# CASE PRESENTATION

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I DO NOT INTEND TO DISCUSS AN UNAPPROVED/INVESTIGATIVE USE OF A COMMERCIAL PRODUCT/DEVICE IN MY PRESENTATION.

# CASE

7 mo F born 37 WGA with recent admission for milk protein allergy presents to ED for persistent bloody stools and FTT despite adequate intake of elemental formula.

# Symptoms

- Bloody stools that vary in amount
- Intermittent fevers, tmax 101 F
- Increased PO intake-22 to 23 ounces/day
- Decreased UOP
- Intermittent macular rash to trunk and face
- Currently on Elecare 26 kcal/ounce

# Differentials

| | | | | |
|---|---|---|---|---|
| Protein-calorie malnutrition | Milk or soy enterocolitis | Anal fissure | Meckel's diverticulum | Anorectal fissures |
| Malrotation with midgut volvulus | Hirschsprung disease with enterocolitis | Coagulopathy | Brisk upper GI bleeding | Vascular malformations |
| Gastric or duodenal ulcer | GI duplication cyst | Intussusception | IBD | Eosinophilic GI disease |

# Exam

## Vitals:

- Temp-98.7 F
- Pulse- 153
- Resp-52
- BP- 99/58
- SpO2- 97% RA

## Physical exam:

- HEENT: atraumatic, normocephalic
- Heart: RRR, no murmurs
- Lung: CTA
- Abd: flat, soft, no tenderness, no guarding
- MSK: FROM
- Skin: mottled and pale
- GU: anal fissure noted at 12 o'clock position

# Pertinent Labs

Hemoglobin: 9.3

Hematocrit: 30.5

CRP: 7.8

ESR: 70

## EGD/COLONOSCOPY RESULTS

- Stomach: chronic and active gastritis with rare mucosal granuloma, possibly secondary to ruptured cryptitis
- Duodenum: chronic and active duodenitis with non-necrotizing mucosal granuloma
- Colon: chronic and active colitis, favor inflammatory bowel disease-type indetermination
- Overall: impression of inflammatory changes through the entire colon characteristic of ulcerative colitis, but histologic changes seen within colon, duodenum and stomach raises the possibility of more widespread involvement by Crohn's disease.

# Case follow up: hospital course

- RVP and BCx negative throughout admission
- Started on IV solumedrol and transitioned to orapred, which she continued at discharge
- Given PPN for 2 days for added nutrition, but discontinued as patient gained weight
- Started on azathioprine
- Discharged with azathioprine, PPI, and steroids

# Case follow up: outpatient course

- Continued on azathioprine, PPI and steroids
- Started on mesalamine
- Follow up labs: immune panel inconclusive, ***calprotectin elevated at 751 (normal <50), pancreatic elastase <40 (normal >200)***
- Now on Puramino 27 kcal/ounce for 32 ounces/day
- Started on creon (pancrealipase) ½ capsule per bottle for pancreatic insufficiency

# Very Early Onset Inflammatory Bowel Disease and Exocrine Pancreatic Insufficiency: A Case and Review of the Literature

# Objectives

- Review inflammatory bowel disease characteristics
- Differentiate between UC and Crohn's disease
- Discuss very-early onset inflammatory bowel disease
- Review pancreatic insufficiency

# IBD General Information

## Signs and symptoms:

- Bloody diarrhea, abd pain, and tenesmus
- Growth failure
- Extraintestinal sxs

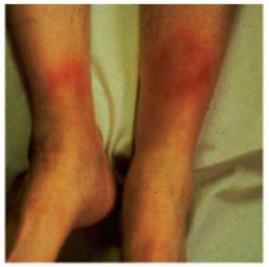
## Lab findings:

- Leukocytosis, anemia, thrombocytosis, hypoalbuminemia, elevated inflammatory markers, elevated calprotectin
- Diagnosis: biopsy

# IBD continued

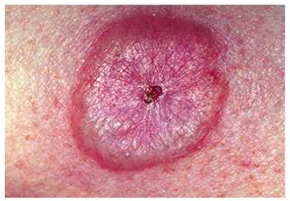
Extraintestinal manifestations:

- Dermal: pyoderma gangrenosum, erythema nodosum, alopecia, oral ulcers, bowel associated dermatosis-arthritis syndrome
- Rheumatologic: arthritis, enthesitis, sacroiliitis, ankylosing spondylitis
- Ophthalmologic: uveitis, episcleritis, iritis
- Hepatic/pancreatic/biliary: primary sclerosing cholangitis, autoimmune hepatitis, pancreatitis, cholelithiasis
- MSK: osteopenia, osteoporosis
- Hematologic: anemia, venous thrombosis
- Urologic: nephrolithiasis
- Oncologic: increased risk of cancer



Patient with inflammatory bowel disease with red r
areas on the shins which are characteristic of eryth
nodosum.

**Pyoderma gangrenosum**



Early lesion in pyoderma gangrenosum presenting as a pustular and violaceous plaque with incipient breakdown.

*Courtesy of Cynthia Magro, MD.*

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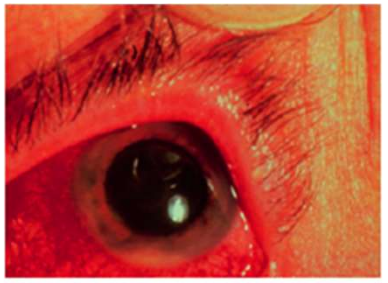
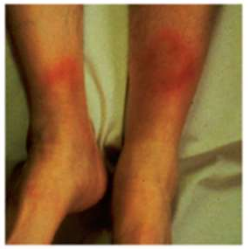


Anterior uveitis in a patient with inflammatory bo
is characterized by injection of the conjunctiva ar
the anterior chamber.

**Erythema nodosum**



Patient with inflammatory bowel disease with red r
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*Courtesy of the American Gastroenterological Association©
cannot be downloaded but may be purchased as part of a s
AGA.*

# UC vs Crohn's

Average age of onset: 15 and 30 years

Small differences in IBD incidence by sex have been reported

Both ulcerative colitis and Crohn disease are more common in Jewish compared with non-Jewish populations

Smoking is a risk factor for Crohn disease but not for UC

# Crohn's disease

Characterized by transmural inflammation

Skip areas: can involve any part of GI tract

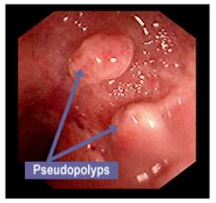
More likely to develop fistulas and/or abscesses

Histology: granulomas

Associated with fibrosis and strictures that can lead to obstruction that is not typically seen in patients with UC

First-degree relatives of patients with IBD are approximately 3- 20 x more likely to develop the disease than the general population

**Colonic pseudopolyps in inflammatory bowel disease**




Endoscopy of pseudopolyps; these lesions are not specific to ulcerative colitis, although they are more common in this disorder than in Crohn disease.

*Courtesy of James B McGee, MD.*

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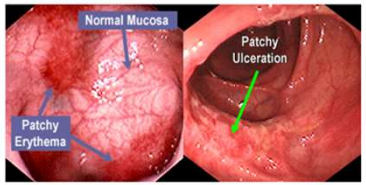
**Asymmetric distribution of lesions in Crohn disease**




Colonoscopy in Crohn disease demonstrates the characteristic patchy erythema (left panel) and ulceration (right panel) that occur next to areas of normal mucosa.

*Courtesy of James B McGee, MD.*

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# Ulcerative Colitis

Chronic inflammatory condition characterized by relapsing and remitting episodes of inflammation limited to the mucosal layer of the colon

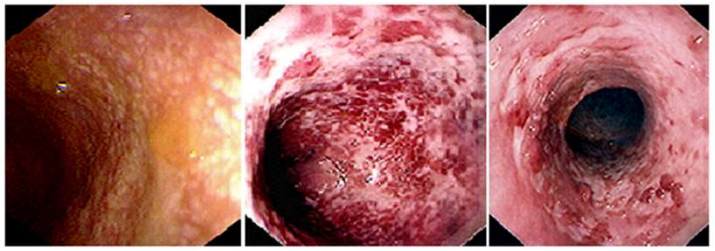
Histology: inflammation, loss vascular markings, crypt abscesses in a continuous fashion

Usually involves the rectum, and the extent often involves more proximal portions of the colon in a continuous fashion

At risk for toxic megacolon

Less likely to have fistula, abscess, or strictures

**Continuous involvement of colonic lesions in ulcerative colitis**



In contrast with Crohn disease, lower endoscopy in ulcerative colitis shows continuous and circumferential involvement, with no normal areas of mucosa.

*Courtesy of James B McGee, MD.*

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# Very Early-Onset IBD

- IBD diagnosed <6 yo (particularly < 2 yo)
- More likely to be due to monogenic mutations and can lead to more severe disease course
  - Monogenic IBD can be related defects in interleukin-10 (IL-10) signaling, or other immunological disease including CVID, CGD, Wiskott-Aldrich syndrome, agammaglobulinemia, hyperimmunoglobulin M syndrome, familial hemophagocytic lymphohistiocytosis, and IPEX or other autoimmune enteropathy syndromes

# VEO-IBD Continued

Concerning signs:

- Family history of IBD and/or immunodeficiency in multiple family members, particularly with male predominance, or consanguinity
- Recurrent infections or unexplained fever
- Associated features of autoimmunity
- Very severe IBD, complex fistulizing disease, and/or resistance to conventional therapies for IBD
- Sxs suggesting HLH: hepatomegaly, fever, cytopenias, high ferritins
- Lesions of the skin, nails, or hair
- Current or past hx cancer hx
- Endoscopic biopsies with tissue eosinophilia, villous flattening

# Pancreatic Insufficiency

- Exocrine pancreatic insufficiency (EPI) refers to inappropriate secretion of pancreatic enzymes
- Can be associated with IBD
  - 4% prevalence Crohn's
  - 10% prevalence UC
- Diagnosis based on fecal elastase test and direct pancreatic function tests
- Treatment includes pancreatic enzyme supplementation

# Association

- VEO-IBD is linked to monogenic mutations and are often immunologic in nature
- Pancreatitis involvement has also been associated with IBD through various mechanisms including medication side effect, anatomic abnormalities, extra-intestinal causes, and immunologic disturbances
- According to Singh et al, pancreatic autoantibodies were found in one third of Crohn's disease patients with pancreatic insufficiency, suggesting that immune causes play a significant role in exocrine function.
- The destruction of pancreatic tissue through either pro-inflammatory cytokines or antibodies can result in pancreatic insufficiency, leading to the need of pancreatic replacement therapy.
- Since there is little evidence on the prevalence of EPI and VEO-IBD, we suggest providers monitor patients with VEO-IBD for concerning signs of pancreatic enzyme deficiency


QUESTIONS?

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