Cardiac Amyloidosis

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Abstract
We present a 54 year old male who presented with congestive cardiac failure and was diagnosed as restrictive cardiomyopathy with mild mitral regurgitation on 2D echocardiography. Cardiac amyloidosis was diagnosed in view of renal biopsy revealing amyloid deposition. Patient did not have any obvious etiology for secondary amyloidosis.

INTRODUCTION
Cardiac amyloidosis occurs most commonly with primary amyloidosis seen with immunocyte dyscrasias. About a third of patients with primary amyloidosis have clinically evident cardiac disease, although most patients have pathological evidence of cardiac involvement at autopsy. In secondary amyloidosis involvement is usually clinically insignificant (perivasular deposits). Familial amyloidosis causes overt cardiac disease in a fourth of patients and is dominated by conduction system involvement.

Here we present a case of cardiac amyloidosis presenting with restrictive cardiomyopathy. Patient did not have any obvious etiology for secondary amyloidosis.

CASE REPORT
A 54 yrs old man, resident of Nanded district in Maharashtra, farmer, presented with history of dyspnea on exertion NYHA class II of 3-4 months duration, which increased to class III over last 4 days. Patient developed orthopnea since 1 day. He also complained of pedal oedema, abdominal distension and facial puffiness for 1 month. There was no history of palpitations, chest pain, or syncope.

There was no past history of hypertension, diabetes mellitus and ischemic heart disease. Also there was no past history of tuberculosis or any other chronic disease. The patient was not on any long term medications.

On examination, he was conscious and oriented, afebrile and had a pulse rate of 116 per minute which was regular and low volume. The patient was tachypneic with a respiratory rate of 30 per minute. His BP was 100/70 mm of Hg in right upper extremity in supine position. There was no evidence of pulsus paradoxus. He had pitting pedal oedema and cold and clammy extremities. Jugular venous pulse was raised and Kussmaul’s sign was positive. He did not have any pallor, icterus, clubbing or lymphadenopathy.

On auscultation, there was normal S1 and S2, with S3 gallop and a soft pansystolic murmur in the mitral area. There was decreased air entry and crepitations bibasally. He also had 3 cm tender hepatomegaly and 2 cm splenomegaly.

On investigation, Hb was 14.2 gm%, TL- 7800 cells/cmm (P74 L24 E1 M1). ESR was 27 mm at the end of 1 hr. FBS- 79mg%, PLBS- 96mg% were normal. Serum creatinine was 1.0 mg%, BUN 25mg%, LFT- WN. ABG showed pH-7.507, pO2 - 98, pCO2 - 24 and HCO3 -19.4. Urine- Albumin 1+ and 24 hr urine protein was 580 mg with no Bence Jones proteins.

XRC revealed normal lung fields with mild cardiomegaly. ECG had no abnormalities except low voltage complexes. USG of abdomen showed hepatomegaly (span 18 cm) with altered echotexture, splenomegaly and mild ascites. Kidneys were of normal size (right kidney- 10.1 cm x 4.4 cm and left kidney- 10.4 cm x 4.2 cm), corticomedullary differentiation maintained and normal pelvicalyceal system.

2D echocardiography showed thickened myocardium with EF 45%, There was severe diastolic dysfunction, mild bialtrial enlargement, mild mitral regurgitation with normal valves and moderate pulmonary hypertension (56 mm), features suggestive of restrictive cardiomyopathy.

A diagnosis of restrictive cardiomyopathy with biventricular failure was made. The patient was treated with routine anti-failure treatment in form of dobutamine infusion, oxygen and diuretics (frusemide and aldactone). The patient improved considerably in terms of decrease in dyspnea and pedal oedema.

Considering that cardiac amyloidosis is the commonest
cause of restrictive cardiomyopathy in adults worldwide and patient having proteinuria, we did a renal biopsy to rule out amyloidosis. The renal biopsy demonstrated amorphous eosinophilic material in arteriolar walls and the interstitium (Fig. 1). This is highly suggestive of amyloidosis. Polarization was not possible due to technical reasons.

On retrospect, the patient did not have any bone pains or backache and did not have any bony tenderness. Radiographs of the spine and skull did not show any ‘punched out’ lesions.

Protein electrophoresis did not show any M band; IgA 110 (82-453 mg/dl), IgM 31.9 (46-304 mg/dl), IgG 949 (751-1560 mg/dl) 949, Kappa 739 (629-1350 mg/dl) and Lambda 400 (313-723 mg/dl). The cause of the systemic amyloidosis could not be elucidated.

Final diagnosis cardiac and renal amyloidosis with restrictive cardiomyopathy with biventricular failure.

**DISCUSSION**

Cardiac amyloidosis is more common in men and is rare before the age of 40 years. Amyloid is deposited in between myocardial fibres, often with significant involvement of papillary muscles and the endocardium. Intramural coronary arteries may also be involved. Our patient had mitral regurgitation most likely due to papillary muscle dysfunction as mitral valve was normal on 2D echo.

The most common manifestation is that of restrictive cardiomyopathy (RCM) due to stiffened ventricles. Right sided findings dominate the clinical picture.

Myopathic involvement produces the characteristic diastolic dip and plateau (square root sign) in the ventricular pressure. There is also an impaired early diastolic filling. Systolic dysfunction occurs late in the course of disease. Angina and myocardial ischemia occurs with coronary involvement. Orthostatic hypotension occurs in 10% of patients and is due to amyloid deposition in the heart, autonomic nerves and vessels.

Exertional syncope is considered a poor prognostic sign and most patients die within 3 months. S4 is uncommon due to atrial amyloid deposits leading to reduced atrial contraction.

Chest radiography usually reveals absence of cardiomegaly, no pericardial calcification. Manifestations of pulmonary venous hypertension and pulmonary congestion are also seen.

Electrocardiography may reveal low voltage, intraventricular conduction delays, bundle-branch blocks, atrial fibrillation, complex ventricular arrhythmias and nonspecific ST-T changes simulating MI (pseudo-infarct pattern). Abnormality of cardiac impulse generation and conduction is least common, the most common arrhythmia being atrial fibrillation. Ventricular tachycardias and blocks are also seen and may be associated with sudden cardiac deaths.

Echocardiography reveals symmetrically thickened walls, small ventricular chambers, dilated atria and thickening of interatrial septum and AV valves. AV valve regurgitations are commonly seen. The cardiac walls have a granular, sparkling texture. Our patient had restrictive cardiomyopathy with mild mitral regurgitation.

Scintigraphy with Technetium99m pyrophosphate is often strongly positive with prominent amyloid deposits, although it may also be falsely negative in some patients. Scanning with Indium labelled antimyosin antibody also detects cardiac amyloid involvement.

Abdominal fat, rectum, kidney, liver and gingivae can be sampled for detection of amyloid. Endomyocardial biopsy is only indicated if any of these sites are inconclusive. We did not do myocardial biopsy in our patient as kidney biopsy was diagnostic.

Treatment of cardiac amyloidosis consists of routine antifailure treatment with cautious use of vasodilators and diuretics. Digitalis is to be avoided as it tends to promote arrhythmias. Calcium channel blockers and beta-blockers are contraindicated. Anticoagulation is given in patients with atrial involvement. Patients with early cardiac involvement may benefit from alkylating agents / stem cell transplantation. Recurrence of amyloid deposition in the transplanted heart can occur. Median survival is less than one year in patients with symptomatic cardiac amyloidosis.

**REFERENCES**


