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Case Report

Understanding amyloid light chain cardiac amyloidosis: a case study emphasising integrated diagnostic techniques

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ABSTRACT

Cardiac amyloidosis, a manifestation of systemic amyloidosis, presents diagnostic challenges due to diverse clinical presentations. A 56-year-old woman with a history of hypertension presented with recurrent chest tightness, palpitations, decreased exercise tolerance, and nocturnal dyspnea. Diagnostic workup revealed reduced left ventricular function, thickened interventricular septum, and left atrial enlargement on echocardiography and electrocardiogram. Immunohistochemical staining confirmed AL amyloidosis in bone marrow biopsy specimens. Treatment involved supportive care and chemotherapy for plasma cell dyscrasia. Prompt identification and intervention for AL amyloidosis are crucial, highlighting the diagnostic role of electrocardiograms and echocardiograms. Management strategies included supportive care and chemotherapy for plasma cell dyscrasia. This case underscores the significance of timely recognition and intervention for cardiac amyloidosis, contributing to improved understanding and management approaches.

Keywords: AL amyloidosis, Cardiac amyloidosis, Multiple myeloma, Integrated diagnostic techniques, Echocardiography, Monoclonal plasma cell

INTRODUCTION

Amyloidosis is a rare condition defined by the mutation or alternation of plasma cells present in the bone marrow. This results in a cluster of diseases characterized by the deposition of fibrillary proteins in the extracellular compartment, leading to the loss of normal tissue architecture.[1,2]

According to the composition of amyloid fibrils, major amyloidogenic protein-related diseases include immunoglobulin light chain amyloidosis (AL), amyloid A protein amyloidosis (AA), and transthyretin amyloidosis (ATTR).[3] AL amyloidosis, also known as amyloid light chain or primary amyloidosis, occurs when there are alterations or mutations in light chain proteins. Light chain proteins are fragments of antibodies produced by the bone marrow's plasma cells that fight infections.[4] Chemotherapy, stem cell transplantation, bone marrow transplantation, or a combination of these treatments can be used to manage amyloidosis based on its severity.[5,6]

The accumulation of amyloid protein in the extracellular matrix of cardiomyocytes is referred to as cardiac amyloidosis (CA).[3] Presentation of symptoms of heart failure with preserved ejection function and diastolic dysfunction is often associated with cardiac amyloidosis.[2] Congo red-staining of myocardial biopsy is regarded as the gold standard for diagnosis, but due to the high risk and high cost of this invasive biopsy, it is not suitable for most patients.[7] Classically, AL amyloidosis is described as the presence of increased interventricular septal thickness, concentric cardiac hypertrophy along with normal to low voltage on the electrocardiogram (ECG).

Immunohistochemical staining has been proven to have clinical benefits in the detection and follow up of patients with AL amyloidosis due to its ability to detect monoclonal plasma cells in bone marrow specimens.[8,9]

This report presents a case of CA diagnosed through electrocardiogram, echocardiogram, and immune-histochemical staining of bone marrow biopsy specimens conducted at our hospital.

CASE REPORT

Patient presentation

A 56-year-old woman presented with a chief complaint of recurrent chest tightness and shortness of breath, persisting for over two years and exacerbated in the past month. The electrocardiogram performed at the local hospital indicated sinus tachycardia, which was left untreated. Five months ago, a PET scan at the local hospital revealed pleural effusion and coronary calcification. Minimal pericardial effusion was also noted, and tuberculosis was ruled out. One month ago, symptoms significantly worsened with a progressive decrease in exercise tolerance and nocturnal dyspnea. The patient also experienced episodes of chest oppression lasting several minutes, accompanied by numbness in the extremities and a rash resembling erythema around the eyes, which led to hospitalization. Since the onset of symptoms, poor appetite and a 4kg weight loss have been noted.

Her past medical history included hypertension spanning 15 years with a maximum blood pressure of 170 mmHg. At present the patient has been taking Bisoprolol sustained-release 23.75 mg and Irbesartan 75 mg, with recent episodes of low blood pressure. Notably, she underwent kidney stone lithotripsy five years ago. The patient denied a history of smoking or alcohol consumption and had no remarkable family history.

Physical examination

Upon examination, the patient displayed a body temperature of 36.3°C, tachycardia (197 beats/min), reduced respiratory rate (18 beats/min), and hypotension (BP 90/55 mmHg, HR 97 beats/min). periorbital purpura is seen around her right eye (Figure 1a). Macroglossia with lateral scalloping of the tongue was observed (Figure 1b). Coarse breath sounds in both lungs, and no crackles or wheezing. Regular heart rhythm with no murmurs detected. Mild symmetric pitting edema and numbness in both lower extremities. Auxiliary examinations revealed cervical spine degeneration (C4/5, C5/6, C6/7). carpal tunnel syndrome was also noted.

Diagnostic findings

Further investigations unveiled multiple findings indicative of cardiac involvement, including low voltage on ECG (Figure 2a). The ultrasound examination revealed a left atrial enlargement (anteroposterior diameter approximately 42 mm) and a slightly small left ventricle (maximum transverse diameter at end-diastole approximately 42 mm). The mid-segment of the interventricular septum and partial left ventricular wall were thickened (approximately 16 mm at the thickest part of the septum and approximately 13 mm at the thickest part of the wall), with uneven signal intensity. There was reduced diastolic movement in various segments of the left ventricle, with an unobstructed left ventricular outflow tract. Mild regurgitation signals were noted in both the mitral and tricuspid valves. Doppler echocardiography indicated a decreased longitudinal strain rate at the basal segment of the left ventricular wall, normal strain rate at the apex, and an ejection fraction (EF) of 54.2%. These findings suggest left atrial enlargement and basal segment thickening of the left ventricular wall, with reduced wall motion (apex preserved) (Figure 2 b and c). They also suggest diastolic dysfunction of the left ventricle (grade III, suggestive of restrictive filling), with normal lower limit of left ventricular systolic function. Imaging revealed bilateral pleural effusion, incomplete subdivision of lower lung segments, and multiple lymph nodes in the hilar and mediastinal regions. Vaginal ultrasound depicted multiple uterine fibroids.

Laboratory and bone marrow assessment

Laboratory tests revealed increased inflammation markers (ESR-40 mm/h), elevated fibrinogen (5.39 g/l), and aberrant immunoglobulin levels (Table 1). Cardiac markers reveal an elevated troponin I level at 0.046 ug/l, with a subsequent recheck showing a further increase to 0.048 ug/l. BNP was elevated at 141.6 ng/l. However, there was an elevated globulin level at 45.1 g/l, and a decreased albumin level at 30.6 g/l. Hematological analysis indicated a hemoglobin level of 114 g/l. Urinalysis showed the presence of urinary protein. Tumor markers included an elevated CA 125 at 117 U/ml, while others remain within normal limits.

Bone marrow biopsy revealed a fat to cell ratio of 60:40%. The specimen exhibited active marrow proliferation with normal granulocytic cell morphology at various stages. Megakaryocytes were present at a rate of 1-3 per high-power field (HPF), and there was significant proliferation of immature plasma cells.

Immunofixation electrophoresis and bone marrow analysis detected abnormal monoclonal plasma cells. The phenotype of these cells includes Kappa: negative (-), Lambda: positive (+), CD138: positive (+), CD38: positive (+), CD34: negative (-), CD117: negative (-). Additionally, MPO and CD15 show partial positivity, CD235a is partially positive, E-cadherin exhibits a few positive cells, CD61 displays scattered positivity, CD20 and CD3 are negative, and CD138 is positive, accounting for 1.51% of abnormal monoclonal plasma cells. Special staining results: Congo red, Reticulin fibers, and Perls are all negative (-).

Table 1: Key laboratory findings, including inflammation markers, cardiac markers, kidney and liver function, blood and urine analyses, bone marrow with reference values.

Tests	Result	Reference Range
Inflammation markers (mm/h)	ESR - 40 mm/h	0-20 mm/h
Fibrinogen (g/l)	5.39	2-4
Immunoglobulin levels	Aberrant	-
Cardiac markers (ug/l)	Troponin I: 0.046 (initial), 0.048 (recheck)	<0.04
Brain Natriuretic Peptide (BNP) (ng/l)	141.6	0-100
Kidney function	Normal	-
Liver function	Normal	-
Globulin (g/l)	45.1	23-35
Albumin (g/l)	30.6	35-50
Hemoglobin (g/l)	114	130-170
Urinalysis	Protein present	-
Tumor markers (U/ml)	CA 125: 117	<35
Bone marrow analysis	Abnormal Monoclonal Plasma Cells	-
Blood light chain	κ chain: 1.57↓, λ chain: 42.7↑	κ: 3.13-7.23; λ: 0-5.35
Urine light chain	λ chain 16.00	0-5.35
Serum immunofixation electrophoresis	IgG Monoclonal, λ-type Monoclonal	-
Urine immunofixation electrophoresis	All Negative	-
Immunoglobulins, complement	IgG: 32.20↑, C3: 0.62↓, C4: 0.15	IgG: 7-16 g/L; C3: 0.82-1.55 g/L; C4: 0.16-0.38 g/L

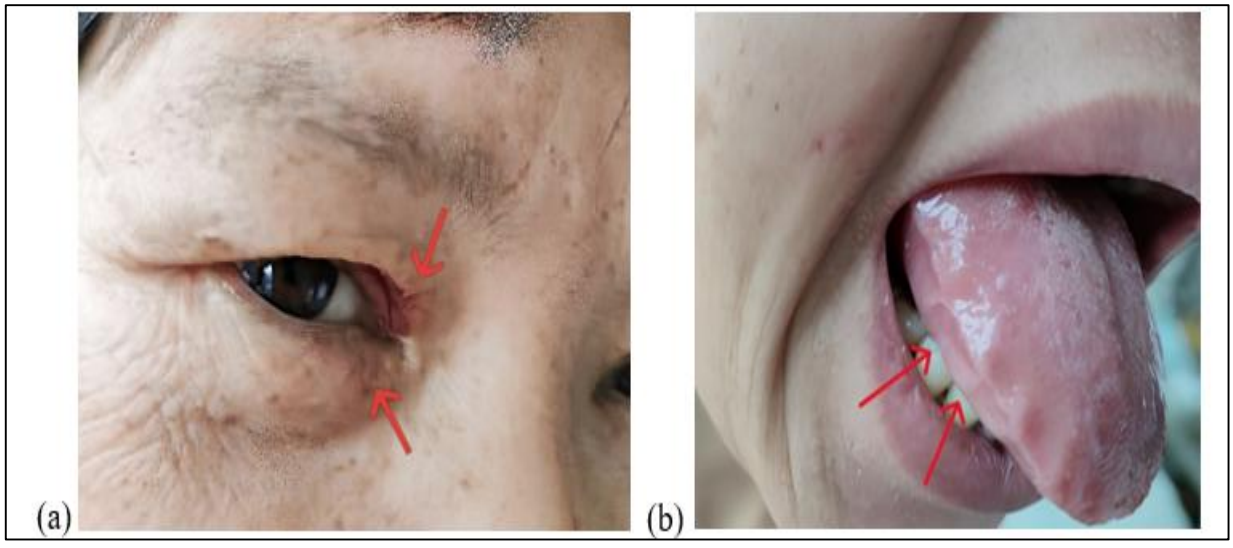

Figure 1: (a) Ecchymosis in the inner corner of the right eye; (b) Macroglossia with lateral scalloping of the tongue.

Blood light chain: κ chain 1.57↓, λ chain 42.7; κ/λ ratio: 0.037 (recommended <0.26 for significance according to guidelines). Urine light chain: λ chain 16.00. Serum immunofixation electrophoresis: Positive for IgG monoclonal immunoglobulin, and λ-type monoclonal immunoglobulin. Urine immunofixation electrophoresis: All negative.

Immunoglobulins, complement: IgG 32.20↑, C3 0.62↓, and C4 0.15↓

Revised diagnosis

It was concluded that the patient had Multiple myeloma, Light-chain (AL) amyloidosis affecting the myocardium, Heart failure with preserved ejection fraction (HFpEF), Pericardial effusion (minimal), Pleural effusion (minimal) and Carpal tunnel syndrome.

Treatment and outcome

Given the diagnosis, upon stabilization, the patient was transferred to hematology department where she underwent PCD chemotherapy (once/week): bortezomib 2 mg+dexamethasone 20 mg+cyclophosphamide (CTX) 300 mg with the addition of daratumumab (DARA) and received diuretics to manage serous cavity effusions. The treatment plan incorporated medications for blood pressure control, lipid-lowering, and symptomatic relief.

Follow-up and discharge

After regular outpatient visits, the patient was discharged with ongoing chemotherapy and scheduled for weekly follow-ups

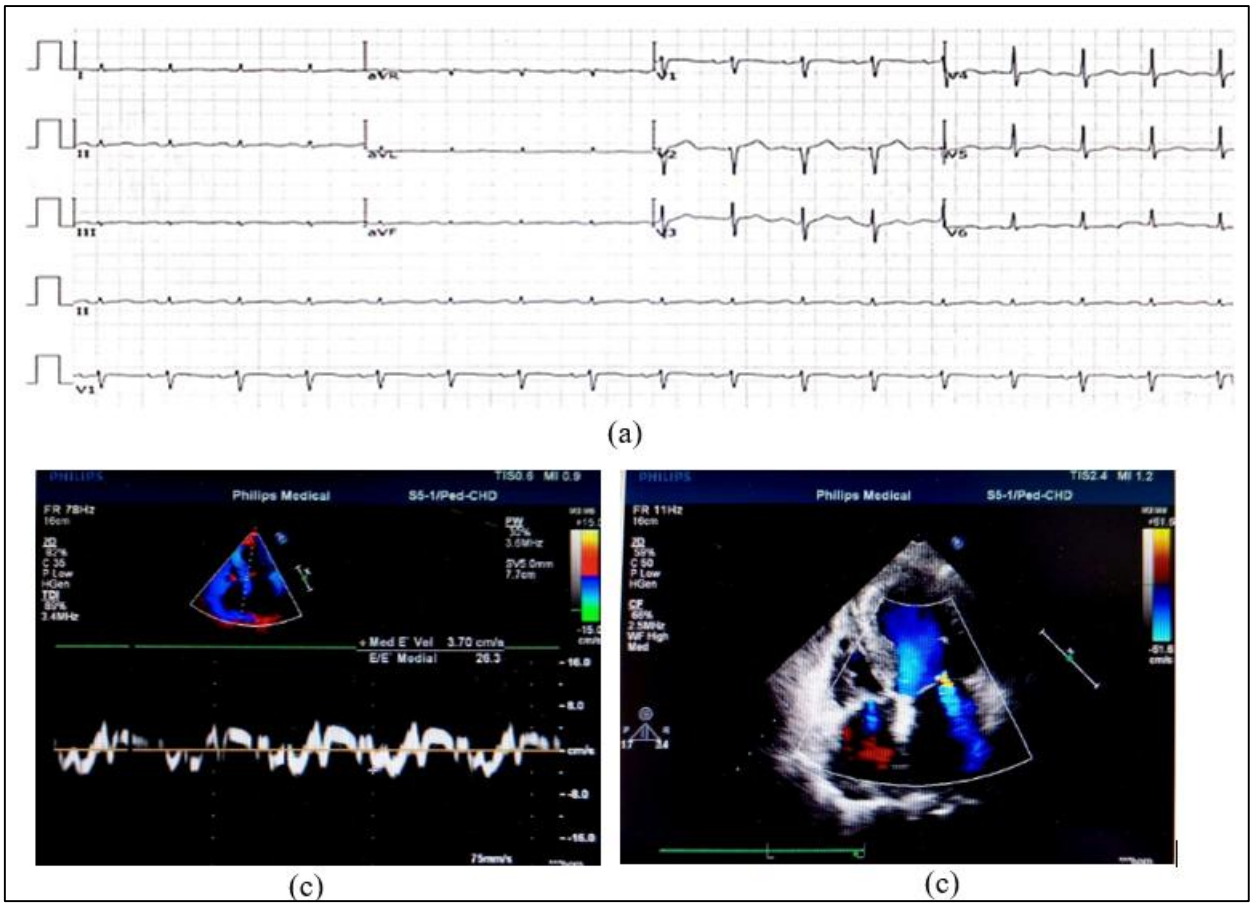

Figure 2: (a) Low voltage electrocardiogram on admission; (b and c) coloured cardiac doppler echocardiography showing left atrial enlargement and left ventricular thickening.

DISCUSSION

In recent years, the integration of echocardiography and electrocardiogram parameters has substantiated the significant clinical advantages as per the several clinical investigations conducted both in China and internationally.[10] This combination of cardiovascular techniques aids in the early identification, diagnosis, and timely intervention, thereby significantly improving patient prognosis in CA.[11]

Detection of monoclonal protein is the first step in the diagnosis of AL amyloidosis, as its absence makes it extremely unlikely. A bone marrow biopsy is the primary method for assessing tissue specimens. AL amyloidosis is marked by the production of insoluble amyloid fibrils from increased serum light chains caused by monoclonal proteins. These abnormal formations result from identical, structured cross β-sheet formations formed by the self-assembly of monoclonal proteins.[12] Flow cytometry is often used to detect the phenotype of plasma cell, abnormal plasma cells express markers like CD38, CD138, and CD45. Moreover, the abnormal expression of immunoglobulins, typically the kappa and lambda light chains, helps in further characterization and identification of these pathological plasma cells.[9]

Cardiac involvement is one of the crucial prognostic markers as it is most common organ involved in AL amyloidosis. Evaluating cardiac involvement involves

assessing concentric hypertrophy and specific filling patterns using echocardiography.[13]

Echocardiography serves as a primary and critical diagnostic tool for cardiac amyloidosis. It reveals myocardial hypertrophy and elevated echogenicity. In cases of AL amyloidosis, moderate to severe LVH is often symmetric while in ATTR, predominant septal hypertrophy is observed. Additionally, echocardiography detects diastolic dysfunction with restrictive filling, bi-atrial enlargement, mild pericardial effusion, and thickening of atrioventricular valves and the interatrial septum.[2,14,15] Classically, Cardiac Amyloidosis is characterized by distinctive features seen on non-invasive imaging, particularly evident in the disproportion between left ventricular wall thickness and QRS voltages, leading to low QRS voltage despite increased LV wall thickness. A cluster of noncardiogenic signs and symptoms, termed "red flags," accompanies CA, including macroglossia, skin bruising, pitting edema, proteinuria, and carpal tunnel syndrome.[2]

Persistently elevated cardiac troponin (cTn) levels signify cardiac amyloid infiltration and serve as a prognostic marker. Higher levels up to >0.035 g/l predict a poor prognosis, while cTn >0.060 g/l or cTnI >0.10 µg/l indicate an increased risk of mortality, particularly in blood stem cell transplantation.[16]

CONCLUSION

The case underscores the importance of early detection and timely intervention in cardiac amyloidosis, as it significantly impacts patient prognosis. The integration of various diagnostic modalities and a comprehensive treatment plan resulted in the patient's stabilization and discharge with scheduled follow-ups. This case highlights the complexities associated with AL amyloidosis and the importance of a multidisciplinary approach in managing such cases.

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