

Isolated Noncompaction of the Ventricular Myocardium

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Noncompacted ventricular myocardium (NCVM), or spongy myocardium, is a rare form of cardiomyopathy that is thought to represent an anomaly of endomyocardial morphogenesis that occurs during cardiac partition. NCVM is anatomically characterized by an excessive number of deep trabeculations in the ventricular wall comprising recesses that link the myocardial wall with the ventricular chamber. Although NCVM is a congenital disorder and is typically diagnosed in pediatric patients, onset of symptoms of cardiomyopathy can be delayed into adulthood. Diagnosis is often established by typical spongiform appearance on 2-dimensional echocardiography and magnetic resonance imaging (MRI). Most presentations involve heart failure, arrhythmias, or nonspecific symptoms that were unexplained by previous examinations. In this article, we report the case of a man who was diagnosed with spongy myocardium in late adulthood and was subsequently found to have a strong familial history of heart disease.

CASE PRESENTATION

A 54-year-old man with a history of palpitations, easy fatigability, and recent mild pedal edema was referred to a cardiologist for evaluation of chest tightness. He was being treated for presumed asthma by his primary care physician without significant relief. The patient had a history of paroxysmal nocturnal dyspnea and mild orthopnea. He denied any history of syncope or dizziness. His social history included 20 pack-years of smoking, smoking cessation 15 years ago, and occasional alcohol use. The patient's father had a myocardial infarction at age 43 years and died following a second myocardial infarction 6 months later. His paternal grandfather and uncle also died in their forties of unspecified cardiac disease. The patient's son had congenital heart disease (CHF) and underwent unspecified cardiac surgery at age 5 years.

On examination, the patient's blood pressure was

118/78 mm Hg, pulse was 62 bpm, and respirations were 12 breaths/min. Head, ear, eye, nose, throat, abdominal, and neurologic examinations were unremarkable. His lungs were clear. Cardiac auscultation and examination revealed a normal heart rate, regular rhythm, presence of S₄, and a grade 2/6 early systolic murmur at the apical area. Mild edema of extremities was observed. Cardiac enzymes, electrocardiogram, and multiple-gated acquisition scan were all normal. However, 2-dimensional echocardiography revealed a left ventricular endomyocardium with heavy trabeculations and deep intertrabecular recesses, which was diagnostic for isolated noncompaction of the ventricular (spongy) myocardium (**Figure**). The left ventricle was globally hypokinetic.

The patient was treated with angiotensin-converting enzyme inhibitors, β -blockers, diuretics, and aldactone for CHF management; aspirin for coronary artery disease prophylaxis; and warfarin for thromboembolism prophylaxis. Twenty-four months later, the patient underwent implantable cardioverter defibrillator (ICD) placement for prevention of cardiac arrhythmias. On follow-up examination, the patient reported symptomatic improvement.

DISCUSSION

NCVM is a rare cardiac malformation that was first reported in 1926. It has an estimated prevalence of 0.05% in adults, with a male to female ratio of 5.7 to 1.^{1,2} Both familial and nonfamilial forms of the disorder have been reported.¹⁻⁵ Right ventricular noncompaction

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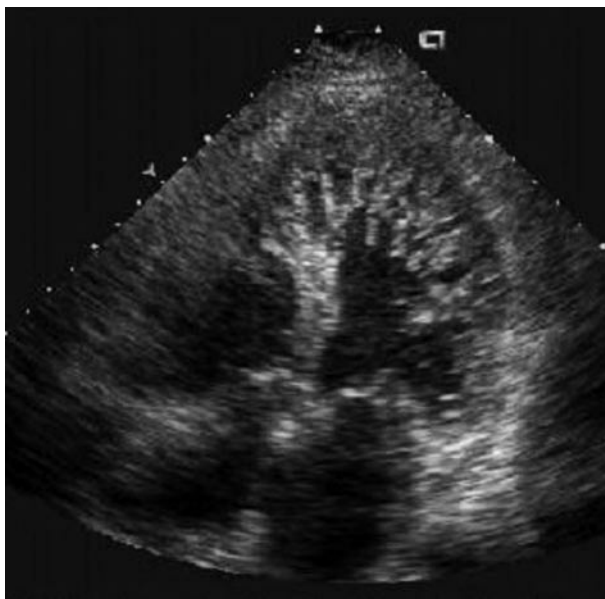


Figure. Transthoracic cross-sectional echocardiography of the left ventricular endomyocardium with multiple trabeculations and deep intertrabecular recesses, illustrating spongy appearance.

occurs less frequently and may accompany left ventricular noncompaction.^{2,6}

Pathogenesis

During normal embryogenesis, a meshwork of early musculature with numerous trabeculations and deep intertrabecular recesses, or “sinusoids,” comprises the ventricular wall and completely fills what will become the left ventricular cavity, producing a spongiform appearance. Normally, during gestational weeks 5 to 8 compaction of the developing myocardium occurs along with the commencement of coronary circulation in the myocardium; during compaction the recesses in the trabecular network are reduced to capillaries.² Compaction failure is thought to result from arrest in endocardial morphogenesis;^{7,8} however, the exact etiology remains unknown. Although the mechanisms for noncompaction are unclear, it has been suggested that NCVM accompanied by other congenital abnormalities may result from pressure overload that impedes regression of embryonic ventricular sinusoids.⁹ The exact cause of arrested development is unknown. The gene responsible for X-linked familial disease has been localized in the Xq28 region, which is in close proximity to other genes associated with systemic myopathies, such as Becker’s muscular dystrophy, Emery-Dreifuss syndrome, centronuclear myopathy, and Barth syndrome.¹⁰ The case patient did not have any symptoms suggestive of

myopathic processes, although familial history of heart disease among male family members was observed. Patient karyotyping was not performed in this case, as genetic screening is not routinely performed.

Prognosis

Morbidity and mortality is substantial at an early age in patients with NCVM. Five-year survival in symptomatic patients may be less than 50%.² In a case series by Ritter et al,² 59% (10 out of 17) of patients with this disorder either died (n = 8) or underwent cardiac transplantation (n = 2) over a 6-year follow-up period. The authors noted that symptomatic patients have a poorer prognosis. Because of the high incidence of heart failure, ventricular arrhythmias, and embolization, early diagnosis with 2-dimensional echocardiography and color Doppler imaging is important.

Clinical Features

Reports of clinical manifestations range from patients who are completely asymptomatic to those presenting with advanced heart failure. Age at development of symptoms varies, and onset of symptoms may be delayed to late adulthood. Patients who were older than age 60 years at diagnosis have been reported.² The extent of myocardial noncompaction and progressive onset of ventricular dysfunction determines when symptoms manifest.

Complications of NCVM are numerous. NCVM can be associated with valvular aortic stenosis, atresia, or obstruction of ventricular outflow tracts.^{2,7,8,11} These abnormalities were not found in the case patient; hence, he was diagnosed with isolated NCVM. The main complications of NCVM are heart failure, arrhythmias, and embolism. Heart failure is related to impaired left ventricular systolic and diastolic function. Both atrial and ventricular arrhythmias have been described.^{2,11} Noncompacted left ventricular myocardium and arrhythmogenic right ventricular dysplasia arrhythmia mechanisms appear to be similar.¹² Ventricular conduction defects, including left bundle branch block, have been described.¹³ Cardioembolic events, related to the high incidence of intraventricular thrombi formation within intertrabecular recesses, and atrial arrhythmias have also been reported.^{11,14}

Diagnosis

Echocardiography is the diagnostic modality of choice.^{2,8,11} Depressed systolic and diastolic functions, dilated ventricle with prominent ventricular trabeculations, and multiple deep intertrabecular recesses communicating with the ventricular cavity are observed with color Doppler imaging. Findings are prominent

in the apical and middle portion of the ventricle.¹¹ Antenatal cardiac malformations, however, are difficult to detect. Echocardiographic diagnostic criteria are subjective with interobserver variability, especially in cases of partial ventricular involvement. Chin et al¹¹ have suggested diagnostic criteria based on calculation of the ratio of X and Y, where X represents the depths of the recesses in the myocardium (distance between the epicardium and the bottom of the intertrabecular chamber) and Y represents the distance between the epicardium and the peak of the trabeculations. These measurements are performed at the levels of the mitral valves, papillary muscles, and apex. Widespread diagnostic application of these criteria is uncommon. MRI is another useful modality for demonstrating spongiform myocardium appearance that theoretically may permit better imaging of the myocardium.

The differential diagnosis for NCVM includes hypertrophic cardiomyopathy with ventricular hypertrophy, dilated cardiomyopathy, left ventricular apical thrombus, and prominent myocardial trabeculations occurring as a normal variant.¹⁵ Definitive diagnosis depends upon laboratory assessment, clinical findings, and imaging results.

Management

The approach to management is generally based upon the patient's symptoms. Strategies include avoiding strenuous activities, treating CHF, and instituting antiarrhythmic therapy, all of which are untested. Optimal control of heart failure medically and heart transplantation may be considered. It should be noted that heart transplantation is the only permanent curative treatment.^{2,3} Long-term anticoagulation is recommended for patients with a high risk for systemic embolization. Screening of first-degree relatives with 2-dimensional echocardiogram and genetic counseling may identify asymptomatic patients. Perhaps the most controversial management option is ICD implantation. Although studies investigating the potential benefit of ICD implantation have not been performed, it seems intuitive that these devices might protect patients from sudden cardiac death.

CONCLUSION

In this article, we reported the case of a patient

presenting with isolated NCVM in the fourth decade of life. The diagnosis was based upon clinical findings of chest tightness and CHF as well as characteristic features of spongy myocardium seen on 2-dimensional echocardiography. NCVM is a rare cardiomyopathy that may cause CHF. Typically, this cardiac anomaly is diagnosed in childhood. This case is unusual because the patient was diagnosed as an adult. **HP**

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