Article published online: 2023-05-08

J. Fetal Med. (March 2018) 5:27–30
https://doi.org/10.1007/s40556-018-0152-8

ORIGINAL ARTICLE

Fetal severe aortic regurgitation due to absence of left coronary leaflet of aortic valve

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Received: 1 November 2017 / Accepted: 3 January 2018 / Published online: 17 January 2018
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Abstract Congenital absence of aortic valve cusps is a rare congenital heart malformation, which is commonly associated with other cardiac defects. Isolated absence of aortic valve or one leaflet is extremely rare and has not been described antenatally. We present the first case of a fetus with isolated congenital severe aortic regurgitation from absent aortic cusp and pathological specimen of this rare entity. On fetal echocardiography at 19 weeks of gestation, there were dilated aortic root and severe aortic regurgitation causing 'to and fro' colour flow signals across left ventricular outflow. This caused severe LV systolic dysfunction, significant ventricular dyssynchrony and cardiomegaly. On fetal autopsy, aortic root was dilated. Left coronary cusp was absent while other cusps showed mild thickening. Left ventricle was dilated and hypertrophied. Pulmonary valve leaflets were normal.

Keywords Aortic valve · Fetal Echocardiography · Aortic regurgitation

Introduction

Absent aortic valve or absence of one of the aortic valve leaflets is usually associated with other cardiac defects and is rarely an isolated lesion. Absence of the aortic valve leaflets was first described in association with double-outlet right ventricle in a pathology specimen [1]. It is commonly associated with absence of pulmonary valve leaflets, conotruncal anomalies, ventricular septal defect and hypoplasia of the left ventricle [2–8]. Absent aortic valve has been described in DiGeorge Syndrome [9, 10].

Absence or dysplasia of one of the leaflets of aortic valve is extremely rare [11]. Only 3 cases have been reported till date. To the best of our knowledge, no case has been diagnosed during antenatal period. We are reporting the first case of absent aortic cusp causing severe aortic regurgitation in antenatal period.

Case report

A 21-year-old, gravida 1, para 0 female with no significant medical history and no family history of congenital heart defects presented at 19 weeks' gestation for obstetric ultrasonography and fetal echocardiography. Anomaly scan did not show any extra cardiac abnormality. On fetal echocardiography, ascending aorta and aortic arch were dilated (Fig. 1a). Aortic valve was not visualized but there was a speck at aortic valve level. Pulmonary valve and pulmonary artery were normal. Colour Doppler ultrasonography revealed "to-and- fro" flow in ascending aorta and arch of aorta, there was prograde flow in the left ventricular outflow tract during systole and retrograde flow into the left ventricle during diastole (Fig. 1b, c) consistent with severe aortic regurgitation.

Pulmonary artery, descending aorta and ductus arteriosus had normal flows. Left ventricle was dilated, globular and hypertrophied. Left ventricle systolic functions were depressed with reduced fractional shortening. Close examination of M Mode of RV and LV showed that cause of systolic LV dysfunction was not depressed LV contractility but ventricular dyssynchrony (Fig. 2). LV posterior wall systolic thickening was normal but there was

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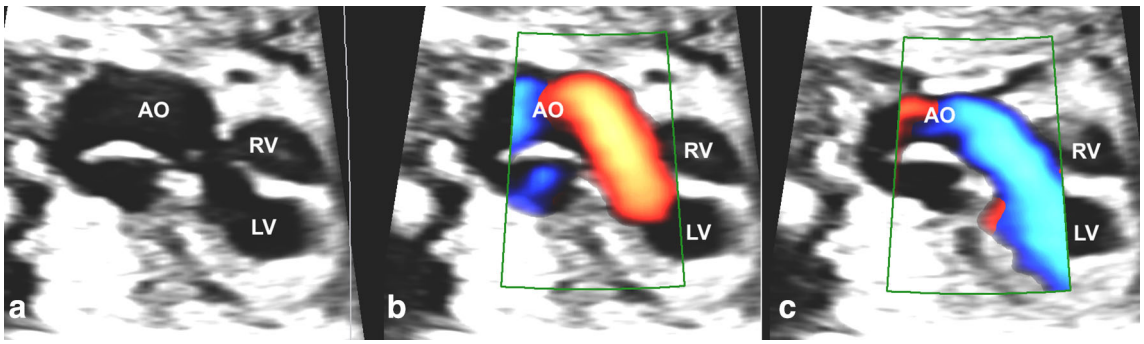

Fig. 1 **a** Shows LVOT view on fetal echo with dilated aorta (AO) **b** Color Doppler of LVOT view showing forward flow into Aorta (AO) from left ventricle (LV) **c** Severe aortic regurgitation shown as blue color from AO to LV

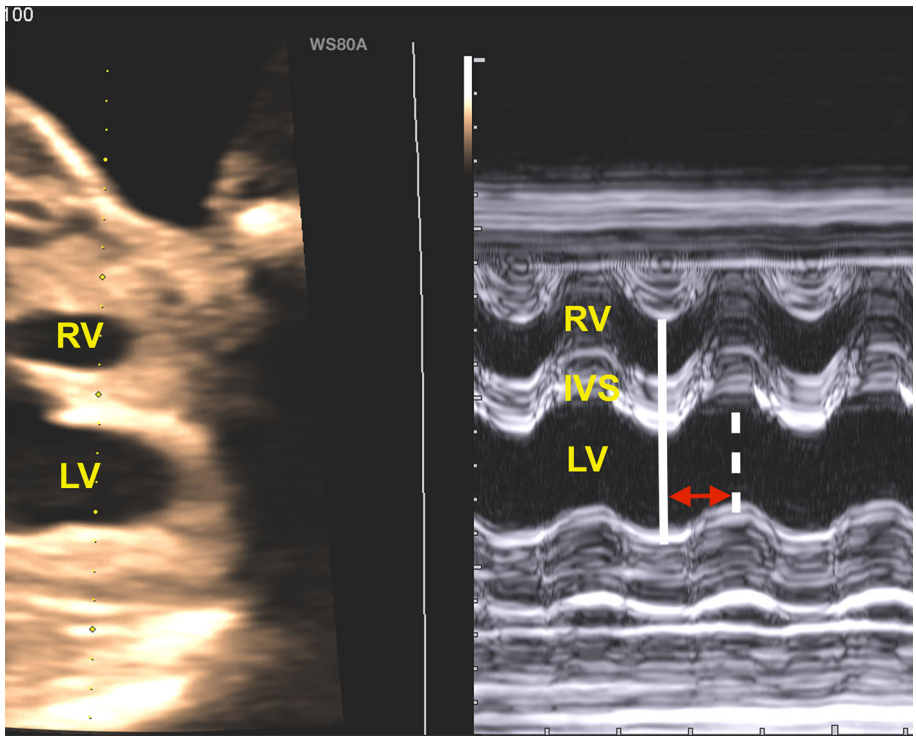

Fig. 2 M mode examination of Left ventricle (LV) and right ventricle (RV) shows normal thickening of both LV and RV free walls. RV and interventricular septum (IVS) contract at the same time (solid white line), while contraction of LV free wall (Dashed line) is markedly delayed (Double sided arrow)

significant delay in left ventricular posterior wall contraction as compared to interventricular septum contraction causing mechanical dyssynchrony between ventricular septum and LV posterior wall. Due to this, LV walls were not contracting synchronously. This dyssynchronous contraction of LV walls was the main cause of LV systolic dysfunction. The right ventricle appeared normal in size with normal global right ventricular functions. RV fraction shortening, Tei index, Ductus venosus flow and IVC flows were normal.

Prenatal counselling was done to discuss high risk prenatal and neonatal mortality associated with severe aortic regurgitation and LV dysfunction at early gestation. Based on these findings, the parents opted for termination of the pregnancy. A dilation and evacuation procedure was performed and was uncomplicated.

On fetal autopsy, there was no extra cardiac abnormality. External appearance of heart was normal except for dilated aortic root and arch of aorta. Dissection displayed normal mitral and tricuspid valves, pulmonary and aortic outflow tracts. Aortic root was dilated. A probe confirmed presence of left and right coronary ostia from respective sinuses of Valsalva. The aortic valve revealed complete absence of the left coronary cusp/leaflet with no rudimentary tissue in its place (Fig. 3). Other two aortic valve leaflets were of normal size but were thickened (Fig. 4). Left ventricle was hypertrophied. Multiple levels of histological sections taken from the ventriculo-arterial junction did not show any definitive valvular tissue at right

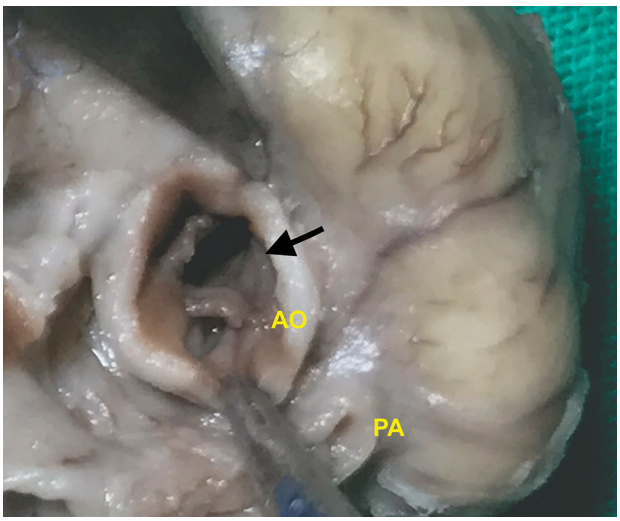

Fig. 3 shows normal relation of Aorta (AO) and Pulmonary Artery (PA) with thickened aortic valves. There is absent left coronary cusp of aortic valve (Arrow)

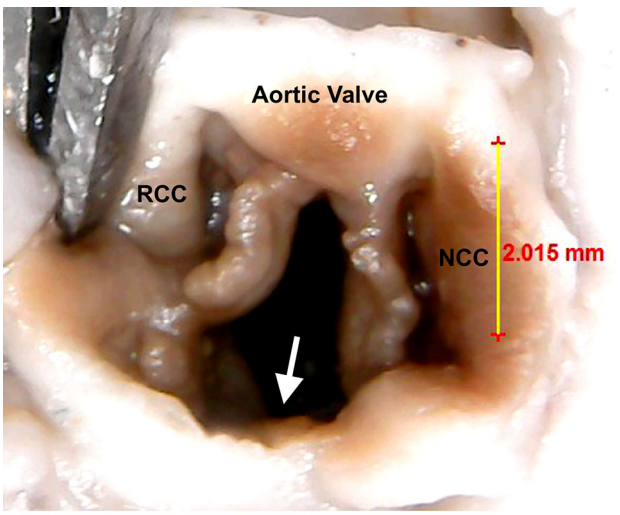

Fig. 4 shows thickened Right coronary cusp (RCC) and non coronary cusp (NCC) of aortic valve. Left coronary cusp is absent (Arrow)

coronary sinus. Other leaflets showed thickening with myxomatous deposits. Karyotyping done from fetal tissue did not show any abnormality.

Discussion

Complete absence of the aortic valve complex (AAV) is an exceedingly rare congenital cardiac anomaly and, till date, less than 30 cases of AAV have been reported [5]. Most commonly, AAV cases are associated with other congenital abnormalities. Isolated AAV is rare and solitary absence of one of the aortic cusp is exceedingly rare. Only 3 cases have been reported and all of them were during postnatal period [11–13] To the best of our knowledge, there is no case report of absent aortic valve leaflet in antenatal period.

Embryologically, aortic and pulmonary valves develop from intercalated cushions. Kramer described the developing outflow tract in terms of the "truncus" and the "conus" but it was not clear about development of arterial valves whether they belong to the "conus" or to the "truncus" [14]. Abnormalities of these valves are not currently considered to be "conotruncal" lesions, despite their location within the middle of the developing outflow tract. As per our understanding as opposed to "conus" and "truncus", embryonic outflow tracts have three parts described as distal, intermediate, and proximal. The distal part is separated to form the intrapericardial portion of arterial trunks, while the proximal part becomes the ventricular outflow tracts. Arterial valves and their supporting valvar sinuses develop form intermediate component. Cusps of arterial valves develop by excavation from distal parts of the cushions that separate the outflow tract along with the intercalated cushions [15].

Congenital absence of the aortic and/or pulmonary valves is an uncommon malformation. Absence of the pulmonary valve alone has been relatively well described while absent aortic valve is very rare. Based on common development of both pulmonary and aortic valves, theoretically, there should be equal numbers of both conditions [16]. Absence of a competent aortic valve results in severe aortic insufficiency which affects the fetal circulation in several ways. In diastole, blood regurgitates back into the ventricle, which leads to a "steal" of blood flow away from the body organs and tissues. The heart dilates due to the volume over load. Cardiomegaly and ventricular dysfunction ensue. Heart failure and fetal hydrops are common. As severe aortic regurgitation is poorly tolerated by the fetus and there is speculation that most fetuses with AAV die early in gestation and, therefore, go unrecognized [17]. This produces inequality in incidence of absent pulmonary and aortic valves.

In our case, there was absence of left coronary cusp which caused severe aortic regurgitation leading to left ventricular hypertrophy and dysfunction. This was the solitary abnormality present in our case. There was severe LV systolic dysfunction primarily due to ventricular mechanical dyssynchrony caused by delayed contraction of free wall of LV. This delayed contraction of posterior wall could have been due to delayed electrical activation of LV due to intraventricular conduction abnormality like left bundle branch block (LBBB) or prolonged diastole due to severe AR and volume overload of left ventricle.

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