CASE REPORT

Unicuspid aortic valve presenting with cardiac arrest in an adolescent

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SUMMARY

Unicuspid aortic valve (UAV) is a rare congenital anomaly typically affecting patients in their fourth and fifth decades and presenting with signs of heart failure. Our case is one of a previously asymptomatic teenage girl with a UAV, who presented with cardiac arrest and was successfully treated. Only two other similar cases have been reported in the literature, both were of slightly older male patients. Our case highlights the morbidity associated with the anomaly supporting the need for careful assessment of the valve in cases where UAV is suspected.

BACKGROUND

Unicuspid aortic valve (UAV) is a rare congenital anomaly characterised by the presence of a solitary aortic leaflet. It commonly presents in males in their 30s–40s with dyspnoea, chest pain and light headedness resulting from heart failure secondary to premature valve calcification.¹ These symptoms may take several years to develop;^{2–7} however, cases of UAV detected on screening of asymptomatic patients have been documented.^{8–14} UAV has a high incidence of concomitant cardiac pathologies including aortic root and ascending aortic dilation. These pathologies are typically more dramatic in younger patients presenting with UAV, suggesting that perhaps patients with who are younger at diagnosis have a more severe disease phenotype.⁸ Additionally, UAV patients typically present with symptoms at an earlier age and, if untreated, die earlier than patients with bicuspid valves.^{15–17} We present an interesting and successful case of the diagnosis and treatment of a UAV in a 15-year-old girl who presented with an out of hospital cardiac arrest.

CASE PRESENTATION

A 15-year-old girl was brought into the emergency department by ambulance after experiencing an out of hospital cardiac arrest. She had been watching television at the time of the cardiac arrest. She had a sudden onset of nausea followed by vomiting and collapse. Cardiopulmonary resuscitation was started immediately by her sister. On arrival of the paramedics, ventricular fibrillation (VF) was found on cardiac monitoring. Defibrillation was successfully performed. On arrival to the emergency department, the patient was intubated and ventilated and induced therapeutic hypothermia was started for cerebral protection. She was extubated 24 h later with no focal neurological symptoms. After resuscitation, a thorough collateral history

Figure 1 The prolapsing valve leaflet is located below the aortic annulus in LVOT (left ventricle outflow tract).

was obtained in the intensive care unit. Physical examination revealed a thin teenage girl with a loud readily audible early diastolic murmur consistent with aortic regurgitation (AR). Heart rate was 60 bpm. Respiratory examination was unremarkable.

After admission, investigations for the aetiology of the cardiac arrest revealed an abnormal ECG with prolonged QT interval. Haematological tests including magnesium, phosphate and calcium, were normal. Transthoracic echocardiogram (TTE) revealed severe eccentric AR with mild aortic stenosis (AS), with a mean gradient of 21 mm Hg, a severely dilated left ventricle with a left ventricular internal diastolic diameter (LVIDd) of 72 mm, mildly impaired left ventricular (LV) function with an ejection fraction (EF) of 40% and mild dilation of the ascending aorta (32 mm) (figures 1–4). The valve morphology was unclear. A bicuspid or rheumatic valve was suspected. Only on

Figure 2 Transthoracic echocardiogram demonstrating severe aortic regurgitation with eccentric jet (long axis view).

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Figure 3 Visible stenotic unicuspid aortic valve with a single commissure ('C'). The 2 raphe are annotated with 'R.'

transoesophageal echocardiogram did a UAV became apparent (figure 5). This was confirmed intraoperatively. An intraoperative photograph is provided in figure 6. Figure 7 is a photograph of the pathology specimen.

On direct questioning, the patient denied dyspnoea, orthopnoea, chest pain, palpitations and peripheral oedema at any time prior to the cardiac arrest. She was an otherwise healthy and active school-going teenager who was not taking any medications or recreational substances. Family history was negative for sudden cardiac death, valvular abnormalities and ischaemic heart disease.

After a thorough discussion between the patient, her parents, her consultant cardiologist and her cardiothoracic surgeon, the decision was made to implant a bioprosthetic valve. Issues discussed included the need for ongoing anticoagulation with a mechanical valve, the teratogenesis of warfarin, which is important in a young female, the reoperation rates and associated risks for both mechanical and bioprosthetic valves, and potential percutaneous options. Ultimately, the patient did not want to take warfarin or other anticoagulants indefinitely, and chose the bioprosthetic valve despite the future need for earlier reoperation and replacement.

She underwent an uneventful aortic tissue valve replacement (St Jude Medical Trifecta Valve, St Paul, USA, size 25) and was discharged on postoperative day 6. Postoperative TTE demonstrated a well seated bioprosthetic aortic valve replacement with a mean gradient of 7 mm Hg and no AR. The ventricle was mildly dilated with low normal LV systolic function.

Figure 4 A severely dilated left ventricle in association with unicuspid aortic valve (UAV) is demonstrated.

Figure 5 Image taken in systole.

OUTCOME AND FOLLOW-UP

On follow-up postoperative TTE at 3 months, a well-functioning tissue aortic valve with a mean gradient of 7 mm Hg with no paravalvular leaks was demonstrated. EF was 50%. LVIDd was 46 mm. The patient attended the outpatients department 12 months postoperatively. She is asymptomatic and remains in class I per the New York Heart Failure Association (NYHA) classification. She has returned to full activities, including sports. She has been closely monitored for her prolonged QT interval, which is shortening, but has not had an internal automated cardiac defibrillator implanted.

DISCUSSION

During normal embryonic development, the aortic valve is formed from three tubercles. Each tubercle develops a cusp with an associated commissure and sinus of Valsalva. Fusion of the cusps results in a unicommissural UAV. Two types of UAV are found (table 1). The unicommissural variant has a lateral attachment to the aorta at the level of the orifice. It is typically slit or teardrop shaped. The acommisural variant, which lacks such an attachment, is due to the underdevelopment of all three cusps and is typically pinhole shaped with a central orifice. The orifice of the unicommissural type is eccentric and larger than in the acommisural type. Thus, signs and symptoms of stenosis typically present later in life while acommisural UAV typically present in early childhood (table 1).

Figure 6 Intraoperative image of unicuspid aortic valve.

Figure 7 Pathology specimen of unicuspid aortic valve.

The incidence of UAV is 0.002% of a population undergoing echocardiography.¹⁸ However, the true incidence in the general population has never been investigated through echocardiography of a completely healthy sampling of patients. In contrast to the more commonly reported and studied bicuspid valve, no consensus on the diagnosis or treatment of UAV presently exists.

In the relatively few cases reported to date, patients with UAV have been reported to present earlier, have increased cardiac pathologies and, if untreated, die earlier than patients with bi and tricuspid valves. Roberts and Ko¹⁵ studied excised stenotic aortic valves from 932 patients and found that the mean age of patients with UAVs was 51 ± 14 years, 67 ± 11 years in patients with bicuspid aortic valves (BAVs) and 74 ± 8 years in patients with tricuspid valves. In a separate necropsy study, Roberts *et al*¹⁹ documented a mean age of death of 48 ± 15 years in those with UAV versus 56 ± 15 years in those with BAV. Additionally, unicuspid valves are typically heavily calcified and therefore are the most stenotic and heaviest, followed by bicuspid, as demonstrated in Roberts' study of the weights of excised stenotic aortic valves.²⁰ Thus valve dysfunction typically manifests 10–20 years earlier in UAV versus BAV.²¹

The commonest concomitant cardiac pathologies found with UAV include AS and/or regurgitation, aortic root and ascending aortic dilatation and aortic dissection. Mookadam's systematic review of 231 cases in 38 articles revealed a fivefold to ninefold higher risk of aortic dissection in a UAV cohort than that in the

general population. Dissection was also shown to occur at a younger age in UAV versus bicuspid and tricuspid patients.¹

Sudden death is an unusual presentation of this anomaly. We report the youngest patient and only female to experience a cardiac arrest in the presence of a UAV in the literature to date. This case is unusual due to the lack of the typical symptoms typically associated with UAV. Typically, patients experience symptoms in their fourth to sixth decades in the case of a BAV and third and fourth decades in the presence of a UAV. The cardiac arrest in this case was most likely due to the severe AR leading to a severely dilated ventricle. This, in conjunction with a reduced EF can cause VF. Additionally, this is the only reported case of a UAV patient who experienced a cardiac arrest with a successful outcome.

Two other cases of sudden death associated with UAV are reported in the literature. The first is a case of a 26-year-old man who died suddenly while dancing in a disco. He had no known medical history and no symptoms of feeling ill prior to his death. UAV was found on autopsy.²² The second case is of a 16-year-old boy with a UAV and 5-year history of increasing fatigue, chest pain and dyspnoea on exertion who died from a VF cardiac arrest.²³ Agnihotri *et al*⁸ found an 'abnormal' pattern of calcification in 11 of 12 patients with UAV. The calcification was of a large volume and extended into the interventricular septum just lateral to its membranous component in six patients. Such calcification may affect the conduction system causing arrhythmias leading to sudden death; however, this is more likely in older patients and is unlikely to have been a contributing factor in our case.

Younger patients with unicommissural UAV and concomitant ascending aortic aneurysm have been found to have more marked dilation when compared to older UAV patients suggesting that those diagnosed at a younger age have a more aggressive disease phenotype with more severe calcification and dilation.^{8 24} However, it is not known how fast the disease progresses in the individual patients and whether or not it is due to genetic and/or environmental factors. Only one case, in which UAV in a 34-year-old man was followed with echocardiography, is documented in the literature. In 5 years, the patient's AR and stenosis progressed from 'mild to moderate' to 'severe'.²⁵ Studies to determine both the true incidence in the general population and the course of the condition in the asymptomatic patient are warranted.

Learning points

- ▶ Unicuspid aortic valve (UAV) is a rare congenital anomaly commonly affecting patients in their third and fourth decades, and presenting with signs of heart failure.
- ▶ Our case is that of a previously asymptomatic teenage girl with a UAV who presented with cardiac arrest and was treated successfully.
- ▶ Only two other similar cases have been reported in the literature, both were of slightly older male patients.
- ▶ Our case highlights the morbidity associated with the anomaly supporting the need for careful assessment of the valve in cases where UAV is suspected.

Correction notice This article has been corrected since it was published online on the 15 July 2015. Two videos has been removed as they included patient specific details.

Competing interests None declared.

Table 1 Unicuspid aortic valve variants

Type of unicuspid valve	Unicommissural	Acommissural
Age of diagnosis	Typically in adulthood (30s–40s)	Typically in childhood (before age 4)
Embryology	Fusion of the 3 cusps	Failure of the development of the 3 cusps
Attachments	1 lateral attachment and 2 raphe where the leaflets should have formed	No lateral attachment to the aorta
Orifice	'Exclamation point' shaped eccentric orifice ⁴	Central stenotic orifice
Typical presentation	Slowly developing heart failure	Severe AS

AS, aortic stenosis.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- Mookadam F, Thota VR, Garcia-Lopez AM, *et al.* Unicuspid aortic valve in adults: a systematic review. *J Heart Valve Dis* 2010;19:79–85.
- Falcone MW, Roberts WC, Morrow AG, *et al.* Congenital aortic stenosis resulting from a unicommissural valve. Clinical and anatomic features in twenty-one adult patients. *Circulation* 1971;44:272–80.
- Murphy BA, Groban L, Kon ND. Diagnosis of a unicuspid aortic valve using transesophageal echocardiography. *J Cardiothorac Vasc Anesth* 2003;17:82–3.
- Ucak A, Inan K, Onan B, *et al.* Left sinus of valsalva aneurysm in an adult with congenital unicuspid aortic valve. *Int J Cardiol* 2011;149:e21–23.
- Ohata T, Miyamoto Y, Takahashi T, *et al.* Sinus of Valsalva aneurysm with congenital aortic stenosis and aortic coarctation. *Asian Cardiovasc Thorac Ann* 2004;12:75–7.
- Nanda S, Longo S, Arastu MI. Unicuspid aortic valve, hand anomalies: a heart-hand syndrome. *Am J Med Sci* 2010;339:296–9.
- Ota T, Okada K, Morimoto Y, *et al.* Unicuspid aortic valve mimicking quadricuspid valve. *Heart Vessels* 2007;22:352–4.
- Agnihotri AK, Desai SC, Lai YQ, *et al.* Two distinct clinical presentations in adult unicuspid aortic valve. *J Thorac Cardiovasc Surg* 2006;131:1169–70.
- Ermis N, Bilgi M, Altay H, *et al.* An asymptomatic unicuspid aortic valve with aortic aneurysm and secundum type atrial septal defect. *Clin Cardiol* 2010;33:E38–9.
- Mohan JC, Tomar D, Shekhar C, *et al.* Unicuspid unicommissural semilunar valvular stenoses: realtime 3D-echocardiographic images. *Indian Heart J* 2010;62:449–50.
- Singh D, Chee TS. Incidental diagnosis of unicuspid aortic valve in an asymptomatic adult. *J Am Soc Echocardiogr* 2008;21:876.e875.
- Karakulak UN, Okutucu S, Evranos B, *et al.* Incidental diagnosis of the unicuspid aortic valve in a patient with ulcerative colitis. *Int J Cardiol* 2012;155:S226.
- 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;19:102–4.
- Fijalkowski M, Jaguszewski M, Pienkowska J, *et al.* Incidental diagnosis of unicuspid aortic valve in an asymptomatic adult. *Kardiol Pol* 2013;71:209.
- 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;111:920–5.
- Roberts WC, Vowels TJ, Filardo G, *et al.* Natural history of unoperated aortic stenosis during a 50-year period of cardiac valve replacement. *Am J Cardiol* 2013;112:541–53.
- Collins MJ, Butany J, Borger MA, *et al.* Implications of a congenitally abnormal valve: a study of 1025 consecutively excised aortic valves. *J Clin Pathol* 2008;61:530–6.
- Novaro GM, Mishra M, Griffin BP. Incidence and echocardiographic features of congenital unicuspid aortic valve in an adult population. *J Heart Valve Dis* 2003;12:674–8.
- Roberts WC, Vowels TJ, Ko JM. Natural history of adults with congenitally malformed aortic valves (unicuspid or bicuspid). *Medicine (United States)* 2012;91:287–308.
- Roberts WC, Ko JM. Relation of weights of operatively excised stenotic aortic valves to preoperative transvalvular peak systolic pressure gradients and to calculated aortic valve areas. *J Am Coll Cardiol* 2004;44:1847–55.
- Schafers HJ, Aicher D, Riodionychewa S, *et al.* Bicuspidization of the unicuspid aortic valve: a new reconstructive approach. *Ann Thorac Surg* 2008;85:2012–18.
- Roberts WC, Roberts CC, Vowels TJ, *et al.* Effect of coronary bypass and valve structure on outcome in isolated valve replacement for aortic stenosis. *Am J Cardiol* 2012;109:1334–40.
- Roberts WC, Morrow AG. Congenital aortic stenosis produced by a unicommissural valve. *Br Heart J* 1965;27:505–10.
- Roberts WC, Vowels TJ, Ko JM. Comparison of interpretations of valve structure between cardiac surgeon and cardiac pathologist among adults having isolated aortic valve replacement for aortic valve stenosis ((plus or minus) Aortic Regurgitation). *Am J Cardiol* 2009;103:1139–45.
- Dandes E, Kirsch J, Novaro G. Congenital rock and a hard place: unicuspid aortic valve with sinus of valsalva aneurysm. *Int J Cardiovasc Imaging* 2012;28:453–4.

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