

Echocardiography evaluation leads to correct etiological diagnosis of amyloid myocardopathy- A Case Report.

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Citation

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Abstract

Amyloidosis is a rare disorder that commonly involves the heart in the light-chain and senile form of the disease. We report an adult patient with a predominant right ventricular heart insufficiency with suggestive echocardiography findings of infiltrative myocardopathy. The invasive coronary angiography showed no significant stenosis and the endomyocardial biopsy confirmed the diagnosis of amyloid myocardopathy.

BACKGROUND

Amyloidosis is a rare disorder of protein conformation and metabolism that results in tissue deposition of proteinaceous fibrils causing organ dysfunction. Heart involvement is a common finding in patients with light-chain, hereditary, and senile forms of the disease (1). The commonest form of amyloid deposition is that produced from clonal light chains, in which the heart is affected in close to 50% of cases, and congestive heart failure is the presenting clinical manifestation in about half of these patients (2). However, clinical presentation varies and may mimic other infiltrative cardiomyopathies or storage disorders, as well as hypertrophic cardiomyopathy. The diagnosis of cardiac amyloidosis is important for prognosis and therapy and may be suggested by characteristic echocardiographic findings, low-voltage ECG complexes and clinical data (3). The endomyocardial biopsy is a safe and reliable procedure for diagnosing cardiac amyloidosis, and immunohistochemical staining of routinely processed biopsy specimens can be performed to classify the type of amyloid present (4). All forms of amyloid are characterized by positive staining with Congo red. When viewed under polarized light, amyloid deposits demonstrate apple green birefringence (5). The natural history of primary amyloidosis is poor, and for patients with symptomatic cardiac involvement, survival is generally less than 6 months. Even among treated patients with amyloid heart disease, survival beyond 5 years is rare (6).

CASE REPORT

We report a 63-year-old white man with sporadic asthma and tobacco abuse that was admitted with a dyspnea on exertion chief complaint. One month ago he noticed an invariable swelling in the course of the day, localized to the middle third of both legs. Two weeks ago the edema becomes more manifest overtaking up the knees, with an associated tiredness and progressively worsening dyspnea on exertion. The physical exam revealed a hard pitting bilateral edema up the knees, enlarged liver, jugular venous distention and positive hepatojugular reflux. The Echocardiography examination revealed a concentric hypertrophy of the left ventricle (LV) arising the interauricular septum with an ECG tracing showing markedly decreased voltage. Increased pericardial echogenicity, minimum pericardial effusion, reduced left ventricular ejection fraction (LVEF=35%) and increased right ventricular lateral wall thickness with systolic dysfunction of the right ventricle (TAPSE=15mm) was also observed. All these findings suggested the diagnosis of infiltrative myocardopathy. The invasive coronary angiography showed no significant stenosis of the coronary arteries. Finally, the endomyocardial biopsy exposed Congo red positivity with apple-green birefringence, confirming the diagnosis of cardiac amyloidosis.

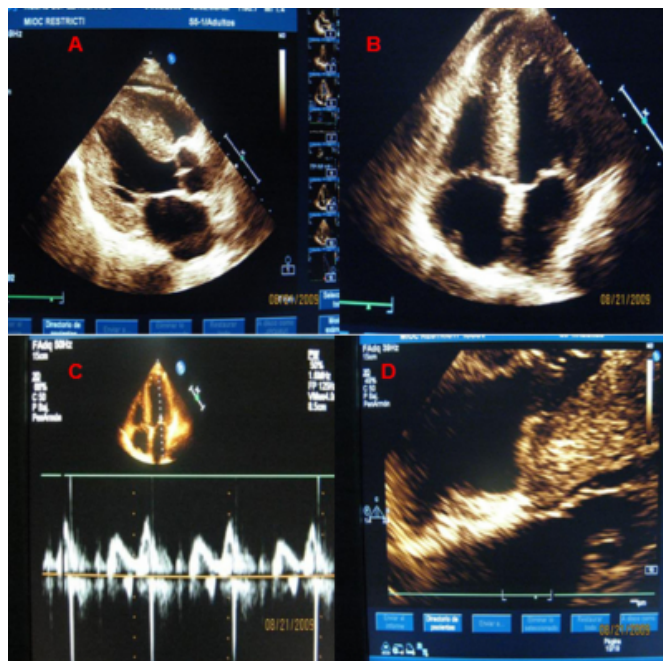
Figure 1

Figure 1. ECG tracing with markedly decreased voltage



Figure 2

Figure 2.



A) Long paraesternal axis view. Important concentric hypertrophy of the left ventricle with a scanty speckled appearance. Observe the increased pericardial echogenicity.

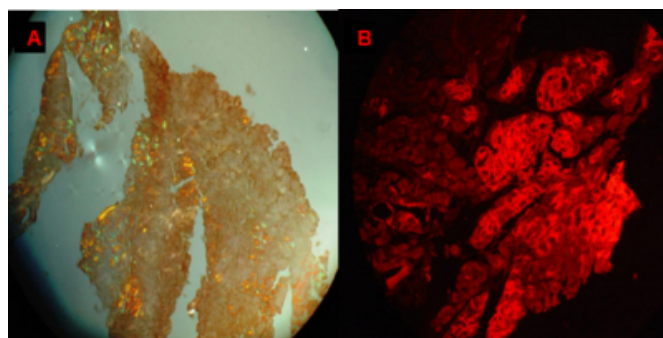
B) Four-chambers apical view. Minimum pericardial effusion in the anterolateral side of the right ventricle. Observe the infiltrative process arising to the lower part of the interauricular septum.

C) Abnormal left ventricular filling typical of diastolic dysfunction type I.

D) Sub-costal view. Increased and echogenic interauricular septum.

Figure 3

Figure 3. Photomicrographs of endomyocardial biopsy



A) In polarized light: Green birefringence identifying part of the amyloid deposition.

B) In fluorescent light: Congo red fluorescence marking amyloid deposition.

DISCUSSION

The diagnosis of cardiac amyloidosis can be difficult because the clinical presentation is similar to that of other cardiomyopathies. The heart is affected pathologically in up to 90% of AL patients, in 50% of whom diastolic heart failure with physical signs of right heart failure is a presenting feature (7). In fact, cardiac amyloidosis frequently presents with rapidly progressive signs and symptoms, such as progressive dyspnoea, peripheral edema, markedly elevated jugular venous pressure and hepatomegaly, almost always associated with evidence of elevated right-sided filling pressure (2), which was the predominant clinical feature of our patient. Other signs and symptoms such as chest discomfort, atrial arrhythmias, dermatological or neurological manifestations were not present.

The most common echocardiographic feature is thickening of the LV wall, particularly in the absence of hypertension. Other features of cardiac amyloidosis include granular myocardial appearance, atrial dilation, thickened valves, small pericardial effusion and diastolic dysfunction progressing to systolic dysfunction. Echocardiography cannot confirm diagnosis in isolation, and the images should be interpreted in the context of the clinical picture and other investigations (7). In that point, we must distinguish that the echocardiography feature of our patient, even if highly suspicious, was not very typical of the entity indeed, perhaps due to the middle stage of the disease.

Added to the echocardiography findings, despite the presence of scanty speckle appearance, and the absence of atrial enlargement (24,5 cm² atrial size) and restrictive transmitral flow pattern, the main clue was the inverse relationship between ECG voltage and LV mass and the continuity of the left ventricular hypertrophy pattern arising the interauricular septum. A low voltage on the ECG and increased septal and posterior LV wall thickness on the echocardiogram are highly specific for cardiac amyloidosis in the setting of biopsy-proven systemic amyloidosis (3). High sensitivity (72% to 79%) and specificity (91% to

100%) have been reported for this combination (7).

Regarding diastolic dysfunction, a study of 36 patients with cardiac amyloidosis showed no restrictive transmitral flow pattern in 22 patients (8) so, it has a variable prevalence and restrictive transmitral flow is observed mainly in advanced stages of the disease.

The reduced by-ventricular systolic function leads us to run a cardiac catheterization in order to search other underlying cause, but the coronary angiography revealed normal epicardial coronary arteries. Even though extensive amyloid deposition in epicardial coronary arteries occurs in patients who have primary cardiac amyloidosis, none obstruction of epicardial vessels by amyloid has been reported (9). The etiological diagnosis of the suspected infiltrative myocardiopathy was finally achieved by the endomyocardial biopsy, which is a safe and relatively simple procedure in skilled hands; and is virtually 100% sensitive because the amyloid is widely deposited throughout the heart (4).

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