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

Souzan Tatari*

Rafik Hariri University Hospital, Beirut, Lebanon

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.
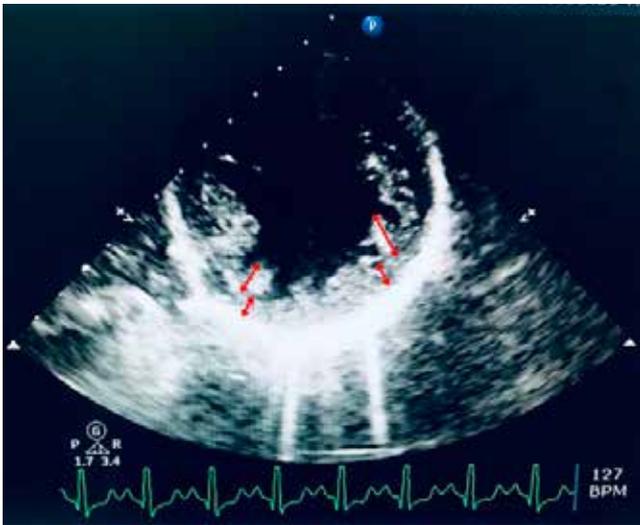
Case Presentation

A 15-year-old deaf male patient presented with a 1.5-year history of intermittent palpitations, shortness of breath, orthopnoea, fatigue, and chest discomfort. The patient reported that he had such symptoms on exertion and also at rest. 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

Figure 1. Noncompacted left ventricle showing prominent trabeculations with intertrabecular recesses. Noncompacted to compacted layers are shown (indicated by red arrows) with a ratio of 2.3
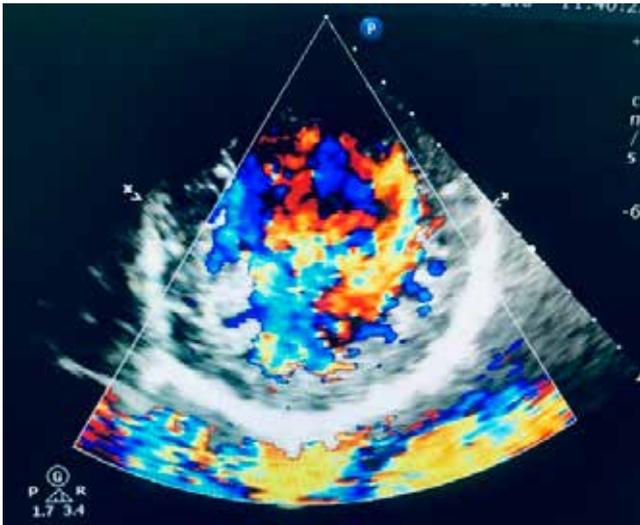

Figure 2. Colour flow Doppler demonstrate how recesses are supplied by intraventricular blood

vascularity, pleural spaces were free of effusion, and dorsal scoliosis was present with convex to the right. Arterial blood gases showed pH-7.4; pO_2-65 mmHg; pCO_2-39 mmHg; O_2sat-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/m^2), 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 hos-
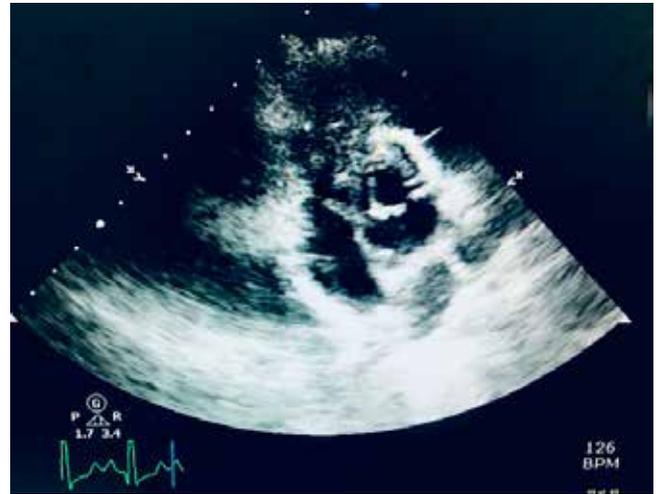
Figure 3. Unicuspid aortic valve in systole

pital 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 pinhole 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

Figure 4. Unicuspid aortic valve in diastole

include echocardiography and MRI. Treatment modalities range from medical management to cardiovascular implantable electrical devices placement and heart transplantation.

Highasi et al. carried out genetic screening in a patient with unicuspid aortic valve and found an association between severe aortic stenosis and dilated cardiomyopathy [14]. Further molecular and functional studies are needed to clarify whether these rare variants are pathogenic [14]. Morita et al. reported that the pathogenicity of newly identified rare variants, even in the known causative genes, should be carefully assessed [15]. In our case, no genetic testing was done, and the presence of severe aortic regurgitation excluded the diagnosis of dilated cardiomyopathy.

Left ventricular hypertrabeculation and its differentiation from LVNC is challenging even using MRI. Kawel et al. analysed MRI findings from 1000 participants of the Multi-ethnic Study of Atherosclerosis (MESA) and found that 43% of patients without cardiac disease or hypertension had at least 1 of 8 regions evaluated with a trabeculated to compacted myocardial ratio > 2.3 [16]. Petersen et al. found that the noncompacted to compacted ratio of > 2.3 in end-diastole had high specificity and negative predictive values when comparing healthy controls to patients with dilated cardiomyopathy, hypertrophic cardiomyopathy, hypertensive heart disease, aortic stenosis, and patients previously diagnosed with LVNC [17]. Jacquier et al. found that LV trabecular mass > 20 % of the global mass predicted the diagnosis of LVNC with a sensitivity and specificity of 93.7% in comparison to other groups [18].

In a literature review, it was found that there was an association between LVNC and other congenital heart diseases such as left ventricular outflow tract abnormalities including uni- or bicuspid aortic valves, aortic coarctations, aortic hypoplasia and subaortic stenosis, Ebstein anomaly, tetralogy of Fallot, and double outlet right ventricle [19]. Regarding the clinical classification of rare cardiovascular diseases and disorders, our case would fall into class III-rare diseases of the heart (cardiomyopathies), group 5 (unclassified cardiomyopathies) and class IV-rare congenital cardiovascular diseases, group 4 (congenital cardiovascular diseases with concomitant organ dysfunction).

Aortic insufficiency leads to volume overload which, over time, causes trabeculae and papillary muscle hypertrophy. This mecha-

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

1. Ward C. Clinical significance of the bicuspid aortic valve. Heart 2000; 83(1):81-85.

2. Novaro GM, Mishra M, Griffin BP. Incidence and echocardiographic features of congenital unicuspid aortic valve in an adult population. J Heart Valve Dis 2003; 12 (6): 674-678.

3. Tutarel O. The quadricuspid aortic valve: a comprehensive review. J Heart Valve Dis 2004; 13(4): 534-537.

4. Arbustini E, Favalli V, Narula N, et al. Left Ventricular Noncompaction A Distinct Genetic Cardiomyopathy? J Am Coll Cardiol 2016; 68 (9): 949-966.

5. Bonou M, Papadimitraki ED, Alexopoulos N, et al. Late appearance of left ventricular non-compaction in a patient with aortic coarctation and bicuspid aortic valve. Int J Cardiovasc Imaging 2016; 32: 741-742.

6. Parra-Ordonez NA, Avila-Vanzzini N, Espinola-Zavaleta N. Left ventricular non-compaction associated with bicuspid aortic valve and aortic coarctation. Arch Cardiol Mex 2018; 88: 507-508.

7. Roberts WC, Ko JM. Frequency by decades of unicuspid, bicuspid, and tricuspid aortic valves in adults having isolated aortic valve replacement for aortic stenosis, with or without associated aortic regurgitation. Circulation 2005; 111(7): 920-925.

8. Singh S, Ghayal P, Mathur A, et al. Unicuspid unicommissural aortic valve: an extremely rare congenital anomaly. Tex Heart Inst J 2015; 42: 273–276.

9. Gibbs WN, Hamman BL, Roberts WC, et al. Diagnosis of congenital unicuspid aortic valve by 64-slice cardiac computed tomography. Proc (Bayl Univ Med Cent) 2008; 21 (2):139.

10. Debl K, Djavidani B, Buchner S, et al. Unicuspid aortic valve disease: a magnetic resonance imaging study [in German]. Rofo 2008; 180(11): 983-987.

11. Kang SD, Seol SH, Park BM, et al. Incidental diagnosis of the unicuspid aortic valve with ascending aortic aneurysm in an asymptomatic adult. J Cardiovasc Ultrasound 2011; 19 (2):102-104.

12. Mookadam F, Thota VR, Garcia-Lopez AM, et al. Unicuspid aortic valve in adults: a systematic review. J Heart Valve Dis 2010; 19(1): 79-85.

13. Zarrouk-Mahjoub S, Finstere J. Noncompaction with valve abnormalities is rarely associated with neurologic or genetic disease. Int J Cardiol 2016; 202: 627-628.

14. Higashi H, Ogimoto A, Inoue K, et al. Eccentric Left Ventricular Hypertrophy in Aortic Stenosis Caused by Unicuspid Aortic Valve. Circ J 2017; 81: 895-897.

15. Morita H, Komuro I. Identification of Pathogenic Mutations for Dilated Cardiomyopathy Accompanied with Unicuspid Aortic Valve. Circ J 2018; 82: 1723.

16. Kawel N, Nacif M, Arai E, et al., Trabeculated (noncompacted) and compact myocardium in adults: the multi-ethnic study of atherosclerosis. Circulation: Cardiovascular Imaging 2012, 5(3): 357-366.

17. Petersen SE, Selvanayagam JB, Wiesmann F, et al. Left ventricular non-compaction: insights from cardiovascular magnetic resonance imaging. J Am Coll Cardiol 2005, 46(1): 101-105.

18. Jacquier A, Thuny F, Jop B, et al. Measurement of trabeculated left ventricular mass using cardiac magnetic resonance imaging in the diagnosis of left ventricular non-compaction. Eur. Heart J 2010, 31(9): 1098-1104.

19. Stahli BE, Gebhard C, Biaggi P, et al. Left ventricular non-compaction: Prevalence in congenital heart disease. Int J Cardiol 2013, 167 (6): 2477-2481.

20. Saidinejad M. Pediatric aortic valve insufficiency. Emedcine.medscape.com. Accessed: Jan 4, 2016.