Cystic fibrosis and coeliac disease: a case report of an unusual association

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Abstract

Coeliac disease (CD) and cystic fibrosis (CF) are well known as the most common causes of chronic intestinal malabsorption in childhood. The coexistence of coeliac disease with cystic fibrosis is uncommon. Here, we describe the case of cystic fibrosis in a patient diagnosed with coeliac disease who failed to respond clinically to a gluten-free diet and had persistent steatorrhea and failure to thrive.

BACKGROUND

Coeliac disease (CD) is a malabsorption syndrome caused by gluten-induced small intestinal damage [1]. The signs and symptoms of CD, such as malabsorption, diarrhea, steatorrhea, and growth failure are nonspecific and may be difficult to distinguish from common symptoms of cystic fibrosis (CF) [2]. In 1938, Dorothy Anderson reported a syndrome which believed to be CD but described a distinct and separate pathology of CF [3].

The coexistent disease should be kept in mind when the diagnosis is not clear or the patient is not improving on appropriate therapy.

CASE REPORT

A 4-year-old Syrian male presented to the pediatric gastroenterology clinic with steatorrhea, failure to thrive, loss of appetite, and recurrent abdominal pain. Since early infancy, he suffered from feeding intolerance, diarrhea; he was passing stool up to 5 times/day, the stool was described as oily, bulky, and foul-smelling. No bilious emesis. He was on breast and bottle-feeding. By the age of 1 year, the family started seeking medical advice. Investigations were performed which showed microcytic hypochromic anemia with fat globules in stool microscopy. The patient was treated with multivitamins without any improvement. He was a full-term infant delivered by spontaneous vaginal delivery. The birth weight was 3.5 kg.

There is no significant familial history of genetic diseases or previous hospital admissions.

On physical examination, his body weight was 12.5 kg (−3.2 SD), the length was 95 cm (−2.5 SD). Vital signs were stable and he was generally well.

Laboratory analysis suggested iron deficiency anemia, with hemoglobin level (Hb) 8.9 g/L, Mean Corpuscular Volume (MCV) 68 fl, ferritin 17.4 ng/mL, serum iron 52.1 mcg/dL. Stool microscopy revealed fat globules.

Other laboratory tests that involved hepatic function, renal function, electrolytes, blood glucose, urine examination, thyroid-
stimulating Hormone (TSH), Insulin-Like Growth Factor-1 (IGF-1), D-xylene, serological immunoglobulin were within the normal range. Anti-gliadin antibodies IgA, IgG (AGA IgA, IgG), Anti-tissue transglutaminase Antibodies IgA (anti-TTG IgA) were markedly elevated (more than 400 U/mL). Esophagogastroduodenoscopy (EGD) was performed that showed normal findings. The duodenal biopsies showed nonspecific changes as mild focal shorting of intestinal villi and elongation of crypts with increased intraepithelial lymphocytes range (30 lymphocytes/100 epithelial cells). That could not exclude CD (marsh 2).

Based on the clinical presentation and hematologic findings, CD was suspected. The patient was started on a gluten-free diet, vitamins, and iron supplements for 3 months with improvement in appetite and weight, and continued steatorrhea. Therefore, elastase in stool was performed which was markedly reduced. On genetic testing, he was homozygous (9 T/9 T) for the F508del (-CTT) mutation. The diagnosis of CD with CF was made, and pancreatic enzyme replacement therapy (PERT) was initiated, at a dose of 1000 U/Kg/per day offered with ursodeoxycholic acid, multivitamins, folic acid, gluten-free diet, and high-calorie rates of 230% for his age. After about 5 months of treatment, his general situation was good with remarkable clinical improvement, normal stool, and weight gain of 4 kg.

We followed the patient for 6 years. During these years, the patient was re-referred to the clinic with abdominal pain and weight loss due to a gluten diet for 2 months. Anti-TTG IgA was elevated (more than 400 U/mL). The gluten-free diet was restored. Currently, the child is in good condition with a normal growth rate.

DISCUSSION
The occurrence of the two diseases in the same patient was first described in 1969 by Hide and Burman [4]. The prevalence of both CD and CF is 1 in 2,000,000–1:5,900,000 [5], around 30 cases described in 1969 by Hide and Burman [4]. The prevalence of both was established with no improvement in treatment. Persistent diarrhea, reduced elastase in stool, and the mutation on genetic testing proposed the co-diagnosis with CF.

CONFLICT OF INTEREST STATEMENT
None declared.

FUNDING
No funding was obtained for this study.

ETHICS APPROVAL AND CONSENT TO PARTICIPATE
This case report did not require review by the Ethics Committee Tishreen University Hospital, Latakia, Syria.

CONSENT FOR PUBLICATION
Written informed consent was obtained from the patient’s parents for publication of this case report and any accompanying images. A copy of the written consent is available for review by the editor.

GUARANTORS
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