European Journal of Echocardiography (2008) **9**, 306–308
doi:10.1016/j.euje.2006.11.007

A rare case of biventricular non-compaction associated with ventricular septal defect and descendent aortic stenosis in a young man

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Received 23 October 2006; accepted after revision 12 November 2006; online publish-ahead-of-print 28 December 2006

KEYWORDS
Noncompaction;
Aortic coarctation;
Ventricular septal defect

Noncompaction of the ventricular myocardium is a cardiomyopathy caused by the arrest of normal embryogenesis of the ventricles. It is classified in isolated noncompaction of the ventricles (most frequently of the left one) and in ventricular noncompaction associated with other congenital anomalies of the endocardium and myocardium, such as obstruction of the right or left ventricular outflow tracts, complex cyanotic congenital heart disease, and coronary artery anomalies. There are controversies regarding the right ventricle noncompaction due to the normally trabeculated shape of its walls. We present a case of severe heart failure with a complex anomaly: biventricular noncompaction, ventricular septal defect and aortic thoracic stenosis.

Case report

A 27-year-old man was admitted in our department in order to assess him for heart transplant. From his history, we noticed a surgical intervention at 2 years old for aortic coarctation (aortic enlargement with patch). After 10 years, slowly began exertional dyspnea and fatigue. At presentation, he complained of severe dyspnea, orthopnea, palpitations and abdominal pain.

The physical examination showed a cachectic young man, with signs of severe heart failure. The auscultation of the heart revealed an irregular rhythm, a rough third degree systolic and diastolic rumble spread all over the heart area and a second degree systolic rumble at the apex. Heart rate was 100 beats/min; blood pressure was 90/60 mmHg and respiratory rate was 28/min. The heart X-ray showed the enlargement of the heart with right pleural effusion. On ECG we saw a sinus rhythm with ventricular extra beats, and left ventricular hypertrophy.

Transthoracic echocardiography (TTE) showed the enlargement of all the heart cavities (right atrium = 61 mm; right ventricle = 49.6 mm; and left atrium = 40 mm) (*Figures 1* and *2*). The dimensions of the left ventricle were 87.8 mm in diastole and 72.5 mm in systole, with an ejection fraction of 23% and shortening fraction of 7.4%. The structure of the walls revealed a compact epicardial layer and an endocardial layer consisting of a trabecular meshwork and deep intertrabecular spaces with the ratio of end-systolic non-compact/compact layer >2 (*Figure 3*). From parasternal short axis view, we can see also the Doppler colour flow between the ventricular cavities and the intertrabecular spaces. The interventricular septum has normal dimensions and presents a small defect in subaortic position with left-to-right shunt (maximum gradient 47.6 mmHg) (*Figure 4*). The mitral and tricuspid valves have normal morphology, with moderate regurgitation of both of them. The pressure in the pulmonary artery (calculated with the velocity of the tricuspid regurgitant jet) was 70 mmHg. From parasternal long axis we noticed a dilated aorta (30.5 mm at the level of the annulus and 74.3 mm the ascending segment) (*Figure 1*). In parasternal short axis, we can see that it is a bicuspid aorta (*Figure 5*), with severe regurgitant jet (*Figure 6*). The longitudinal five chambers confirmed severe aortic regurgitation (pressure half time = 205 ms). From the suprasternal window, the descendent aorta is enlarged with a patch (*Figure 7*) and right below the subclavian artery became stenotic with a turbulent Doppler flow (maximum gradient = 30 mmHg) (*Figure 8*).

His clinical status, the renal insufficiency, and the value of his pulmonary artery resistances (18 UW) were absolute contraindications for accepting him in our heart transplant list. Despite the treatment he was put on, his status became worse in a week, and soon after he died in another cardiology service.

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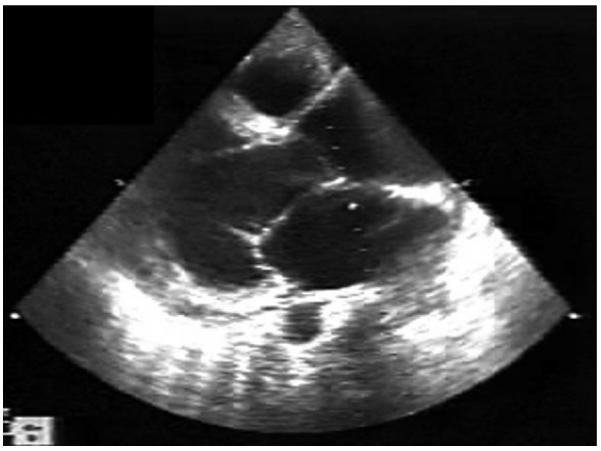

Figure 1 Parasternal long axis shows the enlargement of the left ventricle, left atrium and aortic root.

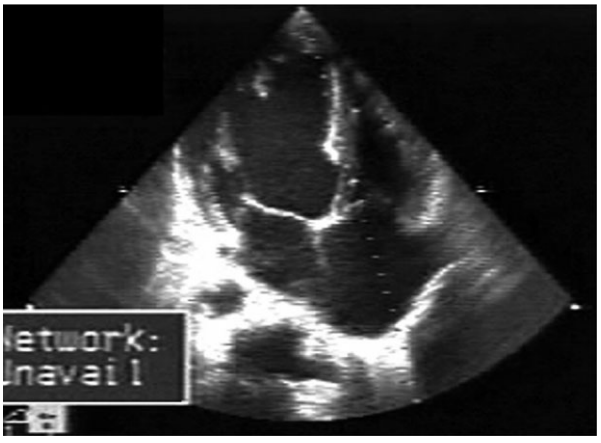

Figure 2 Apical four chamber view showing the enlargement of all the heart cavities and the trabecular structure of the ventricular walls.

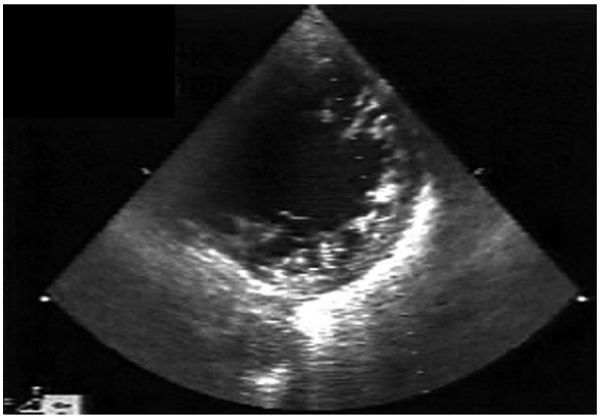

Figure 3 Parasternal short axis view showing left ventricular walls structure with a compact epicardial layer and an endocardial layer consisting of a prominent trabecular meshwork and deep intertrabecular spaces.

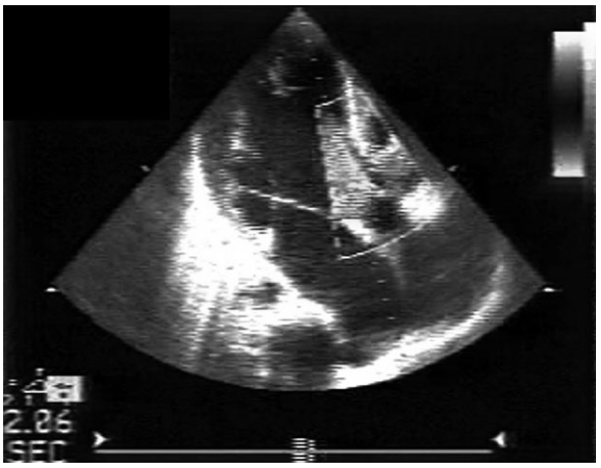

Figure 4 Apical five chamber view shows the small defect in subaortic position with left-to-right shunt.

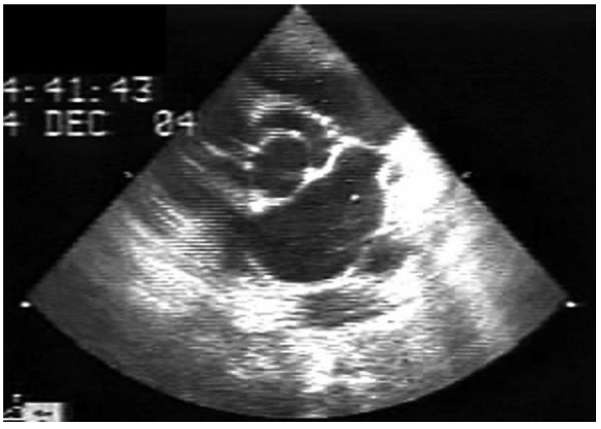

Figure 5 Parasternal short axis view shows the bicuspid aortic valve.

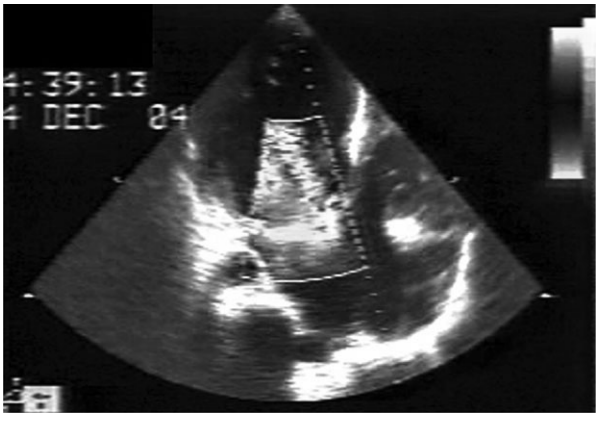

Figure 6 Apical five chamber view showing the regurgitant aortic jet.

Discussions

Ventricular noncompaction is a rare, unclassified cardiomyopathy. 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.[2] It is classified as (1) isolated ventricular noncompaction (INVM) characterized by persistent embryonic myocardial morphology in the absence of other cardiac anomalies (the

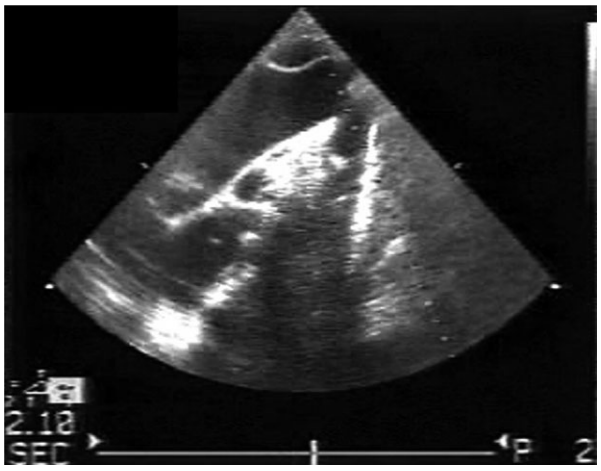

Figure 7 Suprasternal window shows the descendent aorta enlarged with a patch.

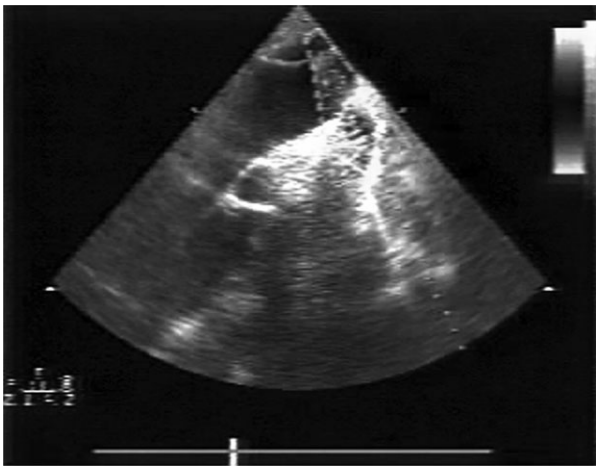

Figure 8 Suprasternal window shows the turbulent Doppler flow in the descendent aorta, right below the left subclavian artery.

recesses are communicating only with the ventricular cavity, not the coronary circulation[3]). In some familial cases was described the mechanism considered responsible: a mutation in the G4.5 gene of the Xq28 chromosome region[4]; and (2) noncompaction of ventricular myocardium (NVM) associated with obstruction of the right or left ventricular outflow tracts, complex cyanotic congenital heart disease and coronary artery anomalies.[1] It is characterized by the persistence of deep intertrabecular recesses in communication with both the ventricular cavities and the coronary circulation.[3] In this case the mechanism involved was novel mutations in the G4.5 gene and mutations in the alpha-dystrobrevin gene, which is associated with muscular dystrophy in humans.[5]

The three major clinical manifestations of this disease are as follows: heart failure, ventricular arrhythmias and thromboembolism. The cardiac insufficiency is both systolic and diastolic and in the greatest number of cases is severe. Especially in the cases of NMV associated with other cardiac diseases the noncompaction is frequently misdiagnosed. 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.[6] The differential diagnosis is made with diseases with similar echocardiographic patterns (apical hypertrophic cardiomyopathy, dilated cardiomyopathy, arrhythmogenic right ventricular dysplasia, endocardial fibroelastosis, cardiac metastases, and left ventricular thrombus). However, both in INMV and NMV the prognosis is bad, and the mortality is high. Until now, the treatment is the classical one for heart failure, and if the patients are diagnosed in the early phases we can consider heart transplant (until now there are few cases described in the literature). In conclusion, NMV is a rare disease, often late diagnosed because of the similarities with other causes of heart failure. As our case illustrated, we must search more carefully the underlying cause of a severe, resistant to treatment heart failure, because may be we can save a life.

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