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Small bowel capsule endoscopy, a modern tool for celiac disease diagnosis - case presentation

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ABSTRACT

Celiac disease is a clinically heterogeneous disease characterized by an inadequate immunological response when patients with specific genetic phenotypes are exposed to gluten. This article presents a case of a young woman diagnosed in Gastroenterology Department of “St. Andrew Apostle” Emergency Hospital of Constanta with celiac disease after multiple admissions into the hospital for unspecific symptoms such as pallor, fatigue, pirois, weight loss and 1-2 soft stools/day. The history with period irregularities and infertility without a known cause, a recent unexplained bone fracture, the muscle weakness, neuropsychiatric symptoms characterized by sleep disturbances and irritability correlated with the biological features characterized by moderate feriprive anemia, Ca and Mg decreased level, thyroid autoimmune impairment and gastrointestinal symptoms raised the suspicion of an autoimmune disorder with multiple targets. The videocapsule endoscopy (VCE) revealed the specific pattern of the celiac disease: villous atrophy of jejunum, scalloping, absent folds and cobblestone mucosal pattern. Results were correlated with immunology tests results. The patient was transferred on a gluten free diet and the clinical and VCE controls

revealed the healing of the jejunum mucosa. The VCE can be the tool for positive diagnosis of an unusual and heterogeneous celiac disease in patients with various symptoms without an apparent cause.

Keywords: celiac disease, unspecific symptoms, diagnosis, videocapsule endoscopy

Introduction

Celiac disease is a chronic inflammatory condition of the small bowel with a not completely known basis. Genetic and environmental factors play a significant role in the disease etiology. It involves a reaction to gluten, found in wheat, barley and rye. A person with the appropriate genetic phenotype (HLA-DQ2 and HLA-DQ8 mutations) generates an immunological response when exposed to gluten, which results in characteristic microscopic features witnessed on biopsy specimens taken from the mucosa of the small intestine. Physicians consider that celiac disease is a “clinical chameleon” — it can manifest with any of more than 100 different symptoms, or it can be totally asymptomatic, the so called “silent celiac disease” The most common aspect of the disease, despite the heterogeneous symptomatic background, are the diagnosis, using

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celiac blood tests and endoscopy, and the treatment, including the gluten-free diet.

Aim

We aimed to present an uncommon case of a woman of 33 years old, who was admitted in the Gastroenterology Department of St Apostle Andrew Emergency Hospital of Constanta County, with unspecific symptoms like: pallor, fatigue, pirosis, some weight loss (7kg in 6 months), and a recent change in her bowel habit, the patient complaining of 1-2 soft stools per day, for no more than 3 months.

Method

We evaluated the medical history of the patient, the clinical aspect, the lab findings and upper GI endoscopy and colonoscopy features. As a final endoscopic investigation, the small bowel video-capsule endoscopy was performed. It was the diagnostic tool who traced the final diagnosis in a patient with heterogeneous, unclassifiable symptoms.

Results

From her medical history we could notice only a recent unexplained fracture of the right arm, two months prior to the present episode, a history of period irregularities and infertility with an unknown cause. She was never hospitalized for any medical condition.

The clinical aspect was of a young pale woman,

with a BMI < 20kg/m², tachycardia (HR= 99/min), slightly protruded eyes, with muscle weakness. Asked about the heat tolerance, she reported heat sensitivity. She also complained of difficulty in sleeping and sudden irritability.

The pathological laboratory findings at the moment of admission in the hospital were as follows: Hb= 9.2g/dl, Ht= 29%, sideremia↓, ferritin ↓, CTLF↑, Ca↓, Mg↓, FT4 ↑, TSH↓;

The diagnosis of hyperthyroidism was made and the patient was referred to the Endocrinology Department for appropriate treatment. Tests for autoimmunity were done and the results of TRAb and TPOAb were positive.

Still, not all symptoms were explained by the endocrine disease. Correlating the autoimmune hyperthyroidism with the patient GI symptoms, we agreed to refer her to the endoscopy examination suspecting an autoimmune disease with more targets.

The upper GI endoscopy didn't reveal any pathological feature and the colonoscopy was also normal. Biopsies from duodenum and colon were negative for a specific disease.

It was suspected a GI autoimmune disease causing malabsorption. The stool calprotectin value was between ranges and we could exclude along with the endoscopic investigation, a certain IBD with an atypical onset.

Knowing from literature that the prevalence of celiac disease is significantly greater in the thyroid disease patients than in general population, we decided to make the small bowel examination using videocapsule endoscopy.

The videocapsule endoscopy (VCE) revealed mucosal changes suggestive of celiac disease, as follows: villous atrophy in jejunum, including scalloping, absent folds and mosaic and cobblestone mucosal pattern (Figure 1).

Figure 1 - Villous atrophy in jejunum.

Figure 2 - Normal appearance of the jejunum.

The diagnosis of celiac disease was setup by VCE, with serology confirmation, the patient being positive for tTG (anti-tissue transglutaminase)-IgA and total serum IgA.

The patient was referred to osteodensitometry exploration (DXA) and the result showed a bone mineral density (BMD) below -2.5 measured with central DXA.

The patient was recommended to keep a gluten-free diet. Referring to “gluten-free diet” it meant to eliminate from the diet all sources of gluten, this making part of wheat, barley and rye (and sometimes oats).

The patient was reexamined after six months of diet restriction to gluten. It was noted a positive clinical evolution and an amelioration of the laboratory findings. VCE examination at the three months interval revealed a proper healing of the jejunum mucosa (Figure 2)

Discussions

The celiac disease is one of the most heterogeneous GI illnesses. The unspecific onset makes the diagnosis sometimes very difficult. In our case, the onset of the disease was unspecific, with general symptoms, our patient complaining of fatigability, pirosis, weight loss, soft stools, sleep disturbances, irritability, heat sensitivity, a recent arm fracture and infertility. These symptoms unspecific for celiac disease, all together, could orientate the physician for a proper diagnosis. It is known that 40% of women reported anemia prior to their celiac diagnoses. It's a common symptom, who could make physicians to test woman patients with unexplained anemia for celiac disease [1]. Although the disease affects the small bowel, only 40% of the women reported diarrhea as one of their first celiac symptoms, and gastroesophageal reflux disease (GERD) often is found alongside these digestive symptoms.

Our patient history was very interesting. Undiagnosed celiac disease significantly raises the risk of osteoporosis [2]. Again, nutritional deficiencies related to problems absorbing nutrients like vitamin D, calcium and magnesium — are the cause. The benefit of osteodensitometry assessment in persons with a history of unexplained fractures can be crucial in the patient evolution. The use of osteodensitometry in patients with celiac disease is already included in the benefits catalogue of the German statutory health insurance [3]. When the celiac disease diagnosis was fixed, the unexplained arm fracture cause was revealed. The history of infertility was also a tool to investigate in our patient. Even if celiac disease appears in about 1% of the overall population, researchers have found it in up to 8% in women with so-called “unexplained infertility,” or infertility that’s not due to the hormonal problems. This could be explained by the nutritional deficiencies, which are common in newly diagnosed celiacs, even those who don’t have gastrointestinal symptoms. Infertility also could result somehow from inflammation. Researchers haven’t yet pointed the exact cause [4]. Regarding the thyroid involvement, our patient presented a particular association of the thyroid autoimmune disorder and the celiac disease. Only 7% of people with autoimmune thyroid disease - including Graves’ disease and Hashimoto’s disease may have celiac disease, and in some of them, the celiac won’t cause any other symptoms. Up to 10% of people with celiac disease have an autoimmune thyroid condition, a far higher rate than in the general population. Meanwhile, between 1.5% and 6.7% of people with autoimmune thyroid disorder also have celiac disease [5,6].

Traditionally, the celiac disease is diagnosed by biopsy of the duodenum in individuals with positive serological tests. Negative biopsies may suggest a latent celiac disease. Our particular case demonstrates that VCE can be used to diagnose celiac disease in patients with suspected celiac disease having a non-diagnostic EGD. Although VCE is used to diagnose celiac disease in clinical practice, previous guidelines have not recommended this approach and our study support its role in diagnosing celiac disease. It is unclear what proportion of patients that have positive celiac disease serology, but a normal EGD (these patients may be classified as having latent celiac

disease and are usually not advised to commence a gluten-free diet), will have a VCE that is diagnostic for celiac disease. One small series did not find celiac disease in this situation [7], but there is growing evidence that celiac disease may sometimes vary in distribution, appearing more distally and patchy in nature [8,9]. However, before embarking on additional testing with VCE, it is important to first confirm the adequacy of the initial diagnostic work up, starting from a meticulous endoscopic evaluation, to taking a sufficient number biopsies, and finally having an experienced gastrointestinal pathologist interpret the pathology [10]. Still, physicians sustain that celiac disease is a growing indication for VCE. The suspicion of celiac disease should be accurately diagnosed with VCE, saving precious time till final diagnosis and more unnecessary lab test to prove the malabsorption.

Conclusions

In patients with heterogeneous symptoms and unusual onset of the celiac disease, the presence of villous atrophy was confirmed by the VCE appearance of the duodenal or jejunal mucosa in a woman patient. Larger and controlled trials are needed to confirm the accuracy of VCE as compared to EGD with biopsy in diagnosing the celiac disease. Further studies documenting the cost effectiveness of VCE as compared to EGD need to be performed.

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