Absent aortic valve and isolated right brachiocephalic artery: A complex congenital heart defect

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Introduction

Absent aortic valve (AAV) is a rare congenital heart anomaly associated with a high mortality rate. AAV is detectable in the perinatal period and is frequently associated with other cardiac anomalies, including double outlet right ventricle (DORV), hypoplastic left heart syndrome, dysplastic pulmonary valve, and mitral atresia (1, 2). The pathogenesis is yet unknown; however, underdevelopment of the endocardial cushion tissue at the ventriculoarterial junction may be the probable etiology (3, 4). AAV is associated with severe aortic regurgitation (AR) that can lead to rapid and progressive heart failure, accompanied by growth retardation in the fetus, and ultimately fetal demise.

Twenty-six cases have been reported since Toews reported the first case in 1975, and most of the reported cases died a couple of weeks after birth (5). In this paper, a newborn diagnosed with AAV associated with DORV, isolated right brachiocephalic artery with severe stenosis in the origin, and severe AR is described as an exceedingly rare clinical presentation.

Case Report

A full-term male newborn weighing 2.7 kg, previously diagnosed with AAV by fetal echocardiography, was admitted to our hospital immediately after birth. On fetal echocardiographic study at 26 weeks of gestation, cardiac anomalies consisting of severe AR, malposed great arteries, a complete commitment of the aortic valve to the right ventricle, large sub-pulmonary ventricular septal defect (VSD), small aorta, and narrowing of the descending aorta without coarctation were identified. In addition, left and right ventricular functions and physiologies were reported to be abnormal.

The pregnancy was unremarkable and he was delivered by cesarean section. Apgar scores at 1 and 10 minutes were recorded as 6 and 9, respectively. On admission, the baby had mild respiratory distress (respiratory rate of 60/min) and a heart rate of 152 beats/min. Oxygen saturation was 90% and blood pressure was 60/40 mm Hg. The axillary and femoral pulses were normal. To-and-fro murmur was heard on cardiac examination. The lungs were clear and the liver was palpable at 2 cm below the right costal margin.

A chest X-ray revealed cardiomegaly with increased pulmonary blood flow. Brain computed tomography (CT) scan showed no intracranial lesion or abnormal echogenicity. The electrocardiogram showed biventricular hypertrophy.

The post-natal echocardiography revealed the following: AAV with vestigial leaflets, DORV, large inlet posterior VSD with anterior extension, severe AR, mild valvar aortic stenosis (AS), malposed great arteries, severe pulmonary hypertension (PH), moderate-to-severe mitral regurgitation (MR), moderate tricuspid regurgitation (TR), isolated right brachiocephalic artery, huge patent ductus arteriosus (PDA), normal systolic function, and left aortic arch (Fig. 1, 2). The CT angiography confirmed the same diagnosis as echocardiography, and additional findings revealed by CT angiography were segmental left ventricle non-compaction with trabecular/compact ratio of 2.61, stretched patent foramen ovale (PFO) with a left-to-right shunt, borderline sized RV, accessory tricuspid valve tissue adhered into the ventricular side of the tricuspid valve (TV) leaflet with protrusion into the right ventricular outflow tract (RVOT), without producing obstruction; moreover, the distance between TV and pulmonary valve (PV) was not appropriate to allow for intra-cardiac repair with baffling of the left ventricle (LV) into the aorta without producing left ventricular outflow tract obstruction.

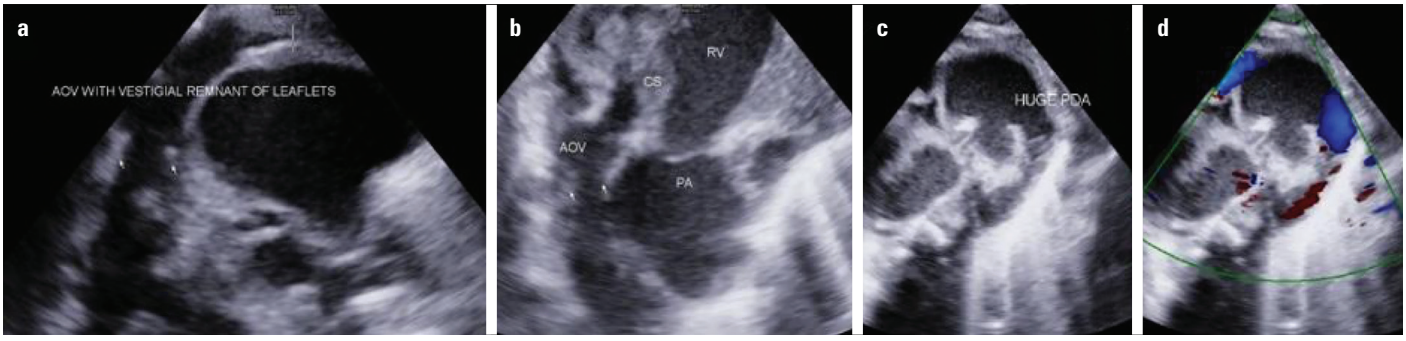

Figure 1. The echocardiographic assessment shows an aortic valve with vestigial remnant of the leaflets (a, b) and huge patent ductus arteriosus (c, d)

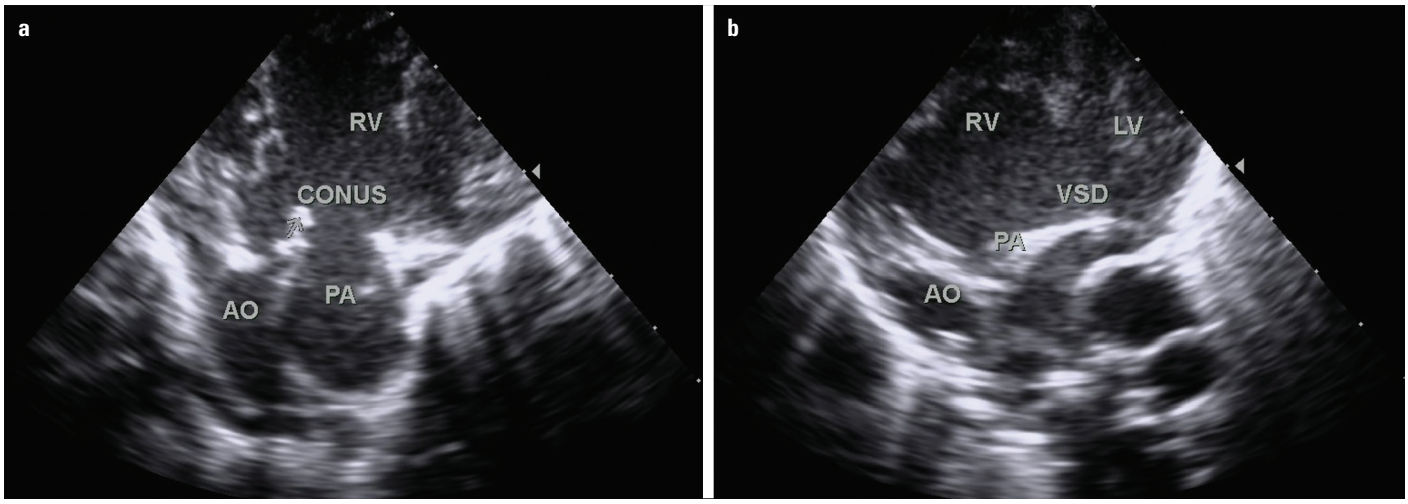

Figure 2. (a) Absent aortic valve with vestigial leaflets and DORV, (b) Large inlet type posterior VSD with anterior extension

Genetic study was also done; a fluorescence *in situ* hybridization reported no deletion of chromosome 22 at 22q11.2 (DiGeorge/VCFS TUPLE1) and no deletion of chromosome 22 at 22q13.3 (22q13.3 deletion syndrome probe).

He was a candidate for the Damus Kaye Stansel procedure, ligation of PDA without VSD closure of the valve conduit between RV and PA (single ventricular approach) versus Blalock-Taussig shunt. The family was consulted regarding the cardiac diagnosis and surgery, but did not consent. Hence, he received furosemide and Spironolactone as medical treatment.

Discussion

To our knowledge, this is the first report of a fetus with AAV and isolated right brachiocephalic artery with severe stenosis in the origin, DORV, and severe pulmonary hypertension. Congenital malformations of the aortic valve include atresia, stenosis, disruption of a sinus cusp, absence of a cusp, and complete absence of all leaflets (6). AAV is described as no valve tissue or only primary fibrous ridges at the aortic annulus, and is consistently associated with other congenital cardiac anomalies such as DORV, mitral atresia, and absent or dysplastic pulmonary valve, hypoplastic left heart syndrome, and DiGeorge syndrome (7). Clinical presentations are cyanosis, respiratory distress, cardiomegaly, and the majority of cases are detected by fetal echocardiography or autopsy. Most reported patients are males, suggesting an X-linked recessive inheritance or other X-linked factors (8).

The absence of aortic valve leads to severely increased left ventricular end-diastolic pressure, which in turn leads to decreased coronary arterial perfusion pressure. Left ventricular hypertrophy causes increased myocardial oxygen consumption and is associated with compromised coronary perfusion. The two aforementioned factors contribute to a high risk for ischemic heart disease, resulting in a high mortality rate during the first weeks after birth (9). However, this case is the first reported case that survived for six months.

No chromosome abnormality, such as DiGeorge syndrome, was detected in this case. In summary, the patient was diagnosed at the prenatal stage and confirmed by an echocardiogram after birth, but his parents did not consent to the proposed surgical repair; therefore, he was prescribed oral medications. The patient died six months after birth due to severe heart failure.

Given the extreme nature of the anomaly, it would be challenging to develop a suitable management protocol for fetuses and neonates with AAV. Fetal echocardiography should provide evidence of the presence and severity of AR, MR, and blood flow via the foramen ovale and left ventricular dysfunction.

Conclusion

AAV is an extremely rare congenital heart defect. AAV is usually associated with other congenital heart anomalies in all instances. Most of the fetuses diagnosed with this anomaly die of heart failure. Prenatal diagnosis provides appropriate counseling for families.

Informed consent: Informed consent was obtained from the patient's parents.

References

1. Clark EB. Cardiac embryology. Its relevance to congenital heart disease. Am J Dis Child 1986; 140: 41-4.
2. Sabati AA, Wong PC, Randolph L, Pruetz JD. Absent aortic valve associated with double outlet right ventricle and aortopulmonary window: physiologic implications of a rare malformation in both the fetus and neonate. Congenit Heart Dis 2014; 9: E98-104.
3. Murakami T, Horigome H, Shiono J, Ishiodori T, Ban Y, Gomi S, et al. Prenatal Diagnosis of Congenital Absence of Aortic Valve: A Report

of Two Cases with Different Outcomes and a Literature Review. Fetal Diagn Ther 2015; 38: 307-14.
4. Sivanandam S, Mackey-Bojack S. Isolated congenital absent aortic valve cusps in a fe- tus: A case report. Indian Journal of Child Health 2015; 2: 86-7.
5. Hartwig NG, Vermeij-Keers C, De Vries HE, Gittenberger-De Groot AC. Aplasia of semilunar valve leaflets: two case reports and developmental aspects. Pediatr Cardiol 1991; 12: 114-7.
6. Toews WH, Lortscher RH, Kelminson LL. Double outlet right ventricle with absent aortic valve. Chest 1975; 68: 381-2.
7. Bertsch EC, Minturn L, Gotteiner NL, Ernst LM. Absent Aortic Valve in DiGeorge Syndrome. Pediatr Dev Pathol 2016; 19: 61-3.
8. Bierman FZ, Yeh MN, Swersky S, Martin E, Wigger JH, Fox H. Absence of the aortic valve: antenatal and postnatal two-dimensional and Doppler echocardiographic features. J Am Coll Cardiol 1984; 3: 833-7.
9. Hibino N, Harada Y, Hiramatsu T, Yasukochi S, Satomi G. Fontan operation for hypoplastic left heart syndrome with absent aortic valve. J Thorac Cardiovasc Surg 2004; 128: 315-6.

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DOI:10.14744/AnatolJCardiol.2020.32323