

Right Ventricular Hypoplasia and Aortic Stenosis: Autopsy Case

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Abstract

Neonatal cardiac malformation cases mostly need surgical repair. It is important to describe the morphological features of these malformations. A 3 months-old baby girl was brought to a local clinic after three days antibiotic therapy. No signs of life were present. Autopsy findings showed adhesion of right ventricular surfaces which caused the chamber volume to significantly decrease. The ventricular wall was thinned, out and become crescent shaped. This rare autopsy case, with right ventricle hypoplasia and severe stenosis of aorta is presented.

Introduction

The great majority of neonatal cardiac malformation cases need surgical repair; failing which they cannot survive. It is crucial to specify the morphological features of these malformations.¹ Among the right ventricular malformations, dysplasia of the right ventricle and Uhl anomaly are well studied and are found to be significantly related to sudden cardiac deaths.^{2,3} Hypoplasia and dysplasia of the right ventricle can present with severe symptoms in early life.^{4,5} In this report we present an

autopsy case of a 3-months-old baby girl with right ventricle hypoplasia and severe stenosis of aorta who died due to pneumonia.

Case Report

A 3 months-old baby girl with 65 cm. height and 5500gr. weight, was brought to a local clinic dead after three days of antibiotic therapy due to cough of three days duration. Autopsy findings showed petechia over the surface of lungs, in cut section oedema, consolidation and mottled discoloration consistent with pneumonia. Minimal pericardial effusion was appreciated, heart was enlarged, weighing 55 gr. Dissection of the heart revealed adhesion of right ventricular surfaces inside which significantly decreased the chamber volume. Ventricular wall was thinned to 0.2 cm, and acquired a crescent shape (Fig 1), Tricuspid and pulmonary valves were normal, as also the exit to pulmonary arteries. On the left side of the heart, left ventricular wall was thick (1.6 cm.) and ventricular chamber was enlarged. Degeneration and stenosis of the aortic valves was appreciated (Fig 2). In the abdomen, mesenteric lymph nodes were prominent. Microscopic evaluations showed; disarray in cardiac muscle bundles,

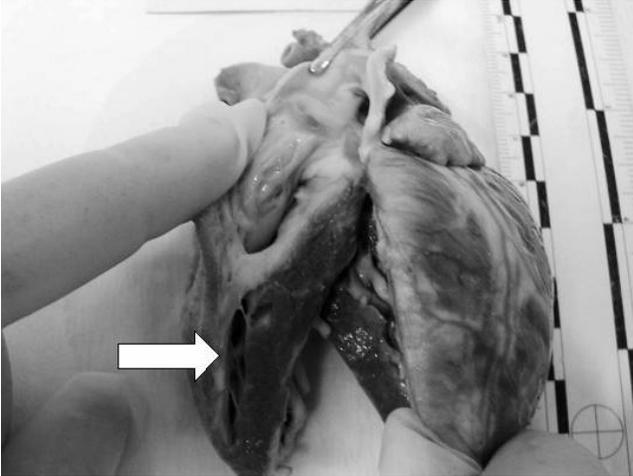


Figure 1: Crescent shaped right ventricle.



Figure 2 Stenosis of the aortic valves.

hyperaemic changes in spleen and kidney, pneumonia in gray hepatization stage, and reactive hyperplasia in mesenteric lymph nodes. The cause of death was reported as cardiac anomaly and pneumonia.

Discussion

The leading arrhythmogenic right ventricular malformations are dysplasias,^{1,2,6} hypoplasias^{4,5} and rarely Uhl anomalies or congenital hypoplasia of right ventricular myocardium.^{2,3} Studies identified new forms of arrhythmogenic dysplasias, but the etiology and pathogenesis of arrhythmogenic right ventricular cardiomyopathy are still unknown.⁷ Literature search revealed no result for right ventricular hypoplasia with

stenosis of aorta as in our case; but right heart dysplasias accompanied by different anomalies such as pulmonary atresia and anomalous coronary artery has been reported.⁸ Significant thickening in the right ventricular wall is consistent with the literature as an important finding in dysplastic cases.^{2,9}

Uhl's anomaly is a very rare anomaly with unknown aetiology, characterized by congenital partial or complete absence of right ventricular myocardium. Associations with other congenital heart diseases, familial occurrence, sudden death and arrhythmia with Uhl's anomaly have been reported.³ The crescent appearance that we described macroscopically has been reported as an echocardiographic finding which may be seen in isolated right ventricular hypoplasia cases.⁴ Histo-pathological studies of D'Amati et al. described different forms of ventricular dysplasias.⁶ Arrhythmogenic right ventricular cardiomyopathy is progressive myocardial atrophy of the right ventricle, which was recently included among cardiomyopathies in the revised WHO classification. The specific gene defects as well as the defective coded proteins have not yet been identified.⁷ Similar to our case, it has been shown that cases with right ventricular dysplasia may exhibit segmental or diffuse loss of myocardial fibers with transmural fatty or fibrofatty replacement, accounting for electrical instability at risk of life-threatening ventricular arrhythmias^{6,7,9} and also it was explained that inflammatory infiltration in right ventricle may be observed.² It has been reported that for unknown reasons, these cases may develop cardiomegaly,³ atrial and ventricular dilation, diverticula formation in their terminal stage. Joy et al reported that isolated hypoplasia of right ventricle can present with cyanosis in childhood, besides underlined that diagnosis and management strategy of isolated hypoplasia of right ventricle in children is difficult.⁵ It has also been documented that during the course of the disease, in addition to right ventricular failure, left ventricle could be involved as well.²

The patients were subjected to surgery for Glenn's operation, Fontan procedures which are reported in literature.^{2,3} Basso et al. reported, that variants of right ventricular dysplasias need to adopt strict diagnostic criteria, warranted not only in the clinical setting but also in the forensic and general pathology arena. Also stated that, when dealing with a case of sudden death, in which the only morphologic finding consists of an increased amount of epicardial or intramyocardial fat, a more convincing arrhythmogenic source such as myocardial inflammatory infiltrates, fibrosis, anomalous pathways, and ion channel disease should be searched, to avoid an over-diagnosis of cases.⁹

Autopsy findings are conclusive in order to

differentiate morphologically diverse cardiac anomalies and postmortem evaluations are important contribution to understand cardiac origin medicolegal sudden deaths in neonatal population.

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