CASE REPORT

Symptomatic unicuspid aortic valve

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SUMMARY

A 22-year-old man with typical angina was seeking medical attention at primary health clinics for a couple of months. Owing to his young age and the absence of coronary artery disease risk factors, he was assured of no serious problem. Proper examination at a referral centre revealed weak peripheral pulses with diminished and delayed carotid upstroke. A normal S1 with a soft S2 were audible. A 3/6 late peaking systolic murmur was best heard in the aortic area radiating to the neck. Symptomatic bicuspid aortic valve disease was suspected. Diagnosis of unicuspid aortic valve was established by transoesophageal and three-dimensional echocardiography. The valve was successfully replaced with a mechanical prosthesis. The patient remains asymptomatic at 1 year follow-up.

BACKGROUND

Chest pain in the young is usually attributed to non-cardiac causes. Anxiety and stress supersede other causes of chest pain in young otherwise healthy patients with normal examination. This young man had been seeking medical attention for some time. In current times there is more emphasis on documentation. Unfortunately, this is at the cost of less time for history-taking and clinical examination. Additionally, due to the availability of multi-modality imaging, clinical examination has become a fading skill. Our patient had a typical history and significantly loud systolic murmur. Though his final diagnosis of unicuspid aortic valve (UAV) is a very rare finding, his presenting signs and symptoms should have prompted early work up. This case illustrates the importance of a basic bedside examination in this high tech era.

CASE PRESENTATION

A 22-year-old otherwise healthy man was referred to our institution, with a history of progressively worsening chest pain. Initially, symptoms occurred only after strenuous activity but, recently, a normal to brisk walk would result in left-sided chest pain radiating to the neck and left shoulder with complete resolution of symptoms at rest. The patient did not have any coronary artery disease risk factors and led a healthy life style. He denied using tobacco, alcohol or recreational drugs. There was no history of dizziness syncope or presyncope. The patient denied any family history of sudden death at a young age. He outplayed his friends in field sports during most of his teenage years. He was not on any regular medication.

Physical examination revealed weak peripheral pulses. Heart rate was 68 bpm. Blood pressure was 110/70 mm Hg in both arms. Body temperature

and saturations were normal; there was no cyanosis or clubbing. Neck examination revealed diminished and delayed carotid upstroke. There was normal jugular venous pressure. The chest was clear to auscultation without any added sounds. Precordium was normal. A normal S1 with a soft S2 were audible. A 3/6 late peaking systolic murmur was best heard in the aortic area radiating to the neck. The murmur became somewhat softer with Valsalva manoeuvre. There was no radiofemoral delay. The rest of the clinical examination was within normal limits.

INVESTIGATIONS

Baseline blood tests revealed normal complete blood count. Metabolic panel, including electrolytes, lipids, liver and kidney function tests, was normal.

ECG: Normal sinus rhythm, normal intervals with voltage criteria for left ventricular hypertrophy.

Chest X-ray was normal. Transthoracic echocardiography showed left ventricular hypertrophy, with normal left ventricular function. Severe aortic stenosis was noted and the valve was suspicious for a unicuspid versus bicuspid valve. Other valves were normal.

Transoesophageal echocardiography (figure 1A, B) and three-dimensional (3D) echocardiography (figure 2) confirmed the UAV with a normal ascending aorta. Coronary CT showed a normal ascending, arch and descending aorta. Coronary arteries were angiographically normal. Other associated congenital anomalies were excluded.

DIFFERENTIAL DIAGNOSIS

Angina with systolic murmur in young adults should raise the suspicion of dynamic or fixed left ventricular outflow tract obstruction. Hypertrophic obstructive cardiomyopathy, aortic valvulopathy from bicuspid, unicuspid and, rarely, rheumatic valve disease and sub or supra aortic membranes, should be excluded.

Angina without cardiac murmur in the young should prompt the physician to investigate for anomalous coronary arteries. Coronary aneurysm from Kawasaki disease may present with anginal symptoms. Aggressive atherosclerotic coronary artery disease may result from metabolic, lipid, glucose, steroid and autoimmune abnormalities.

TREATMENT

Prior to surgery, ample time was spent on patient education and evaluating his social support system. The patient was informed regarding life style

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Figure 1 (A) Transoesophageal echocardiographic mid-oesophageal short axis view (149°). Unicuspid valve with eccentric valvular orifice. AA, ascending aorta; LA, left atrium; UAV, unicuspid aortic valve. (B) Transoesophageal echocardiographic mid-oesophageal short axis view (50°). Unicuspid valve with eccentric valvular orifice. LA, left atrium; RA, right atrium; RV, right ventricle; UAV, unicuspid aortic valve.

changes, the importance of lifelong medication compliance and endocarditis prophylaxis. He successfully underwent mechanical aortic valve replacement surgery. The postoperative period was uneventful.

OUTCOME AND FOLLOW-UP

At 1-year follow-up, he remains asymptomatic and compliant with medical instructions. He is enjoying an active and healthy life style and remains a valuable member of the community.

Figure 2 Three-dimensional transoesophageal echocardiographic mid-oesophageal short axis view (65°). Unicuspid valve with eccentric valvular orifice.

DISCUSSION

UAV is a rare congenital heart anomaly that results from the fusion of the three tubercles during embryonic life. It may occur alone or may be associated with other anomalies such as patent ductus arteriosus, aortic aneurysms and coarctation of the aorta.¹ The reported incidence is about 0.02% in patients referred for echocardiography.² A much higher incidence of 4–6% is observed in patients undergoing surgery for aortic stenosis.³

Two types of UAV have been described.⁴ The acommisural type has a central pinhole-shaped opening and presents with symptoms that require intervention in infancy and childhood.⁵ In the unicommissural UAV, as in our patient, the valve has a slit-like opening.¹ Symptoms usually develop in early to mid-adulthood. Aortic root dilation seems to be caused by medial changes in the aortic tissue.⁶ Aortopathy has a bimodal distribution, older patients without aortic root dilation, and younger with UAV having an aggressive form of aortic root dilation.⁷ Early valve dysfunction, pathological aortopathy and dystrophic calcification are common features in unicuspid and bicuspid valves.

Dyspnoea on exertion, chest pain and syncope are common to all stenotic aortic valves irrespective of the number of cusps. These symptoms develop and progress to necessitate surgery earlier in unicuspid, followed by bicuspid and lastly by tricuspid valves.⁸ UAVs are heavily calcified and weigh twice as much as stenotic tricuspid aortic valves.⁹ This calcification may affect the conduction system of the heart.

Diagnosis is usually established by transthoracic, transoesophageal and 3D echocardiography.¹⁰ Cardiac CT or Cardiac MR are vital to access cardiac and extracardiac associations with this congenital valve problem.^{11 12} Treatment in adults is mostly aortic valve replacement. If aortopathy is found, aortic annulus, root and ascending aorta replacement is performed as well.

Learning points

- ▶ History taking and clinical examination still remain the most important steps in diagnosis and subsequent management of patients.
- ▶ Bicuspid aortic valve is a well-known entity. However, increased awareness of unicuspid aortic valve (UAV) disease with early presenting symptoms is essential. The diagnosis and management of each such reported case adds to our pool of knowledge.
- ▶ Non-invasive, multimodality cardiac imaging obviates the need for invasive angiogram.
- ▶ Patient education and strengthening of their social support system are of key importance in the management of young patients on lifelong medications.

Competing interests None declared.

Patient consent Obtained.

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