

A Delayed Diagnosis of Common Variable Immunodeficiency in a Patient with Chronic Enteropathy

Introduction:

Common Variable Immunodeficiency (CVID) is a primary disorder characterized by decreased B cell differentiation leading to panhypogammaglobulinemia. Diagnosis can be delayed until late adulthood due to the variable systemic presentation of CVID.

Case Description:

47-year-old female from the Dominican Republic, with a complex medical history including severe asthma, bronchopneumonia, anemia, anorexia, and chronic diarrhea, was admitted with worsening shortness of breath, fever, and cough. Physical examination revealed respiratory distress, wheezing, and clubbing. Laboratory findings indicated leukocytosis and elevated lactate. Imaging revealed lung consolidations and chronic changes. The patient was diagnosed with acute hypoxic respiratory failure, asthma exacerbation, and septic shock secondary to extensive bronchopneumonia. Treatment included broad-spectrum antibiotics, prednisone, nebulizers, and oxygen supplementation. Further investigations revealed low iron and vitamin levels indicating malabsorption of iron and fat-soluble vitamins. Undetectable Immunoglobulin levels with normal CD4 levels and CD4/CD8 ratio confirmed CVID. GI was consulted for malabsorption and iron deficiency anemia. Endoscopy showed atrophic gastritis and intestinal metaplasia. The patient improved with treatment and was discharged with gastroenterology and immunology follow-up.

Discussion:

CVID-associated Chronic Enteropathy, though observed in only 10-20% of cases, manifests with chronic diarrhea, weight loss, and deficiencies in fat-soluble vitamins, often accompanied by symptomatic iron deficiency anemia. Pathological findings include low-grade dysplasia, chronic atrophic gastritis, and villous atrophy, leading to malabsorption syndrome. The case underscores the significance of recognizing these gastrointestinal manifestations in CVID, as they contribute to the complexity of the disease. Moreover, the identification of premalignant lesions like chronic atrophic gastritis and intestinal metaplasia highlights the need for regular endoscopic surveillance to mitigate the elevated risk of gastric adenocarcinoma and other complications in these high-risk patients.