RESEARCH LETTER

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An Unexpected Aortic Valve in Trisomy 21

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TO THE EDITOR:

Quadricuspid aortic valve (QAV), a rare congenital anomaly [Hurwitz and Roberts, 1973], was first described by Balington in 1862 [Robicsek et al., 1969]. The prevalence of this entity ranges from 0.013% to 0.043% [Holm et al., 2004]. Aortic regurgitation is more prevalent valvular dysfunction than stenosis in these patients [Tutarel, 2004]. Trisomy 21, also called Down syndrome is the most commonly seen chromosomal abnormality that often associated with congenital heart defect [Stoll et al., 1998]. Some cardiac valvular abnormalities have been reported in patients with Down syndrome but QAV has not been reported previously [Aughton et al., 1995]. Herein we present a patient with Down syndrome and QAV. To best our knowledge this is the first report of co-occurrence of Down syndrome and QAV.

A 6-year-old boy with trisomy 21 was referred to our pediatric cardiology department due to the diastolic murmur on physical examination at outpatient clinic. The patient's medical history was unremarkable for any cardiac abnormality. Physical examination showed appropriate growth for Down syndrome growth charts and cardiac auscultation revealed a early diastolic murmur along the lower left sternal border. Electrocardiography was normal. Transthoracic echocardiographic examination showed structurally normal heart with exception of unexpected QAV with four equal cusps (Fig. 1a,b). Color Doppler imaging showed mild aortic regurgitation (Fig. 1c). Upon this, the patient was followed for aortic regurgitation and given prophylaxis for infective endocarditis.

Different type of congenital cardiac anomalies such as atrioventricular septal defects, atrial septal defects, ventricular septal defects, tetralogy of Fallot and patent ductus arteriosus can be components of the 40–50% of patients with trisomy 21 who have structural heart defect [Stoll et al., 1998]. Isolated bicuspid aortic valve associated with Down syndrome was also reported [Aughton et al., 1995]. But coexistence of isolated QAV and trisomy 21 has not been documented previously.

A QAV is a rare congenital anomaly and is usually detected incidentally [Hurwitz and Roberts, 1973]. The mechanism of this anomaly is still unexplained, abnormal septation of the embryological arterial trunk can lead to QAV [Formica et al., 2004]. Regurgitation is the more commonly seen valvular dysfunction due to abnormal leaflet coaptation which increases the risk of endocarditis. Coronary artery abnormalities can be seen in 10–30% of the patients [Bakirci et al., 2012]. Transthoracic echocardiography is a simple and useful diagnostic tool to detect this anomaly. Seven types of QAV have been described: (A) four equal cusps; (B) three equal cusps with one smaller cusp; (C) two equal smaller cusps and two equal larger cusps; (D) one large, two intermediate and one small cusp; (E) three equal cusps with one large cusp; (F) two equal larger with two unequal smaller cusps and (G) four unequal cusps [Hurwitz and Roberts, 1973]. In patients with severe aortic regurgitation aortic valve replacement or repair can be done. But, due to the accompanying coronary artery anomalies, surgeons should be aware of the coronary ostial obstructions [Bakirci et al., 2012]. In light of these data, our patient is diagnosed as type A QAV (four equal cusps) with mild aortic regurgitation. The patient will be followed up closely for deterioration of the aortic regurgitation.

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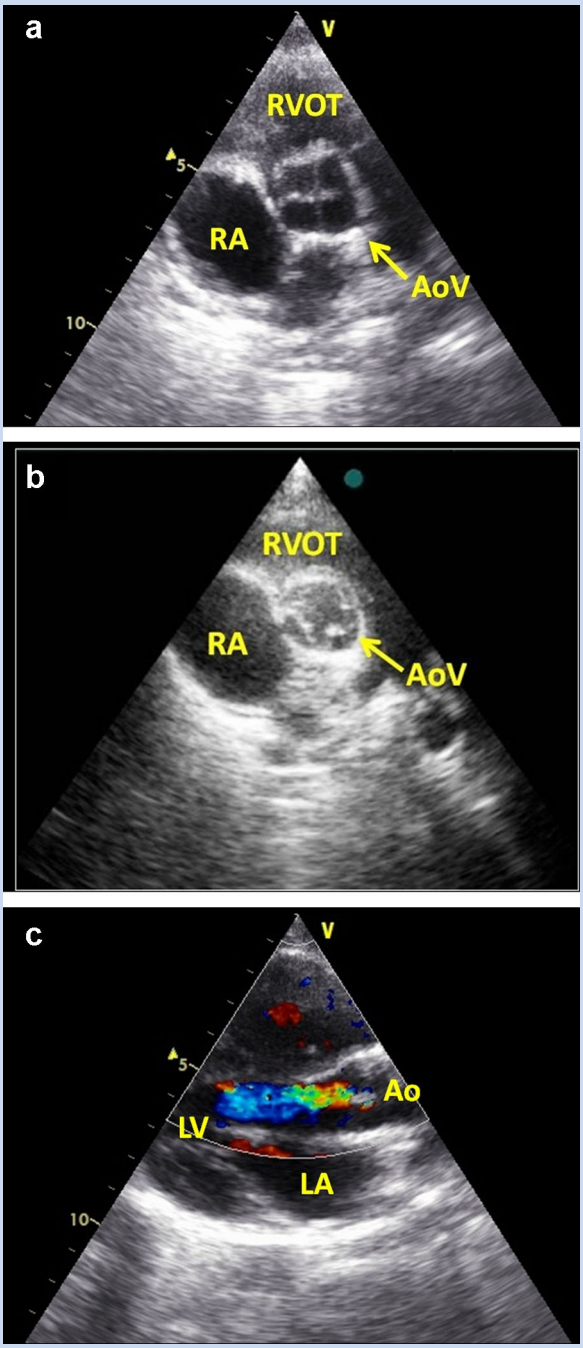

FIG. 1. Parasternal short axis echocardiographic view of quadricuspid aortic valve (a,b) and color Doppler (c) imaging of aortic regurgitation. AoV, aortic valve; LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle; RVOT, right ventricle outflow tract.

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