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CASE REPORT

Left ventricular noncompaction: A rare indication for pediatric heart transplantation☆

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KEYWORDS
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Pediatric heart transplantation;
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Abstract Isolated left ventricular noncompaction is a rare congenital cardiomyopathy, characterized morphologically by a dilated left ventricle, prominent trabeculations and deep intertrabecular recesses in the ventricular myocardium, with no other structural heart disease. It is thought to be secondary to an arrest of normal myocardial compaction during fetal life. Clinically, the disease presents with heart failure, embolic events, arrhythmias or sudden death. Current diagnostic criteria are based on clinical and imaging data and two-dimensional and color Doppler echocardiography is the first-line exam. There is no specific therapy and treatment is aimed at associated comorbidities. Cases refractory to medical therapy may require heart transplantation.

The authors describe a case of severe and refractory heart failure, which was the initial presentation of isolated left ventricular noncompaction in a previously healthy male child, who underwent successful heart transplantation.

PALAVRAS-CHAVE
Ventrículo esquerdo não compactado;
Transplante cardíaco pediátrico;
Insuficiência cardíaca refractária

Ventrículo esquerdo não compactado: causa rara de transplante cardíaco

Resumo A não compactação isolada do miocárdio ventricular é uma causa rara de miocardiopatia congénita primária, caracterizada morfologicamente por dilatação do ventrículo esquerdo, padrão multitrabecular proeminente e recessos profundos intertrabeculares no miocárdio ventricular, na ausência de outras cardiopatias estruturais. Parece resultar da

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cessação intrauterina do processo de compactação miocárdica durante a embriogénese. Clinicamente, apresenta-se com sintomas ou manifestações de insuficiência cardíaca, eventos cardioembólicos, disritmias ou morte súbita. Os critérios de diagnóstico baseiam-se em achados clínicos e imagiológicos, sendo o ecocardiograma bidimensional e *Doppler* o meio complementar de diagnóstico de eleição. Não existe tratamento específico para esta entidade, sendo dirigido às comorbilidades associadas. O transplante cardíaco poderá ser uma possibilidade terapêutica em casos refratários ao tratamento médico.

Os autores descrevem um caso clínico de insuficiência cardíaca refratária como forma de apresentação inaugural de não compactação do miocárdio ventricular numa criança do sexo masculino previamente saudável, submetida com sucesso a transplantação cardíaca.

Introduction

Isolated left ventricular noncompaction (LVNC) or myocardial noncompaction is a rare congenital cardiomyopathy.[1,2] It is thought to be secondary to an arrest of normal myocardial compaction during fetal life and is characterized morphologically by left ventricular (LV) dilatation, prominent myocardial trabeculations and deep intertrabecular recesses, giving the myocardium a spongy appearance.[3,9]

It has been described as an isolated finding or more often in association with other primary heart disease, particularly cyanotic congenital heart defects.[9,10]

The natural history of LVNC is varied; patients may remain asymptomatic or progress to congestive heart failure (HF). At pediatric ages, progression to LV systolic and diastolic dysfunction is almost inevitable, occurring in around 90% of affected children. The associated complications, including systemic thromboembolism and arrhythmias, which are linked to high morbidity and mortality, as well as sudden death, are mainly seen in adulthood.[3,11]

The authors describe a case of this rare disease, which can be difficult to recognize, in a male child with isolated LVNC who underwent successful heart transplantation.

Case report

A 10-year-old boy, white, previously healthy, was admitted to the emergency department due to cough, repeated vomiting and abdominal pain. He reported fatigue with moderate to vigorous exertion (NYHA functional class II) over the previous month. His parents were non-consanguineous, but there was a family history of unexplained sudden death (mother and uncle, grandfather and great-aunt on the mother's side).

At admission, the patient was apyretic, normotensive (blood pressure 100/66 mmHg), tachycardic (heart rate 150 bpm) and tachypneic (respiratory rate 37 cpm). His skin and mucous membranes were pale and there was a grade II/VI systolic murmur over the left sternal border on cardiac auscultation, and hepatomegaly with a palpable liver edge around 3 cm from the right costal margin. No dysmorphic facial features or neuromuscular abnormalities were observed.

The electrocardiogram (ECG) showed narrow-complex tachycardia, with heart rate between 150 and 180 bpm, that did not respond to administration of adenosine, consistent with ventricular tachycardia. The chest X-ray revealed cardiomegaly (cardiothoracic index >0.6, Figure 1) and laboratory tests showed elevated transaminases (GOT 94 UI/l, GPT 72 UI/l) and BNP (742 pg/ml) and hyponatremia (133 mEq/l). Markers of myocardial necrosis were negative. Transthoracic echocardiography (Figure 2A and B) showed LV dilatation and severe systolic dysfunction (end-diastolic diameter 65 mm, z-score +7.8; end-systolic diameter 60 mm, z-score +12.3; ejection fraction 20%; and fractional shortening 12%). Prominent trabeculations and deep intertrabecular recesses filled with blood were visualized in the LV lateral wall and apical region; the ratio between the thickness of the noncompacted and compacted layers

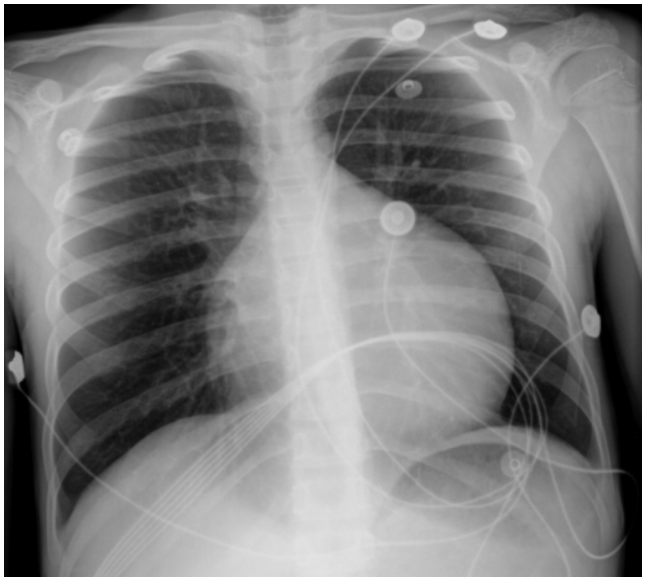

Figure 1 Chest X-ray showing increased cardiothoracic index and signs of pulmonary vascular congestion.