Case Report

Sickle cell disease with left ventricular non-compaction: A rare association

Prashanth Panduranga (MD)*, Mohammed Al-Mukhaini (MD)

Department of Cardiology, Royal Hospital, Muscat, Oman

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Summary
Cardiac abnormalities described in sickle cell disease are pulmonary hypertension, dilated left or right atrium/ventricle, valvular abnormalities, hyperdynamic left ventricle with hypertrophy, and left or right ventricular dysfunction. However, features consistent with left ventricular non-compaction have not been described previously in patients with sickle cell disease. We describe the case of a 21-year-old male with sickle cell disease and left ventricular non-compaction, which is a rare association and discuss the possible mechanisms for such an association.

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Introduction

Sickle cell disease (SCD) is an autosomal recessive disorder involving short arm of chromosome 11. The gene defect is a known mutation of a single nucleotide of the β-globin gene of the hemoglobin, which results in glutamate being substituted by valine at position 6, leading to sickling of erythrocytes and hemolytic anemia. Cardiac abnormalities described in SCD are pulmonary hypertension, hyperdynamic left ventricle (LV), dilated left or right atrium/ventricle, valvular abnormalities, LV hypertrophy, and left or right ventricular dysfunction [1,2]. LV diastolic dysfunction is reported to be common in the SCD population [3]. However, features consistent with left ventricular non-compaction (LVNC) have not been described previously in association with SCD.

Case report

A 21-year-old male with known history of SCD on folic acid was referred for echocardiography in view of a murmur. He had a diagnosis of SCD since childhood with episodes of vaso-occlusive crisis requiring emergency department visits necessitating analgesia. He had undergone exchange transfusion in 2008 for an acute chest crisis with complete recovery. He was asymptomatic at the time of echocardiography. His hemoglobin was 7.7 g/dl, reticulocyte count 3.7%, erythrocyte sedimentation rate 6 mm/h, and other blood investigations including creatinine kinase were normal. The ECG showed sinus rhythm, normal QRS duration, and left ventricular hypertrophy with repolarization changes.

Transthoracic echocardiography demonstrated mildly dilated LV with ejection fraction of 58% and grade I LV diastolic dysfunction along with structurally normal valves.
Figure 1  Transthoracic echocardiogram showing left-ventricular non-compaction. The apical four-chamber view echocardiogram (A) showing prominent trabeculations in apex and mid-lateral wall (arrowheads) and intertrabecular recesses/cavities in inferior wall (arrowheads) (B). LV, left ventricle.

There was trivial tricuspid regurgitation with calculated pulmonary artery systolic pressure of 35 mmHg. However, LV apex, mid-lateral wall, and posterolateral segments of the LV demonstrated a hypertrabeculated appearance with intertrabecular recesses on echocardiography (Fig. 1A and B, arrowheads). The appearance and ratio (≥2.2) of the non-compacted to compacted myocardium were consistent with a diagnosis of LVNC. Color Doppler showed flow within the intertrabecular recesses (Fig. 2). On inquiry, there was no family history of any cardiac or neuromuscular illness.

Discussion

LVNC is a congenital cardiomyopathy characterized by a distinctive ("spongy") morphological appearance of the LV myocardium [4]. LVNC is due to failure of condensation or compaction of myocardial spongy meshwork of trabeculations and intertrabecular recesses during intrauterine life that communicate with the ventricular cavity. LVNC presents as heart failure, thromboembolism, or cardiac arrhythmia. Diagnosis is made with two-dimensional echocardiography, cardiac magnetic resonance imaging, or LV angiography [4].

Figure 2  Parasternal short-axis image demonstrating the hypertrabeculated left ventricle affecting the posterolateral walls (A) and with color Doppler showing flow within the intertrabecular recesses (B). LV, left ventricle.
Subendocardial as well as transmural perfusion defects along with diminished coronary flow reserve correlating with areas of non-compacted myocardium have been described in LVNC [4]. Junga et al. suggested that altered perfusion and coronary flow reserve in LVNC may be related to failure of the coronary microcirculation to grow with the increasing ventricular mass along with compression of the intramural coronary bed by the hypertrophied myocardium, or both processes [5]. A similar pathology may be involved in SCD patients.

LVNC may be isolated or associated with genetic cardiac and noncardiac disorders, particularly genetic neuromuscular diseases [6]. LVNC may be congenital or acquired [7]. Both familial (autosomal dominant/X-linked inheritance) and non-familial cases have been described. In a recent review, Finsterer reported that, most frequently, LVNC is associated with mitochondrial disorders, Barth syndrome, hypertrophic cardiomyopathy, zasopathy, myotonic dystrophy 1, and dystrobrevinopathy [6]. Additionally, LVNC occurs with a number of chromosomal disorders, polymorphisms, and not yet identified genes [6]. Previously, Alter and Maisch reported combined occurrence of LVNC, skeletal myopathy, and hereditary spherocytosis [8]. It has been observed that there is a relationship between congenital diseases with muscular and erythrocyte pathology, such as choreo-acanthocytosis, McLeod syndrome, or hereditary spherocytosis and the homology between spectrin and dystrophin [9]. This patient with SCD demonstrated LVNC and it may be speculated that either a common chromosomal or a mutant gene may be involved. This needs further cytogenetic investigations.

In a recent report, Song [7] described a patient with atrial septal defect with right-sided noncompaction and opined that atrial septal defect results in right heart volume overload followed by neuroendocrine adaptations with increased sympathetic activity and circulating catecholamines resulting in increased contractility and heart rate. Shorter diastolic time due to tachycardia, increased intramyocardial tension from right ventricular dilatation, and increased myocardial oxygen demand, may result in subendocardial ischemia, leading to acquired form of LVNC. This same mechanism may be working in patients with SCD, specifically severe anemia leading to LVNC as in hereditary spherocytosis.

In conclusion, in this patient with SCD, detection of LVNC may be either congenital or acquired and this case illustrates one of the rare associations of LVNC with SCD, of which clinicians and researchers should be aware.

References