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Brief Report

A fatal case of idiopathic restrictive cardiomyopathy

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Abstract We describe the clinical features of idiopathic restrictive cardiomyopathy in a female infant. A marked elevation of left ventricular end-diastolic pressure, and profoundly abnormal myocardial relaxation, were detected with the use of Doppler blood flow echocardiography, coupled with the relatively new technique of Doppler tissue echocardiography. There was no clinical evidence of ongoing heart failure, but she had signs of myocardial ischaemia, and unfortunately died suddenly at the age of 13 months.

Keywords: Childhood restrictive cardiomyopathy; diastolic dysfunction; Doppler tissue echocardiography

RESTRICTIVE CARDIOMYOPATHY HAS BEEN defined as a myocardial disorder characterized by impaired ventricular filling, with either normal or decreased ventricular volume, and normal or increased ventricular mural thickness.[1–3] As with the other cardiomyopathies with predominantly diastolic dysfunction, ventricular systolic function remains unchanged until the later stages of progression of the disease. Idiopathic restrictive cardiomyopathy is a rare primary myopathic disorder, and is diagnosed in the absence of any recognizable underlying myocardial disease.[3] Microscopic examination of the myocardium in this setting demonstrates varying degrees of non-specific interstitial fibrosis, usually combined with myocytic hypertrophy, albeit that the latter feature can be found in isolation.[3] This particular cardiomyopathy is more commonly found in girls, and the prognosis is worse in childhood than in adults.[4] We describe here our experience with a recent case, emphasising the value of Doppler echocardiography, particularly the tissue Doppler technique.

Case report

A 3-month-old girl was referred for cardiac assessment with tachypnoea and a systolic murmur. The physical examination was limited by the irritability of the patient. She was acyanotic and well perfused, with normal pulses in all limbs. Her liver was palpable 2 cm below the right costal margin. Her chest was clear on auscultation. The rate of respiration was 60 breaths/min. She had a gallop rhythm, with third and fourth heart sounds. There was a soft pansystolic murmur heard at the apex, which radiated towards the axilla. Her electrocardiogram showed sinus rhythm, with a rate of 120 beats/min. The tracings showing marked bi-atrial voltage changes, consistent with atrial hypertrophy, and widespread ischemic changes, notably depression of the ST segments and inversion of the T waves. The chest X-ray demonstrated a normal cardiothymic shadow and clear lung fields. Her karyotype was normal.

The echocardiogram showed her left ventricle to be of normal size with preserved systolic function, the ejection fraction being 55%. The left ventricular mural thickness, at 5 mm, was at the upper limit of normal. The myocardium itself was of normal echogenicity, and the left atrium was only mildly enlarged, having a diameter of 17 mm (Fig. 1), with mild mitral regurgitation. The right ventricle was of normal size with good systolic function. There was no pericardial effusion. Doppler indexes of mitral inflow were consistent with an abnormal pattern of relaxation rather than a restrictive pattern of filling (Fig. 2). In addition, velocities of pulmonary venous flow suggested an elevated left ventricular end-diastolic pressure with dominant atrial reversal velocity. Colour M-mode Doppler tissue echocardiography of the left ventricular posterior wall showed a

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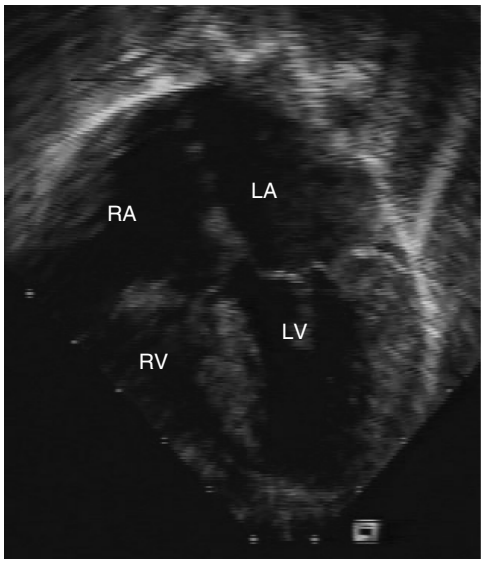

Figure 1.
The apical four chamber cross-sectional echocardiogram. LV: left ventricle; RV: right ventricle; LA: left atrium; RA: right atrium.

marked reduction in the myocardial velocity gradient particularly in the period of rapid left ventricular filling in early diastole just after the opening of the mitral valve. The myocardial velocity gradient was also pathologically positive during the second part of the period of isovolumic relaxation. As in the phase of isovolumic relaxation phase all intracardiac valves are closed, changes in the myocardial velocity gradient are related to the direct active subepicardial motion, which was significantly impaired in our patient. These findings suggest a virtually total lack of active early diastolic left ventricular relaxation, and are consistent with the diagnosis of restrictive cardiomyopathy.[5,6] Interestingly, there was abnormally high and prolonged myocardial velocity gradient in late diastole during atrial contraction, suggesting that left ventricular myocardial relaxation was forced by the external force of "hyper-normal" atrial contraction.

Cardiac catheterisation revealed elevation of both atrial pressures, with the left atrial a wave measured at 29 mmHg, and the right atrial a wave at 15 mmHg.

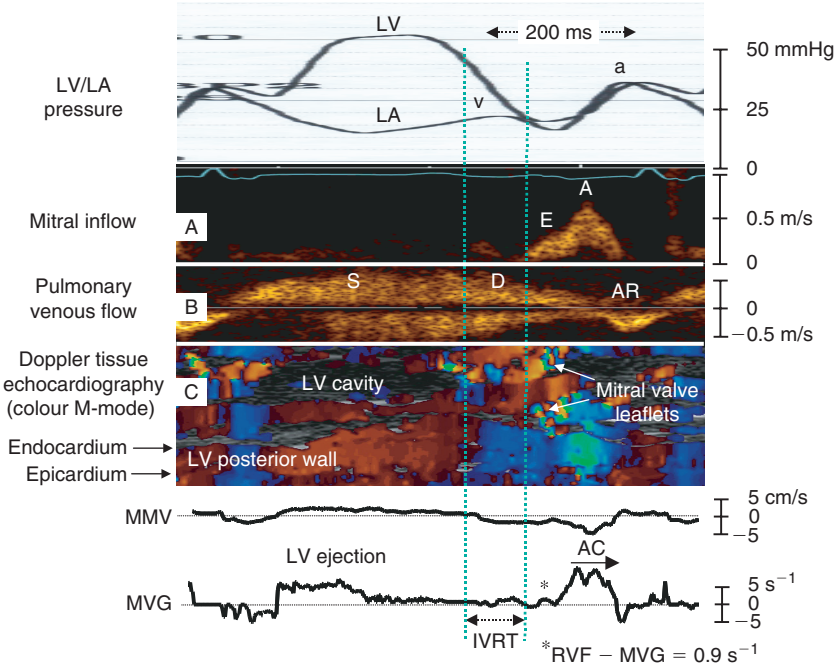

Figure 2.
Top *– Hemodynamic data, with simultaneously recorded left ventricular (LV) and left atrial (LA) pressure tracings during one cardiac cycle. Left ventricular diastolic waveform showed a dip and plateau pattern and the left atrial pressure was markedly elevated with predominant a-wave. There was also prolonged left ventricular pressure decline during isovolumic relaxation.* ***A*** *– Doppler mitral inflow with abnormal relaxation pattern, E/A ratio 0.5 (E-wave, early diastolic and A-wave, late diastolic inflow velocity);* ***B*** *– pulmonary venous flow with high (45 cm/s) and delayed ("shift to the right") atrial reversal flow (AR) (S-wave, systolic, D-wave diastolic velocities) and* ***C*** *– colour M-mode Doppler tissue echocardiography of left ventricular posterior wall with quantification of mean myocardial velocities (MMV) and myocardial velocity gradient (MVG). Note markedly reduced myocardial velocity gradient during rapid ventricular filling (RVF) 0.9 s^{-1} (normal range, 8.0–18.1 s^{-1}) that suggests a virtually lack of early diastolic myocardial relaxation. Myocardial velocity gradient was also abnormal (positive rather than negative) during isovolumic relaxation (1.2 s^{-1}). The myocardial velocity gradient during atrial contraction (AC) was abnormally prolonged and high at 10 s^{-1} (normal range 0–1 s^{-1}) suggesting that left ventricular myocardial relaxation was forced by atrial contraction (external force). Dotted vertical green lines show isovolumic relaxation time (IVRT). Asterisk (*) indicates peak myocardial velocity gradient measured during rapid ventricular filling. mmHg: millimeters of mercury; cm/s: centimeters per second; m/s: meters per second; s^{-1}: second to the power of minus one.*

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Right ventricular systolic pressure was normal, at 32 mmHg, with no systolic pressure drop in the pulmonary trunk or its branches. Left ventricular systolic pressure was relatively low, with no systolic pressure drop to the ascending or descending segments of the aorta. The left ventricular diastolic waveform showed a dip and plateau pattern. Left ventricular end-diastolic pressure was elevated to 20 mmHg. These findings were consistent with marked left ventricular restrictive physiology.[1–5] Oximetry did not demonstrate any shunts. Angiography showed normal coronary arterial anatomy. The aortic arch and aortic valve were normal. Magnetic imaging revealed no evidence of pericardial thickening.

She underwent a number of different examinations, including genetic and metabolic assessments such as a mucopolysaccharide screen. Results of all these examinations were normal, and hence the restrictive cardiomyopathy was considered to be idiopathic. Her clinical status was stable, and repeated echocardiograms did not demonstrate significant changes. Her symptoms of heart failure were minimal, and she required only small doses of diuretics. Sadly she died suddenly at the age of 13 months.

Discussion

The diagnosis of restrictive cardiomyopathy is usually made at an advanced stage of the disease process, typically when the functional and structural changes of the left ventricle are irreversible. Hence, cardiac transplantation has been recommended as the treatment of choice.[7] It is rare for the idiopathic variant of restrictive cardiomyopathy to be diagnosed in the first year of life.[4] Surprisingly, in our patient, the pattern of inflow through the mitral valve showed abnormal relaxation rather that a restrictive pattern.[8] This may be partially explained by the use of diuretics, which may have reduced the filling pressure, and therefore reduced the velocity of the mitral E-wave. Our patient, however, was receiving no medications during the initial echocardiographic examination. The most likely explanation, therefore, is that, during childhood, the haemodynamics of restrictive cardiomyopathy are caused not only by an increased intrinsic stiffness of the ventricular wall, reducing the compliance,[8] but also may result from serious dysfunction and delay of the active myocardial relaxation.[9] The left ventricular pressure curve showed a steady prolonged decline during early-diastolic filling. There was significant left ventricular filling, nonetheless, in mid- and late-diastole, as evidenced by the prominent A-wave seen on the Doppler traces of mitral inflow. This implies that the driving force for left ventricular filling was the increase in the left atrial pressure rather than suction from the left ventricle.[9] The increased velocity of the mitral A-wave, coupled with the high and prolonged myocardial velocity gradient during atrial contraction, emphasized a significant role for atrial systolic function in maintaining both sufficient left ventricular relaxation and adequate left ventricular end-diastolic volume. This suggests that the restrictive cardiomyopathy was in a relatively early stage of its progression, an interpretation also favoured by the mild dilation of the left atrium and the relatively asymptomatic clinical course. Although she was still an infant, we considered her for cardiac transplantation, mainly because of the evidence of extensive myocardial ischaemia seen in her resting electrocardiogram. We were also swayed by a report[10] emphasising the risk of sudden death in girls with restrictive cardiomyopathy who appear well, with no evidence of ongoing heart failure, but with signs or symptoms of myocardial ischaemia. The option of cardiac transplantation was discussed at length with her parents, but was declined. Subsequent to this, unfortunately, she died suddenly at the age of 13 months. There was no post-mortem examination.

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