

שלשול מולד שולל...

ד"ר יעקב אולך

המכון לגסטרואנטロולוגיה



Patient Presentation

- ▶ 52-year-old male
- ▶ Family physician
- ▶ Medical history: mild asthma
- ▶ Medications: symbicort
- ▶ Family history: uneventful
- ▶ Surgical history: appendectomy
- ▶ No smoking, alcohol use



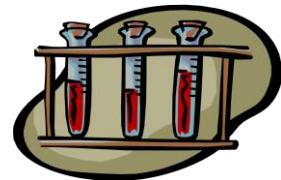


Presentation...

- ▶ Episodic abdominal pain
- ▶ Diarrhea – 5-10 times a day , watery
- ▶ Bloating
- ▶ Normal physical exam



Labs - 2011



| CBC | |
|--------------|--------|
| WBC | 9500 |
| Differential | Normal |
| Hemoglobin | 12 |
| MCV | 88 |
| Ferritin | 6 |

| CHEMISTRY | |
|--------------|--------|
| Electrolytes | Normal |
| LFT | Normal |
| Creatinine | 0.9 |
| Amylase | 84 |
| TSH | 1.2 |
| B12 | 150 |
| Folic acid | 20 |
| CRP | 0.4 |

| | |
|----------|----------|
| Anti TTG | Negative |
|----------|----------|

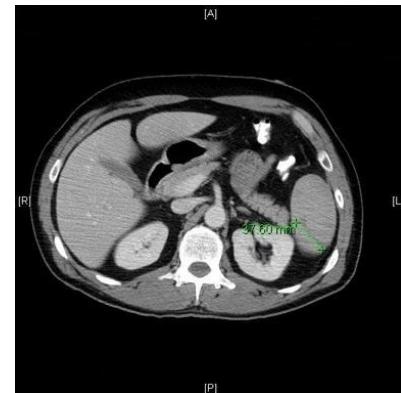
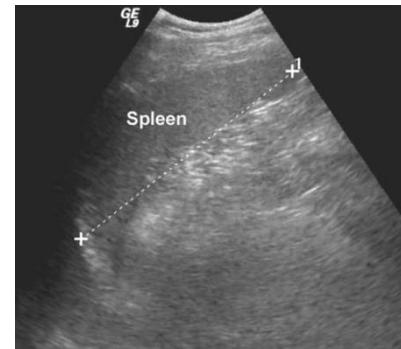
| | |
|---------------|----------|
| Stool Culture | Negative |
|---------------|----------|



Imaging Studies - 2011

- ▶ Abd US :
 - ▶ Mild Splenomegaly

- ▶ Abd CT :
 - ▶ Mild mesenteric lymphadenopathy
 - ▶ Mild Splenomegaly



Endoscopies - 2011

► Gastroscopy :

- ▶ Nodular duodenal mucosa
- ▶ Biopsies:
 - ▶ Duodenum: Nodular lymphoid hyperplasia (NLH), inflammation
 - ▶ Stomach: inflammation. HP negative



► Colonoscopy :

- ▶ Normal including biopsies

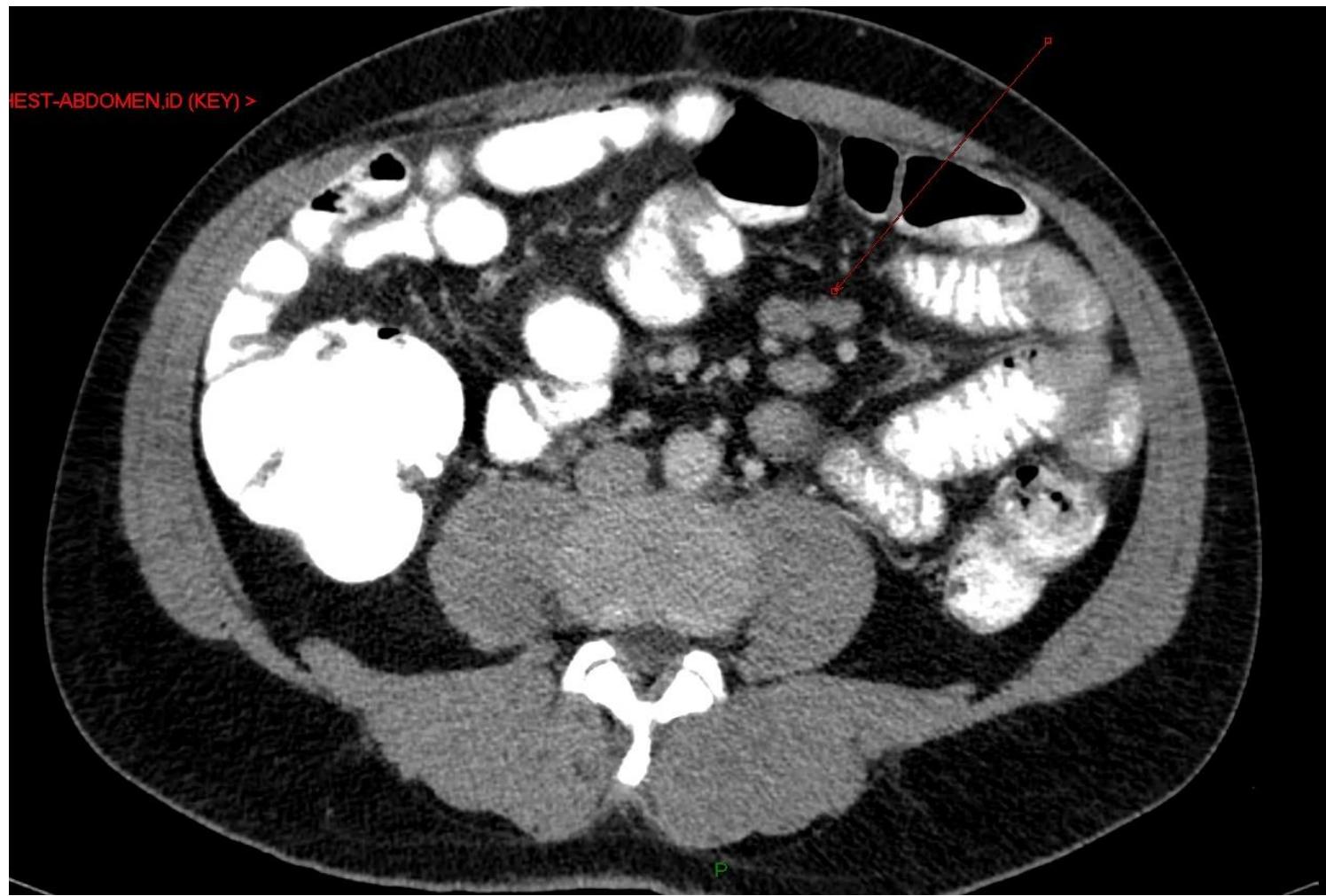


2014...

- ▶ Continued episodes of abdominal pain
- ▶ Persistent diarrhea
- ▶ Weight loss

- ▶ Repeat CT scan (5/2014):
 - ▶ Worsening mesenteric lymphadenopathy
 - ▶ Lymph nodes 2.5*2.5 c”m
 - ▶ Hepatosplenomegaly

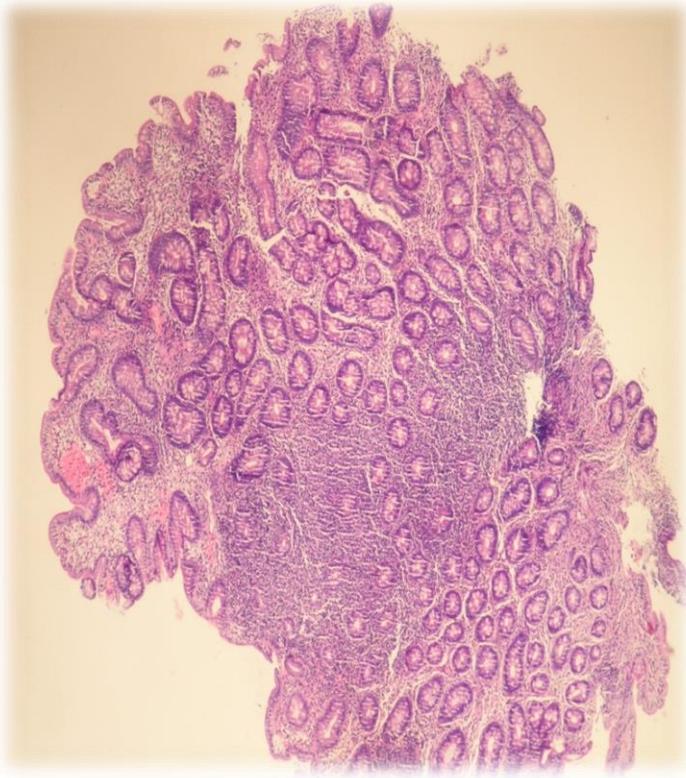




Upper Endoscopy



Duodenal Pathology



Total villous atrophy – Marsh 3c

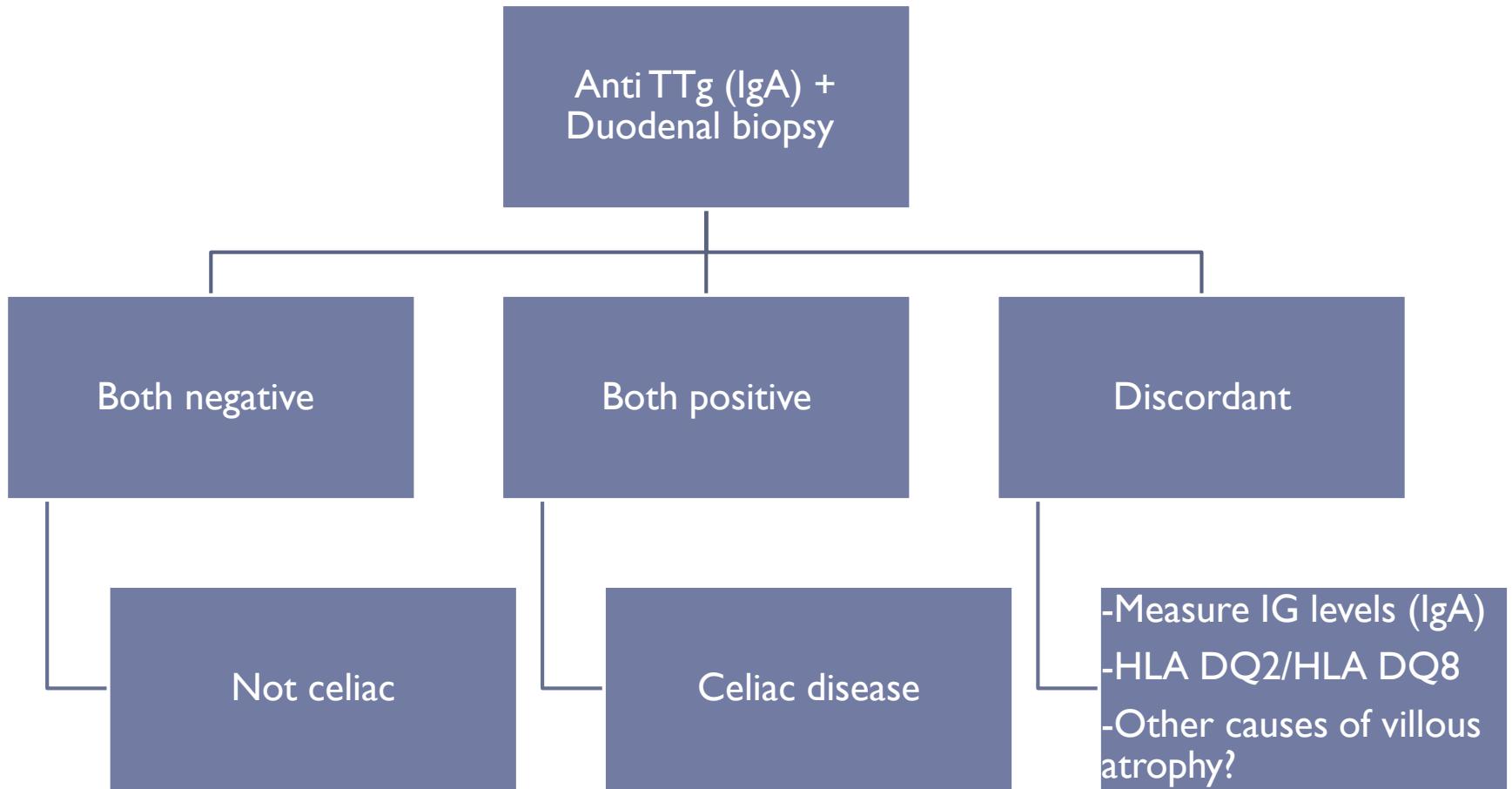


Summary...

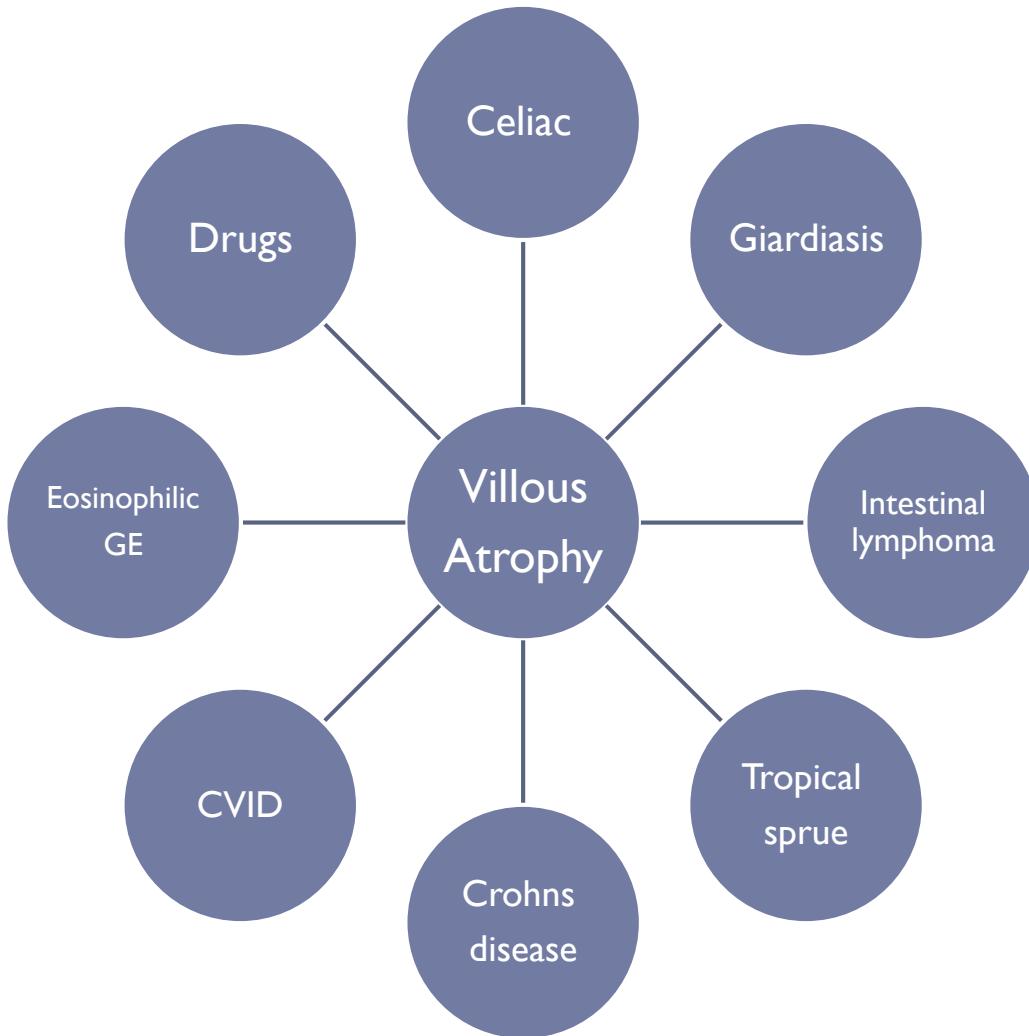
- ▶ Abdominal pain and watery diarrhea
- ▶ Anemia , iron deficiency, low levels of vitamin B12
- ▶ Villous atrophy
- ▶ Negative serology (Anti TTg)



Suspected Celiac Disease



DD of Villous Atrophy



Back to our Patient:

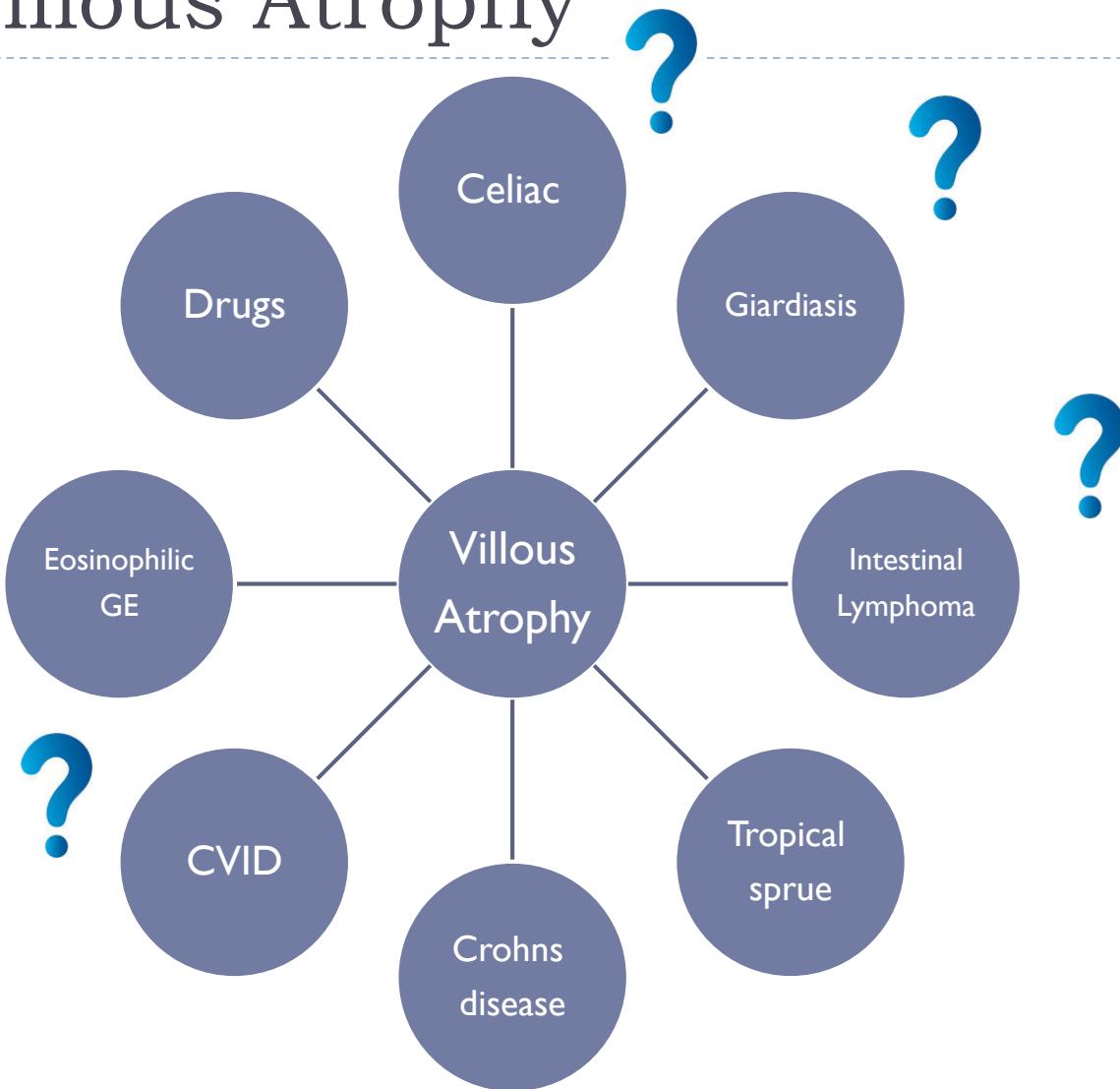
- ▶ Genetic testing:
 - ▶ HLA DQ2 - homozygote
- ▶ Labs:
 1. IgA<24 (70-500mg/dl)
 2. DGP- Normal
- ▶ Stool cultures:
 - ▶ Enteropathogens- negative
 - ▶ Parasites- positive for Giardia Lamblia
- ▶ Other immunoglobulins:
 1. IgG -190 (700-1600 mg/dl)
 2. IgM < 15 (40-230 mg/dl)



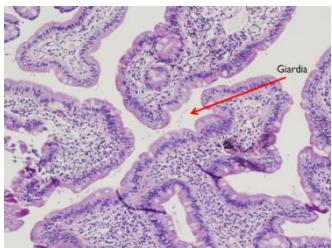
Intestinal Giardiasis



DD of Villous Atrophy



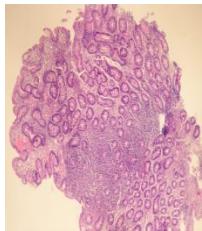
Unifying Diagnosis?



Stool samples

Recurrent
Giardiasis

Histology



Iron
deficiency
B12
deficiency

Labs

Mesenteric
lymph node
enlargement

Imaging

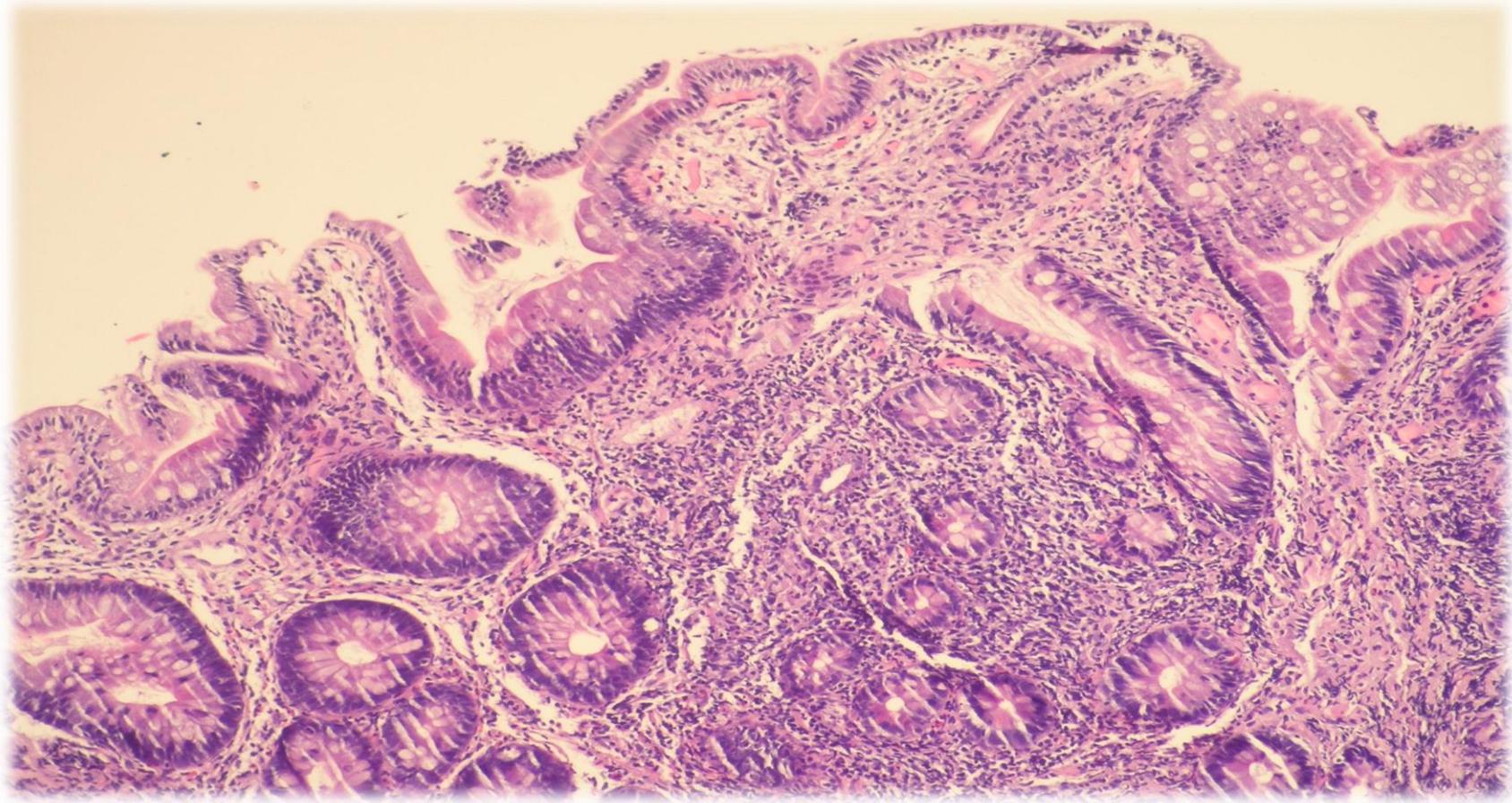


Symptoms

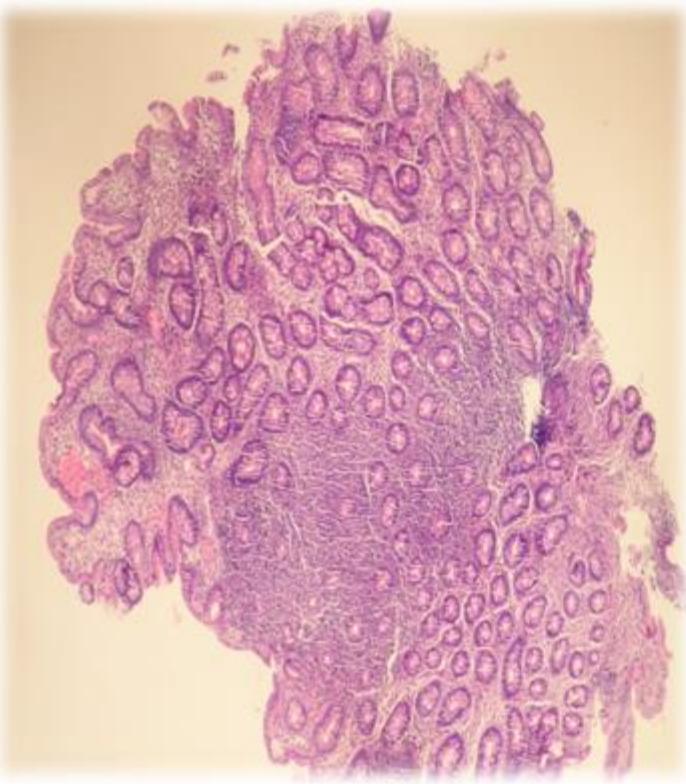
Diarrhea



CD138 (Plasma Cell) Staining



Pathology



Immunophenotype (CD3+, CD8+, CD4-,CD56-) no evidence of a lymphoproliferative process

Bone Marrow exam - Normal



Diagnosis

- ▶ Common variable immunodeficiency (CVID) with Celiac-like enteropathy and recurrent *Giardia lamblia* infection
- ▶ Celiac disease cannot be ruled out



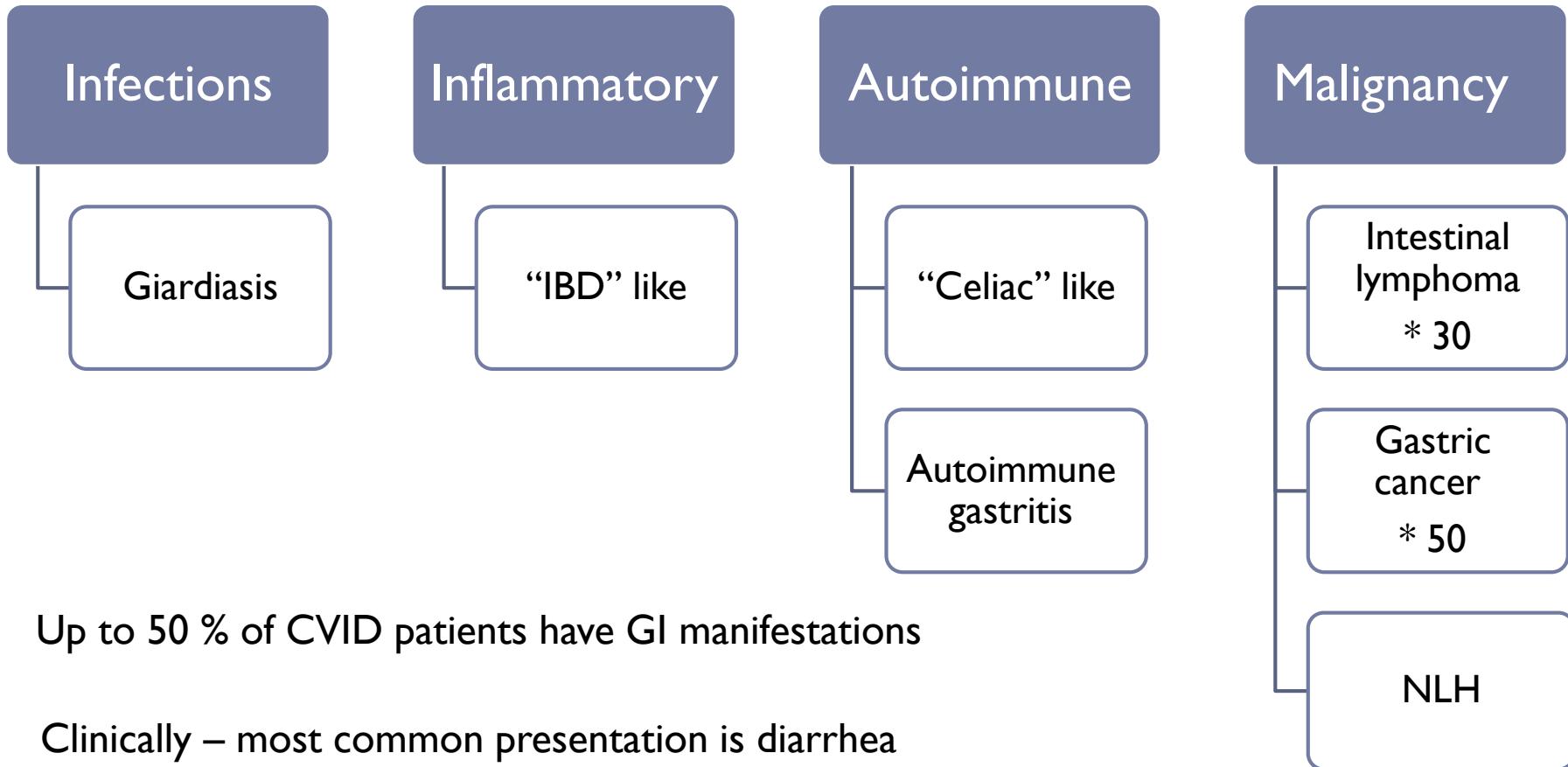
Common variable immunodeficiency (CVID)

- ▶ Impaired B cell differentiation
- ▶ Defective immunoglobulin production

- ▶ Clinical presentation
 - ▶ recurrent infections
 - ▶ chronic lung disease
 - ▶ autoimmune disorders
 - ▶ gastrointestinal disease



GI manifestations of CVID



Up to 50 % of CVID patients have GI manifestations

Clinically – most common presentation is diarrhea causing malabsorption



J Allergy Clin Immunol 2009

Other GI manifestations of CVID

- ▶ Hepatitis
 - ▶ Granulomatous
- ▶ CVID patients may develop **enlarged LN** (neck, chest or abdomen)



Treatment of CVID associated GI disease

- ▶ Gluten free diet - ineffective
- ▶ IVIg – ineffective
- ▶ Corticosteroids including budesonide - effective
- ▶ Treatment of concurrent disease
 - ▶ Giardia – metronidazole , frequent relapses
- ▶ Patients with severe malabsorption - TPN



Back to the patient

- ▶ GFD –partial response
 - ▶ AB (Metronidazole) course – improvement
 - ▶ IVIg treatment
 - ▶ Occasional steroid (budesonide) therapy
-
- ▶ Is doing well occasional diarrhea but works , no loss of weight , no pulmonary infections.



Conclusion

- ▶ To consider immunodeficiency in any patient with intractable diarrhea
 - ▶ Underdiagnosed/ Ds commonly is delayed by 6 to 8 yrs
 - ▶ Multifaceted and puzzling disorder



Thank you

