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Case Report

Isolated congenital absent aortic valve cusps in a fetus: A case report

S Sivanandam, S M Mackey-Bojack

From Department of Pediatrics and Jesse E. Edwards Registry of Cardiovascular Disease, University of Minnesota, Minneapolis, USA

Correspondence to: Shanthi Sivanandam, Department of Pediatrics, University of Minnesota, 2450 Riverside Ave, Minneapolis, Minnesota 55454, USA. Phone: +612-626-2755, Fax: +612-626-2784. E-mail: silv0099@umn.edu

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Abstract

Congenital absence of aortic valve cusps is an extremely rare congenital heart malformation. The absence of aortic valve cusps leads to severe aortic regurgitation with resultant rapid and progressive heart failure and growth retardation in the fetus. Congenital absence of the aortic leaflets in association with other cardiac defects has been reported in the literature, but the isolated anomaly is not reported yet. We present a case of a fetus with isolated congenital severe aortic regurgitation from absent aortic cusps and pathological specimen of this rare entity.

Key words: *Aortic cusps, Aortic regurgitation, Congenital heart disease, Fetal echocardiography, Prenatal diagnosis*

Congenital absence of aortic valve cusps is an extremely rare and fatal congenital cardiac malformation. Severe aortic regurgitation resulting from the absence of aortic valve cusps leads to rapid and progressive heart failure and growth retardation in the fetus and ultimate fetal demise. Only a few cases have been reported in the literature and absence of aortic valve cusps has been associated with other significant structural malformations of the heart in all reported cases in the literature [1]. Herein, we describe in a fetus with isolated congenital absence aortic valve cusps diagnosed at 27 weeks of gestation and a pathological specimen review.

CASE REPORT

A 31 year old healthy gravid 1, para 0 was referred for detailed fetal cardiac assessment at 27 weeks of gestation. Fetal echocardiogram showed severe cardiomegaly, normal atrioventricular and ventriculoarterial connections with severe aortic regurgitation and absence of the aortic valve cusps [Figs. 1 and 2]. There was right ventricular (RV) and left ventricular (LV) dilatation along with mildly diminished bi-ventricular function. Color flow mapping and pulsed Doppler revealed no stenosis across the aorta, and there were antegrade systolic and retrograde diastolic flow patterns in the aorta. There was diastolic runoff from the thoracic aorta. Transverse arch and the aortic isthmus were appearing normal. Furthermore, mitral, pulmonary and tricuspid leaflets were appearing normal without any regurgitation. The great arteries were normally related and appeared dilated.

Level 2 ultrasound showed symmetric intra-uterine growth restriction, extracardiac anomaly consistent with club feet, cerebral hypoplasia, and agenesis of the corpus callosum. Amniocentesis performed showed normal male karyotype (46XY). Subsequently a fetal magnetic resonance imaging (MRI) performed at 27 weeks of gestational age showed pontocerebellar hypoplasia, delayed sulcation pattern, agenesis of the corpus callosum. Cardiac assessment by fetal and MRI was not possible. The family was counseled about the cardiac and extracardiac diagnosis and the pregnancy ended in fetal demise at 29 weeks of gestation.

Autopsy of the heart confirmed the prenatal findings of a dysplastic aortic valve with nodular cusp tissue that was not completely developed and did not coapt [Fig. 3]. LV was dilated with normal appearing aortic arch. No other intracardiac anomaly was seen.

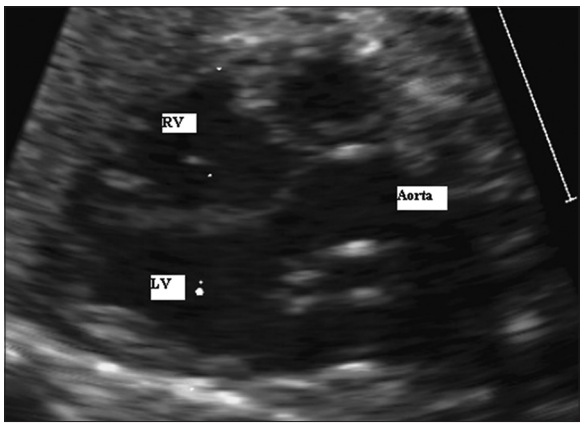

Figure 1: Fetal echocardiogram at 27 weeks of gestation 2D shows absence of the aortic valve and dilated aorta. Left ventricle. Right ventricle

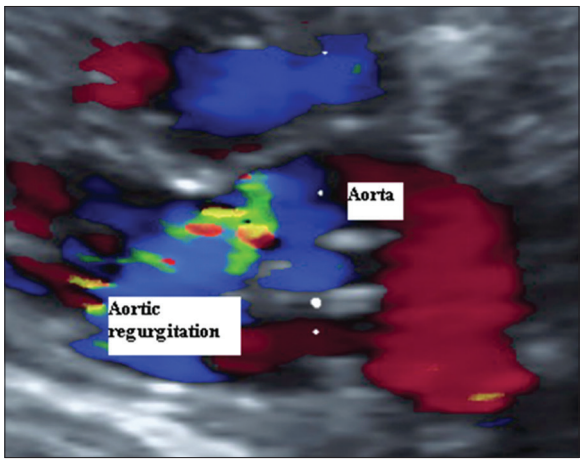

Figure 2: Fetal echocardiogram performed at 27 weeks of gestation demonstrates: Color Doppler across the left ventricular outflow tract shows severe aortic regurgitation

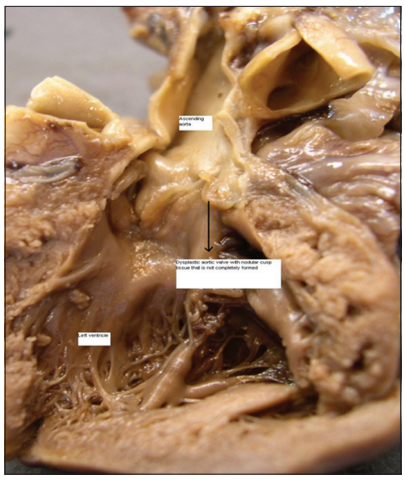

Figure 3: Pathological specimen: Dysplastic aortic valve with nodular cusp tissue that is not completely formed

DISCUSSION

To our knowledge, this is the first report of a fetus with isolated absent aortic valve cusp with no other associated intracardiac lesion. Congenital absence of the aortic leaflets in association with other cardiac defects has been reported and is a rare congenital anomaly [1-4]. Marek et al. in 1996 reported a case of congenital absence of both aortic and pulmonary valves with severe heart failure detected prenatally by cross-sectional and pulsed and color Doppler echocardiography is reported in small for gestational age male fetus in 17th week of gestation. Additional double outlet RV, hypoplastic LV, and ventricular septal defect, as well as multiple extracardiac anomalies, were found by prenatal echocardiographic investigation and confirmed by necropsy examination.

Retrograde diastolic Doppler waveforms retrieved from pulmonary artery, aorta, and umbilical arteries revealed massive insufficiency throughout both the great arteries, which eliminated diastolic placental perfusion, documented by absent anterograde diastolic flow in the umbilical vein [5]. These prenatal echocardiographic findings may contribute to an understanding of the mechanism of rapid and progressive heart failure and growth retardation in the fetus. Severe cardiac failure may explain why the congenital absence of both the aortic and the pulmonary valves has not been described postnatally, and only two fetal cases revealed by necropsy have been published.

CONCLUSION

Congenital absence of the aortic valve cusps is an extremely rare heart malformation. The absent aortic valve is associated with other structural heart malformations in all instances. The majority of fetuses diagnosed with this anomaly die of heart failure. Prenatal diagnosis allows appropriate counseling for the families.

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