Left ventricular noncompaction: case of a heart transplant

Kursat Tigen*, Tansu Karaahmet, Gokhan Kahveci, Bülent Mutlu, and Yelda Basaran

Kartal Kosuyolu Heart Education and Research Hospital, Cardiology Department, Istanbul, Turkey

Received 6 February 2007; accepted after revision 15 April 2007; online publish-ahead-of-print 30 June 2007

Noncompaction of the ventricular myocardium (NVM) is a rare congenital cardiomyopathy characterized by multiple prominent trabeculations with deep intertrabecular recesses resulting from an arrest in normal embryogenesis of the endocardium and myocardium. The major clinical manifestations are depressed left ventricular systolic and diastolic function, systemic embolism, ventricular tachyarrhythmias, conduction disorders and neurologic abnormalities. We present a 21-year-old female who was diagnosed as dilated cardiomyopathy due to isolated noncompaction of the left ventricle and underwent cardiac transplantation.

KEYWORDS
Noncompaction; Dilated cardiomyopathy; Transplantation

Introduction
Noncompaction of the ventricular myocardium (NVM) is a rare congenital cardiomyopathy characterized by multiple prominent trabeculations with deep intertrabecular recesses resulting from an arrest in normal embryogenesis of the endocardium and myocardium which can either be isolated or associated with complex congenital heart disease.1 The prevalence of NVM differs from 0.06 to 0.24%/y with a male dominancy.2–4 The major clinical manifestations are depressed left ventricular systolic and diastolic function, systemic embolism, ventricular tachyarrhythmias, conduction disorders and neurologic abnormalities.5,6 Medical management of heart failure and heart transplantation when clinically indicated, management of arrhythmias, prevention of systemic embolism and echocardiographic screening of first-degree relatives are treatment strategies.4,5,7,8 We present a 21-year-old female who was diagnosed as dilated cardiomyopathy due to isolated noncompaction of the left ventricle and underwent cardiac transplantation.

Case history
A 21-year-old female with heart failure during adolescence was admitted to Kosuyolu Heart and Research Hospital with symptoms of severe congestive heart failure. On physical examination the patient was orthopneic and jugular venous distention and bilateral leg edema were present.

Blood pressure was 95/65 mm/Hg. The cardiac apex was displaced laterally. On auscultation, there was tachypnea and moderate retraction of the chest wall and moist rales over the entire lung fields. The first and second heart sounds were normal and a S3 sound was prominent. A grade 3/6 holosystolic murmur was heard at the apex. Her liver was palpable 4 cm below the right costal margin. Sinus rhythm with a heart rate of 102 pulse/min, normal QRS duration and nonspecific ST–T wave changes were present on the electrocardiogram. Chest X-ray showed mild cardiac enlargement with a cardiothoracic ratio of 0.65, prominent central pulmonary arteries and pulmonary venous congestion. Fasting blood glucose: 88 mg/dl, BUN: 45 mg/dl, creatinine: 0.97 mg/dL, AST: 44 U/L, ALT: 32 U/L, LDH: 388 U/L, haemoglobin: 9.1 g/dL, hematocrit: 27.9%, WBC: 4500/μL, platelet count: 118 000/μL, INR: 1.1, total protein: 6.8 g/dL, and albumin: 3.5 g/dl were found in the biochemical assessment. Plasma NTproBNP levels were also severely elevated (1595 pmol/L). A transthoracic echocardiographic examination was performed with a GE Vivid System Five ultrasonographic unit. Echocardiography revealed a markedly dilated left ventricle and severely impaired left ventricular systolic function (left ventricular ejection fraction by Teicholtz’s method was 17%). Multiple, prominent muscular trabeculations were present in the left ventricular apex and lateral wall above the papillary muscle insertion (Figure 1). Parasternal long and short axis views revealed a spongy appearance of the posteriobasal left ventricular wall (Figure 2). Deep intertrabecular recesses communicating with the left ventricular cavity were evident on color Doppler imaging. A mild degree of tricuspid and severe mitral regurgitation was present on color
Figure 1  Apical four chamber views of left ventricular cavity. Muscular trabeculations and deep invaginations are present.

Figure 2  Parasternal long and short axis views showing spongy appearance of the left ventricular wall.

Figure 3  Macroscopic specimens of native heart: prominent trabeculations and invaginations at the left ventricular apex and lateral wall.
Doppler examination. Estimated pulmonary artery systolic pressure was 65 mmHg. Mitral inflow pattern was consistent with restrictive physiology (mitral E velocity of 85 cm/s, mitral A velocity of 35 cm/s and deceleration time of 100 ms). A mild degree of spontaneous echocontrast was also present in left ventricular cavity. The right ventricle was mildly enlarged with normal trabeculations and systolic function was reduced. Both atrias were detected enlarged. Coronary vessels were found normal at coronary angiography and heart catheterization revealed moderately elevated chamber pressures. The findings were consistent with isolated noncompaction of the ventricular myocardium. Neurological consultation of the patient revealed completely normal. Echocardiographic screening of the first-degree relatives was also within normal ranges. During hospitalization period, despite vigorous treatment, patient’s condition progressively deteriorated requiring intravenous inotropic support for adequate systemic perfusion. Due to poor functional capacity (NYHA Class IV) despite intensive therapy with evidence of severe left ventricular dysfunction the patient has been considered for heart transplantation and underwent orthotopic heart transplantation within two months. The patient is in her second year after transplantation and being follow-up under appropriate manner. In pathologic examination the explanted heart was dilated and weighed 420 g. The coronary arteries were normal. Prominent trabeculations and invaginations especially at left ventricular apex were found impressive (Figure 3). Areas of noncompaction were markedly thickened when compared with compacted areas. Histologic examination shows endothelial lining of the recesses communicating with left ventricular cavity. Rhabdomyocyte hypertrophy, multinucleation, anisochromacy, interstitial musinosis and fibrosis, subendocardial musinosis and fibrosis were detected (Figure 4). These pathologic findings confirmed the diagnosis of isolated left ventricular noncompaction.

Discussion

Noncompaction of the ventricular myocardium which was first described in 1984 by Engberding and Bender, is a rare congenital cardiomyopathy characterized by multiple prominent trabeculations with deep intertrabecular recesses resulting from an arrest in normal embryogenesis of the endocardium and myocardium. This abnormality can either be isolated, first described by Chin et al in 1990, or associated with complex congenital heart disease. In early embryonic period human myocardium is a loose meshwork of interwoven myocardial fibers which is characterized by excessively prominent trabeculations and deep intertrabecular recesses. By gradual compaction, the fetal myocardium condenses causing the large spaces within the trabecular meshwork to disappear. This process typically progresses from epicardium to endocardium and from the base of the heart toward the apex. NVM is the result of an arrest in the normal process of myocardial compaction that is characterized by persistence of excessive prominent ventricular trabeculations and deep intertrabecular recesses. The prevalence of NVM differs from 0.06 to 0.24%/y with a male dominancy. Familial forms have been described. The major clinical manifestations are impaired left ventricular systolic and diastolic function, systemic embolism, ventricular tachyarrhythmias, conduction disorders and neurologic abnormalities. Our case also admitted with symptoms of severe left ventricular systolic and diastolic dysfunction, no history of systemic embolization was present and we were not able to document an arrhythmia during the hospitalization period. Neurological consultation also revealed normal results. Echocardiographic examination revealed prominent trabeculations that perfused from the left ventricular cavity on color Doppler at apical and lateral left ventricular wall. Confirmation of the diagnosed NVM was also made by pathologic examination. Echocardiographic examinations of the first-degree relatives of our case were within normal range. The prognosis of NVM is quite poor. Ritter and colleagues reported that 59% of patients with isolated left ventricular noncompaction either died or underwent heart transplantation during 6-year follow-up and observed worse prognosis in patients and with higher functional classes. Oechslin and colleagues reported that 35% of the patients with isolated left ventricular noncompaction were dead and 12% had
undergone heart transplantation during 40 ± 40 months follow-up. A review of the literature identified only eight patients with isolated left ventricular noncompaction who underwent cardiac transplantation. Our patient’s clinical status was also progressively deteriorated against aggressive medical management and she had undergone cardiac transplantation within two months. Because of high mortality rates, patients with NVM should be followed-up closely with appropriate medical management and considered for heart transplantation with high priority. With all these data she is presented as a case report because she is the first patient in our country and the 9th patient in the literature who had undergone heart transplantation due to isolated left ventricular noncompaction related dilated cardiomyopathy.

References