Atypical Presentation of Celiac Disease in asymptomatic adult with no significant medical history : A rare diagnostic challenge

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Abstract

Background: Celiac disease mainly presents with gastrointestinal symptoms, but atypical presentations, including liver dysfunction, are increasingly recognized.

Case Presentation: We report the case of a 49-year-old asymptomatic healthy gentleman, having mildly elevated cholesterol managed through healthy lifestyle. He had no history of alcohol or drugs intake and a negative family history of Celiac or liver disease. Patient was incidentally found to have markedly elevated liver transaminases on routine blood tests. Subsequent detailed workup for elevated liver transaminases including Celiac serology was normal.

Conclusion: Celiac disease must be considered in the differential diagnosis of the patients with unexplained liver enzyme abnormalities without gastrointestinal symptoms and negative serological tests.

Keywords: Celiac disease (CD), Seronegative Celiac disease (SNCD), Liver enzymes, Gluten-free diet

Introduction

Celiac disease (CD) is an immune-mediated enteropathy precipitated by gluten in genetically susceptible individuals, characterized by villous atrophy and lymphocytic inflammation of the epithelial layer covering the mucosa (1).

Celiac disease (CD) is classically known for gastrointestinal symptoms such as diarrhea, weight loss, and abdominal pain. However in many patients especially adults, extraintestinal manifestations (hepatic abnormalities, dermatological, hematological, skeletal and endocrine) are increasingly recognised (2). These can often be the first or only signs of the disease. Recognizing these can help clinicians diagnose CD earlier and prevent its complications.

Liver involvement in CD, often termed 'Celiac hepatitis', can present as elevation of liver transaminases and can occur without clinical evidence of CD, occasionally leading to significant diagnostic delays (3) if CD is not considered.

Here, we present a rare case of asymptomatic celiac disease diagnosed during investigation of elevated liver transaminases in a person with normal celiac serology.

Case Presentation

A 49-year-old man, physically active and with no significant medical history aside from diet-controlled hypercholesterolemia, underwent routine blood tests revealing:

- ALT: 547 IU/L (normal <50)
- AST: 245 IU/L (normal <45)
- ALP: 145 IU/L (normal <120)

Bilirubin, albumin, clotting profile, viral hepatitis panel (HAV, HBV, HCV, HEV), autoimmune markers (ANA, ASMA, AMA), metabolic workup (iron studies, ceruloplasmin, alpha-1 antitrypsin) were all within normal limits. The Celiac screen serology including TTG IgA and TTG IgG were negative and patient had normal total serum IgA levels (ruling out IgA deficiency which can cause a false negative serological test). HLA typing was not performed. Imaging studies including abdominal ultrasound and MRI of the liver and biliary tree were unremarkable. Ultrasound-guided liver biopsy demonstrated no specific histological features and showed mild portal and lobular inflammation.

In light of persistent liver enzyme elevation an esophagogastroduodenoscopy (OGD) was performed, revealing:

- Normal esophageal and gastric mucosa
- Erythema and erosions in D1
- Scalloping of the folds in D2
- Negative CLO test for H. pylori

Targeted duodenal biopsies with the above findings were obtained and showed villous atrophy, crypt hyperplasia, and intraepithelial lymphocytosis, corresponding to Marsh classification 3B, confirming celiac disease (4).

Patient's liver enzymes improved significantly after six months of gluten-free diet, further strengthening the diagnosis of coeliac disease. The patient remained asymptomatic.

Discussion

This case illustrates an atypical presentation of CD manifesting solely as marked liver enzyme abnormalities in a clinically well patient. While mild transaminase elevation is common in CD, severe liver enzyme elevation is rare and may resemble viral or autoimmune hepatitis (3,5).

Seronegative celiac disease (6) though rare, especially in early or patchy disease or in IgA deficiency (7) can occur and necessitates biopsy for definitive diagnosis. Mechanisms proposed for hepatic involvement include immune dysregulation and increased intestinal permeability leading to systemic inflammation.

Celiac disease should be considered in the differential diagnosis of unexplained liver enzyme abnormalities, even in patients without gastrointestinal symptoms or positive serology.

This case illustrates the diagnostic importance of duodenal biopsy in unexplained abnormal liver function tests and the potential reversibility of liver dysfunction with dietary intervention (8).

Conclusion

Isolated liver enzyme abnormalities can be a rare presentation of silent celiac disease. Negative serology does not exclude the diagnosis. Clinicians must maintain a high index of suspicion and consider duodenal biopsy in persistent unexplained liver enzyme abnormalities. Early diagnosis and gluten-free diet initiation can reverse liver dysfunction and prevent further complications.

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