An Extremely Rare Congenital Association: Unicuspid Aortic Valve with Left Ventricular Noncompaction (RCD code: III-5A.1.o)

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Abstract

Unicuspid aortic valve is a rare congenital abnormality with a prevalence of 0.02% in the adult population. Left ventricular noncompaction is another congenital anomaly with an incidence of 0.05%. Unicuspid aortic valve with severe aortic regurgitation and left ventricular noncompaction was found in a 15-year-old deaf male patient who presented with shortness of breath, orthopnoea and intermittent palpitations. JRCD 2019; 4 (4): 106–108

Key words: rare disease, congenital heart disease, echocardiography, magnetic resonance imaging

Introduction

The aortic valve is normally tricuspid, with unicuspid, bicuspid, and quadricuspid aortic valves being congenital malformations. Bicuspid aortic valve is the most common abnormality with a prevalence of 0.9 – 2% [1], followed by unicuspid aortic valve with a prevalence of 0.02% [2], and quadricuspid aortic valve having a prevalence of 0.01% [3]. Diagnosis is often made by transthoracic (TTE) or transoesophageal (TEE) echocardiography. Treatment consists of surgical replacement of the valve whenever there is significant stenosis or regurgitation.

Left ventricular noncompaction (LVNC) is characterised by the presence of prominent left ventricular (LV) trabeculae, a thin compacted layer, and deep intertrabecular recesses [4]. LVNC may occur as an isolated disease or it may also be present in various types of congenital heart diseases. Its link to congenital heart disease accounts for 12% of LVNC cases, a significant proportion of which are left ventricular outflow tract abnormalities [5]. The echocardiographic diagnosis is made based on the criteria of Jenni et al. The end-systolic ratio of noncompacted to compacted layers should be greater than 2:1 [6]. Multidetector computed tomography angiography is useful when cardiac magnetic resonance imaging (MRI) is contraindicated or when echocardiography and cardiac MRI provide discordant data [4]. Medical management does not differ between patients with LVNC and genetic cardiomyopathy.

Case Presentation

A 15-year-old deaf male patient presented with a 1.5-year history of intermittent palpitations, shortness of breath, orthopnoea and fatigue. He was seen by a paediatric cardiologist 1.5 years prior who performed an echocardiographic examination and initiated treatment with furosemide, captopril, and spironolactone. However, the patient was not compliant with the treatment regimen. He presented this time to our emergency room with shortness of breath, palpitations, and oedema of the lower extremities. Physical examination was significant for a diastolic high-pitched murmur that was loudest at the left sternal border while the patient leaned forward on expiration. Pulse pressure was wide, with a systolic blood pressure of 105 mmHg and diastolic blood pressure of 57 mmHg. The patient was admitted to the cardiac care unit for further management. Electrocardiogram revealed sinus tachycardia with left axis deviation and voltage criteria for left ventricular hypertrophy. Chest radiography revealed an enlarged cardiac silhouette and clear lung fields with normal...
vascularity, pleural spaces were free of effusion, and dorso-scoliosis was present with convex to the right. Arterial blood gases showed pH-7.4; pO₂-65 mmHg; pCO₂-39 mmHg; O₂sat-93.5%. TTE revealed LV dilatation (End-diastolic dimension was 61mm) with mildly impaired LV function (ejection fraction was 45%), increased LV mass index for body surface area (129 g/m²), normal LV wall thickness, and prominent trabeculations with intertrabecular recesses in the LV with a noncompacted to compacted layer thickness ratio of > 2 (Figure 1). The recesses were supplied by intraventricular blood as demonstrated by colour flow Doppler (Figure 2). A unicuspid aortic valve (UAV) with severe aortic regurgitation with a vena contracta width > 6 mm and with holodiastolic flow reversal (end diastolic velocity > 20 cm/sec) was noted (Figures 3, 4). Cardiac MRI was ordered and the patient was scheduled for a planned aortic valve replacement at another hospital but he did not appear for further follow-up visits. Multiple attempts to contact the parents and the paediatric cardiologist were unsuccessful, hence, none of the MRI images or valve images from the surgery were available to us.

**Discussion**

UAV exists in two forms, acommissural and unicommissural, both with a predisposition to aortic dilatation. Most patients with this congenital abnormality will eventually require therapeutic intervention during infancy or childhood. UAV develops due to failure of the three aortic cusps to separate before birth [7]. The difference between acommissural and unicommissural UAV is the absence or presence of an attachment to the aorta, hence, they appear as pin-hole or slit-shaped accordingly [8]. The diagnosis of UAV can be made with the use of two-dimensional or three-dimensional TTE or TEE [8], cardiac computed tomography [9], or cardiac MRI [10]. Abnormalities associated with UAV include aortic aneurysm [11], aortic regurgitation, aortic dissection, coarctation of the aorta, patent ductus arteriosus [12], and aortic dilatation.

LVNC is a congenital cardiomyopathy, which is often first diagnosed in adults. This condition can be found in isolation with an incidence of 0.05% or in association with other cardiac abnormalities. An excessively prominent trabecular meshwork and deep trabecular recesses characterise myocardial noncompaction. Although LVNC is congenital, it has been well established that in some patients, particularly those with neuromuscular disorders, in professional athletes, and in pregnant females, LVNC may be non-congenital (acquired LVNC) [13]. The echocardiographic criteria for noncompaction have been well described and include a thick noncompacted layer and a thin compacted layer, displaying a maximal noncompacted to compacted thickness ratio of greater than 2. Genetic testing is available to search for mutations in the genes known to cause cardiomyopathy. All first-degree relatives of an individual with cardiomyopathy should undergo routine cardiac evaluation. Screening is recommended for those family members shown to also carry the gene mutation. The best screening tools
nism cannot continue for a prolonged period of time, and thickness to chamber ratio will deteriorate, resulting in LV dilatation and decreased ejection fraction and stroke volume [20]. Therefore, it cannot be excluded that unicuspid aortic valve was the primary cause of the left ventricular noncompaction in our patient.

Conclusion

This case reported on an extremely rare combination of UAV and LVNC in a deaf patient without excluding the possibility that UAV was the cause of LVNC in such a patient.

References