GI Amyloidosis Masquerading as Complicated Diverticulitis

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Background

Amyloidosis is a rare disease with an incidence of 1 in 100,000(1). It is characterized by the extracellular deposition of insoluble misfolded proteins causing architectural distortion and organ dysfunction.
Gastrointestinal infiltration of these proteins can be seen in different layers of the gastrointestinal tract. The clinical manifestations of GI amyloidosis vary with the amount and location of amyloid deposits.

Case Presentation

71 y/o female presented to the colorectal surgery clinic for evaluation of a presumed diverticular abscess, refractory to extensive courses of IV antibiotics. Synchronous symptoms included fecal incontinence, hematochezia, and diarrhea. Follow up imaging revealed a persistent 7cm intramural sigmoid collection and a 4cm distal ileal mass. Preop CTA chest for a known abdominal aortic aneurysm revealing multiple pulmonary nodules. Biopsy pertinent for large aggregates of amyloid deposition consistent with amyloidosis. Colonoscopy was performed with findings of pan colonic diverticular disease. No masses, strictures, or mucosal abnormalities were identified.

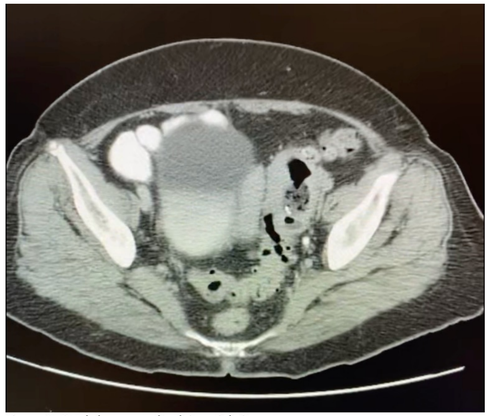


Figure 1: CT abdomen and pelvis, axial view

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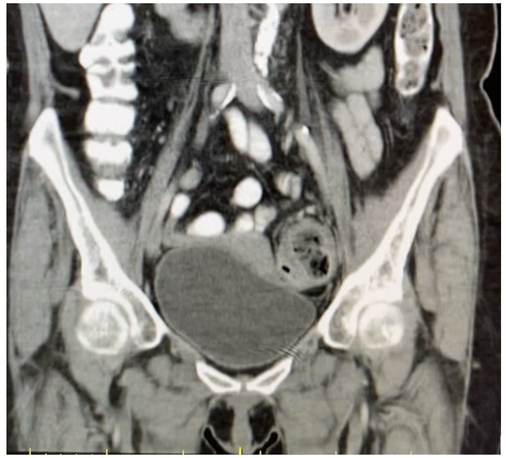


Figure 2: CT abdomen and pelvis, coronal view

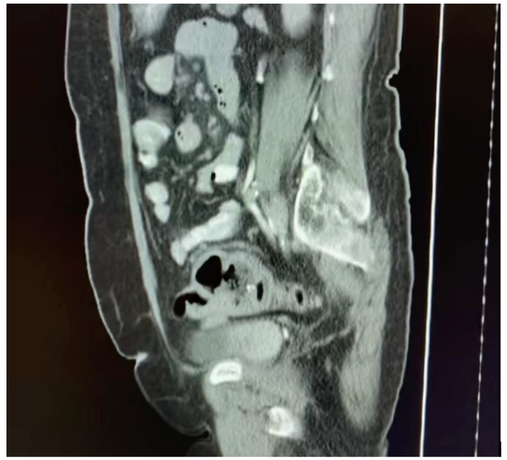


Figure 3: CT abdomen and pelvis, sagittal view

Management

Patient underwent surgical resection of sigmoid colon and distal ileum. Surgical pathology confirmed bulky amyloid deposits at both intestinal sites. Following surgical resection, patient reported complete resolution of her GI symptoms.

Pathology

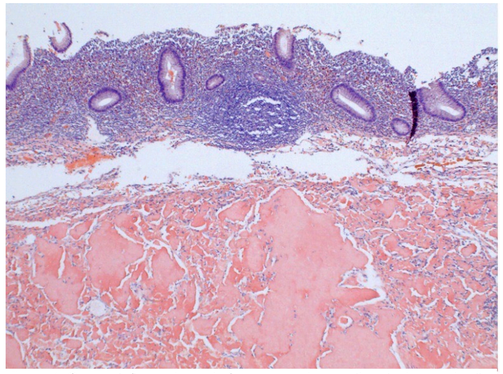


Figure 4: Chronic Inflammation with foreign-body type multinucleated giant cells

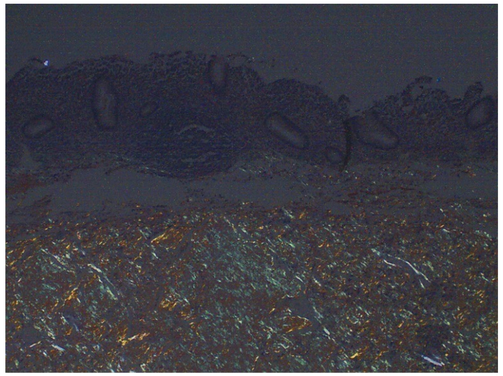


Figure 5: Congo red stain under polarized light shows green birefrigrence

Discussion

Gastrointestinal amyloidosis is defined as the presence of GI symptoms coincident with biopsy proven amyloid deposits. This is a relatively rare reported occurrence. Retrospective studies in known amyloidosis patients report that only 16.8% of patients develop GI signs and symptoms(2). Of those, only 3.3% of patients have biopsy proven gastrointestinal amyloidosis(3).
Amyloidosis affects all areas of the GI tract including small intestine, stomach, colorectum and esophagus. AL Amyloid deposition may occur in the muscularis propria, muscularis mucosa and submucosa leading to protrusions that may cause bowel obstruction. AA amyloid deposition is seen mainly in the mucosa and may lead to friability and ulcerations presenting with diarrhea and malabsorption.
Neuromuscular infiltration can affect the intrinsic nerve plexus and muscularis externa leading to abnormal peristalsis and dysmotility(4). Bleeding may result from mucosal lesions, vascular friability, and ischemia. Patients may even develop pneumatosis intestinalis or present with bowel perforation.

Conclusion

Diagnosis requires a high degree of clinical suspicion given the non- specific presentation. It should be suspected in patients with chronic GI symptoms, amyloidosis-associated conditions, or familial history of amyloidosis. Imaging is usually non-specific. A definitive diagnosis requires tissue biopsy with Congo Red (CR) staining which shows characteristic green birefringence on polarized light microscopy. Treatment for gastrointestinal amyloidosis involves symptomatic management and surgical excision of localized disease in patients presenting with severe or recurrent symptoms.

References

1.Dahiya, Dushyant Singh, et al. "Gastrointestinal Amyloidosis: A Focused Review." World Journal of Gastrointestinal Endoscopy, vol. 13, no. 1, 2021, pp. 1–12., https://doi.org/10.4253/wjge.v13.i1.1.
2.Yen, T., et al. "Clinical Implications of Gastrointestinal Symptoms in Systemic Amyloidosis." Neurogastroenterology & Motility, vol. 30, no. 4, 2017, https://doi.org/10.1111/nmo.13229.
3.Cowan, A. J., et al. "Amyloidosis of the Gastrointestinal Tract: A 13- Year, Single-Center, Referral Experience." Haematologica, vol. 98, no. 1, 2012, pp. 141–146., https://doi.org/10.3324/haematol.2012.068155.
4.Battle, William M., et al. "Gastrointestinal Motility Dysfunction in Amyloidosis." New England Journal of Medicine, vol. 301, no. 1, 1979, pp. 24–25., https://doi.org/10.1056/nejm197907053010105.
5.Alshehri, Saeed Ali, and Mahmoud Rezk Hussein. "Primary Localized Amyloidosis of the Intestine: A Pathologist Viewpoint." Gastroenterology Research, vol. 13, no. 4, 2020, pp. 129–137., https://doi.org/10.14740/gr1303.
6.Alshehri, Saeed Ali, and Mahmoud Rezk Hussein. "Primary Localized Amyloidosis of the Intestine: A Pathologist Viewpoint." Gastroenterology Research, vol. 13, no. 4, 2020, pp. 129–137., https://doi.org/10.14740/gr1303.

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