Combined atresia of one left-sided and one right-sided cardiac valve in a premature newborn

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Described herein is the heart of a 2-day-old newborn, the product of a 25-week gestation, with atresia of two cardiac valves, one on the right side and one on the left side, apparently a previously undescribed entity.

The worst heart disease—the one allowing the shortest survival—is aortic valve atresia, the most common cause of death in the first month of life (1, 2). About 25% of these newborns with aortic valve atresia also have mitral valve atresia. The second most common condition associated with an atretic cardiac valve is pulmonic valve atresia. The occurrence of one right-sided atretic valve and one left-sided atretic valve in the same heart must be incredibly rare, but such was the case in the newborn described herein.

CASE DESCRIPTION

A 2-day-old female newborn after a 25-week gestation weighed 550 g. An electrocardiogram shortly after birth disclosed a prolonged P-R interval and sinus bradycardia. A technically difficult echocardiogram disclosed a dilated right ventricle, a normal-sized left ventricle, a large atrial septal defect with bidirectional flow, a large ventricular septal defect with bidirectional flow, a small (1 mm) patent ductus arteriosus with left-to-right flow, severe “pulmonic stenosis,” and an unobstructed aortic valve and aortic arch. The echocardiographic findings were interpreted as being consistent with tetralogy of Fallot.

The newborn died in the intensive care unit on the second day of life. At necropsy, the heart weighed 4.15 g. The cardiac findings are illustrated in the Figure. Both the mitral and pulmonic valves were atretic, the atrial septum was absent, and a ventricular septal defect was located caudal to the aortic valve, which arose from the dilated right ventricle. A narrowed patent ductus was present, and it was the only source of blood to the lungs.

In addition to the cardiac anomalies, a cleft lip was present, and it extended to involve the entire hard and soft palate. Cytogenetic SNP microarray analysis performed on an ante-mortem blood sample disclosed a normal female chromosome pattern with no deletions or duplications of known or potential clinical significance. Postmortem chromosome analysis also showed a normal female 46 XX karyotype. No numerical or structural abnormalities were found.

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structural aberrations were seen at the 500 G-band resolution. Examination of the placenta disclosed a three-vessel cord, premature villous architecture, severe acute chorioamnionitis, and mild acute funisitis.

DISCUSSION

Our patient had combined pulmonic and mitral valve atresia. Such a combination, to our knowledge, has not been reported previously. Death was probably the consequence of progressive narrowing of the ductus arteriosus.