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Case Report

Gastric Amyloidosis: A Rare Culprit Behind Persistent Dyspeptic Symptoms

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Abstract

Gastric amyloidosis is a rare, localized form of amyloidosis that presents with non-specific gastrointestinal symptoms, often mimicking other conditions. A 56-year-old woman presented with dyspeptic symptoms, including postprandial bloating, heartburn, and weight loss. Endoscopy revealed nodular gastric lesions, and biopsies confirmed localized gastric amyloidosis (AL subtype) via Congo red staining. Systemic involvement was excluded. Despite initial symptom control with proton pump inhibitors, symptom recurrence and lesion progression led to a laparoscopic total gastrectomy. The patient had an uneventful recovery and remains asymptomatic three months post-surgery.

Gastric amyloidosis is exceedingly rare, with fewer than 35 cases reported over six decades. It predominantly presents with non-specific gastrointestinal symptoms, such as dyspepsia, hematemesis, or hematochezia, and lacks distinctive endoscopic features. Diagnosis relies on histological confirmation via Congo red staining and polarized light microscopy. Exclusion of systemic involvement is critical for accurate disease classification and management. While systemic amyloidosis often requires chemotherapy, localized forms are best managed surgically, with favorable outcomes.

This case highlights the diagnostic challenges of localized gastric amyloidosis and the importance of a multidisciplinary approach. Surgical management can offer excellent outcomes in this rare condition.

Keywords: Amyloidosis; Gastric Amyloidosis; Minimally Invasive Surgery; General Surgery; Laparoscopy; Gastrectomy

Introduction

Amyloidosis is the name given to a heterogeneous group of disorders, all of which have the hallmark of extracellular deposition of insoluble fibers in various tissues of the body, resulting from alterations in the mechanisms of protein formation.

This disease affects men more than women, and the median age of diagnosis is 65 years [1]. This spectrum of diseases is rare and can be fatal [2,3]. Since amyloidosis corresponds to a spectrum of diseases, it can be associated with a panoply of signs and symptoms, many of which are non-specific and can mimic other pathologies.

There are various types of amyloid deposits, which are categorized according to their origin. Actually, the disease is named with two capital letters, the first one A stands for Amyloid, and the second is for the type of fibril deposited, which in turn can be L for light-chain (primary amyloidosis), A for A protein, TTR for transthyretin and Aβ2M for amyloid β2 microglobulin (secondary amyloidosis) [1-6]. Amyloidosis can be further classified into systemic and localized, depending on the distribution of the amyloid in the body.

Systemic amyloidosis is more common, with a prevalence of 51 cases per million inhabitants, and can be acquired or hereditary

[1,2,5]. Localized amyloidosis, on the other hand, is substantially rarer, accounting for 12% of all diagnosed cases of amyloidosis [3].

Case Report We present the case of a 56-year female, with no relevant medical history, who presented to her doctor complaining of postprandial bloating, heartburn and weight loss. Based on these symptoms, she underwent an upper digestive endoscopy in July 2023, which described 'nodular elevations and raised erosions in the fundus-body transition. In the distal portion of the great curvature, on the slope of the posterior wall, irregular and/or amputated folds'. Biopsies were taken, which showed nodular areas of hyaline material, positive Congo red. *Helicobacter Pylori* negative. No other alterations.

The rest of the investigation was carried out, but no further foci of amyloid deposition were found; echocardiogram without any suspicion of amyloid infiltrate, renal and bladder ultrasound as well, negative search for amyloid in abdominal fat.

He also underwent a computerized tomography scan, which apart from the gastric thickening did not detect any other abnormal thickening of the digestive tract (Figure 1 and 2).

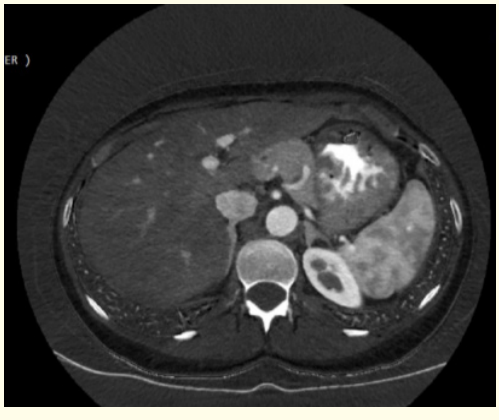

Figure 1: CT scan showing gastric Thickening.

The decision was made to keep her under surveillance and medicate her with a proton pump inhibitor, with apparent symptomatic control for a few months.

About 9 months later, due to worsening complaints, she underwent another endoscopy, which this time reported: Cardia without lesions. Mucosa of the distal body/transition of the great curvature and anterior face with violet nodular lesions measuring around 30 mm; in the great curvature, an extensive infiltrative and nodular lesion occupying a large area. Posterior face of the antrum with a 15 mm violet nodular lesion, which was biopsied. Centered and permeable pylorus.

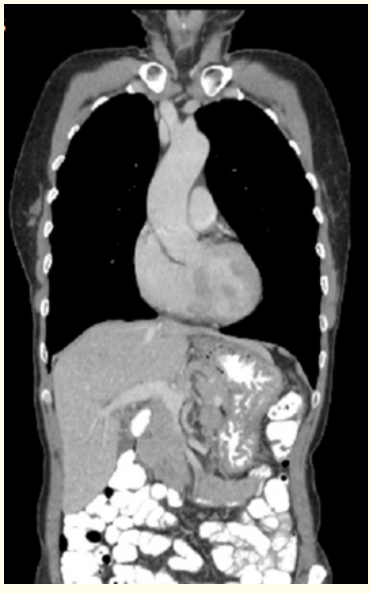

Figure 2: CT scan showing gastric Thickening.

The histology describes: fragments of gastric mucosa, of the antral and corpus types, where areas with a pink amorphous substance are observed, which, using Congo Red staining, becomes birefringent and dull green in color under polarized light - aspects compatible with amyloid deposition. No dysplasia or neoplastic tissue in the sample. *H. pylori* negative.

Mass spectrometry was performed in the presence of DJ1/PARK. Non-immunoglobulin protein pattern, dominated by the presence of ApoAII and in the background ApoAI. Clear predominance of lambda chains, suggesting AL.

Given the rarity of the case, the fact that it was amyloidosis localized to the stomach, and the patient's symptoms, after discussion at a multidisciplinary meeting, the patient was offered laparoscopic total gastrectomy, which she accepted.

The patient underwent the procedure in August 2024, and the surgery and post-operative period were uneventful, with the patient being discharged on the 5th post-operative day, tolerating a diet.

The histopathological assessment of the surgical specimen agreed with the initial findings of gastric amyloidosis (Figure 3 - 9).

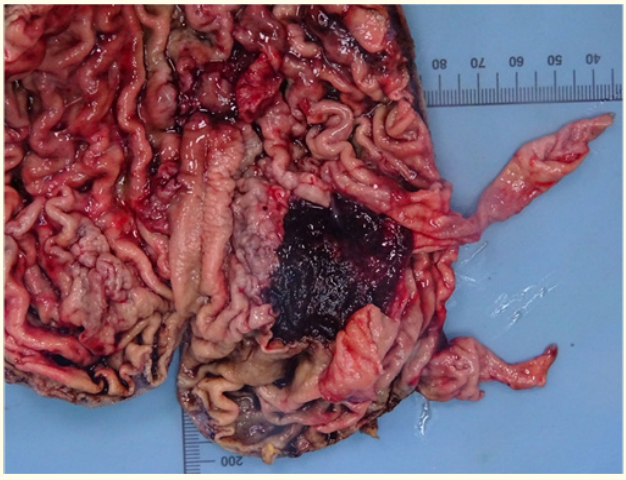

Figure 3: Macroscopic appearance of the stomach showing an extensive infiltrative and nodular violet lesion.

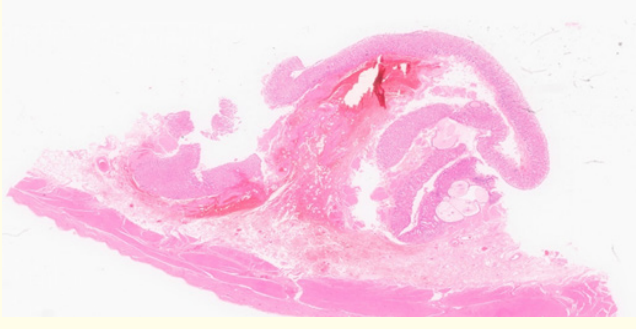

Figure 4: Gastric wall with mucosal ulceration and submucosal hemorrhage (H&E, 2x objective).

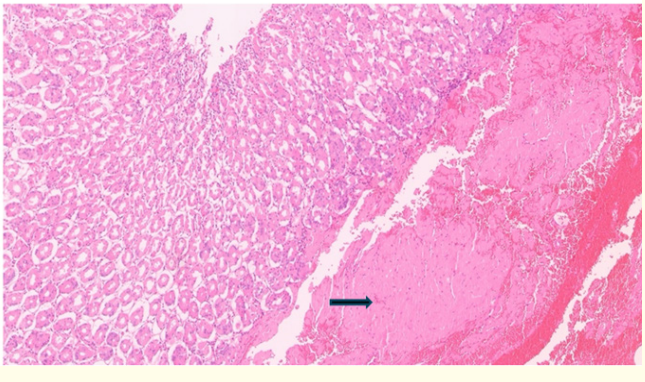

Figure 5: Extensive pale pink homogenous areas of amyloid deposition (arrow) in between submucosal fibers (H&E, 10x objective).

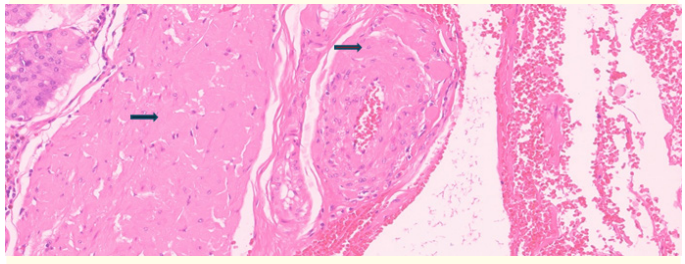

Figure 6: Extensive pale pink homogenous areas of amyloid deposition in submucosal layer (black arrow) and surrounding blood vessels (red arrow) (H&E, 20x objective).

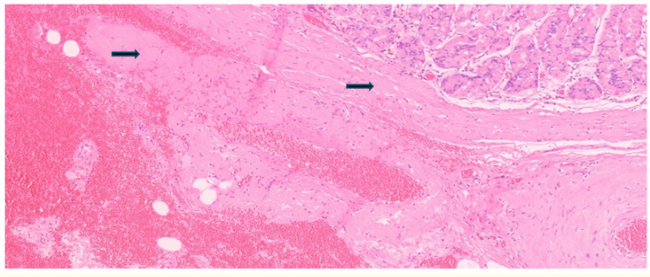

Figure 7: Gastric wall with extensive amyloid (H&E, 20x objective).

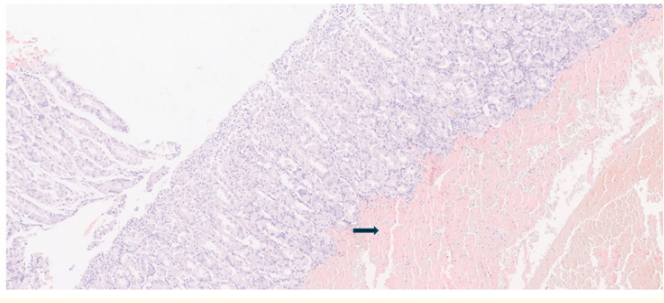

Figure 8: Gastric wall with extensive amyloid, Congo Red, 10x objective. Under polarizing filter, this area demonstrates apple green birefringence.

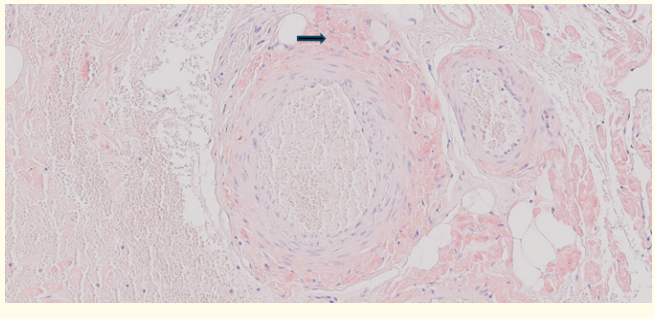

Figure 9: Pale pink homogenous areas of amyloid deposition in submucosal vessels (red arrow), Congo Red, 20x objective. Under polarizing filter, this area demonstrates apple green birefringence.

Three months after the operation, the patient is well and has no complaints.

Discussion

Gastric amyloidosis, which is a form of localized amyloidosis, is extremely rare [3,5]. When we searched Pubmed for 'gastric amyloidosis' we found only 33 cases reported in the last 62 years.

As stated before, localized amyloidosis is a rare disease, but most cases are AL amyloidosis, which is the most common type of amyloidosis. These light-chain proteins, also named Bence Jones proteins, can be related to plasma-cell dyscrasias, and that's why it is utterly important to exclude other causes of light-chain immunoglobulins production [6].

Once the diagnosis of amyloidosis is made, it is essential to rule out the involvement of other organs, namely the heart and kidney (the main organs affected by amyloid deposits), and the presence of amyloid in the abdominal fat, to help us classify the disease burden and adjust treatment. Also, when there is gastrointestinal involvement, in the majority of cases, we are speaking of systemic amyloidosis (79%), and only 21% correspond to localized disease [3].

The biopsy and staining with Congo Red is the gold-standard for diagnosis, showing green-apple birefringence on polarized light, but when stained with hematoxylin-eosin the amyloid deposits are visible as amorphous eosinophilic hyaline material [3].

The signs and symptoms are non-specific and can mimic almost any disease that affects the stomach. The patient can experience dyspeptic symptoms, heartburn, hematemesis, hematochezia, among other [7]. Also, endoscopic findings are unspecific, which can include ulcers, folds, elevation, cancer like-lesions, erosions or hematomas [3,6,8]. This information highlights the need for a high level of suspicion and multidisciplinary approach.

Concerning the treatment options, when we refer to systemic amyloidosis, either AL or AA, chemotherapy followed by hematopoietic stem cell transplant is the main option, for localized amyloidosis the option is different, and surgery has been the mainstay treatment, with very good prognosis [5]. However, given the small percentage of cases of this disease, there are no guidelines for treatment defined yet, so the options depend on a multidisciplinary decision, case by case.

Conclusion

This case of localized gastric amyloidosis underscores the diagnostic challenges posed by this rare disease, particularly given its non-specific symptoms and endoscopic findings. Our patient's dyspeptic symptoms, initially controlled with conservative management, progressed over time, compelling a multidisciplinary approach to confirm the diagnosis and determine the optimal treatment.

The absence of systemic involvement in this case highlights the importance of thorough diagnostic workup, including imaging, histopathology, and mass spectrometry, to classify amyloidosis accurately. Total gastrectomy proved to be a definitive and curative intervention, with excellent postoperative outcomes and symptom resolution.

This report emphasizes the need for a high index of suspicion for localized amyloidosis in atypical presentations of dyspepsia and illustrates the critical role of a multidisciplinary team in managing such rare cases. Further research is needed to standardize treatment guidelines for localized amyloidosis and improve outcomes for affected patients.

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