Case Report

A Rare Case of Immunodeficiency Diagnosed During Upper Gastro Intestinal Endoscopy

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Abstract

Common variable immunodeficiency (CVID) is the most prevalent primary immunodeficiency disease. It is defined as hypogammaglobulinemia with normal B cell phenotype and recurrent episodes of infection. CVID can mimic Familial Adenomatous Polyposis (FAP) on endoscopy. Here we are reporting rare case of Immunodeficiency diagnosed during Upper Gastro Intestinal endoscopy.

Abbrevations : CVID (Common variable immunodeficiency) , NLH (Nodular lymphoid hyperplasia) , IVIG (Intravenous immunoglobulin).

Introduction

Common variable immunodeficiency (CVID)is defined as hypogammaglobulinemia with normal B cell phenotype and recurrent episodes of infection.[1,2] CVID are sporadic mostly however an autosomal dominant & recessive pattern of inheritance also exits. The various gastrointestinal manifestations of CVID are atrophic gastritis, pernicious anemia, chronic giardiasis, nodular lymphoid hyperplasia (NLH), and intestinal malabsorption.[3] Predominant symptoms in CVID can be diarrhea, weight loss, dyspepsia, sinopulmonary infections and malabsorption.[4] Majority of the gastrointestinal manifestation in CVID are due to T-cell-mediated defects.[11]

Case report

A 25-year-old male presented with dyspeptic symptoms to our institute with no significant past history. On physical examination, patient is thin built and routine laboratory data and HIV Serology were unremarkable. Patient underwent upper gastrointestinal endoscopy which showed numerous polyps like a carpet in the second part of duodenum and even extending to third part (Fig 1). Multiple biopsies were taken from duodenal mucosa to rule out familial adenomatous polyposis and patient planned for sigmoidoscopy and genetic markers. However, there is no history of polyps in the family. The specimens were sent to the pathology department in 10% buffered formalin and processed and microscopic examination showed mild villous atrophy, with lamina propria showing reactive lymphoid follicles. No malignant cell / plasma cell/granuloma cell/parasites seen (Fig 3,4). Histology report was discussed with pathologist and the possibilities of nodular lymphoid hyperplasia, immunodeficiency and celiac sprue were also discussed. IHC has a role for work up of lymphoma, provided histopathology picture shows plasma cells. However, in our patient plasma cells were absent. So, immunohistochemistry was not done.

Sigmoidoscopy was undertaken as a screening purpose. Nodular Lymphoid Hyperplasia is associated with left sided colonic polyp. Hence, only limited colon was studied. For further workup patient was subjected to sigmoidoscopy which was normal. As sigmoidscopy was normal, so further work up done for CVID. Total immunoglobins were sent and the results of laboratory tests are listed in Table 1. CVID was diagnosed by reduction of serum IgA, IgG & IgM levels in this patient and patient plan for the immunoglobin therapy with dosage of 300-600mg/kg subcutaneous monthly for life long. However, for personal reason, patient is being treated elsewhere. Patient was advised for family screening as 5% to 25% cases are familial, with an autosomal-dominant pattern of inheritance being more frequently observed.

Total Immunoglobins	Normal Range (mg/dl)	Patient Value (mg/dl)
IgA	70-400	=<26.2
IgG	700-1600	290
IgM	40-230	22.6

Table 1 - Level of various total immunoglobins

Discussion:

We are presenting a patient with uninvestigated dyspepsia who was found to have numerous polyps on routine endoscopy and further work up revealed CVID.

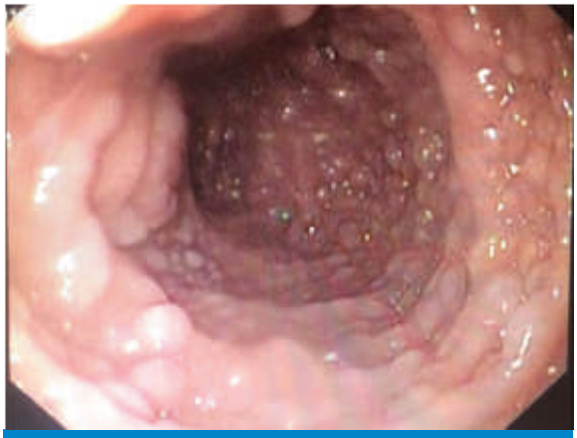

Fig 1: Endoscopy picture showing numerous polyps in D2.

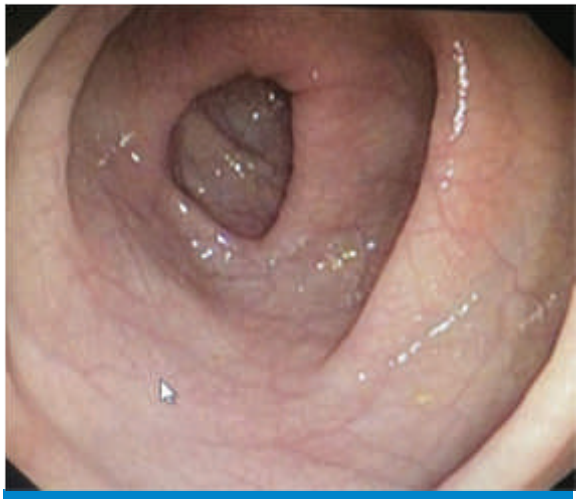

Fig 2: This picture shows normal colon mucosa.

CVID is the most prevalent primary immunodeficiency disease along with antibody deficiencies. The defects in cellular immunity are responsible for CVID2,[5–7] In severe cases, malabsorption of vitamins, dietary fat, carbohydrates and folate is seen.[7] In CVID patients, nodular lymphoid hyperplasia along with villous

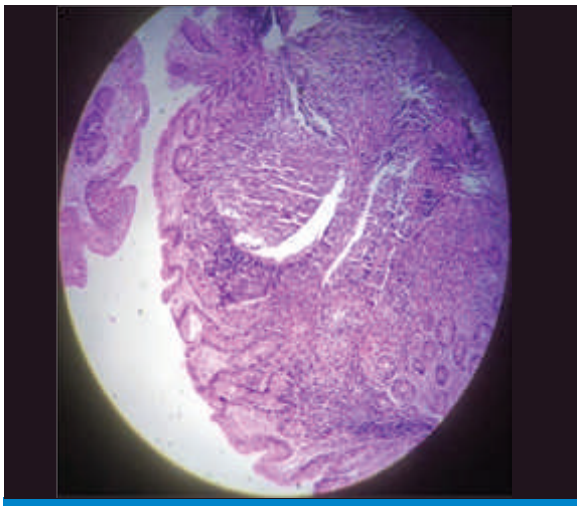

Fig 3: Histopathology picture shows Mild villous atrophy with underlying lamina propria showing prominent lymphoid follicles.

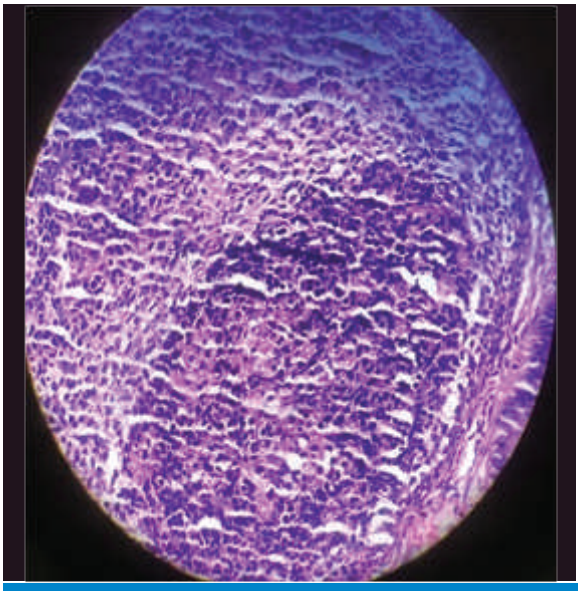

Fig 4: Histopathology Picture showing Reactive Lymphoid Follicles

atrophy is the predominant finding in mucosal biopsy. Similar findings may be seen in celiac sprue and lymphoma.[8] However, this finding does not correlate with malabsorption and plasma cells are not present in the extra mantle zones and similar finding seen in our case.[8] The major concern was a mismatch in clinical diagnosis of FAP, as per endoscopy finding, though no adenomatous polyp or dysplasia was found.

This misinterpretation is due to polypoid appearance of large lymphoid follicle present in the lamina propria. Thus, high index of suspicion is required to evaluate CVID in patients with multiple polyps having histologically picture of nodular lymphoid hyperplasia and good communication is required with histopathologist for meticulous work up.

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