

## #137 - CLINICAL, ENDOSCOPIC AND HISTOLOGIC FEATURES OF COMMON VARIABLE IMMUNODEFICIENCY

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### 1 Introduction

Common variable immunodeficiency (CVID) is a rare illness with estimated prevalence of 1/25,000 individuals. Studies describing endoscopic and histopathological findings, and therapeutic options are lacking.

### 1 Objectives

To describe gastrointestinal (GI) symptoms, endo and histo findings, and real-world experience treatment of patients with CVID enteropathy (CVIDe).

### 1 Methods

Retrospective review of patients ≥16 years of age diagnosed with primary CVID who underwent endo evaluation for GI symptoms at a major three site academic medical center. Patients were identified by searching our institution's database. Demographics, GI symptoms, endo and histo findings, and treatments were abstracted. Descriptive statistics were used to analyze the results.

### 1 Results

A total of 57 patients were included; 53% female, median age at CVID diagnosis was 32 years (range 4-66). A total of 147 procedures were included. The most common GI symptoms leading to endo evaluation included diarrhea (50%), abdominal pain (29%), nausea (23%), weight loss (17%), vomiting (14%), and less frequently GI bleeding, bloating, anorexia/early satiety, heartburn, dysphagia, iron and vitamin B12 deficiency. Endoscopic and histopathological findings are described in Table 1. Twelve patients were diagnosed with CVIDe and received treatment with budesonide (9), prednisone (1), mesalamine (1), vedolizumab (2), infliximab (2), ustekinumab (1), sirolimus (1), abatacept (1), cholestyramine (1) in addition to intravenous or subcutaneous immunoglobulin. One patient underwent bone marrow transplant for medically refractory disease.

### 1 Conclusion

GI symptoms are frequent in patients with CVID. While endoscopic and histopathological findings may be normal, decreased plasma cells and apoptosis are characteristic of CVIDe. In our series, 21% of the patients were diagnosed with CVIDe, similar to 15-20% as described by the literature. Most patients were treated with budesonide and 25% of patients eventually required biologics. Despite the lack of randomized controlled trials, patients with CVIDe may benefit from treatment with immunomodulators/biologics to reduce morbidity and mortality.

Table 1. Endoscopic and histopathological findings

Type of procedure	Number of procedures n = 147 (%)
- EGD	73 (50)
- Upper DBE	1 (1)
- Colonoscopy	63 (43)
- Flexible sigmoidoscopy	9 (6)
- Lower DBE	1 (1)
<b>EGD/Upper DBE endoscopic findings (n=74)</b>	
- Normal	30 (41)
- Esophagitis	5 (7)
- Gastric erythema and/or erosions/ulcer	23 (31)
- Gastric atrophy	4 (5)
- Gastric polyp	4 (5)
- Duodenal erythema	2 (3)
- Scalloping duodenum/Villous blunting	16 (22)
- Nodular mucosa duodenum	4 (5)
- Ulcers duodenum/jejunum	1 (1)
<b>Colonoscopy/Flexible sigmoidoscopy/Lower DBE endoscopic findings (n=73)</b>	
- Normal	41 (56)
- Nodular mucosa terminal ileum	2 (3)
- Atrophic mucosa terminal ileum	1 (1)
- Aphtha/erosion/ulcer terminal ileum	3 (4)
- Granularity terminal ileum	1 (1)
- Inflammatory changes in 1 or more colonic segments	11 (15)
- Inflammatory changes ileocolonic anastomosis	1 (1)
- Pseudopolyps	2 (3)
- Other polyps/polypoid lesions	17 (23)
<b>EGD/Upper DBE histopathology findings (n=74)</b>	
- Normal	16 (22)
- Apoptosis	2 (3)
- Decreased/absent plasma cells	21 (28)
- Decreased goblet and Paneth cells	1 (1)
- Villous blunting	19 (26)
- Crypt distortion	7 (9)
- Brunner gland hyperplasia	1 (1)
- Lymphoid aggregates	2 (3)
- Prominent lymphoid follicle/lymphoid hyperplasia	4 (5)
- Increased intraepithelial lymphocytes (duodenum)	14 (19)
- Active duodenal inflammation	3 (4)
- Chronic duodenal inflammation (peptic and non-peptic)	8 (11)
- Increased eosinophils	1 (1)
- Reactive gastropathy	14 (19)
- Active chronic gastritis	2 (3)
- Chronic gastritis	13 (18)
- Lymphocytic gastritis	3 (4)
- Autoimmune gastritis/atrophic gastritis	6 (8)
- Gastric mucin cell or foveolar metaplasia	6 (8)
- Multinucleated giant cells	1 (1)
- Increased epithelial lymphocytes (esophagus)	4 (5)
- Active inflammation esophagus	2 (3)
- Active chronic inflammation esophagus	1 (1)
- Giardia in the duodenum	3 (4)
- Positive Helicobacter pylori	1 (1)
- No biopsies	2 (3)
<b>Colonoscopy/Flexible sigmoidoscopy/Lower DBE histopathological findings (n=73)</b>	
- Normal	21 (29)
- Apoptosis	7 (10)
- Decreased/absent plasma cells	13 (18)
- Villous blunting terminal ileum	2 (3)
- Lymphoid aggregates	7 (10)
- Prominent lymphoid follicle/lymphoid hyperplasia	3 (4)
- Active ileitis	6 (8)
- Active chronic ileitis	1 (1)
- Active colitis	13 (18)
- Active on chronic colitis	2 (3)
- Chronic colitis/crypt distortion	5 (7)
- Increased subepithelial collagenous band	4 (5)
- Increased intraepithelial lymphocytosis	14 (19)
- Decreased goblet cells	1 (1)
- Pseudopolyps	1 (1)
- CMV	2 (3)
- Other polyps: TA, serrated, hyperplastic	11 (15)

EGD = upper endoscopy, DBE = double balloon enteroscopy, CMV = Cytomegalovirus, TA = tubular adenoma.  
 \*Only 45% of the procedures with normal endoscopic appearance had normal histology.