

Tricuspid aortic stenosis with systolic dysfunction in a 14-year-old boy: Critical stenosis, noncompaction or secondary fibroelastosis?

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A bicuspid aortic valve is the most common cause of isolated valvular aortic stenosis in the pediatric age group; another cause is annular hypoplasia, with the tricuspid valve being the least commonly involved. In aortic stenosis, causes of systolic dysfunction are critical stenosis, secondary fibroelastosis and noncompaction. Critical stenosis is diagnosed in the usual fashion. Secondary fibroelastosis is characterized by endocardial thickening, systolic dysfunction and distinctive bright echoes originating from the endocardium.

Noncompaction of the ventricular myocardium, or 'spongy myocardium', is a rare, morphologically distinct primary genetic cardiomyopathy characterized by prominent ventricular trabeculations and deep intertrabecular recesses. It is believed to represent an arrest in endomyocardial morphogenesis. Noncompaction may present as an isolated cardiomyopathy or may be associated with complex congenital heart lesions (1). These deep recesses are lined by endocardium and are continuous with the ventricular endocardium. Noncompaction of the left ventricle (LV) has been described in infants with aortic stenosis or atresia; however, its presence beyond infancy with aortic valve disease is exceedingly rare (2,3). We describe the coexistence of left ventricular noncompaction and tricuspid aortic valve stenosis in a 14-year-old boy. This is also interesting because stenosis in this age group is mostly because of bicuspid aortic valve. The echocardiographic appearance of the ventricular myocardium was characteristic of noncompaction and unlike that observed in a hypertrophied ventricle. Noncompaction was also demonstrated by computed tomography angiogram of the LV.

CASE PRESENTATION

A 14-year-old boy presented with a nine-month history of progressive exertional dyspnea. Clinical examination revealed a blood pressure of 98/80 mmHg in the right upper limb while supine, with a heart rate of 96 beats/min; his pulse was regular, low volume with a delayed peak with no radioradial or radiofemoral delay, and all peripheral pulses equally palpable. The jugular venous pressure was raised 4 cm above the angle of Louis with a prominent c-v pattern. Bilateral pedal edema was present. The apical impulse was displaced in the sixth intercostal space, 2 cm outside the midclavicular line, heaving in character. Auscultation revealed a muffled S1, an S2 that was paradoxically split with a loud P2 component, and an audible left ventricular S3. There was no ejection sound. Basal ejection murmur of grade IV/VI in intensity,

Noncompaction is a congenital cardiomyopathy due to embryonic arrest of the normal development of the myocardium, leading to persistence of fetal myocardium in postnatal life. Although described in infants with aortic stenosis or atresia, the presence of noncompaction beyond infancy associated with aortic valve disease is exceedingly rare. The echocardiographic appearance of the ventricular myocardium in noncompaction is the distinctive presence of deep intramyocardial recesses. A case of left ventricular noncompaction in association with a tricuspid aortic valve with severe aortic stenosis with systolic dysfunction in a 14-year-old boy is reported.

Key Words: Bicuspid aortic valve; Bright echoes; Noncompaction; Secondary fibroelastosis; Systolic dysfunction; Tricuspid valve

crescendo-decrescendo with delayed peaking at aortic area was heard radiating to both the carotids. There was pansystolic murmur along the left lower sternal border. Auscultation over the lung fields revealed basal crepitations. The liver was tender and palpable 3 cm below the costal margin. Electrocardiography revealed normal sinus rhythm, left atrial enlargement and left ventricular hypertrophy (Figure 1). A chest x-ray revealed cardiomegaly with mild pulmonary venous hypertension. Two-dimensional echocardiography in apical four-chamber view

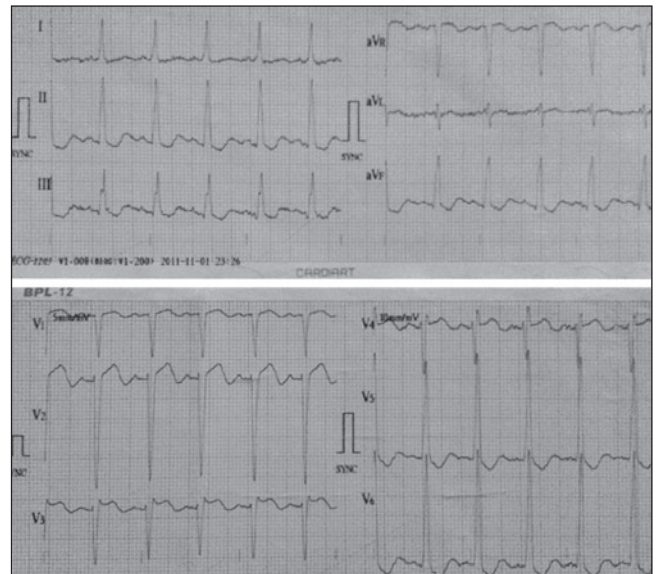


Figure 1) Electrocardiogram showing normal sinus rhythm, left atrial enlargement and left ventricular hypertrophy with strain pattern

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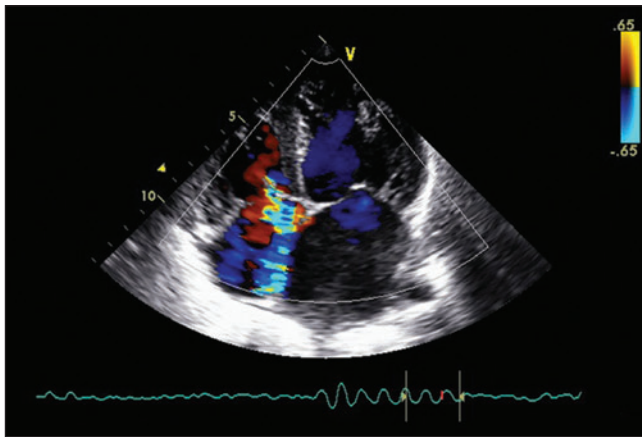


Figure 2) Two-dimensional echocardiogram in apical four-chamber views in end-diastole showing deep intertrabecular recesses in the apical segments and along the lateral wall

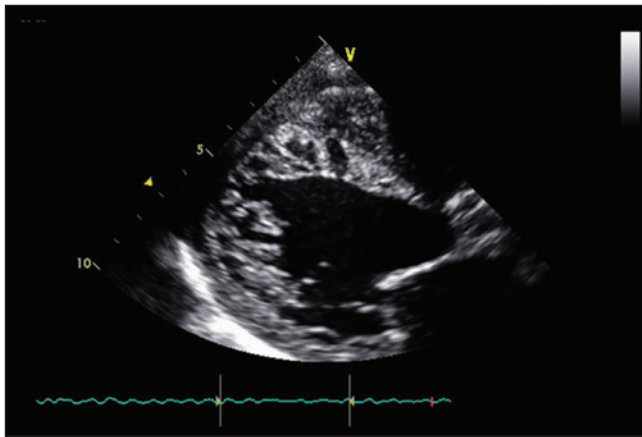


Figure 3) In parasternal short-axis view, prominent trabecular meshwork is observed circumferentially

revealed a dilated LV (LV end-diastolic diameter 56 mm) and left atrium (4.1 cm.) with global hypokinesia, and an ejection fraction of 30% with mild mitral regurgitation (Figure 2). The epicardium was thin while the endocardium was markedly trabeculated and spongy (Figure 3). Noncompaction was observed in all segments of the LV but was more marked in the apical, lateral and inferior segments. The diastolic wall thickness of the basal anteroseptal and basal posterior segments in the parasternal long axis view were 8 mm each, without trabeculations. Deep intertrabecular spaces communicating with the main LV cavity were evident on two-dimensional imaging. The aortic valve was trifoliate, noncalcified and showed restricted opening with an effective orifice area of 0.8 cm^2 . Doppler examination revealed a peak and mean gradient of 100 mmHg and 63 mmHg, respectively, across the aortic valve (Figure 4). There was mild aortic regurgitation. The right ventricular endocardium was normal. Moderate tricuspid regurgitation was present with an estimated right ventricular systolic pressure of 49 mmHg and the inferior vena cava was dilated. Computed tomography angiography revealed a thin epicardium while the endocardium was markedly trabeculated and spongy with deep recesses (Figure 5). The patient was referred to surgery for aortic valve replacement after medical stabilization.

DISCUSSION

Although aortic stenosis can be present in children secondarily to a small valve orifice, the valve usually has no or mild degree of obstruction

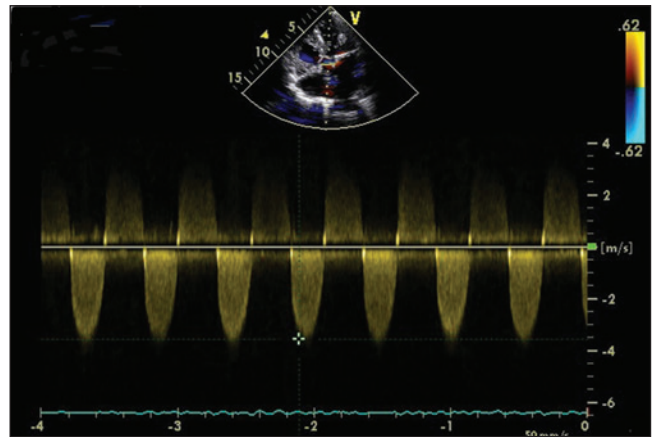


Figure 4) Continuous-wave Doppler interrogation of the left ventricular outflow tract showed a peak velocity of 4.1 m/s

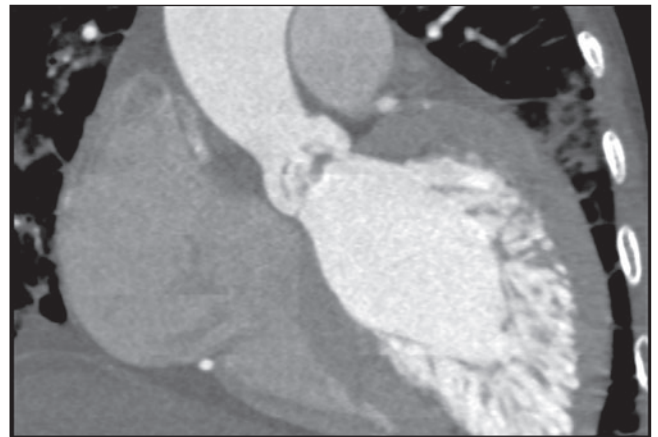


Figure 5) Computed tomography angiogram with contrast in coronal view showing deep intertrabecular recesses of the left ventricle

in initial years and progressively worsens over time because of turbulence and hemodynamic stress. The rate of aortic stenosis progression is highly variable and appears to be age related. With increasing age, progression slows; therefore, progression is fastest in infants, moderate in children and slowest in adolescents. It is probably related to the inability of the valve orifice to increase in proportion to somatic growth. Bicuspid aortic valve is the most common cause of isolated valvular aortic stenosis in the pediatric age group; other causes include annular hypoplasia and trifoliate valve (the least common cause). As stenosis progresses, the LV compensates as per Laplace's Law. When its contractile reserve depletes, the LV begins to dilate and sets the stage for systolic dysfunction; however, this occurs over many years. In the present case, systolic dysfunction appeared relatively early and was, therefore, unlikely to be a result of stenosis only. Other reasons for early appearance of systolic dysfunction are secondary fibroelastosis and noncompaction. Endocardial fibroelastosis is primarily a disease of infancy and early childhood, with rare occurrences in young adulthood (4). Secondary endocardial fibroelastosis occurs secondary to congenital heart diseases, especially hypoplastic left heart syndrome or LV obstructive lesions. Endocardial fibroelastosis is characterized by endocardial thickening due to overzealous fibroblastic proliferation. As it affects papillary muscles and chordae tendineae, other valvular abnormalities become apparent. It presents clinically as LV dysfunction, congestive heart failure, thrombembolism and arrhythmias. Distinctive bright echoes originating from the endocardium are the

diagnostic hallmark and, thus, it was excluded in our case. Noncompaction is the embryonic arrest of development of myocardium and is characterized by the presence of deep intramyocardial recesses, which gives it the characteristic 'spongy' appearance. Noncompaction has a reported prevalence of 0.045% (4-6). Although a congenital disorder, noncompaction may present late in adult life. It is likely related to variations in the extent of myocardial involvement, with progressive myocardial dysfunction caused by subendocardial hypoperfusion and microcirculatory dysfunction playing a role. Because the patient was symptomatic for only the past two years, we believe that noncompaction of the ventricular myocardium was silent until the progressive hemodynamic burden of the associated aortic stenosis resulted in significant systolic and diastolic dysfunction of the LV. The hallmark signature is the echocardiographic appearance of the ventricular myocardium, which is unlike that observed in a hypertrophied ventricle. Our patient also had severe aortic stenosis, for which the clinical presentation is similar to that of a case of isolated noncompaction. The distinctive clinical and echocardiographic features of noncompaction in the present case are similar to those described in one of the largest series on isolated noncompaction of the LV in adults by Oechslin et al (6). Patients with noncompaction have a high incidence of heart failure, arrhythmias, thromboembolism and sudden cardiac death (6). Coexistence of critical aortic stenosis with noncompaction further adversely alters the prognosis; such case reports are rare. Treatment of these patients also poses a therapeutic dilemma and the prognosis after aortic valve replacement is not clear. However, it

has been suggested that cardiac transplantation is probably a better option for these patients (6).

DISCLOSURES: The authors have no financial disclosures or conflicts of interest to declare.

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