

An Unusual Presentation of Myopathy in Celiac Disease

by John R. Stroehlein, Martin Poliak, Huamin Wang

Although celiac disease in adults is more often manifest by non-gastrointestinal pathology or nutritional deficiency, indeterminate gastrointestinal symptoms combined with hepatopathy and myopathy can be an indication of celiac disease despite a normal anti TTG and serum IgA. This case illustrates a.) the importance of small bowel biopsy to render a diagnosis of celiac disease and b.) the potential for myopathy to occur without overt myositis, obvious nutritional deficiencies or metabolic bone disease. Celiac disease should be considered and pursued as a diagnostic possibility when the cause of weight loss and myopathy is indeterminate.

Although rare, the development of combined myopathy and hepatopathy from celiac disease is of clinical importance and requires consideration when gastrointestinal symptoms are associated a combination of markedly elevated creatine kinase and loss of muscle mass. This is illustrated by the following case which calls attention to the possibility of celiac disease as the basis for myopathy and hepatopathy.

A 66 year-old Caucasian female with a history of breast cancer developed acute onset of diarrhea beginning in February 2011. Her history included stress related irritable bowel symptoms, not necessarily with diarrhea, and reflux managed with a proton pump inhibitor. Recent upper and lower endoscopy were reported to be normal. She was empirically treated with ciprofloxacin and metronidazole by her primary physician. Her diarrhea improved but did not resolve and was followed by myalgia, fatigue, noticeable muscle loss without muscle tenderness and general weakness without electrolyte abnormalities. Subsequent treatment with a yogurt based probiotic, hyoscyamine and rifaximin were unsuccessful. Notable physical exam findings were limited to loss of muscle mass.

She was referred for evaluation 6 months later; at that time, stool evaluation (*C. difficile*, ova and parasites and culture) was negative but lactoferrin was positive. Colonoscopy with biopsies performed elsewhere in October 2011 was negative for microscopic colitis. Her

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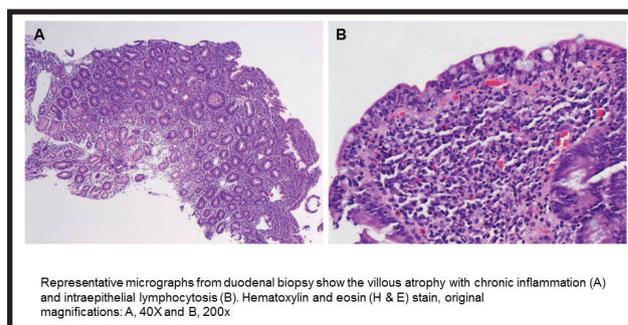


Figure 1.

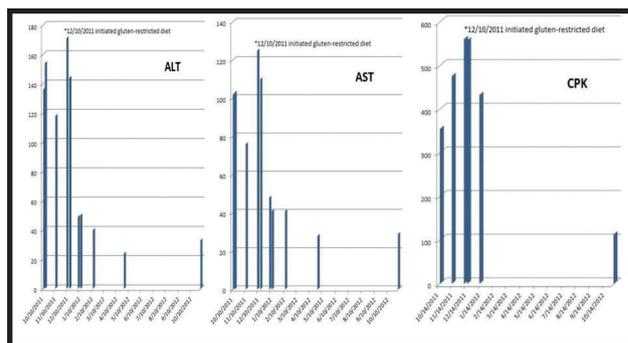


Figure 2.

erythrocyte sedimentation rate and C-reactive protein were normal, total serum IgA was 136 mg/dl (normal) and the anti-tissue transglutaminase was less than 1.2 (normal). Laboratory studies initially revealed a normal CBC, glucose, creatinine, electrolytes, magnesium, albumin (4.3 gm/dl), liver enzymes, folate, B12, iron, TIBC and vitamin D. Serum carotene was 11 mcg/dl. The ANA was 1:160 and DNA double strand antibody was normal.

Between October and December 2011, her alanine aminotransferase ranged between 136 and 171 IU/L and aspartate aminotransferase from 102 to 125 IU/L

with consistently normal alkaline phosphatase and bilirubin. A normal gamma glutamyl transpeptidase (GGT) and negative viral serology prompted evaluation of the creatine kinase which proved to be 355 U/L and rose to 561 U/L as of December. Duodenal biopsies obtained during esophagogastroduodenoscopy (EGD) revealed chronic inflammatory change, villous atrophy and intraepithelial lymphocytosis, suggestive of celiac disease (Figure 1A & 1B). A gluten free diet was initiated and was followed by prompt normalization of all biochemical abnormalities with subsequent 35 pound weight gain and restoration of muscle mass. All of the patient's symptoms resolved and have remained resolved, and the liver enzymes and creatine kinase have remained normal (Figure 2).

This index case exemplifies the possibility for myopathy to occur as a consequence of celiac disease. The possibility of myopathy was initially considered when the GGT was normal despite significant elevation of the ALT and AST. Additionally, duodenal biopsies are often necessary to establish the diagnosis of celiac disease in the absence of hypokalemia or other electrolyte abnormalities.¹ Prior case reports of celiac-induced myopathy have been reported in children.²⁻³ Celiac disease associated myopathy may be more commonly encountered alongside other manifestations of gluten-sensitive enteropathy including metabolic bone disease⁴⁻⁶ and polyneuropathy⁷ or, even more rare, myelopathy. There has been interest in the association of myotonic dystrophy and celiac disease although the response to gluten restriction is not predictable. Our case was not characterized by the aforementioned associations nor was there evidence of an inflammatory myopathy, which is often associated with higher levels of creatine kinase and elevation of the sedimentation rate. Although response to gluten restriction is not assured, the mere fact that there is no specific treatment for otherwise idiopathic inflammatory myopathy renders the association with celiac disease to be of more than passing interest.⁸

The association between celiac disease and neuromuscular disorders is not well defined; however, it is clear that celiac disease can manifest primarily if not exclusively with neuromuscular pathology. Myopathy is the third most common neuromuscular disorder associated with sprue and second only to ataxia and peripheral neuropathy.⁹ In addition to a clinical association between celiac disease and neuromuscular manifestation is the presence of IgA deposits against

transglutaminase 2 observed in muscle in a patient with celiac disease.¹⁰ Interestingly, the antigen that endomysial antibodies recognize is tissue transglutaminase type 2. It is important to recognize the possibility that celiac disease can present with extra intestinal manifestations including unexplained myopathy. ■

References

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Answers to this month's crossword puzzle:

1	S	T	E	A	T	O	S	I	S	5	V	E	6	G	F				
	W		N		E		A				I		7	U		A			
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