Unusual association of aortic valve stenosis with ventricular septal defect and pulmonary atresia: Differentiation from truncus arteriosus with truncal valve stenosis

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ABSTRACT

Stenosis of systemic semilunar valve in cyanotic congenital heart defects is rare. It can happen in truncus arteriosus with truncal valve stenosis and the very rare anomaly of tetralogy of fallot with aortic valve stenosis. Here we describe a neonate with pulmonary atresia, ventricular septal defect and associated aortic valve stenosis and discuss the points of differentiation from truncus arteriosus.

Keywords: Echocardiography, pseudo truncus, truncus

INTRODUCTION

Systemic semilunar valve stenosis in cyanotic congenital heart disease is rare but is well known with truncus arteriosus. Tetrology of Fallot with pulmonary atresia can rarely be associated with aortic valve stenosis. Differentiating between these two conditions is important for planning optimal surgical strategy.

CASE REPORT

Two weeks old, 3,110 grams male baby was referred for evaluation of cyanotic congenital heart disease. Examination of the neonate showed mild central cyanosis, cardiomegaly, single 2nd heart sound and an ejection click followed by grade 3 ejection systolic murmur at left 2nd intercostal space. Based on these clinical features, the possibility of truncus arteriosus with truncal valve stenosis was considered. Child

was subjected to trans-thoracic echocardiographic evaluation.

Echocardiography showed situs solitus, levocardia, normal systemic and pulmonary venous connections, small ostium secundum atrial septal defect and atrio-ventricular concordance. There was no atrio-ventricular valve stenosis or regurgitation. There was a large malaligned subarterial ventricular septal defect with bidirectional shunt. One great artery was arising from the ventricular mass and was continuing as the arch of aorta [Figure 1, Video 1]. The semilunar valve of this great vessel was tricuspid and markedly dysplastic [Figure 2, Video 2] with peak instantaneous gradient of 58 mm Hg across the same. There was also moderate regurgitation across this valve. The pulmonary arteries, which were confluent, arose from the undersurface of the arch which is the typical location of patent ductus arteriosus in duct dependant pulmonary circulation. However, patent ductus arteriosus is uncommon in truncus arteriosus with normal aortic arch. Hence, careful evaluation of the right ventricular outflow tract was performed, which demonstrated the pulmonary annulus and the atretic pulmonary valve [Figures 3 and 4]. This established the diagnosis of the rare entity of pulmonary atresia with ventricular septal defect (PA-VSD) with aortic valve stenosis and ruled out the more common truncus arteriosus.

DISCUSSION

Association of aortic valve stenosis with pulmonary atresia is extremely rare and is restricted to a few case

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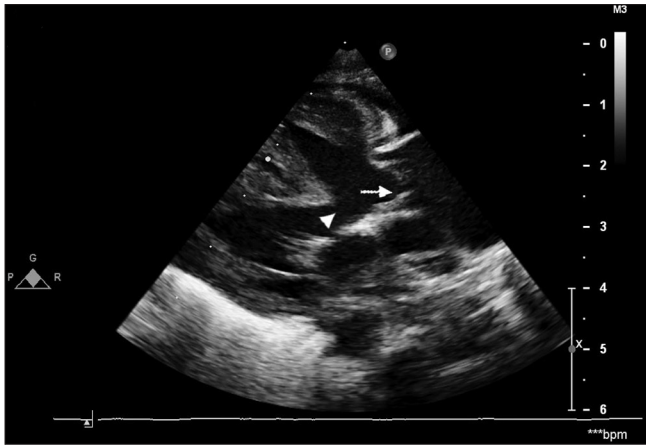
Figure 1: Parasternal long axis view demonstrating an arterial trunk with dysplastic valves (arrow) arising from the ventricles. Arrow head denotes the unrestrictive VSD
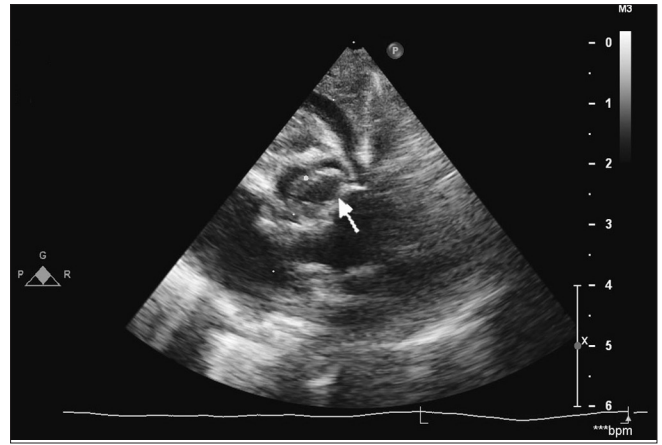

Figure 2: Parasternal short axis view demonstrating the dysplastic aortic valve (arrow)
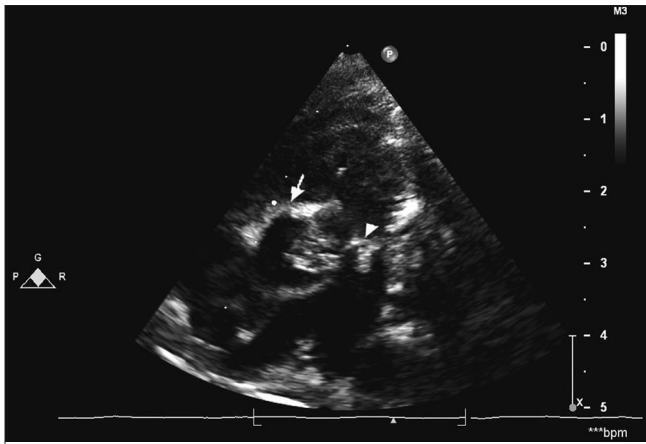

Figure 3: High parasternal short axis view showing the atretic pulmonary valve (arrow head) giving rise to confluent pulmonary arteries (oval). Dysplastic aortic valve is also shown (arrow)
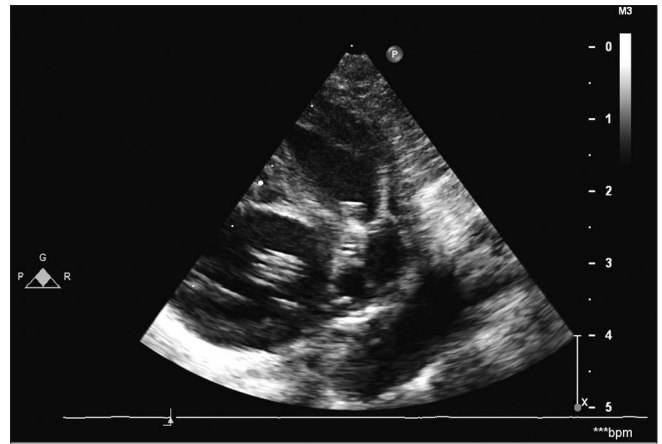

Figure 4: Low parasternal short axis view demonstrating the right ventricular outflow tract and atretic pulmonary valve

reports.[1-3] It can be easily confused with the more common truncus arteriosus with truncal valve stenosis.[4,5]

Differentiating PA-VSD with aortic valve stenosis from truncus arteriosus is important as the surgical approaches to these conditions are different. While truncus arteriosus often requires homograft conduit for surgical reconstruction, valvular pulmonary atresia with ventricular septal defect can be corrected without a conduit. In addition, the high variability in the sources of pulmonary blood supply and the branch pulmonary artery anatomy in PA-VSD, necessitate individualization of surgical plan, usually with unifocalization being part of the surgery.

Morphogenesis of PA-VSD is different from truncus arteriosus.[6,7]

PA-VSD results from extreme anterior and cephalad deviation of the fused outflow endocardial cushions, obliterating the right ventricular outflow leading to pulmonary atresia. This also results in concomitant failure to commit the aorta to the left ventricle, leaving a large ventricular septal defect with overriding aorta. Truncus arteriosus is caused by deficiency of the intercalated cushion which divides the embryologic truncus into intrapericardial arterial trunks and valves, explaining the single arterial valve, common trunk from which systemic, pulmonary and coronary vessels arise and the large ventricular septal defect. This may also explain the high incidence of arterial valve abnormalities seen with truncus, compared with tetralogy of fallot. Spiral septum divides the aortic trunk from the pulmonary trunk, contributing to the normal cup and sausage orientation of the great vessels. Defects in this septation cause aorto-pulmonary window.

In truncus arteriosus, pulmonary artery origin follows a described pattern, most commonly from the proximal ascending aorta. In PA-VSD, the blood supply to the lungs is quite variable and can be from the ductus arteriosus, systemic-to-pulmonary collateral arteries, coronary arteries, bronchial or pleural arteries either alone or in combinations.

The truncal valve can be tricuspid in 69%, quadricuspid in 22%, bicuspid in 9%, pentacuspid in 0.3%, and unicommissural in 0.3%.[7] In PA-VSD, the systemic semilunar valve is usually tricuspid, with rarely described bicuspid aortic valve. Stenosis of the truncal valve can occur in about 11% of cases, whereas stenosis of aortic valve in PA-VSD is extremely rare. Regurgitation occurs in about 10-60% of truncal valves.[6,7] It has been noted at surgery that valves with 4 or more cusps are usually regurgitant, whereas bicuspid truncal valve is usually stenotic.[8]

Dilation of the solitary arterial trunk and right aortic arch are common with both truncus and PA-VSD. Interrupted aortic arch and retroesophageal origin of the right subclavian artery occurs in 11% to 19% of patients with truncus, but is rare in PA-VSD.[6,7] Unlike PA-VSD, in which without exception the coronary arteries arise from one or both of the aortic valvular sinuses facing the pulmonary trunk, there is no constant pattern of origin of coronary arteries in the of truncus arteriosus. The two coronary arteries can arise from any of the truncal valvar sinuses.[8] If one pulmonary artery is absent, in truncus arteriosus it is most frequently absent on the side of the aortic arch. In PA-VSD, the pulmonary artery is more frequently absent on the side opposite to the aortic arch.

In spite of the above mentioned differences, the most important finding which distinguishes between the two conditions is the presence or absence of an atretic pulmonary annulus. If a separate pulmonary annulus can be demonstrated, the diagnosis of truncus arteriosus can be excluded. Occasionally, cases with no demonstrable pulmonary annulus and central pulmonary arteries may be encountered. These may be better described as solitary arterial trunks, although in terms of clinical presentation and treatment, such patients have more affinity with PA-VSD. Hence, in a majority of cases, careful echocardiographic evaluation is sufficient to distinguish truncus from PA-VSD and to plan surgical correction, obviating the need for other imaging modalities and cardiac catheterization.[9] This child is planned for surgical correction without conduit with aortic valve repair on follow-up.

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