CASE REPORT

DOI: 10.4274/cmj.galenos.2023.70299
J Cau Med Sci 2023;1(3):48-50

Celiac Disease's Early Diagnosis: A Case Report

Marina Fedossina[1], Zhaniya Kanash[2], Ainash Tanabayeva[2], Gaukhar Kozhakeeva[3], Jamilya Kaibullayeva[3], Alma Khabizhanova[4], Indira Khamitova[5], Eugene Enin[6]

[1]JSC Research Institute of Cardiology and Internal Diseases, Department of Functional and Ultrasound Diagnostics, Almaty, Kazakhstan
[2]JSC Research Institute of Cardiology and Internal Diseases, Department of Clinical Trials, Almaty, Kazakhstan
[3]JSC Research Institute of Cardiology and Internal Diseases, Department of Therapy No. 1, Almaty, Kazakhstan
[4]JSC Research Institute of Cardiology and Internal Diseases, Department of Endoscopy and Interventional Gastroenterology, Almaty, Kazakhstan
[5]Regional Clinical Hospital, Endoscopic Center, Karaganda, Kazakhstan
[6]National Scientific Center of Surgery named after A.N. Syzganov, Pathoanatomic Laboratory, Almaty, Kazakhstan

ABSTRACT

Celiac disease (gluten-sensitive enteropathy) occurs because of an immune-mediated reaction to gluten in genetically predisposed individuals. In clinical practice, there are often difficulties in diagnosing celiac disease, particularly in patients without specific gastrointestinal complaints. Rarely, celiac disease can present with acute symptoms, delaying diagnosis and exposing these patients to possible life-threatening complications. In this article, we present a review of the literature and a case report demonstrating the usefulness of intestinal ultrasound in the primary diagnosis of celiac disease, followed by complete confirmation of the diagnosis by enteroscopy with biopsy and serologic diagnostics.

Keywords: Celiac disease, gluten-sensitive enteropathy, intestinal ultrasound

INTRODUCTION

Celiac disease is a multisystem immune-mediated disease caused by gluten ingestion in genetically predisposed individuals. The primary target organ of the autoimmune response against the tissue transglutaminase (TG2) enzyme is the small intestine, where a gluten-related inflammatory cascade causes progressive mucosal damage leading to severe villous atrophy. The gold standard for diagnosing celiac disease and/or gluten-related hypersensitivity is endoscopic examination with a series of multiple biopsies (to detect Marsh-Oberhüber duodenal mucosal atrophy) and serological confirmatory methods, including antibodies to TG2, deamidated gliadin peptides, and gliadin [1]. From a clinical perspective, celiac disease is a multifaceted chronic disease that exhibits several intestinal (ranging from mild irritable bowel syndrome-like symptoms to severe malabsorption symptoms) and extraintestinal manifestations affecting multiple tissues and organs (eg. skin, endocrine/exocrine glands, nervous system, joints/muscles). Only 20-30% of patients suffer from the ordinary active form of the disease (malabsorption, diarrhea, overall weakness anemia weight loss). Non-diagnosed, non-active forms of the disease constitute up to 70-80% of cases in adults [2]. Delay in diagnosis is common in clinical practice, especially in patients without gastrointestinal complaints; the median delay in diagnosis was 2.3 months for the group with gastrointestinal symptoms and 42 months for the non-gastrointestinal group ($p<0.001$) [3]. Therefore, celiac disease remains a difficult condition to diagnose, leading to a significant delay in initiating appropriate therapy and an increase in associated morbidity [4].

CASE PRESENTATION

A 47-year-old woman complained of diarrhea up to 5-6 times a day without pathological impurities during defecation, bloating, and general weakness. The patient reported that the complaints had been bothering her for 5 years; the onset of the disease was associated with stress. The patient did not note a clear connection between symptoms and food intolerance. She was examined several times and received symptomatic therapy. Exacerbations were associated with stressful situations. The patient self-stopped the symptoms by taking loperamide. During the physical examination, signs of skin and mucosal paleness and abdominal bloating were detected. The NRS 2002 scale was used to evaluate the nutritional status of the patient; the total score was 2 points. From the anamnesis of life, the

Address for Correspondence: Ainash Tanabayeva MD, JSC Research Institute of Cardiology and Internal Diseases, Department of Clinical Trials, Almaty, Kazakhstan
Phone: +7 701 600 75 65 E-mail: ainashik@list.ru ORCID ID: 0000-0003-3228-9796
Received: 09.09.2023 Accepted: 22.11.2023

patient suffered from viral hepatitis A in childhood. At the time of the examination, she was periodically observed by a gynecologist for uterine myoma. Laboratory studies revealed mild anemia (hemoglobin: 96 g/L), thrombocytosis (platelets: 490 x 10 x 9/L), elevated D-dimer -1139 µg/mL, erythrocyte sedimentation rate acceleration up to 21 mm/h. In addition, the patient had hypoproteinemia (total protein: 57.12 g/L). Studies of stool samples revealed mild excretory pancreatic insufficiency (pancreatic elastase: 159.26 µg/g) and normal levels of calprotectin (11.86 ng/m). Taking into account the duration of the disease and violation of nutritional status, the patient was hospitalized in the therapeutic department for additional examination and selection of therapy. During hospitalization, the infectious nature of the pathology was excluded (fecal tests). Routine abdominal ultrasound without any features The patient was decided to undergo an intestinal ultrasound, which revealed an expansion of the ileal cavity with clearly defined circular folds - jejunization (Figure 1), in connection with which the patient was suspected of enteropathy.

For a more accurate assessment of the condition of the gastrointestinal tract, the patient underwent endoscopic examinations with biopsy sampling. Ileocolonoscopy revealed sluggish peristalsis in the colon in all its departments without mucosal defects. However, thickening and shortening of the villi in the ileum were observed in the small intestine, and more proximal, in the jejunum, confluent lesions with low, inactive villi were visualized, indicating focal atrophy of the small intestine. On esophagogastroduodenoscopy with enteroscopy, the esophagus and stomach were unremarkable. The mucosa of the duodenal bulb was thinned, and the velvet was sharply smoothed out with linear areas of atrophy; this process extended to the subbombous sections. The small intestine was examined down to the lower third of the jejunum. The mucosa was thinned over the entire examined area, and the villi were thickened, low, and immobile (Figure 2). Folding was preserved. Based on the described picture, an endoscopic conclusionis as follows: "atrophic duodenitis, jejunitis" was made, and biopsy material was taken.

After endoscopic examination, celiac disease was suspected, so serological diagnostics were performed, as a result of which positive antibodies to TG2, gliadin, and endomysium were detected.

On pathohistological examination of biopsy specimens of the jejunum, the villi were dilated, flattened, and lined with rimmed enterocytes with a moderate number of goblet cells; the number of intraepithelial lymphocytes was up to 50 per 100 enterocytes; the ratio of the height of the villi to the depth of the crypts was 2:1; the crypts were deep and filled with border enterocytes, goblet cells, and Paneth cells; the lamina propria was moderately infiltrated by lymphocytes and plasmocytes with an admixture of eosinophils [4]. Histopathological conclusion: "the morphological picture of chronic atrophic lymphocytic duodenitis corresponds to gluten-sensitive enteropathy type 3b, according to Marsh-Oberhuber (Figure 3)." Thus, the patient was diagnosed with celiac disease and was transferred to a gluten-free diet and discharged with further recommendations. Taking into account the duration of the disease, systemic steroids (methylprednisolone) were prescribed for up to 12 weeks.

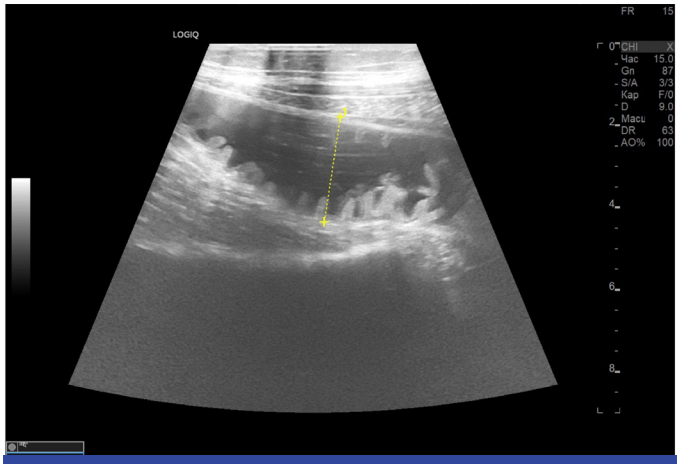

Figure 1. Fragment of the ileum, with a slightly enlarged lumen and clearly defined circular folds- jejunization; during fasting, an increased amount of fluid in the lumen of the small intestine and its increased peristalsis are determined

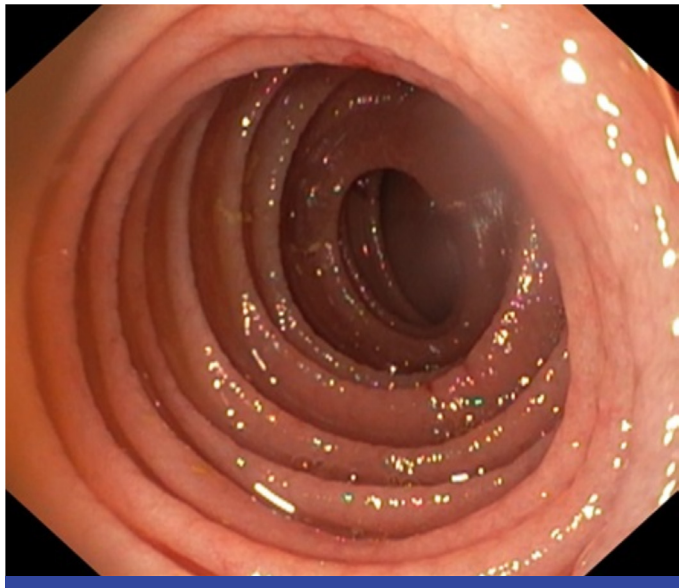

Figure 2. Endoscopic photo of the small intestine showing villous atrophy

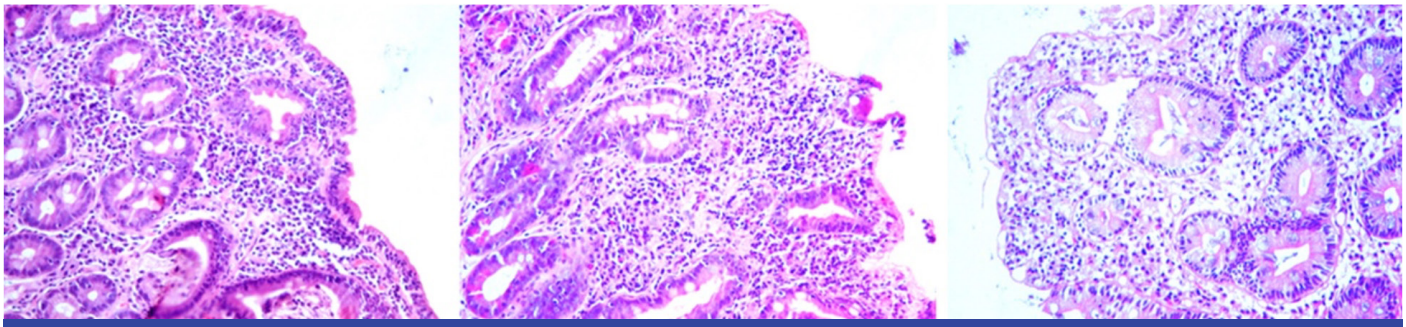

Figure 3. Biopsy of the jejunum, magnification 200. Staining: hematoxylin and eosin. In these preparations, there is an expansion of the lamina propria due to abundant lymphoplasmacytic infiltration, a decrease in villi, and an increase in intraepithelial lymphocytes

DISCUSSION

Today, ultrasound examination of organs is widely used in medicine for screening before the appointment of invasive and expensive diagnostic procedures. Intestinal ultrasound is mainly used to diagnose and monitor patients with inflammatory bowel disease. Various ultrasound features associated with celiac disease have already been described in the literature, and the recent advent of high-frequency transducers (5-12 MHz) has made it possible to assess the condition of the loops of the small intestine and abdominal cavity using this method [5]. Using ultrasound as the first (non-invasive) line of diagnosis, including in clinically asymptomatic patients with untreated celiac disease, it is possible to determine several sonographic signs characteristic of this disease [6]:

• Increased fluid content in moderately dilated intestinal loops (25-35 mm) with fasting hyperperistalsis;

• Slightly thickened wall of the small intestine (3-5 mm) and thickened cuspid valves, most often observed in patients with untreated celiac disease;

• A decrease in the number of folds of the jejunum and an increase in the folds of the ileum (jejunization of the ileum);

• Periodic invasion due to hyperperistalsis;

• Presence of slightly enlarged mesenteric lymph nodes.

Therefore, intestinal ultrasonography may help avoid delaying the diagnosis, especially in patients with atypical clinical manifestations of celiac disease. This is a promising method with high sensitivity and specificity. Although the gold standard for diagnosing celiac disease is histological confirmation of bowel involvement in serologically positive individuals, expanding the range of diagnostic options for clinically asymptomatic patients with untreated celiac disease is possible through ultrasound diagnostics. The limitation may be the lack of experts with the skill of ultrasound diagnostics of the pathology of the abdominal cavity and intestines.

Ethics

Informed Consent: Obtained.

Peer-review: Externally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: M.F., Z.K., A.T., G.K., J.K., A.K., I.K., E.E., Concept: M.F., Design: G.K., Data Collection or Processing: Z.K., Analysis or Interpretation: A.T., Literature Search: M.F., Writing: Z.K., A.T.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

References

1. Velikova T, Taneva G, Chourchev S, Tankova L, Vladimirov B. A case report of a patient with gluten enteropathy: the importance of immunological markers for diagnosis and follow-up. ARC Journal of Hepatology and Gastroenterology. 2018;3:22-4.
2. Bartusek D, Valek V, Husty J, Uteseny J. Small bowel ultrasound in patients with celiac disease. Retrospective study. Eur J Radiol. 2007;63:302-6.
3. Paez MA, Gramelspacher AM, Sinacore J, Winterfield L, Venu M. Delay in diagnosis of celiac disease in patients without gastrointestinal complaints. Am J Med. 2017;130:1318-23.
4. Guarino M, Gambuti E, Alfano F, Strada A, Ciccocioppo R, et al. Life-threatening onset of coeliac disease: a case report and literature review. BMJ Open Gastroenterol. 2020;7:e000406.
5. Fraquelli M, Colli A, Colucci A, Bardella MT, Trovato C, et al. Accuracy of ultrasonography in predicting celiac disease. Arch Intern Med. 2004;164:169-74.
6. Kralik R, Trnovsky P, Kopáčová M. Transabdominal ultrasonography of the small bowel. Gastroenterol Res Pract. 2013;2013:896704.