Endoscopic Presentation of Amyloidosis


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ABSTRACT

A 80 year old female presented to the ED with a 4 day history of increased lethargy, confusion, disorientation, and weakness. The patient was noted to have decrease in P.O. intake and diarrhea for the prior two days as well as guiac positive stool. Endoscopic evaluation revealed the presence of gastric and duodenal mucosal abnormalities identified as amyloid deposits.

Reason for Presentation

The endoscopic presentation of amyloidosis in the digestive tract is quite variable. Awareness of the different potential endoscopic presentations of this disease may assist gastroenterologists in making prompt and accurate diagnosis especially in patients without typical clinical signs or symptoms of amyloidosis. This presentation seeks to highlight the need to consider amyloidosis in the differential diagnosis of any endoscopic abnormality of the digestive tract.

Conclusions and Recommendations

Due to the variable presentation of intestinal amyloidosis, any suspicious lesions involving the stomach, duodenum, small bowel or even the colon should raise suspicion of the possibility of amyloidosis whether or not the patient has typical clinical features of amyloidosis.

INTRODUCTION

Amyloidosis is a group of diseases that is characterized by extracellular deposition of insoluble protein fibrils in tissues and organs. (1) The common property shared among this group of disorders is a common beta pleated sheet combined with various combinations of misfolded proteins that leads to its characteristic staining properties. Amyloidosis may be classified as Primary, in which there is no apparent causative process or related disease, Secondary, in which there is an associated chronic infectious or inflammatory process, localized or tumor forming, familial and finally, multiple myeloma related.

Amyloidosis occurs at an incidence of 12 cases per million people annually with gastrointestinal involvement and symptoms occurring in 30-60% of patients. Common symptoms of amyloidosis include: diarrhea, weight loss, macroglossia, malnutrition, abdominal pain, nausea, vomiting, poor intestinal motility, occult or overt gastrointestinal hemorrhage, ischemia, obstruction or even perforation. (2) The endoscopic presentation of amyloidosis in the digestive tract is quite variable. (3)

- There may be thickened irregular gastric folds that demonstrate hypoechoic mucosa and submucosa with a loss of sonographic wall layers
- Endoscopic loss of rugal folds
• Ulcers with clean bases or with irregular overhanging edges resembling carcinoma
• Arteriovenous malformations
• Granular-appearing mucosa
• Plaque-like lesions
• Ulcerative gastritis
• Gastroparesis

The variable endoscopic presentation of this disease presents a diagnostic challenge for the novice as well as experienced Gastroenterologist alike.

This case report presents a rare intestinal endoscopic presentation of amyloidosis. Knowledge of the various endoscopic presentations of this disease may help in the accurate diagnosis of this disease process. This diagnosis is rarely considered when endoscopic mucosal lesions are encountered in the digestive tract, especially when lesions resemble other subcutaneous lesions such as Gastrointestinal Stromal Tumors (GIST), lipomas, lieyomas, lieomyosacromas, adenomas and other nodular lesions of the digestive tract.

Case Presentation

A 80 year old female presented to the ED with a four day history of increased lethargy, confusion disorientation, and weakness. The patient was noted to have decrease in P.O. intake and non-bloody diarrhea for the prior two days. Past medical history included Hypertension, Depression, Spinal Stenosis, Asthma and Chronic Back pain. Physical exam revealed a well developed female with lethargy and Guiac positive stool. All other physical parameters were unremarkable. Laboratory exam showed a creatinine of 7 and a BUN of 90, hemoglobin of 10.1 g/dL with MCV of 87.9 and RDW of 15.4, serum iron of 64, TIBC 298 and ferritin 86.9, coagulation indices showed INR 1.1, PT 14.3 and PTT 32.8. A Computed Tomography of the abdomen and pelvis revealed thickening second portion of the duodenum. The patient subsequently underwent an upper endoscopy to evaluate CT scan abnormality.

DISCUSSION

There has not been much literature recording the gastrointestinal manifestations of amyloidosis nor the frequency of the different endoscopic presentations. Though uncommon, there are specific patterns clinically seen with deposition of amyloid in the gastrointestinal tract. Endoscopically, the gastrointestinal mucosa may have multiple yellowish-white polyoid protrusions as in our case, prominent folds, fine granular appearance, small mucosal hemorrhages, shallow or deep ulcers, mucosal friability or simply just waxy plaques. (5, 6) Amyloid can be seen by routine haematoxylin stain and display apple green birefringence under polarized light.

A postmortem study by Gilat et. al. with 70 patients with systemic amyloidosis showed that amyloid deposits were found in the inner layer which includes the intima or media of blood vessels in all cases, as well as, the outer layer including the adventitia in most cases. Infiltration of all layers of the small intestine wall may also be seen, however, submucosal involvement was most prominent. In cases of Primary Amyloidosis (AL) and amyloidosis involved with multiple myeloma, the deposits were always
found in the outer layer of the vessels, and parenchymal deposition was found in the muscularis. (4) On the other hand, in amyloidosis associated with Familial Mediterranean Fever and Secondary Amyloidosis (AA) the amyloid was involved in the inner coat of small blood vessels and parenchymal deposition were mainly in the mucosa. Based upon the location of deposition, it was proposed that certain clinical manifestations could occur. For example, amyloidosis that lead to muscular damage could lead to impaired motility, where as involvement of the mucosa could lead to malabsorption (4).

One of the primary manifestations of amyloidosis is occult or overt gastrointestinal bleeding. The etiology of bleeding is multifactorial. First, small vessel fragility due to direct amyloid infiltration or from related ischemia plays a large role as was seen in our case. Secondly, bleeding may be increased by coagulation abnormalities as a result of hepatic involvement leading to factor X deficiency. (7) Thirdly, other factors that lead to elevated prothrombin time can be disseminated intravascular coagulation, malabsorption or decreased intake of vitamin K. (7)

The definitive diagnosis of amyloidosis may be made by tissue biopsy with Congo Red Dye. Abdominal fat pad aspirate is the easiest way of obtaining tissue. Tissue biopsy of duodenal or colorectal mucosa however are more sensitive than fat biopsy, but more invasive.

**Conclusion**

In summary, our patient presented with guiac positive stool and mucosal nodularity which represented a rare endoscopic presentation of amyloidosis. Endoscopists should be aware of the many possible endoscopic appearances as it may be the only initial manifestation of gastrointestinal amyloidosis in the absence of gastrointestinal symptoms.

**Citations**

2. Tada S., Mitsuo S. et. al., Amyloidosis of the Small Intestine: Findings on Double contrast Radiographs. AJR. April 191. 741-744
1. Large firm erythematous mucosal lesion with ulceration

2. Multiple yellow tinged nodular mucosal lesion in the second portion of the duodenum

3. Patchy erythematous friable gastric mucosa
A. Pink amorphous subcutaneous deposit on eosin stain

B. Vasular amyloid infiltration

C. Apple green birefringence on Congo Red stain