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CLINICAL CASE

Restrictive cardiomyopathy: a pediatric case report

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Summary

Introduction: Restrictive cardiomyopathy (RCM) is a rare condition with a poor prognosis in pediatric patients. It is the least common variant within cardiomyopathies and, in most cases, its etiology is unknown. It has a dire prognosis unless a heart transplant can be performed.

Case: Here, we report a 1-year-old boy with a history of respiratory infections and dyspnea when suckling his mother's breast. He was admitted with dyspnea, cyanosis, and edema. Physical examination describes the presence of a grade IV/VI systolic murmur at the left sternal border. The respiratory rate was 60 per minute with the presence of intercostal, subcostal, suprasternal retraction as well as hepatomegaly.

Evolution: Laboratory data showed the presence of leukocytosis and normochromic normocytic anemia. Chest radiography showed grade IV cardiomegaly. The echocardiogram showed atrial growth and the presence of patent foramen ovale. There was preserved systolic function with a restrictive relaxation pattern and pulmonary hypertension (47 mmHg). Magnetic resonance imaging of the heart revealed enlargement of the atrial chambers with normal myocardial thickness. The subject was managed as restrictive cardiomyopathy with furosemide, spironolactone, and antiplatelet agents. The patient was discharged with the same indications for outpatient consultation.

Conclusion: Radiography and electrocardiogram can provide important data for diagnosis. The echocardiogram remains the best study for this diagnosis. The use of functional measurements such as tissue Doppler can help to show early diastolic failure.

Key words: Cardiomyopathy, Restrictive; Endomyocardial Fibrosis; Heart failure; Child; Case Reports.

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Introduction

Restrictive cardiomyopathy (RCM) is a primary disease of the myocardium [1]. It is characterized by diastolic dysfunction with preserved systolic function, atrial dilation, and the absence of ventricular hypertrophy or dilation [2, 3]. Its etiology can be idiopathic or secondary to systemic diseases [4].

It is the rarest form of cardiomyopathy because it constitutes around 5% of cases. It has a poor prognosis [1, 5].

Clinical case

We present a one-year-old male patient from Santo Domingo de los Tsachilas-Ecuador who had the following personal history: Prenatal History: 40-year-old pregnant mother with no pathologies during gestation and with 3 pregnancies, 1 abortion, and 1 caesarean section. A fetal heart rate of 144 bpm was determined during an obstetric evaluation at the last trimester. The gestational age was 39 weeks, and the placenta was maturation grade III. A caesarean section was indicated due to transverse fetal position with fetal macrosomia. The postnatal history was Apgar 9/9, weight 3200 grams, and height 50 cm. He received the full immunization schedule. He was hospitalized for pneumonia at 10 months of age.

No relevant pathological family history was reported for his parents or siblings.

The mother reported that the patient has a history of frequent respiratory infections and recurrent episodes of dyspnea when suckling the mother's breast since he was 5 months of age.

The reason for consultation is cough with two weeks of evolution. One week later the clinical picture worsened with dyspnea, fever, cyanosis, weakness, and edema in the extremities.

Physical examination showed polypnea with intolerance to decubitus position. The heart rate was 110 beats per minute, heart sounds are rhythmic, and a third heart sound is auscultated. A grade IV/VI systolic murmur at the left sternal border was described.

The respiratory rate was 60 per minute with the presence of intercostal, subcostal, and suprasternal retraction. The respiratory sounds were decreased with wheezes and crackles; hepatomegaly was palpated in the abdomen.

Diagnostic workshop

Complementary tests were performed upon admission:

- Biochemistry: urea 35 mg/dl; creatinine 0.4 mg/dl; TGO 61 U/L (H); TGP 92 U/L; bilirubin T/D/I: 0.7/0.5/0.2 mg/dl; alkaline phosphatase 158 IU/L; sodium 135 mEq/L, potassium 4.6 mEq/L, chloride 100 mEq/L.
- Hematic biometry: Leukocytes 14.2 x 103 cells/µl, eosinophils 1.1%, basophils 0.4%, neutrophils 55.4%, lymphocytes 37.4%, monocytes 5.7%, red blood cells 4.4 x 106 cells/mm3, hemoglobin 11.6 g/dl, hematocrit 37.2%, mean corpuscular volume 84 fL, mean hemoglobin concentration 26.1 pg, mean globular diameter 14.1%, platelets 500 x 103 cells/mm3, mean platelet volume 8.7 µm3.

A chest radiography was performed (**Fig. 1**) in which cardiomegaly was reported at the expense of atrial enlargement and impaired pulmonary flow due to passive congestion. In addition, the electrocardiogram (**Fig. 2**) reported biatrial hypertrophy.

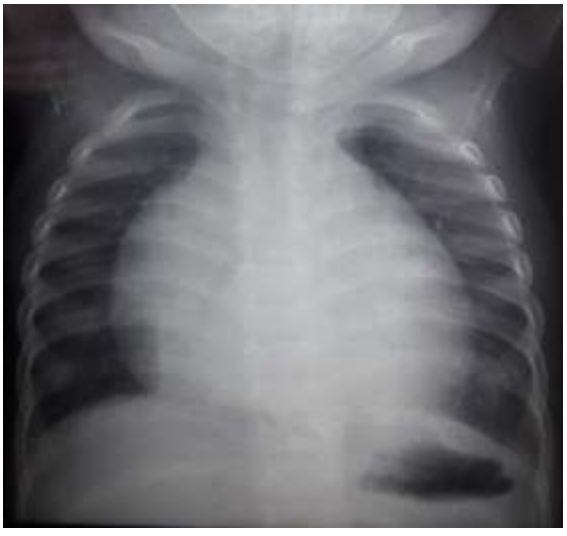

Fig. 1 Chest x-ray at admission showed cardiomemegaly.

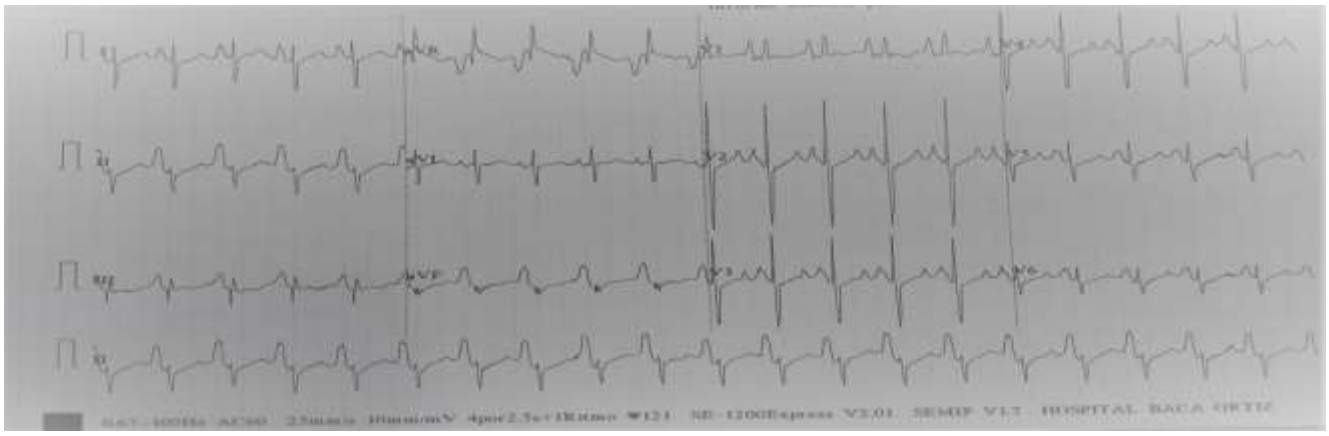

Fig. 2 Electrocardiogram: sinus rhythm, biatrial hypertrophy and giant P waves

The echocardiogram showed an increase in the size of the atrial cavities (**Fig. 3, 4 and 5**) with normal sized ventricles: The right atrium was 37 mm with an area of 15.2 cm^2, and the left atrium was 42 mm with an area of 17.5 cm^2. The inferior vena cava did not have inspiratory collapse. The presence of an interatrial defect was detailed in the form of a patent foramen ovale. Biventricular systolic function was preserved. There was a restrictive relaxation pattern, and the presence of mild pulmonary hypertension with pulmonary artery systolic pressure of 47 mmHg. The mitral and tricuspid atrioventricular valves presented moderate insufficiency and the presence of mild pulmonary insufficiency is detailed.

Simple and contrasted nuclear magnetic resonance imaging of the heart and great vessels revealed the growth of atrial cavities; the right atrium measured 4.5 x 5.1 cm in its transverse and anteroposterior axes; the left atrium measures 4.5 x 4.7 cm on these same axes respectively. The thickness of the myocardium was maintained. Pathological impregnations suggesting deposition disease or fibrosis were not seen. Discreet amounts of pericardial fluid were highlighted. The pericardium had a normal thickness without pathological enhancement.

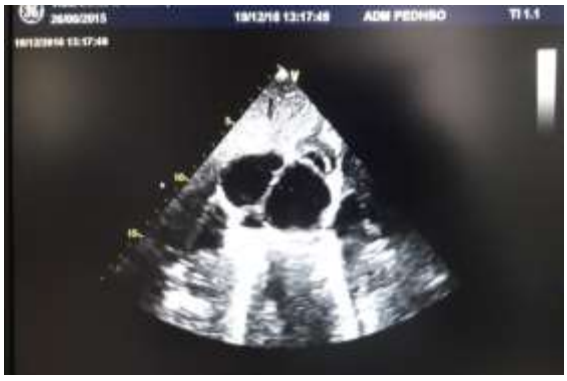

Fig. 3 Echocardiogram: Atria. Two-dimensional image showing biatrial hypertrophy.

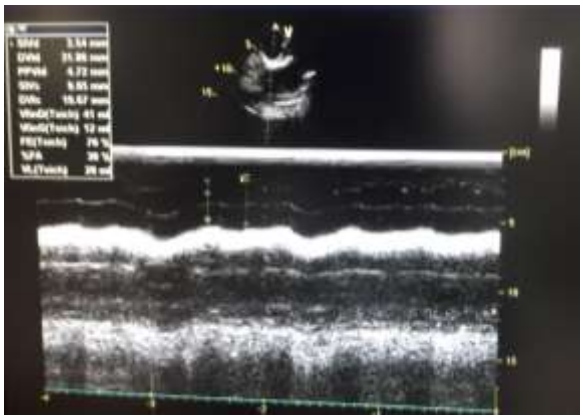

Fig. 4 M-mode ultrasound.

Fig. 5 Mitral Doppler study evaluating diastolic function.

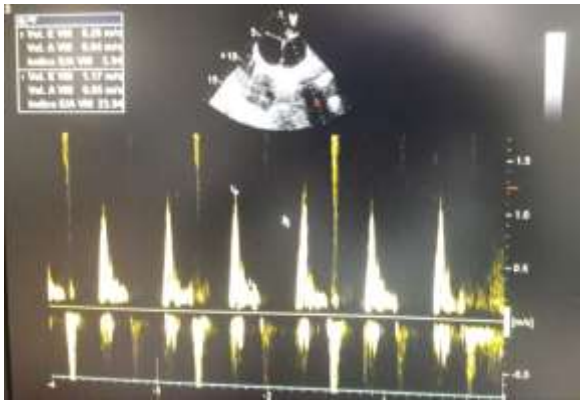

Fig. 5 Mitral Doppler study evaluating diastolic.

Clinical evolution

Symptoms of heart failure were the reason for hospital admission. Complementary studies including diagnosis of restrictive cardiomyopathy were established so that treatment with diuretics could be started. Intravenous Furosemide until clinical stabilization was performed first. This then continued orally with spironolactone. Prophylactic antiplatelet therapy was also initiated due to the presence of dilated atria and predisposition to thrombus formation.

Outcome

The patient was discharged with diuretics: furosemide, spironolactone, and aspirin. Outpatient consultations were established, and a heart transplant was considered.

Discussion

RCM is a rare pathology that represents between 3% and 5% of all cardiomyopathies. It is characterized by restrictive filling of both ventricles with diastolic dysfunction and preserved systolic function. The most common etiology in the pediatric age is idiopathic. During adulthood, the causes secondary to systemic diseases are more common including amiloidosis, sarcoidosis, Andersen's disease, Fabry disease, and Löffler's syndrome [4].

In cases of idiopathic and familiar restrictive cardiomyopathy, there are mutations in the genes TNNI2, TNNI3, and ACTC associated with sarcomeric protein diseases [5].

In RCM, there is biventricular diastolic alterations with preserved systolic function. The filling of the ventricles is altered, which leads to a disproportionate increase in ventricular end-diastolic pressure in response to small volumes of blood; this in turn causes dilation of both atria.

Macroscopically, dilated atria and ventricles of normal size are observed as demonstrated by echocardiography and cardiac magnetic resonance in our patient.

The ventricular wall of these patients is abnormal with a rubbery texture and with thickening of the endocardium. Microscopically, changes dependent on the infiltration material are seen in secondary causes [6], which did not occur in here.

The clinical picture of this pathology is variable with predominant findings of systemic venous congestion [7] with varying degrees of pulmonary hypertension due to impairment of the left atrial emptying [8, 9]. The presence of symptoms such as dyspnea, tachypnea, and tachycardia are common and are often confused with respiratory symptoms [9].

This coincides with a history of repeated respiratory processes in our subject who even had a hospitalization for pneumonia.

Physical examination showed clinical signs of heart failure; hepatomegaly is the most constant sign as well as auscultation of the third and fourth heart sounds.

Radiography can characterize the heart. As the disease progresses, cardiomegaly appears due to atrial dilation (the most frequent finding) [5, 6]. The current case was considered advanced disease because it had approximately 7 months of evolution.

The electrocardiogram shows biatrial dilatation manifested by high and biphasic P waves, which was a striking feature on the patient's ECG. The QRS is narrow and does not present alterations. Some patients show ST segment depression without evidence of ischemia and the T wave that may be negative [6]. Supraventricular arrhythmias may appear due to atrial dilation such as atrial extrasystoles or atrial fibrillation.

The echocardiogram must rule out any structural abnormality and differentiate it from other cardiomyopathies. Typical findings are atrial dilation, diastolic

dysfunction, preserved or close to normal systolic function, and the absence of ventricular dilation. Some authors refer to these data as "cavity inversion" [7-9].

Treatment should focus on alleviating congestive symptoms with diuretics and water restriction. The use of digitalis is reserved for cases with impaired systolic function and atrial fibrillation. The use of anticoagulants should be considered due to the high frequency of thrombi associated with atrial fibrillation and atrial dilation [8]. Cases secondary to metabolic diseases should receive specific treatment.

The prognosis of the disease is very poor; survival rates two years after the diagnosis is lower than 50% and mortality increases in cases with syncope [9]. Other markers of poor prognosis are radiographic evidence of pulmonary congestion and alteration of the ratio of the diameter of the left atrium and the aortic root [10, 11].

In idiopathic cases (most frequent in children), transplantation is often the only option. Survival rates at one year have been reported at 86% and 50% at two years after diagnosis [12, 13]. Some studies indicate a mortality of 100% [14] or the need for transplantation in the first 5 years after diagnosis [15, 16]. Sudden death occurs in up to 28% of cases with an annual mortality of 7% [8].

Conclusions

Our data show are similar to those described in the literature. The symptoms of heart failure such as dyspnea and hepatomegaly are typical of this disease. Radiography and electrocardiography can provide important diagnostic data. Echocardiogram remains the best study for diagnosis. The use of functional measurements such as tissue Doppler can help to show early diastolic failure. In addition, it is important to perform laboratory studies or biopsies of different organs especially the myocardium to rule out tissue storage pathologies (storage diseases).

As the disease progresses, the most obvious findings are atrial dilation and the restrictive pattern. Heart transplantation could be the main therapeutic alternative and this may be the most suitable option here.

Abbreviations

RCM: Restrictive Cardiomyopathy.

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Authors' contributions

MDCFD: Research idea, article writing, critical analysis, editorial corrections.
DIRR: Data compilation, Bibliographic review.
AMPM: Research Idea, Article Writing, Critical Analysis.
All authors read and approved the final version of the manuscript

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Availability of data and materials

Data sharing is not applicable.

Ethical statements

Protection of persons

The authors declare that the procedures followed were in accordance with the ethical standards of the responsible human experimentation committee and in accordance with the World Medical Association and the Declaration of Helsinki.

Confidentiality of the data

The authors declare that they have followed the protocols of their work center on the publication of patient data.

Consent for publication

The authors have obtained the informed consent from the guardians of the patient referred to in the article. This document is in the possession of the corresponding author. The parents have signed the authorization for publication of this case.

Competing interests

The authors have no competing interests to declare.

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