Diagnosis of Cystic Fibrosis and Celiac Disease in an Adult: One Patient, Two Diseases, and Three Reminders

Israel Rabinowitz MD

This case report describes the uncommon occurrence of celiac disease and cystic fibrosis in an obese adult patient. Apart from its rarity, the case serves to highlight the elusive nature of these 2 diseases when presenting with atypical clinical features in an adult. Key words: celiac disease, cystic fibrosis, obese.

Introduction

Celiac disease is an immune-mediated enteropathy triggered by dietary gluten (present chiefly in wheat, barley, and rye) in genetically susceptible individuals. Its prevalence is approximately 0.4% in the United States, Europe, and North Africa. Iron deficiency anemia is often a presenting sign. A positive test for anti-endomysial antibody carries a sensitivity of 85–98% and a specificity of 97–100% for celiac disease.

Case Summary

A 42-year-old man had been diagnosed with iron deficiency anemia (hemoglobin 9.2 g/dL, mean corpuscular volume 62 femtoliters, red cell distribution width 18.2%; ferritin 8 ng/mL) in another clinic, after which a test for occult blood, a gastrointestinal endoscopy, and a colonoscopy were performed, all with normal findings. He received iron supplementation and his hemoglobin improved.

In addition he described a recurring productive cough and frequent bulky stools, both of several years duration.

The patient was a land surveyor, married, and father to 7-year-old twins. He did not smoke or consume alcohol and denied any chronic illnesses.

His height and weight were 1.82 m and 112 kg, respectively (body-mass index 33.8 kg/m²), and he had marked abdominal obesity, with a waist circumference of 137 cm. His blood pressure was 140/85 mm Hg, with a heart rate of 76 beats/min and a respiratory rate of 16 breaths/min.

A post-nasal drip was noticed. The physical examination was otherwise unremarkable. A radiograph performed 3 months earlier revealed bilateral maxillary-sinus opacities.

Levels of glucose, creatinine, urea, serum electrolytes, liver enzymes, thyroid-stimulating hormone, and vitamin B₁₂ were all in the normal range. The folic acid level was low (3.1 nmol/L), and so was the total cholesterol (134 mg/dL).

The iron deficiency and low folic acid and cholesterol levels suggested intestinal malabsorption, and a test for anti-endomysial antibody (immunoglobin A type) was ordered. The result was positive, at a titer of 1:80; a jejunal biopsy confirmed the diagnosis of celiac disease. A gluten-free diet was instituted.

In order to determine the duration of the patient’s anemia, he was asked to bring in any past medical documentation he possessed. He brought in an 8-year-old file from a fertility clinic, the study of which revealed 3 points of interest. The first was a result of a complete blood count (hemoglobin 12.2 g/dL, mean corpuscular volume 73 femtoliters, red cell distribution width 17%) compatible with longstanding iron deficiency. The second was a diagnosis of obstructive azoospermia, which led to surgical retrieval of spermatozoa, successful in vitro fertilization, and the birth of twins. The third was the result of the clinic’s routine screening for cystic fibrosis, which was positive for cystic fibrosis transmembrane conductance regulator mutations W1282X and the ribonucleic acid splicing variant IVS8–5T/5T (5T allele).

The patient had now been on a strict gluten-free diet for 3 months. His hemoglobin was 13.3 g/dL and he felt stronger, but his weight increased dramatically, by 16 kg, be-
cause of the increased intestinal absorption, and he re-
ported symptoms consistent with obstructive sleep apnea.
His productive cough continued. A chest radiograph was
interpreted as normal and a sinus radiograph revealed bi-
lateral maxillary sinusitis.

The presence of azoospermia and chronic sinusitis in a
patient with cystic fibrosis mutations led to the perfor-
manence of a sweat test. The result was highly positive, with
a sodium chloride concentration of 86 mEq/L and 83 mEq/L
on 2 separate occasions. On spirometry, his forced expi-
ratory volume in the first second was 51% of predicted,
with a 17% increase after β agonist inhalation. A sputum
culture yielded normal flora.

The level of pancreatic elastase in a stool specimen was
473 μg/g of stool (normal > 200 μg/g), ruling out pan-
creatic insufficiency.

On the basis of sino-pulmonary disease and obstructive
azoospermia in a patient with transmembrane conductance
regulator mutations and a highly positive sweat test, a
diagnosis of cystic fibrosis was made.

Discussion

Obesity of the degree reported in this patient is a rare
finding in celiac disease patients. Diagnosis of cystic fi-
brosis in adults, although much more uncommon than ce-
liac disease, is increasingly being made, and adult disease
is usually milder than the pediatric form. The Rosenstein
diagnostic criteria for cystic fibrosis (persistently elevated
concentrations of electrolytes in sweat plus characteristic
clinical findings, which include typical gastrointestinal or
pulmonary disease and perhaps obstructive azoospermia
or a family history) were fulfilled in this patient. The
combination of the 2 diseases is indeed a rarity, with only
15 cases reported in the literature and none in an obese
adult.

Apart from its uniqueness, the case offers 3 reminders.

The first is that cystic fibrosis presents atypically in
adults and is probably under-diagnosed. The 5T allele, for
example, carries a variable phenotype, including asthma-
like symptoms and chronic sinusitis. Therefore, a sweat

The second, always to be kept in mind, is that curing
one disease can exacerbate another. In this case the gluten-
diet, through its positive effect on intestinal absorp-
tion, led to marked weight gain and symptoms of obstruc-
tive sleep apnea in a patient with limited respiratory reserve.
Thus, a holistic view of the patient’s health status should
always be maintained.

The third is the role of the primary physician as inte-
grator of data derived from specialist settings and their
interpretation in the context of the patient’s clinical course.
In an era of highly specialized medicine, this role is more
important than ever.

The patient is currently feeling better and is under joint
care of a cystic fibrosis clinic, a dietitian, and his family
physician.

REFERENCES

with untreated celiac disease. J Pediatr Gastroenterol Nutr 2001;
32(2):226.
3. Rosenstein BJ. Interpreting sweat tests in the diagnosis of CF. J
Coexistence of cystic fibrosis and celiac disease. Description of a
clinical case and review of the literature. Pediatr Med Chir 1999;21(5
5. Kerem E, Rave-Harel N, Augarten A, Madgar I, Nissim-Rafinia M,
Yahav Y, et al. A cystic fibrosis transmembrane conductance regu-
lator splice variant with partial penetrance associated with variable
cystic fibrosis presentations. Am J Respir Crit Care Med 1997;155(6):
1914–1920.