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Case Report

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Colonic perforation caused by secondary systemic amyloidosis

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Introduction

Amyloidosis is a rare condition characterized by the accumulation of amyloids in various tissues. These amyloid proteins are usually misfolded and form insoluble fibrils that deposit in the extracellular spaces of tissues, disrupting their normal structure and function. Amyloidosis can either be acquired or hereditary, systemic or localized.

The most affected organs include the kidneys, heart, nervous system, or gastrointestinal tract [1]. Intestinal amyloidosis can introduce diagnostic challenges in clinical practice and may be misdiagnosed or confused with other digestive tract diseases.

Intestinal-specific damage can indeed be the sole manifestation of amyloidosis in some cases, although it is more commonly a component of systemic amyloidosis where multiple organs are involved. The gastrointestinal tract can be affected in a significant percentage of patients diagnosed with amyloidosis, typically ranging from 3% to 28% of cases. AL amyloidosis is the most diagnosed form of amyloidosis based on gastrointestinal biopsies (52.8-83.3%), followed by AA (1.5-16.2%) and ATTR amyloidosis (4.2-12.5%) [1].

The clinical manifestations of intestinal amyloidosis may vary from asymptomatic to complitated forms [2].

We present a case of perforation of the colon in a patient caused by amyloidosis associated with multiple myeloma who underwent total colectomy with fatal evolution.

Case report

A 69-year-old woman was diagnosed with a light chain multiple myeloma. She underwent treatment with an autologous stem cell transplant and remained in remission for 9 years (with the help of maintenance treatment). However, her condition relapsed recently, marked by kidney injury that required corticosteroid therapy. About a month later, she experienced a new symptom of bloody diarrhea, which she had never encountered before. During her assessment, vital signs appeared normal, with a temperature of 37.7°C, a heart rate of 65 beats per minute, blood pressure of 130/70 mmHg, and oxygen saturation of 98% on room air. Physical examination revealed abdominal tenderness but no abdominal distension, palpable masses, or organomegaly.

Laboratory tests showed a normal white blood cell count of 4000/µL. However, her C-reactive protein level was elevated at 44 mg/L (normal range: 0-5 mg/L). Additionally, she had hypercalcemia (calcium level of 2.7 mmol/L), hypoalbuminemia (albumin level of 30 g/L), and significant proteinuria (7 grams of protein in a 24-hour urine sample).

The patient's colonoscopy revealed patchy erythematous and ulcerated mucosa throughout the colon. The histology findings indicate the presence of eosinophilic deposits on the surface and around the wall of the vessels, with these deposits showing a brick-red color when stained with congo red. Additionally, under polarized light, these deposits exhibit a yellow-green polarization (Figure 1). During his hospital stay, the patient's condition progressively worsened, manifesting with the

sudden onset of fever, hemodynamic instability, polypnea, and intense abdominal pain accompanied by peritoneal signs. A biochemical blood examination revealed an elevated level of C-reactive protein, measuring 240 mg/l, as well as elevation in creatinine and white (blood cell count. Abdominal CT imaging revealed intra peritoneal air compatible with sigmoid perforation.

Therefore, she underwent an exploratory laparotomy, which revealed the presence of purulent fluid within the peritoneal cavity. A perforation was identified in the transverse colon, and the entire colon showed signs of tissue devitalization. To address the situation, a total colectomy was performed, and peritoneal lavage was carried out. A double stomy was created to divert the gastrointestinal tract. Unfortunately, the patient's postoperative course was complicated of multisystem failure and she dead two days later. A pathological examination of the colon confirmed the presence of colonic amyloid deposits.

Discussion

The involvement of the GI tract in amyloidosis varies among different types of the disease. While cardiac, renal, and neurological manifestations are commonly recognized in amyloidosis, the GI tract can, often from the early stages of the disease, also be affected, albeit to a lesser extent. The prevalence of GI involvement in diagnosed patients ranges from 3% to 28%. As a result, the gastroinyestinal tract is considered a preferential site for biopsy due to its procedural safety and the increased likelihood of detecting amyloid deposits [1]. Intestinal involvment in amyloidosis can present with a wide range of symptoms, including diarrhea, weight loss, abdominal pain and malabdorption [1]. However, the clinical diagnosis of intestinal amyloidosis can be challenging, particularly in the absence of a known history of the condition. Most types of amyloidosis may not display distinctive findings during endoscopy (fine granular appearance, erosions, ulcerations, mucosal friability). The endoscopic appearance of amyloid deposits in the GI tract is often nonspecific and can mimic other GI disorders. This further emphasizes the need for histopathological examination of biopsied tissue to confirm the presence of amyloid deposits. Regarding the types of amyloidosis diagnosed based on gastrointestinal biopsies, the literature suggests that AL (immunoglobulin light chain) amyloidosis is the most commonly identified form in the GI tract, accounting for 52.8% to 83.3% of cases. AA (amyloid A) amyloidosis is reported in 1.5% to 16.2% of GI biopsies, while ATTR (transthyretin) amyloidosis is seen in 4.2% to 12.5% of cases.

For diagnosing GI tract amyloidosis, an intestinal biopsy is typically performed. The tissue sample is then stained with Congo red, a dye that binds to amyloid fibrils. Under polarized light microscopy, the stained amyloid deposits exhibit a characteristic green birefringence. This finding confirms the presence of amyloid in the tissue sample.

In AA amyloidosis, which is typically associated with chronic inflammatory conditions, the amyloid deposits are mainly found in the lamina propria mucosae and perivascular walls in the submucosa of the gastrointestinal tract. This distribution may result in endoscopic findings such as a fine granular appearance and mucosal friability.

On the other hand, in AL amyloidosis, which is caused by the deposition of abnormal immunoglobulin light chains, the amyloid tends to accumulate massively in the muscularis mucosa, submucosa, and muscularis propria. Endoscopically, this can manifest as multiple polypoid protrusions and thickening of the folds.

While gastrointestinal involvment is a known feature of systemic amyloidosis, including amyloid deposits in the intestinal wall, the occurrence of perforation is uncommon. However, when perforation does happen, it can be a significant and sometimes the initial manifestation of the disease [1].

In intestinal amyloidosis, the submucosal blood vessels are often the primary site of amyloid deposition. Overtime, amyloid fibrils gradually accumulate in both the blood vessels and the intestinal wall. This deposition can lead to the narrowing of vascular lumen, compromising blood flow to the affected areas [1,3].

Patients with GI amyloidosis who experience perforation have a significantly high postoperative mortality rate, ranging from 50% to 69%, regardless of multiple myeloma. Several factors contribute to this elevated mortality, including preoperative organ damage caused by amyloid deposits in vital organs such as the heart, kidney, and liver. Additionally, the occurrence of disseminated intravascular coagulation as a preoperative complication further exacerbates the risks [2,3].

Conclusion

Given that the clinical presentation of intestinal amyloidosis often lacks specific features, it is crucial to conduct a comprehensive assessment involving meticulous endoscopic, radiologic, and histopathological evaluations to establish an accurate diagnosis. Intestinal amyloidosis may present with more varieties of clinical symptoms than previously thought.

The occurrence of life-threatening complications such as intestinal perforation, ischemic disease, or a combination of both should be recognised as potential gastrointestinal complications associated with amyloidosis. Early diagnosis and rapid surgical intervention should be considered.

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