

**HEMOCHROMATOSIS REVEALED BY DILATED CARDIOMYOPATHY: REPORT
CASE**

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ABSTRACT

Hemochromatosis is a systemic disorder characterized by the excessive deposition of iron in multiple organs. Myocardial iron-overload whatever its cause, can induce dilated or restrictive cardiomyopathy and cardiac rhythm disorder, often presents with a restrictive cardiomyopathy and usually heart involvement is a very rare presentation. A 39 year old male followed for dilated cardiomyopathy for 7 month presented to Emergency Room in state of respiratory distress. He complained of a respiratory gene that had been evolving for 15 days, orthopnea and leg edema. After intensive medical treatment, the patient was stabilized and benefited from explorations diagnosing hemochromatosis and he is program for heart transplant.

KEYWORDS: Hemochromatosis, dilated cardiomyopathy, Heart failure.

INTRODUCTION

Hemochromatosis is a heritable heterogeneous disease, characterized by an increase in iron absorption from the gut inappropriate to body iron stores, resulting in a progressive accumulation of iron in tissues, especially in the liver and pancreas. Cardiac involvement is less frequent, nevertheless can also occur, causing arrhythmias and in rare cases restrictive or cardiomyopathy dilated. Knowledge and understanding of the early features of the condition, often nonspecific, and of the diagnostic route are necessary to detect iron overload and diagnose hemochromatosis before irremedial damage has been done. We report here a rare case of a 39-year-old patient with haemochromatosis, unfortunately diagnosed late, whose cardiac function did not improve even after medical treatment and he suffered from recurrent heart failure.

CASE REPORT

A 39-year-old man with a 4-year history of type II diabetes mellitus treated with insulin with periodic check-ups, and 7-

month history of dilated cardiomyopathy treated with diuretics, came to the emergency service after 2 week of progressive dyspnea, orthopnea, leg edema, and increasing abdominal girth. On physical examination, the patient had brownish-gray skin pigmentation. On admission, he was tachypneic with blood pressure of 9/6 mmHg and regular heart rate of 82 beats per minute. He had jugular venous distension, bilateral pulmonary crackles, IV / VI systolic murmur heard at the apex, ascites, and pitting edema in both lower legs. The ECG showed sinus tachycardia, frequent ventricular extrasystoles and repolarization abnormalities. The echocardiogram showed a dilated left ventricle with severely compromised systolic function (left ventricular ejection fraction, 10%) and severely mitral insufficiency without elevation of the pressures of the right cavities (figure 1). A chest radiograph (Figure 2) revealed cardiomegaly and pleural effusion.

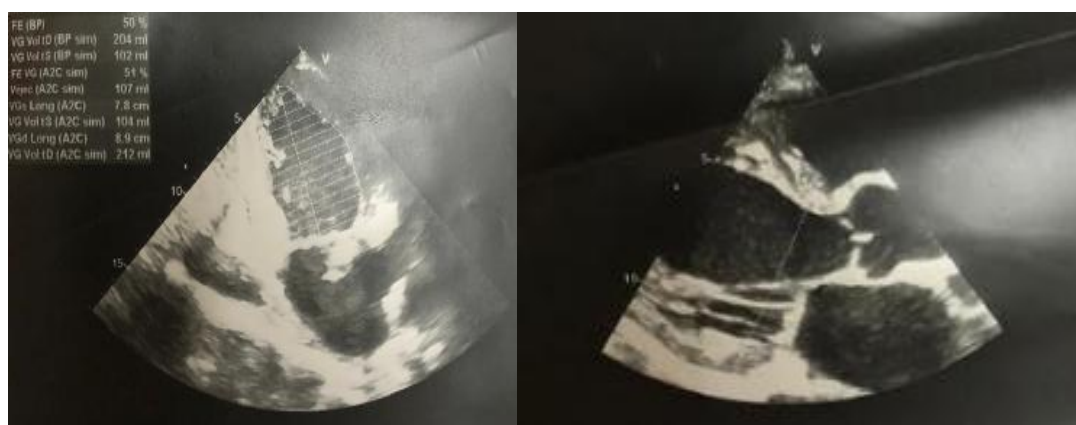


Figure 1: The echocardiogram showed a dilated left ventricle with severely compromised systolic function and severely mitral insufficiency without elevation of the pressures of the right cavities.



Figure 2. Chest radiographs at first admission showing cardiomegaly and pleural effusion.

Laboratory findings included increased levels of urea 11.4 (2.5–6.4) mmol/L, with normal levels of troponin hemoglobin 14g/dl, glycemia 0,78 g/l and creatine 9,9mg/l. Hepatobiliary enzymes were elevated as follows; aspartate aminotransferase 121 U/L, alanine aminotransferase 64 U/L, alkaline phosphatase 372 U/L, gamma-glutamyl transpeptidase 81 U/L and total bilirubin 21 (<17) μ mol/L. Procalcitonin and C-reactive protein was normal 0,28 ng/ml and 8mg/l. Serum ferritin, serum iron, and transferrin saturation levels were markedly elevated to 6208 ng/mL, 216 μ g/mL, and 87%, respectively. Iron overload was treated by phlebotomy, without improvement of cardiac function and the patient is programmed for cardiac transplantation.

DISCUSSION

HH is a common heritable disease, characterized by an increase in iron absorption from the gut inappropriate to body iron stores, resulting in a progressive accumulation of iron in tissues, especially in the liver and pancreas. Cardiac involvement is less frequent, nevertheless can also occur, causing arrhythmias and in rare cases restrictive or DCM. The most frequent form of primary hemochromatosis is inherited as an autosomal recessive disease, often caused by the HFE (C282Y) gene mutation. Family screening is recommended for all first degree relatives of an individual with the disease. Secondary hemochromatosis occurs due to ineffective erythropoiesis secondary to a defect in hemoglobin synthesis and chronic and massive iron overload such as blood transfusion. Unfortunately, the means are limited in our case, we could not genetically prove this heterozygous mutation. The most common symptoms of HH are subjective and non-specific, such as fatigue, lethargy and arthralgia. In cases of significant iron overload, individuals may present with organ specific symptoms such as those related to cardiac disease. In this case, he was suspected to be hemochromatosis based on the findings of iron

metabolism including elevated plasma iron and serum ferritin levels, and especially marked elevation of saturation of transferrin. Iron overload may be identified as part of the evaluation of an asymptomatic individual with abnormal liver function tests. Additionally, an increasing number of cases are detected as a result of cascade family screening. Cardiac hemochromatosis usually occurs in advanced disease and its severity depends on the amount of iron deposited in the myocardium. Treatment of HH with clinical iron overload involves a combination of phlebotomy and/or chelation therapy. Cardiac transplantation should be considered for patients with HH-related decompensated cardiopathy. The prognosis is poor due to progressive congestive heart failure and refractory arrhythmias. Early initiation of iron chelation therapy has been reported to improve the outcome.

CONCLUSION

Hereditary hemochromatosis is characterized by an excessive absorption and progressive accumulation of iron in the liver, the pancreas, the heart, and the joints. In summary, we report a rare case of fatal cardiac hemochromatosis associated with HS. The possibility of cardiac hemochromatosis needs to be considered in cases of heart failure or arrhythmia in patients with HS.

Disclosures

None.

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