Gastroenterological Manifestations in a Patient with Common Variable Immunodeficiency

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Introduction

Gastrointestinal manifestation of common variable immunodeficiency (CVID) can present with protein-losing enteropathy (PLE). A wide spectrum of liver injuries can occur with CVID, including nodular regenerative hyperplasia (NRH).

Case Description

41 y.o. female with PMH of CVID on IgG, chronically elevated LFTs, and nutritional deficiencies. Patient presented with 8-year history of diarrhea, bloating, weight loss, abdominal pain, hepatic encephalopathy (HE), new ascites, and shock. Previously was diagnosed with celiac disease and has strictly followed a gluten-free diet with no improvement.

EGD on the current presentation showed visible changes in duodenal mucosa including villous flattening. Biopsies this time revealed villous flattening and absence of plasma cells consistent with CVID enteropathy.

Liver biopsy 5 years prior to admission showed no liver disease. Serologic workups, including anti-gp210 and anti-sp100, were negative. Biopsy showed nodular regenerative hyperplasia and non-cirrhotic portal hypertension, which was confirmed by reticulin stain.

Patient was started on budesonide 9 mg daily for treatment of PLE, and marked improvement of diarrhea was noted. Non-cirrhotic portal hypertension was managed with furosemide and spironolactone. HE was managed with oral lactulose. The patient was started on enteral and parenteral supplements.

Discussion

Duodenal villous atrophy which is a diagnostic criterion for celiac disease can be seen in multiple other conditions including immune disorders such as CVID. In CVID patients with gastrointestinal symptoms, up to 80% of biopsies from different sites in the GI tract will show histological abnormalities. Given their common presentation, celiac disease can be initially diagnosed. The absence of plasma cells and apoptosis presence in duodenal biopsy can suggest PLE diagnosis. Serological studies are not reliable for celiac disease diagnosis due to deficiency of immunoglobulin production in CVID. Steroids can be used for the treatment of PLE in CVID.

Hepatic manifestations in CVID are extremely varied and often misdiagnosed given they can mimic other diseases. NRH is a potentially fatal complication of CVID. Recognition of this complication allowed for symptom resolution within two days of starting lactulose therapy. NRH in CVID represents an autoimmune-like liver disease that might be driven by a local vasculopathy, and thus severe cases can warrant a trial of immunosuppression.

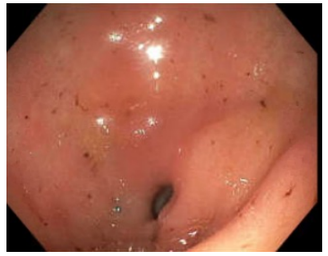

Figure 1. Moderate antral gastritis

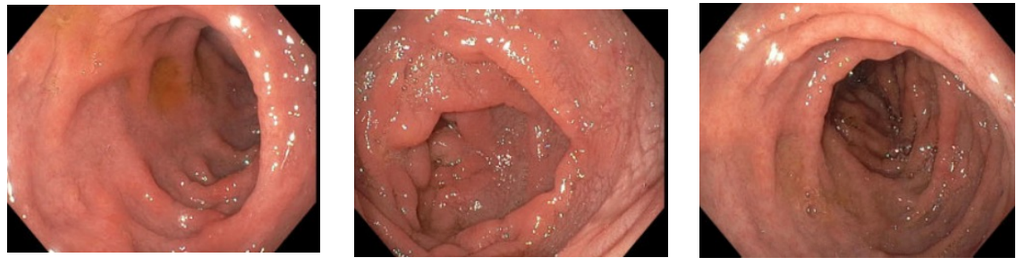

Figure 2-4. Diffuse moderate mucosal changes characterized by congestion, flattening and nodularity

Laboratory testing

MCV 81

Protein 4.4
Albumin 2.4
AST 81
ALT 67
Bili 0.6
ALP 293

Low IgG, IgA, IgM

Fecal fat Increased
Pancreatic elastase 206

9.0
2.3 155
27.8

Vitamin A 5.1
Vitamin D 25 <4
Vitamin E 0.8
B12 394
Zinc 36
Copper 22
Ferritin 23.6
Iron 29
%sat 20
TIBC 143

EGD 6 years prior to presentation showed celiac disease on duodenal biopsy
Celiac serologies negative
Celiac HLA DQ2/DQ8 negative

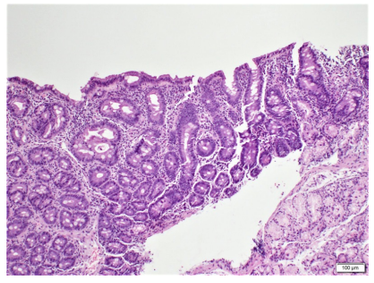

Figure 5. The biopsy from duodenum shows diffuse villous flattening.

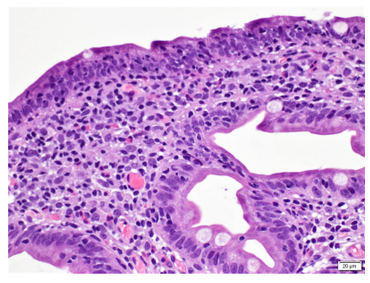

Figure 6. The lamina propria contain moderately increased lymphocytes associated with lymphocytic infiltrate in superficial epithelium and glands.

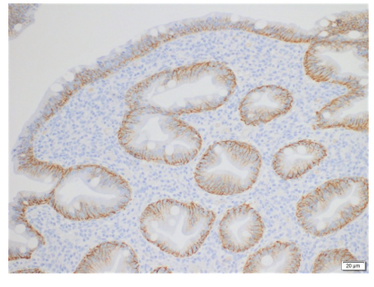

Figure 7. Immunostaining for CD138 demonstrates absence of plasma cells in lamina propria

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