newborn period
- Up to 90% infants may not pass meconium in the first day of life
- Abdominal distension
- Biliary vomiting
- Stools are thin
HD

- Older infants: FTT
- PE: tight anus, with empty, collapsed rectum
- Occurrence of explosive diarrhea, fevers, abdominal distension, can indicate enterocolitis-> toxic megacolon-> shock
- Rectal biopsy: gold standard test: absence of ganglion cells
- Rx: Surgical
Open Discussion

• What are some symptoms or signs that could be considered red flags? How to differentiate between serious and non-serious causes?
• How can you efficiently assess the patient?
• How can I prepare my practice to better treat GI cases?
• How do I proceed if my patient has an allergy such as milk protein?
• What resources can I provide patients to educate on these conditions?
• What processes can my practice put in place to make the referral process seamless?
• What does the follow up process look like?