Unusual right ventricle aneurysm and dysplastic pulmonary valve with mitral valve hypoplasia

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ABSTRACT

We report a newborn with an unusual combination of aneurysmally dilated thin-walled right ventricle with hypertrophy of the apical muscles of the right ventricle. There was narrow pulmonary annulus, pulmonary regurgitation, and hypoplasia of the mitral valve and left ventricle. We propose that this heart represents a partial form of Uhl`s anomaly.

Keywords: Absent pulmonary valve, right ventricle aneurysm, Uhl's anomaly

INTRODUCTION

Aneursymal dilatation of the right ventricle can be seen in several malformations like congenital aneurysm, Uhl's anomaly, arrhythmogenic right ventricular dysplasia, atrialized portion of the Ebstein's anomaly, absent right pericardium, or post-ischemic aneurysms.[1] Such aneurysms are being increasingly diagnosed prenatally.[2]

We report a case having an aneurysmatically dilated right ventricle resembling the Uhl's Anomaly, with an unusual combination of lesions. Cases with similar features like this anomaly have been very rarely reported.[3,4]

CASE REPORT

An aneurysmally dilated right ventricle was suspected in the fetal echo at the 24-week gestation in a baby, but a detailed evaluation was not done. The baby was delivered by a planned Cesarean section without any complications. He weighed 3 kg; the APGAR scores at the first and fifth minutes were 5 and 7, respectively. The oxygen saturation was 85%. A pansystolic murmur was heard. The chest X-ray showed marked cardiomegaly and a cardiothoracic ratio of 85%. Echocardiography showed an aneurysmally dilated right ventricle [Figure 1, Videos 1-3]. The entire right ventricle except the apical portion was dilated. There was apical muscular hypertrophy of the right ventricle. [Figure 1] The tricuspid valve was mildly hypoplastic (Z score -0.95) and prolapsing, but not displaced. The pulmonary valve was dysplastic and doming [Figure 2], with mild pulmonary regurgitation [Figure 3]. Although the pulmonary annulus was not very small, the anatomy resembled absent pulmonary valve. The left ventricle was compressed by the dilated RV and measured 18 mm in the end-diastole (Z score - 0.13), but the mitral valve was only 5.5 mm. There was a large patent ductus arteriousus flowing left to right, with a low velocity flow. There was no family history of any cardiac illness. Multislice computerized tomography revealed right ventricle aneurysm. In addition, it showed a very thin RV wall measuring 1.4 mm in some places. A cardiac catheterization done on day 10 revealed pressures of the right atrium: 7 mmHg, right ventricule: 67/0 – 9 mmHg, pulmonary artery: 62/21, (mean 34 mmHg). Aorta: 75/55 (mean 60 mmHg). Aortic and pulmonary artery saturations were 97 and 93%, respectively. The baby continued to require mechanical ventilator support and inotropes, and died on the fifteenth day of life. An autopsy was not done.

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DISCUSSION

Uhl's anomaly is characterized by a complete or partial absence of the myocardium of the right ventricle, which is replaced by a parchment-like endocardial and

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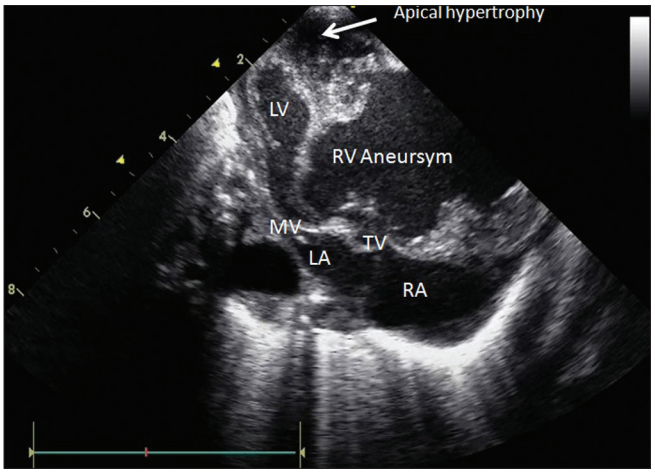

Figure 1: Aneurysmal dilatation in the rest of the right ventricle that was compressing the left ventricle

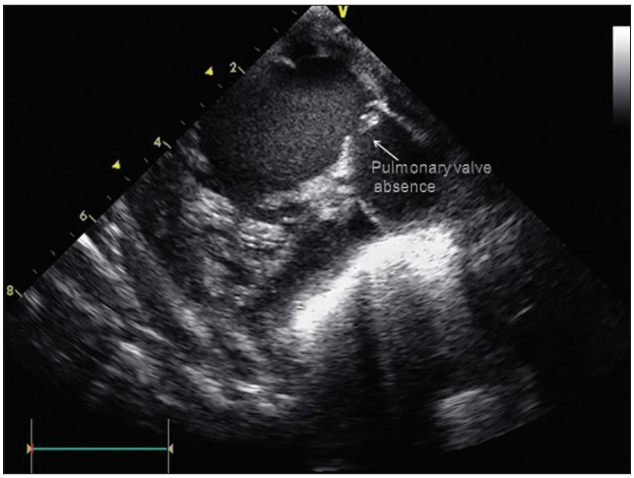

Figure 2: Dysplastic pulmonary valve

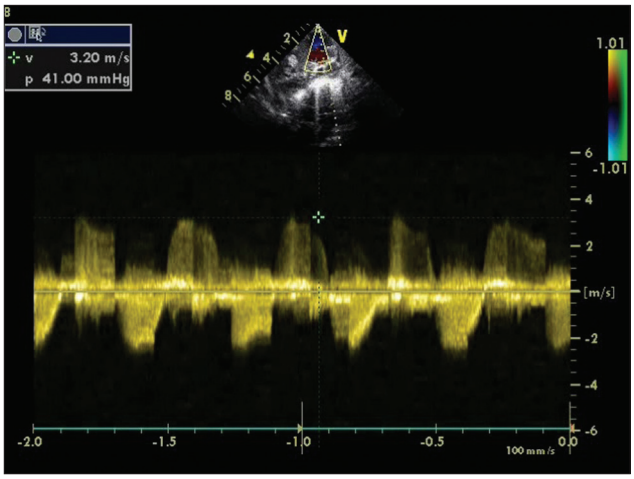

Figure 3: Doppler echocardiography showing pulmonary regurgitation

epicardial tissue.[5] Uhl`s anomaly is a very rare disorder, and needs to be distinguished from Ebstein`s anomaly, Arrythmogenic Right Ventricle Dysplasia (ARVD), and other forms of RV cardiomyopathy. Patients with Pulmonary Atresia, with an intact ventricular septum, sometimes have a thinned out RV, but this is possibly secondary to atresia. Arrythmias and fibrofatty replacement that characterize ARVD are usually not seen in Uhl`s anomaly, but the decision may be difficult with an overlap in some situations.[6] Uhl`s anomaly has a poor prognosis, although survival into adulthood[7] and treatment with total cavopulmonary connections have been reported.[8]

The muscle loss in Uhl`s anomaly results from apoptosis rather than from lack of development.[5] The septal components, septomarginal trabeculations, and papillary muscles of the tricuspid valves are normally muscularized in Uhl's anomaly. This may suggest a different embryological origin of these tissues that are spared in Uhl`s anomaly. Partial forms of Uhl's anomaly may not be well recognized.[9,10] There may be embryological or other reasons for some components of RV being muscularized. An analysis of the partial forms of the Uhl's anomaly might be of interest and shed more light on the pathogenesis of the disorder. Cases similar to ours, with aneurysms involving RV outflow tracts have been previously reported.[3,4,10] We hypothesized that our patient represents a variant of Uhl's anomaly. In the absence of histological confirmation, this assertion remains less than established. In any case, the present case has a very unusual anatomy and combination of lesions. The remarkable thin-walled RV supports the likely possibility of a partial Uhl's anomaly.

In conclusion, we report a neonate with an aneurysmally dilated right ventricle with thinned out walls, but an apical hypertrophy, dysplastic pulmonary valve, and hypoplastic mitral valve. The lesion may represent a partial variant of the Uhl's anomaly.

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