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Amyloidosis light chain (AL) amyloidosis with gastrointestinal involvement: diagnostic value of endoscopy in systemic diseases

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Dear Editor,

Intestinal amyloidosis is a rare manifestation of systemic amyloidosis, characterized by extracellular deposition of misfolded proteins (immunoglobulin light chains) in the wall of the gastrointestinal tract, causing structural and functional damage. Its clinical presentation is heterogeneous, with nonspecific symptoms and overlap with other digestive disorders, which contributes to its underdiagnosis[1].

A 76-year-old woman with no relevant medical history was admitted for new-onset heart failure presenting with anasarca and associated pleural and pericardial effusion. Transthoracic echocardiography revealed marked interventricular septal hypertrophy, a finding suggestive of a deposition disease. Serum protein immunoelectrophoresis

was requested, revealing a monoclonal peak in the beta region and very high levels of kappa free light chains (2117.95mg/L; 3.30-19.40mg/L). On the fifth day of admission, the patient presented melena and anemia. An upper endoscopy was performed, revealing patchy areas of erythematous and edematous tissue in the third portion of the duodenum, friable to touch, without complete circumferential involvement, and with preserved interlesional mucosa (Fig.1A-1B). Biopsies showed submucosal deposits of amorphous material with apple-green birefringence on Congo red staining, and immunohistochemistry demonstrated expression of kappa light chains, findings compatible with AL amyloidosis (Fig.1C-1D). With the diagnosis of AL amyloidosis confirmed by endoscopy, and given the suspicion of an underlying hematologic neoplasm, a bone marrow biopsy was performed revealing diffuse infiltration by plasma cells (27.8%) and deposits of amorphous material compatible with amyloid substance, establishing the diagnosis of multiple myeloma with AL amyloidosis involving advanced cardiac involvement and intestinal infiltration.

Primary amyloidosis, or AL amyloidosis, is a rare disease caused by abnormal deposition of monoclonal immunoglobulin light chains, and it is the most common form of systemic amyloidosis[2]. This deposit predominates in the muscularis mucosae and submucosa, unlike secondary amyloidosis where the amyloid deposit (AA protein) predominates in the lamina propria, resulting in different endoscopic and histological patterns[3]. Although it usually presents in isolation, up to 10-15% of cases are associated with plasma cell dyscrasias, especially multiple myeloma, which implies a worse prognosis. Diagnosing this association can be complex, especially when it presents initially with cardiac or gastrointestinal involvement. Gastrointestinal tract involvement, although described in systemic amyloidosis, often goes unnoticed or is underdiagnosed due to the nonspecific nature of its symptoms, including gastrointestinal bleeding[4]. In these cases, gastroscopy with biopsies is the fundamental technique to identify findings suggestive of amyloid infiltration and to confirm the histological diagnosis through Congo red staining and immunohistochemistry for light chains[5].

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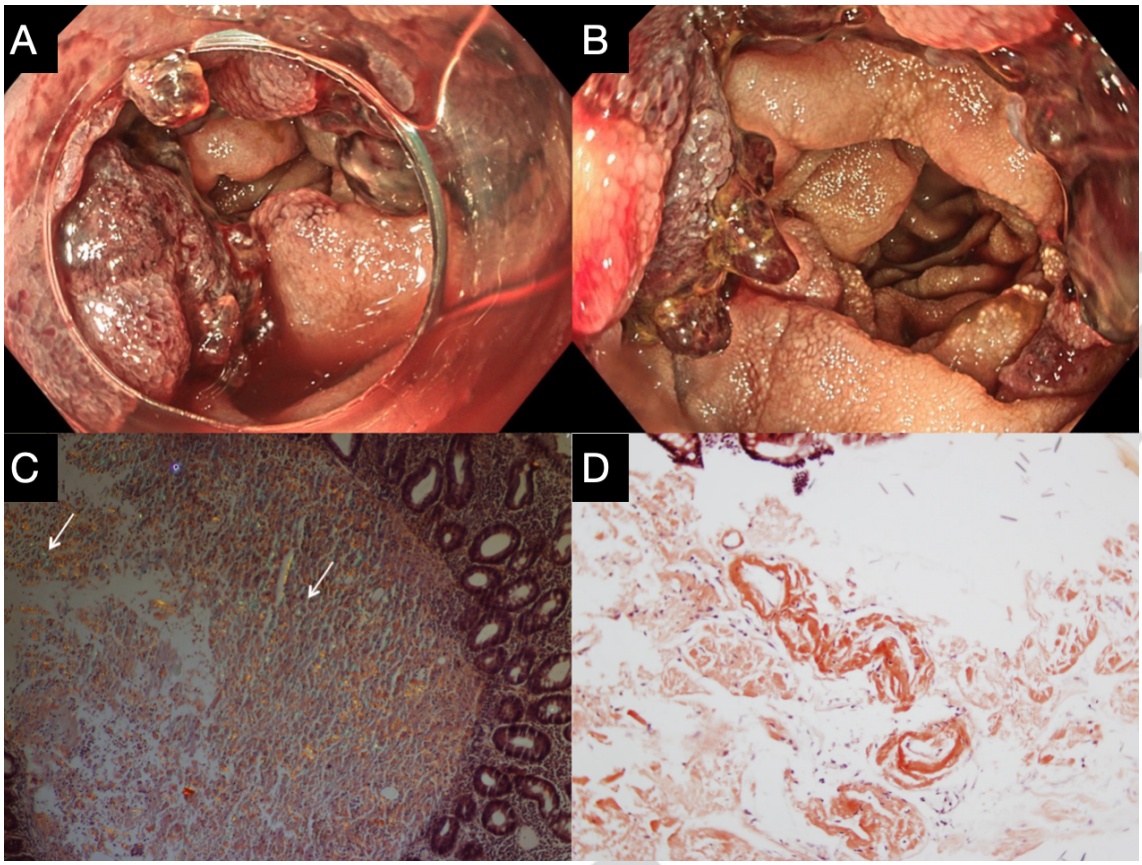

Fig. 1. Endoscopic images of the duodenal mucosa and histological images of duodenal biopsies. (A) and (B) Patchy areas of edematous and erythematous excrescent tissue with a nodular appearance, friable to the touch, and preserved surrounding mucosa. (C) and (D) Histological sections of duodenum with Congo red immunohistochemistry showing focal apple-green birefringence in the extracellular amorphous material and in the vessels, findings compatible with amyloid substance.