

## Original Article

# Primary amyloidosis mimicking Crohn's disease: a case report

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**Abstract:** Amyloidosis is an uncommon disease that results from the extracellular deposition of abnormal fibrillary protein. This paper reports a case of primary amyloidosis with predominant involvement of the gastrointestinal tract and heart as a mimicker of Crohn's disease in a sixty-seven years old man admitted with repeated diarrhea and fatigue. This patient poorly responded to 5-aminosalicylic acid and quickly developed dyspnea and hypotension. The further laboratory test revealed a monoclonal protein detected by serum protein electrophoresis. Biopsy of abdominal wall fat pad revealed amyloid substance deposited and positive Congo red staining, which was diagnosed as primary amyloidosis.

**Keywords:** Primary amyloidosis, diagnosis, Crohn's disease

### Introduction

Amyloidosis is a group of diseases, characterized by the extracellular deposition of an abnormal fibrillary protein, which disrupts tissue structure and function [1]. The wide range of presenting symptoms encountered and the lack of specific radiological characteristics make rapid clinical diagnosis difficult. Here we report an unusual case of primary amyloidosis, which was misdiagnosed as a Crohn's disease.

### Case presentation

A 67-year-old male presented with repeated diarrhea for 1 year. In addition to gastrointestinal symptoms, the patient also suffered with fatigue. He had no past history of tuberculosis, rheumatoid arthritis or collagen vascular disease. Physical examination revealed blood pressure: 105/51 mmHg, heart rate: 58 bpm/min. Palpation of abdomen revealed no abnormalities with respect to pain, tenderness or masses. The laboratory findings revealed hemoglobin 104 g/L (normal 131-172 g/L), total protein of 39.8 g/L (normal 64.0-83.0 g/L), albumin of 20.5 g/L (normal 35.0-55.0

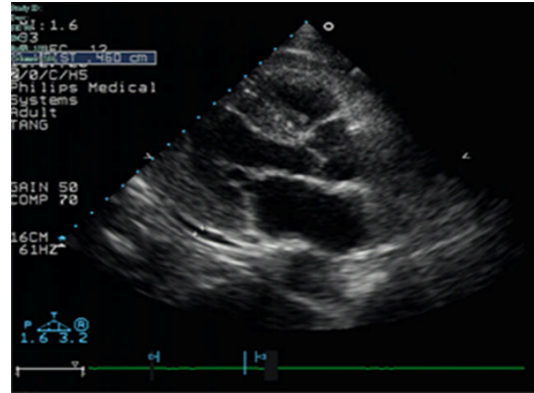
g/L), brain natriuretic peptide of 2742 pg/mL. ECG showed low voltage in the limb leads, right axis deviation and ST-T changes. Echocardiography showed left ventricular systolic dysfunction (ejection fraction 31%), mitral and tricuspid mild regurgitation and localized little pericardial effusion. It is considered heart failure. Transverse unenhanced computed tomography (CT) image showed partial small bowel wall thickening, dilatation and mesenteric infiltration (**Figure 1**). Bowel inflammation was suspected. Initial colonoscopy was negative for any lesions. Capsule endoscopy showed a punctiform ulcer of upper small intestine and multiple mucous membrane erosions of lower small intestine. The diagnosis was considered as Crohn's disease.

This patient was initially treated with 5-aminosalicylic acids to which he poorly responded. Later, he developed dyspnea and hypotension. Ten days later, his blood pressure dropped to 65/40 mmHg and dopamine infusion was administered to restore blood pressure. The patient received cardiology consultation and was diagnosed as myocarditis, and then he was referred to department of cardiology for further

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**Figure 1.** Transverse unenhanced computed tomography (CT) image showed partial small bowel wall thickening and dilatation.



**Figure 2.** Echocardiography showed obvious thickening of the left ventricular walls.

treatment. Laboratory test results revealed a monoclonal protein detected by serum protein electrophoresis. Bone marrow biopsy showed plasma cell hyperplasia. Cardiovascular magnetic resonance imaging (CMR) showed left ventricular systolic and diastolic dysfunction, thickened left ventricular walls and pericardium. Repeated echocardiography showed large left atrium and large right atrium, thickened ventricular walls, left ventricular systolic dysfunction (ejection fraction 31.7%), mitral and tricuspid regurgitation, localized little pericardial effusion (**Figure 2**). It is considered cardiac amyloidosis. Biopsy of abdominal wall fat pad was collected and stained with hematoxylin eosin, showed necrosis of adipose tissue with fat dissolution and amyloid substance deposited, especially positive Congo Red staining. The case was diagnosed as primary systemic amyloidosis. Unfortunately, he responded poorly to treatment of heart failure and conventional treatment for amyloidosis. The cardiac involvement resulted in progressive heart failure and hypotension. After consulting the patient and family, further active treatment was withdrawn and he was transferred to a hospice for palliative care where he died shortly afterwards.

### Discussion

Amyloidosis falls into several major categories: primary, secondary, dialysis-associated, and a variety of hereditary forms [2]. Primary amyloidosis is the most common form of amyloidosis, which has been associated with deposition of excess immunoglobulin light chains in multiple myeloma, lymphoma, and other lymphoprolif-

erative disease [3, 4]. Secondary amyloidosis is associated with deposition of serum amyloid protein A in inflammatory, infectious, and neoplastic diseases. Clinically apparent disease usually presents in middle or old age. Both genders are nearly equally affected with slight predominance of men over women [5]. Amyloidosis can be acquired or hereditary, and systemic or localized to a single organ.

Clinically, amyloidosis may be categorized as localized or systemic [4]. Most forms exist in both localized and systemic forms. The clinical manifestations and imaging features of amyloidosis are diverse and nonspecific. It is often mistaken for other diseases [6]. In the gastrointestinal tract, amyloidosis usually appears as ulcers, nodules, or polypoid masses, and it may have ischemic, hemorrhagic, or dysmotility presentations [7]. The most common radiological finding in cases of intestinal amyloidosis is of a symmetrical thickening of the folds, which represents oedema caused by ischaemia due to vascular deposition [8]. In this case, gastrointestinal amyloidosis was misdiagnosed as Crohn's disease due to similarities in gastrointestinal symptoms and gross appearance as revealed by capsule endoscopy examination and other diagnostic modalities, such as CT scan. Accurate diagnosis is only possible with histological examination of the involved tissues, Congo red staining of which is the only reliable means to confirm diagnosis [9].

Amyloid deposition in the gastrointestinal tract leads to mucosal friability, erosions and autonomic neuropathy. Clinical symptoms vary in accordance with the layer of bowel wall involved.

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Mucosa predominant disease is manifested as malabsorption, whereas muscularis predominant disease presents as obstruction. Autonomic neuropathy may affect gut function, which presents as diarrhea, constipation and early satiety [8, 10]. The heart is frequently the predominant organ affected by primary amyloidosis. The worst outcome was associated with overt congestive heart failure. Myocardial deposition of amyloid causes progressive thickening of ventricular walls that primarily results in the deterioration of ventricular filling with later worsening of global systolic function in advanced stages [11]. So some degree of fatigue, dyspnea and hypotension are common presenting signs and symptoms of amyloid cardiomyopathy.

In fact, the onset of clinical signs or symptoms at different times considered to be nonspecific will become decisive factors for the orientation towards a correct diagnosis, if the doctor could relate them to each other. Panels of differential diagnoses are certainly very useful. Primary amyloidosis should be considered in the light of a differential diagnosis of gastrointestinal symptoms with involvement of multiple tissues and organs, even when we see what seems to be a familiar disease pattern, and especially when a patient has associated chronic inflammatory disease, multiple myeloma, or monoclonal gammopathy [7]. Biopsies stained with Congo red play a role in the initial diagnosis of amyloidosis and should be considered for any patient with unexplained diarrhea, malabsorption, weight loss, or gastrointestinal bleeding, particularly in association with a monoclonal gammopathy or symptoms or signs of other organ disease. Early diagnosis of primary systemic amyloidosis is important because most cases are diagnosed late and have a grave prognosis. The survival rate may be improved by early diagnosis and early therapy.

### Disclosure of conflict of interest

None.

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