CASE IN POINT

Gastrointestinal Bleeding Caused by Large Intestine Amyloidosis

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Background: Amyloidosis is a rare disorder caused by abnormal folding of proteins, leading to the dysfunction of normal tissues. Amyloid deposition can affect several organs, but deposition in the large intestine is rare.

Case Presentation: A 79-year-old man presented with gastrointestinal bleeding and nonspecific symptoms of weight loss, dry heaves, dysphagia, and weakness. The patient underwent esophagogastroduodenoscopy and colonoscopy and a biopsy confirmed the diagnosis of intestinal amyloidosis.

Conclusions: This case report highlights the importance of a strong differential when working up gastrointestinal bleeding that includes amyloidosis. Early identification and multidisciplinary involvement are crucial for management and tailored care to each patient's needs.

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Gastrointestinal (GI) bleeding is a common cause of hospital admissions. The yearly incidence of upper GI bleeding is 80 to 150/100,000 people and lower GI bleeding is 87/100,000 people.[1,2] The differential tends to initially be broad but narrows with good history followed by endoscopic findings. Getting an appropriate history can be difficult at times, which leads health care practitioners to rely more on interventional results.

Amyloidosis is a rare disorder of abnormal protein folding, leading to the deposition of insoluble fibrils that disrupt normal tissues and cause disease.[3] There are 2 main types of amyloidosis, systemic and transthyretin, and 4 subtypes. Systemic amyloidosis includes amyloid light-chain (AL) deposition, caused by plasma cell dyscrasia, and amyloid A (AA) protein deposition, caused by systemic autoimmune illness or infections. Transthyretin amyloidosis is caused by changes and deposition of the transthyretin protein consisting of either unstable, mutant protein or wild type protein. Biopsy-proven amyloidosis of the GI tract is rare.[4] About 60% of patients with AA amyloidosis and 8% with AL amyloidosis have GI involvement.[5]

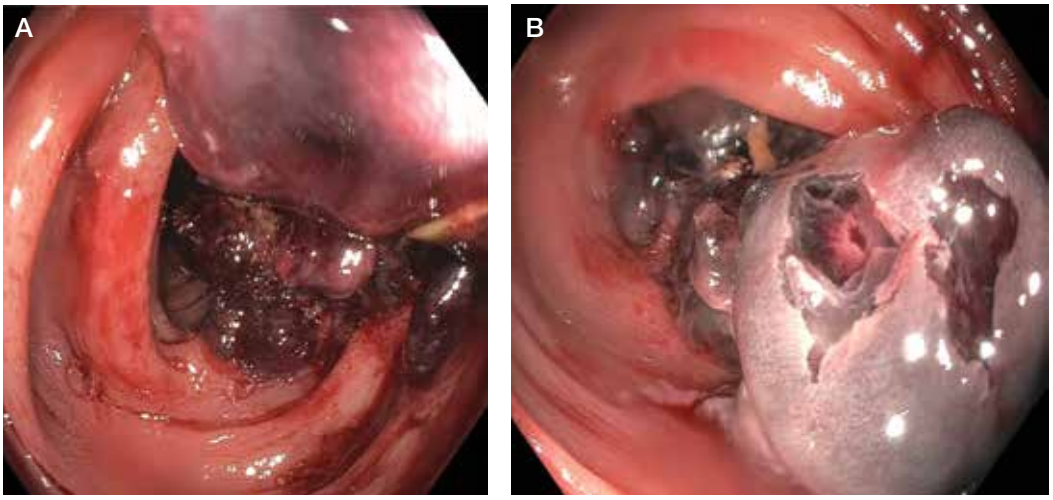
We present a case of nonspecific symptoms that ultimately lined up perfectly with the official histologic confirmation of intestinal amyloidosis.

CASE PRESENTATION

A 79-year-old man with a history of type 2 diabetes mellitus, congestive heart failure, hyperlipidemia, obstructive sleep apnea, hypothyroidism, hypertension, coronary artery disease status postcoronary artery bypass grafting, and stent placements presented for 3 episodes of large, bright red bowel movements. He reported past bleeding and straining with stools, but bleeding of this amount had not been noted prior. He also reported dry heaves, lower abdominal pain, constipation with straining, early satiety with dysphagia, weakness, and decreased appetite. Lastly, he mentioned intentionally losing about 35 to 40 pounds in the past 3 to 4 months and over the past several months increased abdominal distention. However, he stated he had no history of alcohol misuse, liver or intestinal disease, cirrhosis, or other autoimmune diseases. His most recent colonoscopy was more than a decade prior and showed no acute process. The patient never had an esophagogastroduodenoscopy (EGD).

On initial presentation, the patient's vital signs showed no acute findings. His physical examination noted a chronically ill–appearing male with decreased breath sounds to the bases bilaterally and noted abdominal distention with mild generalized tenderness. Laboratory findings were significant for a hemoglobin level, 9.4 g/dL (reference range, 11.6-15.3); iron, 23 ug/dL (reference range, 45-160); transferrin saturation, 8% (reference range, 15-50); ferritin level, 80 ng/mL (reference range, 30-300); and carcinoembryonic antigen level, 1.5 ng/mL (reference range, 0-2.9). Aspartate aminotransferase level was 54 IU/L (reference range, 0-40); alanine transaminase,

FIGURE 1 Clot and Mass Seen on Colonoscopy



Bulky clot-like masses can be seen protruding through the mucosa with patchy colitis and ulceration. A, Image taken approaching the mass. B, Largest mass areas and ulceration within it can be seen.

24 IU/L (reference range, 7-52); albumin, 2.7 g/dL (reference range, 3.4-5.7); international normalized ratio, 1.3 (reference range, 0-1.1); creatinine, 1.74 mg/dL (reference range, 0.44-1.27); alkaline phosphatase, 369 IU/L (reference range, 39-117). White blood cell count was 15.5 × 10^9/L (reference range, 3.5-10.3), and lactic acid was 2.5 mmol/L (reference range, 0.5-2.2). He was started on piperacillin/tazobactam in the emergency department and transitioned to ciprofloxacin and metronidazole for presumed intra-abdominal infection. Paracentesis showed a serum ascites albumin gradient of > 1.1 g/dL with no signs of spontaneous bacterial peritonitis. Computed tomography of the abdomen and pelvis with contrast was suspicious for colitis involving the proximal colon, and colonic mass could not be excluded. Also noted was hepatosplenomegaly with abdominopelvic ascites.

Based on these findings, an EGD and colonoscopy were done. The EGD showed mild portal hypertensive gastropathy. The coloscopy showed patchy colitis in the cecum, ascending colon, and transverse colon with a mass vs clot adherent to the mucosa and areas of ulceration next to the masslike structures with oozing (Figure 1).

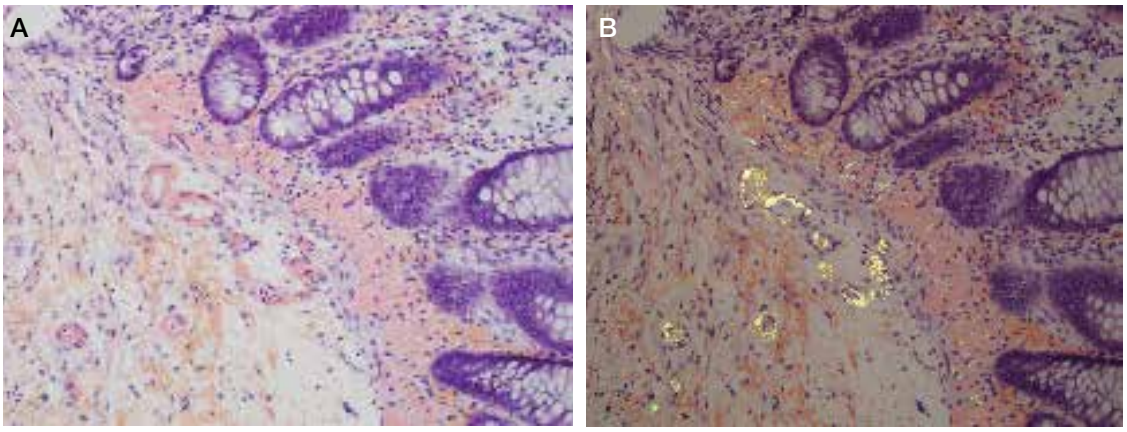
After the biopsy results, the patient was officially diagnosed with intestinal amyloidosis (Figure 2). His hemoglobin level stabilized, he was to complete his antibiotic treatment outpatient, and there were plans to follow up with gastroenterology, hematology/oncology, nephrology, and his primary care physician for further management.

He returned to the gastroenterology clinic 2 months later. At that point, he had worsening symptoms, liver function test results, and international normalized ratio. He was admitted for further investigation. A bone biopsy was done to confirm the histology and define the underlying disorder. The biopsy returned showing Waldenstrom macroglobulinemia, and he was started on bortezomib. Unfortunately, his clinical status rapidly worsened, leading to acute renal and hepatic failure and the development of encephalopathy. He eventually died under palliative care services.

DISCUSSION

Amyloidosis is a rare disorder of abnormal protein folding, leading to the deposition of insoluble fibrils that disrupt normal tissues and cause disease.[3] There are several variations of amyloid, but the most common type is AL amyloidosis, which affects several organs, including the heart, kidney, liver, nervous system, and GI tract. When AL amyloidosis involves the liver, the median survival time is about 8.5 months.[6] There are different ways to diagnose the disease, but a tissue biopsy and Congo Red staining can confirm specific organ involvement as seen in our case.

FIGURE 2 Biopsy of Masslike Structures



A, Before Congo Red staining. B, With Congo Red staining (original magnification × 200).

This case adds another layer to our constantly expanding differential as health care practitioners and proves that atypical patient presentations may not be atypical after all. GI amyloidosis tends to present similarly to our patient with bleeding, malabsorption, dysmotility, and protein-losing gastroenteropathy as ascites, edema, pericardial effusions, and laboratory evidence of hypoalbuminemia.[7] Because amyloidosis is a systemic illness, early recognition is important as intestinal complications tend to present as symptoms, but mortality is more often caused by renal failure, cardiomyopathy, or ischemic heart disease, making early multispecialty involvement very important.[8]

CONCLUSIONS

Health care practitioners in all specialties should be aware of and include intestinal amyloidosis in their differential diagnosis when working up GI bleeds with the hope of identifying the disease early. With early recognition, rapid biopsy identification, and early specialist involvement, patients will get the opportunity for expedited multidisciplinary treatment and potentially delay rapid decompensation as shown by the evidence in this case.

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Disclaimer

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Ethics and consent

Consent was obtained by the patient's next of kin.

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