CASE REPORT

ISOLATED VENTRICULAR NONCOMPACTION: CASE REPORT AND LITERATURE REVIEW

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Summary

A case of isolated ventricular noncompaction of the myocardium (IVNC) is presented in this article. IVNC is a rare cardiomyopathy resulting from the failure of trabecular compaction of the developing myocardium. Echocardiography is a method of choice to diagnose this disease. A characteristic feature to diagnose IVNC is a two-layered myocardial wall structure with both a thin epicardial compacted zone and an extremely thickened endocardial noncompacted zone with deep recesses filled with blood from the ventricular cavity.

According to the report of the World Health Organization/International Society and Federation of Cardiology Task Force in 1995 on the definition and classification of cardiomyopathies, isolated ventricular noncompaction of the myocardium (IVNC) is considered an unclassified cardiomyopathy [1]. It is a rare cardiomyopathy, which results from the arrested normal compaction of the ventricular endomyocardium during cardiac embryogenesis. IVNC is characterized by prominent myocardial trabeculations and deep intertrabecular recesses which lie in continuity with the left ventricular endocardium. IVNC was first described over a decade ago [2]. It is a rare congenital disorder, and it has been reported that the incidence of this unique cardiomyopathy is diagnosed in 0.05 percent of adults [3].

The most important clinical manifestations of IVNC are: heart failure (53%), ventricular tachycardia (41%), sudden cardiac death (35%), cardioembolic events (24%) and syncope (18%) [4].

Because of a lack of knowledge, the rarity of this pathology and the relatively recent description of IVNC, this unclassified cardiomyopathy is often left undiagnosed, despite its important prognostic implications for patients.

In this article we have presented a case of IVNC first diagnosed in our department. This is the first published case of IVNC reported by Lithuanian authors.

A 49-year-old man had experienced worsening dyspnea for the last six months. The patient was treated in a regional hospital, where dilated cardiomyopathy was diagnosed. Then he was admitted to our hospital because of the signs of progressive heart failure. There was no history of heart failure or cardiomyopathy in his family.

Clinical examination in our cardiology department revealed a heart rate of 86 beats per minute and blood pressure of 110/80 mmHg. There were no râles in the lungs, no enlargement of liver, no peripheral edema.

The results of routine laboratory analysis were normal. The patient’s 12 lead electrocardiogram showed sinus rhythm, left atrial enlargement, left ventricular hypertrophy. Mild cardiomegaly was observed on chest radiograms. Echocardiography showed the slightly enlarged left ventricular cavity dimensions. The left ventricular end-diastolic diameter was 63 mm. Ejection fraction was reduced to 30%. The left ventricular wall was thickened with prominent muscular trabeculations seen in the apical region and in the superior and mid-ventricular segments of the lateral wall. There were deep recesses penetrating the myocardium seen in the affected segments of the left ventricle. The left ventricular myocardial wall was thick, with two layers of myocardial wall structure. The thin external zone of the myocardium was compacted and its thick...
Figure 1. Parasternal short axis view of the left ventricle (LV) at the apical level (A) and at the level of the papillary muscles (B). There is a two-layered myocardial wall structure with both a thin epicardial compacted zone and an extremely thickened endocardial noncompacted zone with deep recesses filled with blood from the ventricular cavity (arrows).

Figure 2. Apical four chamber view. The structural alterations were predominantly localised to the left ventricular (LV) mid lateral wall and to the apex (arrows). LA – the left atrium; RV – the right ventricle; RA – the right atrium.

Figure 3. Colour Doppler study showed deep recesses filled with blood from the left ventricular (LV) cavity. LA – the left atrium; RV – the right ventricle.

The internal zone had prominent muscular trabeculations with deep endomyocardial spaces (Figures 1, 2). The end-systolic thickness ratio of the non-compacted to the compacted zone was 2.1/0.8 = 2.6. A colour-flow Doppler study showed forward and reversed blood flow from the ventricular cavity into spaces between the prominent trabeculations throughout the cardiac cycle (Figure 3). Moderate mitral regurgitation with structurally normal mitral valve leaflets was present. The right ventricle was not enlarged, with normal trabeculations.

Typical features of IVNC, including global hypokinesia and the thickening of the myocardium, numerous prominent trabeculations, and deep inter-trabecular recesses were observed on echocardiography.

Ramipril (2.5 mg daily), spironolactone (25 mg daily), furosemide (40 mg two to three times per week) were administered to the patient. Because of a high risk of thromboembolic events during this disease, oral anticoagulation treatment with warfarin was started. Small doses of carvedilol (3.125 mg twice a day because of low blood pressure) were started several days later.

Discussion
The cause of IVNC is not fully elucidated. The disease is thought to be a morphogenetic abnormality involving the arrest of compaction of the loose myocardial meshwork during fetal ontogen-
esis (normally during the first month of fetal life) in the absence of any coexisting congenital lesion [3,5]. Although a congenital disorder, the delayed onset of symptoms in isolated noncompaction and late presentation in adulthood are well recognized. The explanation for the late onset of symptoms is not clear but may relate to variations in the extent of myocardial involvement and progressive myocardial dysfunction caused by chronic myocardial ischemia superimposed on the primary process [4].

Both the familial [4,6] and sporadic [2] forms of IVNC were described. The familial form was observed in 18% of adult population with IVNC [4]. Because of the risk of familial occurrence, the first-degree relatives should be screened by echocardiography to identify asymptomatic patients.

Echocardiography is a method of choice to diagnose IVNC. There are four echocardiographic diagnostic criteria established for IVNC: (1) the absence of coexisting cardiac abnormalities; (2) a two-layered structure of the left ventricular wall, with the end-systolic ratio of the noncompacted to the compacted layer >2.0; (3) finding this structure predominantly in the apical and mid-ventricular areas; (4) blood flow directly from the ventricular cavity into deep intertrabecular recesses as assessed by Doppler echocardiography [5].

Although other modalities such as computer tomography, magnetic resonance imaging, and ultrafast computed tomography may be also helpful, no diagnostic criteria for these modalities have been proposed yet [5,6].

Prominent left ventricular trabeculations can be found in up to 68% of healthy hearts and can be observed in hypertrophic hearts secondary to dilated, valvular or hypertensive cardiomyopathy [4]. The end-systolic ratio of the noncompacted to compacted layers of >2.0 is a diagnostic criterion for IVNC and allows differentiation from hypertrophic cardiomyopathy and dilated cardiomyopathy or left ventricular hypertrophy. In hearts with prominent trabeculations due to other causes, the thickness ratio between trabeculated and normal zones never reaches the ratio of >2.0 [4]. By contrast to prominent trabeculations secondary to arterial hypertension or valvular disease, segmental rather than diffuse thickening or hypertrophy is present in patients with IVNC.

As confirmed in a large series of patients, features of IVNC are found predominantly in the apical and mid-ventricular segments of the left ventricle [4]. Most commonly, the apical and mid-ventricular segments of both the inferior and lateral walls are affected in more than 80% of patients which are different from the prominent trabeculations found in normal or hypertrophied hearts. Prominent trabeculations as variants of normal hearts most frequently (85%) course from the free wall to the ventricular septum.

In our case there were signs of heart failure and all the four echocardiographic diagnostic criteria of IVNC. A two-layered structure of the left ventricle was found in the apical region and superior and mid-ventricular segments of the lateral wall. Deep intertrabecular spaces communicating with the main left ventricular cavity were evident on both 2D and color flow echo imaging. The end-systolic ratio of the noncompacted to the compacted layer was 2.6. There were no coexisting cardiac abnormalities. The noncompaction of the ventricular endocardium primarily affects the left ventricle, but may also involve the right ventricle, although distinguishing this form from normal anatomy is more difficult [7]. Right ventricular noncompaction may accompany left ventricular noncompaction – in less than 50% of patients [8].

Cardioembolic events have been reported in many patients with IVNC. Long-term follow-up of 34 adults with IVNC cardioembolic events have been reported in 24% of patients with IVNC [4,6]. It was independent of the left ventricular size or function [2]. Endomyocardial morphology in IVNC predisposes to the development of mural thrombi within deep intertrabecular spaces [9]. Oral anticoagulation is recommended for all adult patients in whom IVNC is diagnosed, irrespective of the left ventricular size and function [4]. In our case, we started oral anticoagulation with warfarin when IVNC was diagnosed. Ventricular arrhythmias are a major and sometimes fatal complication in patients with IVNC [4,6]. Ventricular tachycardia has been observed in 41% of patients with IVNC. Other arrhythmias (atrial fibrillation, ventricular premature beats) were also found in patients with IVNC. There were no rhythm disturbances in our case.

Long-term follow-up showed a high incidence of heart transplantation and death [4]. The prognosis of IVNC is poor and about 50% of adult patients die suddenly. The prognosis is worse in patients with heart failure NYHA class III–IV, the left ventricular end-diastolic diameter >60 mm, the left bundle branch block and chronic atrial fibrillation.

There is no specific treatment option for non-compaction cardiomyopathy and the treatment includes all that is available for the treatment of heart failure. A more aggressive approach to the diagnosis and treatment of ventricular arrhythmias may be justified.

Since the disease often has familial character, all the first-degree relatives of this patient will be screened by echocardiography to identify asymptomatic disease.
References


