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Case Report Open Access

A Rare Case of Biventricular Non Compaction with Associated Pentad – An Echocardiographic Diagnosis

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Abstract

Noncompaction of the ventricular myocardium is a cardiomyopathy caused by the arrest of normal embryogenesis of the ventricles. It is classified as isolated noncompaction of the ventricles and ventricular noncompaction associated with other conditions. The left ventricle is the most affected site, but right ventricular involvement has also been reported. As the right ventricle is normally trabeculated, there are controversies regarding noncompaction of right ventricle. We present a case of biventricular noncompaction associated with severe aortic coarctation and arch hypoplasia, patent ductus arteriosus, bicuspid aortic valve, coronary cameral fistula and atrial septal defect.

Keywords: Spongy myocardium; Intertrabecular recesses; Trabeculations

Case Report

This 3 month old baby presented to us with cough and chest retractions of 1 month duration without any improvement on routine medications. The physical examination showed a sick infant, with signs of intractable heart failure. Heart rate was 140 beats/min and respiratory rate was 68/min with chest retractions; blood pressure was 102/56 mmHg in upper limb and 54 mmHg (systolic) in lower limb; peripheral saturation 99% in UL and 96% in LL. The auscultation of the heart revealed a regular rhythm, loud pulmonic component of second heart sound and a short systolic murmur at left sternal border. The chest X-ray showed cardiomegaly with pulmonary plethora as well as pulmonary venous hypertension and electrocardiogram showed biventricular hypertrophy with sinus rhythm.

In transthoracic echocardiography, views had to be modified to get clear image and enhanced information. In the parasternal short axis view at the level of cross section of ventricles, the walls of both the ventricles revealed a compact epicardial layer and an endocardial layer consisting of a prominent trabeculae and deep intertrabecular recesses with the ratio of end-systolic non-compact/compact layer being 3.2. Doppler colour flow in the intertrabecular recesses was in continuity with the flow in ventricular cavity (Figure 1, Movie). The mitral, tricuspid and pulmonary valves had normal morphology, (Figure 2a) with mild pulmonary regurgitation seen in modified parasternal long axis view; a coronary - cameral fistula {Left Anterior Descending artery (LAD) to Right ventricle (RV)} was visualised in the same view (Figure 2b). Modified parasternal short axis view revealed a bicuspid aortic valve and a non restrictive patent ductus arteriosus (PDA), 4 mm shunting right to left (Figure 3a). From the suprasternal window, the aortic arch was visualised, which showed discrete coarctation with a turbulent Doppler flow (maximum gradient 42 mmHg) and arch hypoplasia (Figure 3b). Subcostal and apical four chamber views showed enlargement of all the cardiac chambers with small LV cavity accounting for diastolic dysfunction and a large secundum atrial septal defect (O.S. ASD) of size 1 cm which was shunting left to right (Figure 4). The patient was lost to follow up and was not traceable due to incorrect records, so the clinical evolution and further course could not be assessed.

Discussion

Despite improvements in medical therapy and increased availability of cardiac transplantation, cardiomyopathy remains one of the leading cardiac causes of death in children. The incidence of pediatric cardiomyopathy is not known because of a lack of clinical recognition and underreporting. The diagnostic evaluation of children with cardiomyopathy of genetic origin is complicated by the large number of rare genetic causes, the broad range of clinical presentations and the array of specialized diagnostic tests [1].

Ventricular noncompaction is a rare genetic cardiomyopathy with an incidence of 0.05% in general population. The importance of its diagnosis lies especially in asymptomatic patients, screening relatives of index cases in order to focus on their follow-up and searching for criteria warranting prophylactic anticoagulation, implantable cardioverter defibrillator and anti-remodelling drugs such as angiotensin-converting inhibitors [2].

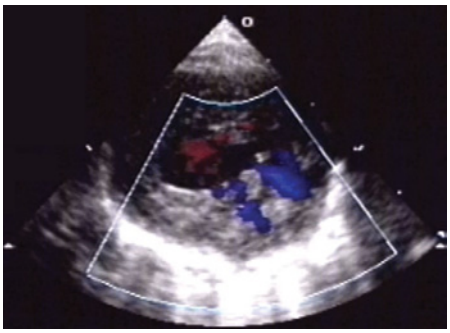

Figure 1: PSAX view showing cross section of LV and RV with prominent trabeculations, deep intertrabecular recesses with colour flow (seen as blue) continuous with the ventricular cavity.

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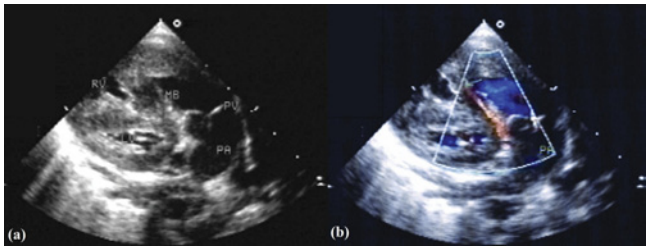

Figure 2: Modified PLAX view showing.

(a) Thick ventricular myocardium and moderator band (MB), dilated pulmonary artery (PA) and a normal pulmonary valve (PV), small left ventricular (LV) and right ventricular (RV) cavity.
(b) LAD to RV fistula (seen as a red colour flow with arrow).

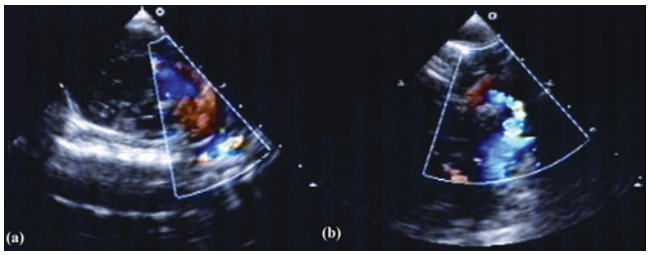

Figure 3: (a) Modified PSAX view showing PDA (asterisk) shunting right to left. **(b)** Suprasternal long axis view showing hypoplastic aortic arch with area of turbulent flow followed by dilated segment in descending aorta.

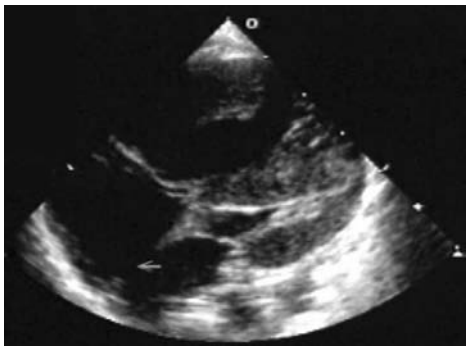

Figure 4: Apical four chamber view showing O.S. ASD (arrow), dilated RA and RV and small LV cavity.

It was initially described in 1984 by Engberding and Bender [3].

The main cause of this disease is due to an intrauterine arrest of normal myocardial development with lack of compaction of the loose myocardial meshwork [4]. Isolated noncompaction of ventricular myocardium (INVM) is characterized by persistent embryonic myocardial morphology in the absence of other cardiac anomalies (the recesses are communicating only with the ventricular cavity, not the coronary circulation) [5]. In some familial cases, the mechanism considered was a mutation in the G4.5 gene of the Xq28 chromosome region [6]. Noncompaction of ventricular myocardium (NVM) which is associated with obstruction of the right or left ventricular outflow tracts, complex cyanotic congenital heart disease and coronary artery anomalies is characterized by the persistence of deep intertrabecular recesses in communication with both the ventricular cavities and the coronary circulation [5]. In this case the mechanism involved is novel mutations in the G4.5 gene and mutations in the alpha-dystrobrevin gene, which is associated with muscular dystrophy in humans [7].

It presents as heart failure, ventricular arrhythmias and thromboembolism. There is both systolic as well as diastolic dysfunction of the ventricles. It can present at any age, but it is still underdiagnosed when present with other conditions [8-10]. In adults, associated thromboembolism is known but can be seen in pediatric age group also as high as 9% of patients [11].

The quantitative evaluation for the diagnosis of NVM could be done by determining the ratio of maximal thickness of the noncompacted to compacted layers (measured at end systole in a parasternal short axis view), with a ratio >2 diagnostic of NVM [9,12]. The specific criterion for the diagnosis of the right ventricle noncompaction has not been proposed as very few cases reported [13], though few people use the extrapolation of criteria proposed for isolated noncompaction of left ventricle for the same. Two-dimensional and color Doppler echocardiography has been the diagnostic procedure of choice, but the diagnosis is often missed because of the limitations of near field imaging, especially in cases with focal involvement. Echo contrast imaging improves the endocardial border definition and could improve the detection of this rare type of cardiomyopathy, which could otherwise be misdiagnosed [14]. Non-compaction is more common in the apical segments (91%), as compared with the mid-cavity levels (78%) and the basal segments (21%). Non-compaction is most common in the anterior segment, becoming less frequent in successive segments as viewed in a clockwise direction [15].

Both INVM and NVM carry a poor prognosis and high mortality with limited treatment options which include standard medical therapy for heart failure and heart transplant with limited success. According to recent data, middle-term prognosis appears to be better than that previously reported [16].

In conclusion, non compaction is a rare cardiomyopathy and biventricular involvement, though controversial, is even rarer. It is often diagnosed late if other conditions coexist. As our case illustrated, we must search carefully the underlying cause of intractable heart failure, along with a search for associated malformations.

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