Case report - Congenital
Aarskog syndrome with aortic root dilatation and sub-valvular aortic stenosis: surgical management

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Received 4 May 2004; received in revised form 30 June 2004; accepted 20 September 2004

Abstract

Aarskog syndrome is a familial condition associated with craniofacial anomalies, genital malformations and short stature. Affected children have significantly higher chance of having congenital heart disease (CHD) than the general population. We report the case of a child afflicted with progressive aortic root dilatation and sub-valvular aortic stenosis, successfully managed with aortic root and valve replacement. Given the association between Aarskog syndrome and CHD, cardiac surveillance should be undertaken in all affected children.

Keywords: CHD; Aarskog syndrome; Aortic root surgery

1. Introduction

Aarskog syndrome is an X-linked recessive developmental disorder [1] characterised by short stature, round facies, hypertelorism, widow’s peak, short anteverted nose, hyperextension of the proximal interphalangeal joints, cutaneous syndactyly, inguinal hernia(s) and genital malformation [2]. This disorder is thought to be causally linked to congenital heart disease and reports of atrial septal defects, ventricular septal defects, pulmonary stenosis, mild coarctation of the aorta and aortic valve stenosis and insufficiency have appeared in the literature [3].

We report the presence and successful management of sub-valvular aortic stenosis and aortic root dilatation in a child with Aarskog syndrome and propose that in view of the higher incidence of CHD in Aarskog syndrome, cardiac surveillance should be undertaken in all affected children.

2. Case report

A male newborn was noted to have hypertelorism and low-set ears soon after birth. There was no relevant family history and the pregnancy and delivery were uncomplicated. Syndrome work-up was undertaken and Aarskog syndrome was diagnosed. Cardiac workup showed a small patent ductus arteriosus, a non-stenosed bicuspid aortic valve but no other abnormalities. The baby was pink with good peripheral pulses and no further action was taken.

At 2.5 years a routine examination showed ejection systolic murmur most audible over the left sternal edge and a chest X-ray showed abnormal cardiac contour. Echocardiogram revealed the aortic root to be dilated to 3 cm. He was under paediatrician review and grew well.

During his next cardiac investigation 4.5 years later the ejection systolic murmur radiating to the carotids with slight aortic regurgitation a non-stenotic valve, slight left ventricular hypertrophy on ECG and worryingly, an aortic root of 5.08 cm on echocardiogram. This prompted Magnetic Resonance Imaging of the upper mediastinum (Fig. 1), which showed a root dilatation of 4.2 cm, and a subvalvular aortic stenosis, attributable to a non-intrusive membrane. The imaging was repeated 6 months later, by which time the aorta had dilated to 4.8 cm.

He was referred for surgery, which was carried out when he was 7 years old. At operation the root dilatation was found to extend approximately 1.5 cm below the origin of the innominate artery down to the aortic root, a functionally bicuspid valve with fusion of right and left coronary commissures with a central gap leading to moderate regurgitation, and a sub-aortic ledge extending underneath the left coronary cusp. Cardiopulmonary bypass was established and the patient was cooled to 28 °C. The fibrous aortic membrane was resected and the aortic annulus was enlarged using a pericardial gusset. The aortic root was excised and was replaced with a tube graft and a size 21 CarboMedics prosthetic valve (CarboMedics, USA). In addition, the coronary arteries were reimplanted using the Bentall’s procedure. He made an uneventful recovery and was discharged 7 days later on long-term warfarin.

Histopathology showed focal diffuse intimal thickening, with the media showing no evidence of vascularisation, fibrosis, inflammation or medio-necrosis, but there was some elastin fragmentation in the outer third of the media. There was no atheroma. He is under annual cardiac review and was last reviewed in 2003 aged 14. He has remained well on warfarin therapy and has ‘normal’ prosthetic valve sounds and the valve and replaced root function normally.

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on echocardiography. The ECG remains unchanged with sinus rhythm, normal axis and borderline left ventricular hypertrophy on voltage criteria. He has normal exercise tolerance but is advised to avoid strenuous exercise and competitive sports.

3. Discussion

The Aarskog syndrome is a rare X-linked recessive condition, which classically manifests by its effects on the genital and skeletal systems. It is believed to be the result of mutations in gene FGD1 [1,4], that is first expressed in the onset of skeletal ossification in embryogenesis [5]. While there is no obvious theoretical framework to link Aarskog syndrome with congenital heart disease, strong association between the two have led some to believe that Aarskog syndrome causes CHD. A number of CHD conditions have been reported, including atrial and ventricular septal defects, coarctation of the aorta and aortic valve stenosis and incompetence [3], but we are not aware of any reports of aortic root dilatation occurring with this condition, and this report may present the first case.

Given the fact that Aarskog syndrome is rare and the link with CHD is not fully established, affected children are not routinely offered cardiac surveillance. In this case, the child was originally referred for the investigation of patent ductus arteriosus, and was incidentally found to have a bicuspid aortic valve. His second cardiology referral as a toddler was also coincidental and upon noting an abnormal radiological appearance in a chest X-ray taken as part of his craniofacial treatment. The discovery of progressive aortic root dilation and its successful timely management therefore owed much to serendipity.

Fernandez et al. noted four cases of CHD in 30 Japanees and two CHD in 169 non-Japanese reported cases of Aarskog syndrome, which is in excess of the 0.8% general population risk [3]. We therefore propose that all patients with Aarskog syndrome be screened for congenital heart disease.

References