Recognizing a common disease hiding in plain sight

It is a dictum of diagnostic medicine that we see unusual presentations of common diseases more often than we see common presentations of rare diseases. Yet even when patients come in with a relatively common systemic disorder, we sometimes don't make a prompt diagnosis. There are many reasons for this.

In my subspecialty, I find that polymyalgia rheumatica often goes unrecognized early in its course despite a fairly characteristic presentation once it "settles in." I believe this happens, in part, because clinicians don't realize how often this entity occurs in older patients.

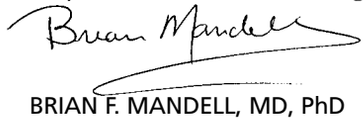
On page 209 in this issue, Harrison et al discuss celiac disease, an even more common yet often unrecognized entity. Celiac disease is estimated to occur in 1 of every 100 people in the United States, with lower but still significant prevalences in other countries with different genetic compositions. The disease is particularly intriguing because it is an extrinsically triggered, immune-mediated disorder that is treated by dietary adjustment, not by corticosteroids. We learned about celiac disease in medical school, perhaps as nontropical sprue. In my school it was lumped with other gastrointestinal disorders, and thus has been compartmentalized in my mind with the other chronic diarrheal illnesses.

But as Harrison et al remind us, diarrhea need not be the major manifestation of celiac disease. Whereas we may miss polymyalgia rheumatica because we believe it to be rare, we often miss celiac disease because we don't realize how *common* it is, and because we fail to recognize its many manifestations other than diarrhea.

Because celiac disease is associated with autoimmunity and nutritional deficiency, its manifestations are truly myriad, and they can also be subtle and confusing. It often hides among bone pain, premature osteoporosis, fatigue, patchy pruritis, weight loss, and an anemia that we ultimately (but not initially) recognize as due to iron deficiency. Diarrhea—the manifestation that I most readily recall from medical school—is quite often absent.

The clinical picture is often misleading. For example, alopecia with some combination of rash, anemia, pains, and fatigue in a young woman frequently prompts a search for lupus, and an antinuclear antibody titer is obtained. Alternatively, the fatigue, pains, dysesthesias, and ill-defined bowel symptoms prompt the diagnosis of fibromyalgia with irritable bowel syndrome. In adolescent women, celiac disease may readily be confused with a primary eating disorder.

The fact that celiac disease is associated with diabetes, thyroid disease, other autoimmune disorders, and infertility virtually guarantees that we will all see patients with this disease. Hopefully, after reading this article, I will be more adroit at recognizing the celiac patient in the midst of others with similar comorbidities. And I may be able to do something about it.



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