Restrictive Cardiomyopathy in a Child

Shan-Miao Lin¹, Haw-Kwei Hwang¹, Ming-Ren Chen¹,²*

¹Department of Pediatrics, Mackay Memorial Hospital, Taipei, Taiwan
²Mackay Medicine, Nursing and Management College, Taipei, Taiwan

Received: Jan 11, 2008
Revised: Feb 14, 2008
Accepted: Mar 25, 2008

KEY WORDS:
cardiac catheterization; child; restrictive cardiomyopathy

1. Introduction

Restrictive cardiomyopathy is a rare entity in childhood and carries a poor prognosis. Because it is so uncommon, most of our understanding of its clinical and hemodynamic manifestations derives from observations in adults. However, there may be certain distinct features that are specific to its presentation in early childhood. Here, we report a pediatric patient who presented with hepatomegaly as an initial manifestation of restrictive cardiomyopathy.

2. Case Report

An 18-month-old girl suffered from progressive abdominal distension and poor feeding for 2 months. Her perinatal course was uneventful, and there was no history of prior viral infection causing heart failure. Physical examination revealed hepatomegaly that was confirmed by abdominal ultrasound. No heart murmur was audible, and there was no significant peripheral edema. Chest radiography showed slight cardiomegaly, but electrocardiography revealed severe bilateral atrial enlargement and QRS-T discordance (Figure 1). Two-dimensional echocardiography showed dilatation of both atria and a slightly thickened interventricular septum (Figure 2A). The left ventricular (LV) ejection fraction was 0.58, and the left atrial-to-aortic root ratio (LA:Ao) was 1.69. Both ventricles were of normal size (Figure 2B). A small pericardial effusion was present, and the inferior vena cava and hepatic vein were markedly dilated. There was no systolic anterior motion of the mitral valve. Doppler interrogation revealed moderate tricuspid and mitral regurgitation, decreased deceleration of the mitral (70 ms) and tricuspid (80 ms) inflows, faster than normal isovolumic relaxation of the right ventricle (55 ms), and increased E/A ratios of both the mitral (Figure 2C) (E, 91.1 cm/s; A, 26.6 cm/s; E/A, 3.42) and tricuspid (E, 56.8 cm/s; A, 22.2 cm/s; E/A, 2.56) inflows. Cardiac magnetic resonance imaging (MRI) showed no thickening of the pericardium. The child had mild liver dysfunction (aspartate aminotransferase, 46 IU/L) with a prolonged prothrombin...
time of 15.6 seconds. Cardiac catheterization showed elevated end-diastolic pressure in both ventricles (right ventricle [RV], 13 mmHg; LV, 24 mmHg). The RV pressure tracing demonstrated a characteristic square root sign (an early diastolic dip with a rapid rise to an elevated plateau) and lack of respiratory variation (Figure 3). The pulmonary artery pressure was 21/16 mmHg. No coronary artery abnormality or other cardiac anomaly was detected. Results of metabolic screening tests were normal. We could not identify any infiltrative disease that would explain the restrictive cardiomyopathy this child had, although we did not perform endomyocardial biopsy because the patient was so young.
She was maintained on aspirin to prevent atrial thrombus while awaiting cardiac transplant.

3. Discussion

The mean ages at diagnosis of restrictive cardiomyopathy in two small pediatric series (each with 8 patients) were 6.3 and 4.0 years, respectively, with the youngest child in the latter series presenting at the age of 1.3 years.1,2 All of the reported patients presented with evidence of congestive heart failure, either systemic venous congestion or pulmonary edema. Other common initial symptoms of restrictive cardiomyopathy in children include failure to thrive, fatigue and syncope.3 Our patient was diagnosed at 1.5 years of age and, aside from nonspecific symptoms, was unusual in having only abdominal distension.

Restrictive cardiomyopathy accounts for only approximately 2–5% of all pediatric cardiomyopathies.2,4 Because the number of reported patients is so small,1,2 findings in adult disease are often extrapolated to guide clinical evaluation and management of pediatric patients. There are, however, some important differences. Adults with idiopathic restrictive cardiomyopathy have a relatively prolonged course and reasonably good 5-year survival.5,6 However, in the two reported pediatric series mentioned above, the average 2-year survival was <50% from the time of diagnosis.1,2 This difference in survival rate may be due to delayed detection of the disease in children who already have overt congestive heart failure when they present, in contrast to adults, who tend to be diagnosed earlier.

The diagnostic criteria for restrictive cardiomyopathy are: (1) marked bialtral dilatation; (2) normal LV chamber size and LV ejection fraction >50%; (3) LV end-diastolic pressure >15 mmHg; and (4) LV end-diastolic pressure at least 5 mmHg greater than RV end-diastolic pressure.1 Doppler study reveals diastolic dysfunction consistent with restrictive physiology, namely the typical square root sign. Our patient’s findings were completely compatible with this diagnosis, and she had no evidence of pericardial thickening that would suggest constrictive pericarditis.7 She did not yet have significant pulmonary hypertension at the time of diagnosis because of early detection. Other entities to be considered that may reduce ventricular diastolic compliance include specific infiltrative disorders of the myocardium such as glycogen storage disease, mucopolysaccharidosis, hemochromatosis, amyloidosis, sarcoidosis, and endomyocardial fibrosis.5,8 Our patient’s absence of multiorgan involvement, a history not suggestive of such disease, and the cardiac MRI helped to exclude these possibilities, although we did not do endomyocardial biopsy to rule them out completely.

Pediatric restrictive cardiomyopathy carries a high risk for rapidly progressive elevation in pulmonary vascular resistance9 and for sudden death.10 Poor prognostic factors that have been identified include cardiomegaly, age <5 years, thromboembolism, pulmonary venous congestion, syncope, or chest pain at diagnosis.10,11 An increased echocardiographic LA to Ao ratio at initial presentation is also a strong predictor of poor outcome.3 Because of the poor prognosis of this disease, it is recommended that children with restrictive cardiomyopathy be given preferential status on cardiac transplant waiting lists.10 Our patient’s young age and increased LA to Ao ratio put her at high risk, and she is therefore listed for a cardiac transplant.

References