CASE REPORT

Quadricuspid aortic valve and anomalous origin of the left circumflex coronary artery in a young man with Down syndrome

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Background
Quadricuspid aortic valve accounts for 0.008–0.033% of all the cases of congenital heart diseases in autopsy series, and 0.013–0.043% in echocardiographic examinations. It was first described by Balington (London Medical Gazette 1862) [1].

In accordance to the classic Hurwitz and Roberts classification, quadricuspid aortic valve can be subdivided into seven types. Type A (four equal cusps) is the most common one, whilst type G (four unequal cusps) is the rarest [2]. A further subtype, called type H, which comprised two equal-sized smaller cusps and two unequal-sized larger cusps, has been described [3]. Aortic insufficiency is the most frequent dysfunction associated with quadricuspid aortic valve, while stenosis is rare [2].

Quadricuspid aortic valve usually appears as an isolated cardiac defect. However, in 18% of the cases, other associated cardiac defects have been described: abnormal origin of the coronary arteries (10%), atrial septal defect, ventricular septal defect, patent ductus arteriosus, mitral valve prolapse, tetralogy of Fallot, partial atrio-ventricular canal defect, subaortic stenosis, nonobstructive hypertrophic cardiomyopathy, aneurismatic dilatation of the ascending aorta [2].

Down syndrome, also called Trisomy 21, is the most common chromosomal abnormality associated with congenital heart diseases. With respect to that, about 40–50% of Down syndrome patients suffer from cardiac abnormalities, such as atrial septal defect, ventricular defect, atrio-ventricular defect, tetralogy of Fallot, patent ductus arteriosus [4]. Some cases of isolated bicuspid valve associated with Down syndrome have been described, as well as an only recent case report about the coexistence of isolated quadricuspid aortic valve and Down syndrome [5].

We report the case of a man with Down syndrome and the concomitant presence of quadricuspid aortic valve and anomalous origin of the left circumflex coronary artery. To the best of our knowledge this is the first description of the concomitant presence of the above stated three conditions. Informed written consent for the
publication of this case report has been obtained from the patient’s parents.

Case Presentation

A 36-year-old man with Down syndrome was admitted to our Cardiology Department for a diastolic murmur at the lower left sternal border during cardiac auscultation in outpatient clinic. The patient’s medical history was unremarkable for any other cardiac abnormality. However, notwithstanding the presence of Down syndrome, interestingly echocardiographic examination had not previously performed on this subject. A physical examination showed appropriate growth for Down syndrome reference nomograms and a 2/4 diastolic murmur over the left sternal edge. Systemic blood pressure was 116/74 mmHg.

Investigations

Electrocardiogram showed sinus rhythm with a heart rate 68 beats/min. Chest X-ray was normal as well.

Transthoracic echocardiography detected a quadricuspid aortic valve with a mild central insufficiency. Furthermore, the left circumflex artery arose from the proximal right coronary artery. The course of the right coronary artery was normal, whereas the left circumflex artery had a retroaortic course. The patient’s parents did not give their consent to perform a coronary angiographic study in this subject.

A better definition of the aortic valve was achieved through transoesophageal echocardiography, which displayed the presence of four equal cusps, thus allowing to classify the quadricuspid aortic valve as type A according with the Hurwitz and Roberts classification (Fig. 1).

Treatment and Follow-Up

Since the patient was asymptomatic even during the watt test, with no electrocardiographic signs of inducible ischaemia (left circumflex coronary artery coursed between the great vessels, making this unusual case a low-risk one) and the possible impairment of the aortic insufficiency as time passes, he was enrolled in a close clinical and echocardiographic follow up. Beta blocker (metoprolol) and acetylsalicylic acid were administered, and antibiotic prophylaxis against infective endocarditis was suggested as well. Upon editing this paper, the patient is still in good clinical condition.

Discussion

This is the second case report in literature concerning the association of quadricuspid aortic valve with Down syndrome, and the first one reporting the concomitance of these two abnormalities with the anomalous origin of the left circumflex coronary artery from the right coronary artery.

Quadricuspid aortic valve, as in the present case, is generally detected incidentally during echocardiography or angiography, or at autopsy [2]. The average age at diagnosis is 46-50 years, with a slight prevalence of male gender [2].

The mechanism inducing the development of quadricuspid aortic valve is still unexplained: probably it originates from an anomalous septation of the embryological arterial trunk [2].

Only a minority (16%) of the quadricuspid aortic valves have a normal valvular function. As stated above, insufficiency is the most common valvular dysfunction associated with this anomaly (50–75%), due to the progressive age-related fibrotic thickening of the cusps related to the valvular dishomogeneous mechanical stress, which increases the risk of endocarditis. On the contrary, stenosis is rare [2].

The need of quadricuspid aortic valve surgical correction occurs usually between the fifth and sixth decade of life [2]. This is why our patient, asymptomatic and with a mild aortic insufficiency, was enrolled in a close follow up. Echocardiography is a simple and useful diagnostic tool to detect this anomaly.

Valvular replacement is the main surgical treatment for quadricuspid aortic valve. However, only for selected cases, surgical repair may be possible [6]. Furthermore, owing to the possible presence of associated coronary arteries abnormalities, surgeons should be aware of the coronary ostial obstructions [6].

As stated above, quadricuspid aortic valve is at increased risk of infective endocarditis, with no relationship with quadricuspid aortic valve subtype [2].
In the light of this consideration, though not strictly recommended from the Guidelines, an antibiotic prophylaxis was suggested.

With regard to the anomalous origin of the left circumflex coronary artery from the right coronary sinus, it is a well-known anatomical variant, which can be detected in approximately 0.37–0.7% of all patients undergoing coronary angiography [7]. As a general rule, a coronary anomaly is known to be at high ischemic risk when the anomalous artery originates ectopically from the opposite sinus and courses between the aorta and the pulmonary artery.

In our patient, the left descending coronary artery had an anterior free wall course. The course of the right coronary artery was normal as well, while the left circumflex coronary artery had a retroaortic course. It means no coronary artery coursed between the aorta and the pulmonary artery, making this rare case a low-risk one, as testified by the fact that the watt test of this patient was normal. However, although this anomaly was classified as benign and asymptomatic, a medical therapy with beta-blocker and acetylsalicylic acid was instituted. In fact, regarding the possible association between atherosclerosis and coronary artery anomalies, an analysis from the Coronary Artery Surgery Study showed that anomalous left circumflex coronary artery arteries had a significantly greater degree of stenosis than that in non-anomalous arteries in age- and gender-matched control patients \((P = 0.02)\) [8]. Taking into account both these considerations and the small area supplied by the left circumflex coronary artery, the Authors decided to treat the patient conservatively and he did well on medical therapy.

**Conflict of Interest**

None declared.

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