Reminder of important clinical lesson

Gastric amyloidosis presenting with severe weight loss

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Summary

A previously well 59-year-old lady with 70 kg weight loss and chronic diarrhoea over a 28-month period presented following collapse and subsequent diagnosis of pulmonary embolism. Previous investigations for this weight loss included normal gastroscopy and colonoscopy, CT and MRI abdomen, barium follow through and octreotide scan.

She underwent echocardiogram which revealed myocardial speckling and asymmetrical left ventricular hypertrophy. Repeat oesophago-gastro-duodenoscopy and colonoscopy for rectal bleeding was performed. Colonoscopy revealed intramucosal haematomas and electron microscopy (EM) of the gastric biopsies confirmed amyloid deposition. Amyloidosis of the gastrointestinal (GI) tract and heart were confirmed on serum amyloid protein scan.

GI amyloid is rare and symptoms include weight loss, diarrhoea, GI bleeding and gut dysmotility.¹ GI amyloidosis should be considered as a diagnosis and sought when other common causes have been excluded. The greatest yield is by Congo red staining or EM of rectal specimens.

BACKGROUND

Gastrointestinal (GI) amyloidosis is very rare but should be considered as a diagnosis in a patient with malabsorption when commoner causes have been excluded. This was a fascinating case of a lady with significant weight loss and deterioration in quality of life who had undergone many tests over 2 years before the final diagnosis was made. This is a diagnosis which should often be revisited when all investigations prove normal even when amyloidosis is initially 'excluded' as the case here.

CASE PRESENTATION

A 59-year-old woman with body mass index 46 (weight 137 kg) presented with a 2-month history of diarrhoea and weight loss. There was no significant past medical history apart from atrial fibrillation controlled with digoxin. Clinical examination was normal and there was no abdominal tenderness or masses. Blood tests including full blood

count, liver function tests, amylase, thyroid function, immunoglobulins, serum electrophoresis and short synacthen test were normal. Stool cultures were negative. Serum albumin was 24 g/dl without proteinuria. Gastroscopy and colonoscopy were normal and biopsies showed no evidence of inflammatory bowel disease or coeliac disease. Abdominal ultrasound showed gallstones and a non-dilated biliary tree.

She had persistent diarrhoea and weight loss of 70 kg over the next 26 months but remained well. Barium follow through revealed slow intestinal transit but no focal abnormality. Faecal elastase and *p*-amino benzoic acid were normal. CT abdomen was normal and there was no intra-abdominal lymphadenopathy. Fasting gut hormone profile (vasoactive intestinal peptide, pancreatic polypeptide, gastrin, glucagon, chromogranin A&B, somatostatin) was normal. Octreotide and SeHCAT scans were normal, as was endoscopic ultrasound. Lactose breath test was positive.

Figure 1 Narrow band and white light images of the gastric body: prominent thickened gastric mucosa.

Figure 2 Intramucosal haematomas of sigmoid colon.

Figure 3 Electron microscopy of gastric biopsy, magnified $\times 4600$: peri-vascular amyloid deposition.

She was admitted to hospital as an emergency following a collapse and was diagnosed with a pulmonary embolism. An echocardiogram revealed asymmetric left ventricular hypertrophy with slight speckling of the myocardium. During her inpatient stay she developed melaena and fresh rectal bleeding. Repeat oesophago-gastro-duodenoscopy showed prominent thickened gastric mucosa (see figure 1). Colonoscopy showed intramucosal haematomas in the sigmoid (see figure 2).

Congo-red staining of the gastric biopsies was negative, but electron microscopy was strongly positive for amyloid

(see figures 3 and 4). She was referred to for a serum amyloid protein scan which confirmed amyloid of the stomach, small intestine, colon and heart.

INVESTIGATIONS

See above within 'Case Presentation'.

DIFFERENTIAL DIAGNOSIS

Commoner causes of malabsorption such as coeliac, inflammatory bowel disease, chronic pancreatitis, underlying GI

Figure 4 Electron microscopy of gastric biopsy, magnified $\times 130\,000$: amyloid protein filaments.

malignancy, hyperthyroidism. Rarer causes such as metabolic disease, for example, Fabry's disease.

TREATMENT

Total parenteral nutrition (TPN) was given both in hospital and at home and the patient's weight improved. Unfortunately shortly after diagnosis this patient died from sepsis secondary to a TPN line infection.

Treatment of primary GI amyloid is usually symptomatic with pro-kinetics and TPN feeding. Therapy for systemic amyloidosis with GI involvement may be beneficial but there is limited data and includes steroids and melphalan in reactive cases.

OUTCOME AND FOLLOW-UP

Shortly after diagnosis this patient died from sepsis secondary to a TPN line infection.

DISCUSSION

There is one published case report of gastric amyloidosis presenting in a similar way with chronic diarrhoea, in a 64-year-old lady previously diagnosed with irritable bowel syndrome.² There was again GI and cardiac involvement as the patient had restrictive cardiomyopathy. There is otherwise little published about GI amyloid and thus I feel it is important to highlight this case.

Learning points

- ▶ Know the common causes of malabsorption.
- ▶ Think about sending GI biopsies for Congo red staining and electron microscopy.
- ▶ In the case of continuing weight loss despite normal investigations, consider GI amyloidosis.

Competing interests None.

Patient consent Obtained.

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