

Amyloidosis Diagnosed by the GI Doctor

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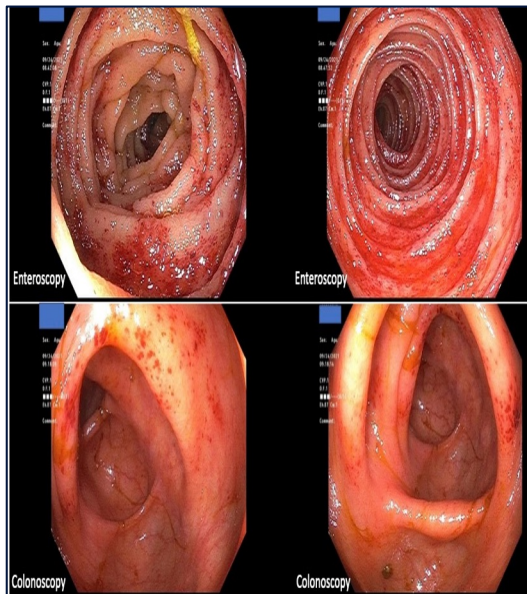
Introduction

Amyloid light-chain (AL) amyloidosis is the most common form of amyloidosis and is the main type to involve the gastrointestinal tract. Monoclonal Gammopathy of Undetermined Significance (MGUS) is a Plasma Cell Dyscrasia (PCD) with potential to precipitate amyloidosis. Our patient presented with a constellation of signs and symptoms that lead to his diagnosis.

Case Description

A 59-year-old man with a history of Hemoglobin S-C presented with recurrent admissions to our hospital for new-onset orthostasis, weight loss, and dyspnea progressing over a 9 month period. Physical exam was unimpressive at the first presentation. Labs were significant for a NT-Pro BNP of 17,100 pg/ml. Echocardiography and cardiac MRI showed a restrictive filling pattern with preserved ejection fraction, however a specific etiology was not identified. Serum immunofixation revealed monoclonal IgA light chain proliferation. Bone marrow biopsy confirmed MGUS. The patient later developed epigastric pain with associated chronic watery diarrhea, peripheral edema, and hypoalbuminemia. A CT demonstrated colonic wall thickening with trace abdominal ascites. Enteroscopy revealed severe inflammation of the duodenum and jejunum characterized by edema, friability, scalloping, and diffuse petechiae. Colonoscopy showed similar but patchy involvement throughout. Histology was initially inconclusive but our suspicion remained high so slides were sent to a GI specialty lab. Pathology ultimately demonstrated immunoglobulin light chain lambda amyloidosis. He developed nephrosis as well. A work-up for autoimmune, infectious, and malignant causes was negative. The patient was ultimately transferred to a nearby hospital with specialty care services.

Images



Description of images in clockwise fashion beginning with the top left image: 1. Patchy inflammation of the duodenum, 2. Diffuse inflammation of the jejunum, 3 and 4. Patchy inflammation of the colon

Discussion

AL amyloidosis develops as a result of monoclonal light chain deposition in various tissues. The cardiac and renal systems are most commonly involved. The main causes include PCDs and autoimmune illnesses. The diagnosis is often delayed. It should be suspected in a patient who presents with restrictive cardiomyopathy or otherwise unexplained heart failure. Our patient's illness manifested as restrictive cardiomyopathy and likely protein losing enteropathy. MGUS can precede amyloidosis by about 12 years so this was an important finding. Initial GI biopsies were inconclusive but ultimately cinched the diagnosis, highlighting the importance of remaining skeptical. Treatment is aimed at the underlying cause.

References

1. Dahiya, D. S., Kichloo, A., Singh, J., Albosta, M., & Wani, F. (2021). Gastrointestinal amyloidosis: A focused review. *World journal of gastrointestinal endoscopy*, 13(1), 1–12. <https://doi.org/10.4253/wjge.v13.i1.1>
2. Engelberg, Rachel MD1; Niec, Rachel MD, PhD1; Wan, David MD2 Gastrointestinal Amyloidosis Presenting as Hyponatremia, *American Journal of Gastroenterology*: October 2018 - Volume 113 - Issue - p S1395-S1396
3. Rowe, K., Pankow, J., Nehme, F., & Salyers, W. (2017). Gastrointestinal Amyloidosis: Review of the Literature. *Cureus*, 9(5), e1228. <https://doi.org/10.7759/cureus.1228>

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