

Title:

Isolated colonic amyloidosis: a rare condition mimicking malignancy

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Isolated colonic amyloidosis: a rare condition mimicking malignancy

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Dear Editor,

We present the case of a 54-year-old male smoker admitted to our institution for painless rectal bleeding without anemia. An initial colonoscopy showed a neoplastic looking stenosing lesion in the descending colon, with necrotic and ulcerated areas (Fig1). However, the pathological results showed no evidence of malignancy and demonstrated amyloid deposits (Congo Red-positive staining with apple-green birefringence under polarized light). A subsequent CT scan revealed colonic wall thickening extending 15 cm distal to the splenic flexure, suggesting an inflammatory-infectious process or, less likely, a neoplastic lesion. Given this discrepancy, a second colonoscopy was performed, confirming ulcerations with erythematous and elevated mucosal areas but with no necrosis in this occasion (Fig1). Histopathological analysis again identified amyloid deposits. The patient was being previously followed by hematology for monoclonal gammopathy of undetermined significance (MGUS) as his only significant previous medical history. Further assessment by hematology and internal medicine ruled out primary (AL) amyloidosis and systemic involvement. A diagnosis of isolated or localized colonic amyloidosis was established.

Discussion

Gastrointestinal amyloidosis is rare and most often presents within a systemic context. Localized colonic involvement may manifest with nonspecific symptoms, including diarrhea, abdominal pain, malabsorption, obstruction, weight loss, or gastrointestinal bleeding [1,2].

Although our patient had monoclonal gammopathy of undetermined significance, further assessment by internal medicine as well as hematologic and genetic testing ruled out AL amyloidosis, confirming that the amyloid deposits did not derive from light-chain misfolding and that the process was limited to the colon. MGUS may raise suspicion of AL amyloidosis, but in this case, advanced testing confirmed a localized form of amyloidosis unrelated to the monoclonal protein.

Endoscopic and radiologic findings are often nonspecific, underscoring the importance of biopsy and Congo Red staining to confirm amyloid deposition [1,3]. Management depends on both the extent of disease and any underlying condition. In localized cases without systemic involvement, therapy may be limited to symptomatic measures or surgical resection if complications arise. Systemic disease, however, requires multidisciplinary care to address its etiology. Interestingly, Setake et al. [4] recently reported two different types of gastrointestinal amyloidosis (AL and AA), each with unique endoscopic findings and clinical outcomes, emphasizing the importance of early recognition. Our case further highlights how hematologic and genetic evaluations are critical to accurately classify amyloid deposits and guide appropriate management.

In conclusion, given the rarity of localized gastrointestinal amyloidosis, this case underscores the value of thorough evaluation in patients with atypical colonic findings, even when confounding factors such as MGUS is present. The biopsy is key in detecting the amyloid, but a comprehensive hematologic and genetic assessments determine whether amyloid deposition is isolated or part of a systemic process, guiding appropriate management and avoiding unnecessary therapies.

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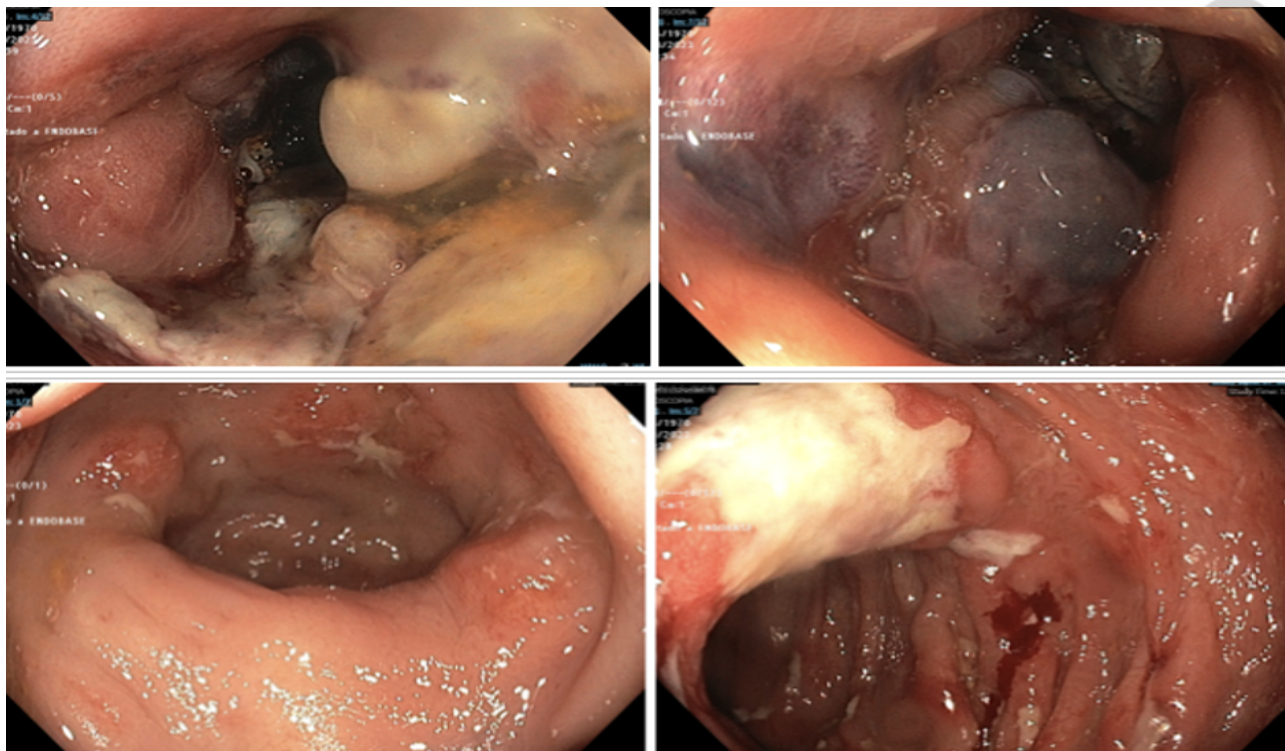


Figure 1. Upper images (first colonoscopy): a lesion with a neoplastic appearance, featuring violaceous nodules and seemingly necrotic areas. Lower images (second colonoscopy): fibrin-covered ulcerations with inflamed, erythematous mucosal areas interspersed with normal mucosa.