INTRODUCTION

Absent pulmonary valve syndrome (APVS) is a unique and rare cardiac defect that occurs in 1% of fetuses with congenital heart defects and 0.2% to 0.4% of live births. It entails a rudimentary and dysplastic pulmonary valve with severe regurgitation, leading to a dilated main pulmonary artery. There are two phenotypes that are differentiated by the presence of a ventricular septal defect (VSD). More common is Fallot type, which entails a VSD, has more severe pulmonary dilatation, and usually has no patent ductus arteriosus (PDA). Non-Fallot type, on the other hand, has an intact ventricular septum, is extremely rare, and has less pulmonary dilatation secondary to an in utero PDA that allows runoff into the systemic vasculature. Single-ventricle physiology has been reported specifically with tricuspid atresia. We present a complex cardiac case report of APVS in the setting of double-outlet right ventricle (DORV) with unbalanced complete atrioventricular canal. APVS with this single-ventricle anatomy was documented once in a 1980 surgical review article, but a full detailed depiction of the clinical course has not yet been reported. Fetal echocardiography and magnetic resonance imaging were used in the evaluation to anticipate the degree of respiratory compromise at birth. Postnatal transthoracic echocardiography confirmed the diagnosis.

CASE PRESENTATION

At 16 weeks’ gestation, a male newborn was prenatally diagnosed using fetal echocardiography to have APVS in the setting of a DORV and right-dominant atrioventricular canal defect with no PDA. The mother was 32 years of age, G3P2, with a pertinent comorbidity of possible lupus. The family history was negative for congenital heart defects. Because this particular cardiac defect is known to have dilated branch pulmonary arteries that can then cause bronchial compression, a fetal magnetic resonance imaging was performed at 31 weeks’ gestation to quantitatively assess lung volumes and indirectly predict the degree of respiratory compromise at birth. For this fetus, there was a bilateral symmetric increase in lung volume by approximately 35% (Figure 1). This information was equivocal for trapped fluid in the fetal lung, as there are no prospective studies that have quantified fetal lung volume to predict risk for respiratory distress. Furthermore, the fact that it was a symmetric increase in lung volumes made it difficult to predict respiratory complications. Because of the uncertainty, the fetus was scheduled to be delivered at the closest birthing center. At 38 weeks’ gestation, the mother underwent elective cesarean section. The delivery was uncomplicated. Birth weight was 2.865 kg. Apgar scores were 8 and 8 at 1 and 5 min, respectively. He was pink and vigorous and had spontaneous respirations without any increase in work of breathing. Breath sounds were clear, with a 3/6 to-and-fro murmur. He was placed on 2 L of oxygen via nasal cannula to maintain saturation >90%. Additional respiratory support was not required. He was then stabilized and transferred to the cardiac intensive care unit.

Postnatal transthoracic echocardiography (Video 1) demonstrated DORV {S,D,D} with right-dominant complete atrioventricular canal defect with small atrial septal defect and VSD. The left-sided atrioventricular valve had a parachute deformity and no antegrade flow. The dysplastic pulmonary valve had moderate stenosis in the setting of severe regurgitation (Video 2). The main and branch pulmonary arteries were markedly dilated (Figure 2), and there was no PDA. There was an unusual right aortic arch and left descending aorta with mirror-image branching pattern. The mildly dilated right ventricle had mildly depressed systolic function. The moderately hypoplastic left ventricle had qualitatively normal systolic function. There were bilateral superior vena cavae with no interconnecting vein (Figures 3 and 4).

After arrival, laboratory workup and chest radiography were unremarkable. The infant remained on room air and was started on oral ad lib feeding. Because of the complex anatomy, he was deemed best suited for single-ventricle palliation. Thus, on day 5 of life, he underwent nonemergent surgical intervention with a 3.5-mm central shunt placement, atrial septectomy, and pulmonary artery transaction (Figure 5). By creating pulmonary atresia, the pulmonary regurgitation would be eliminated and the pulmonary arteries would not continue to dilate. The perioperative course was pertinent for an episode of ventricular fibrillation requiring electrical shock and lidocaine administration. The postoperative course was complicated by a large left-sided pleural effusion causing acute respiratory failure, reintubation, and chest tube placement. After aggressive diuresis and pulmonary rehabilitation, the infant was successfully extubated on postoperative day 17. He went home at 1 month of age. At home, he thrived well, with no evidence of failure to thrive or cyanosis. He returned for bilateral bidirectional Glenn surgery at 5 months of age, with an uneventful postoperative course. Currently, he remains clinically stable and will later undergo the Fontan operation to complete the single-ventricle palliation.
DISCUSSION

APVS is unique, intricate, and one of the rarest cardiac defects, with profound physiologic effects during fetal life. The anomaly consists of complete agenesis or dysplasia of the pulmonary valve leaflets, subsequently causing severe pulmonary regurgitation with varying degrees of stenosis. It commonly occurs in association with tetralogy of Fallot, though it can occur in isolation with an intact ventricular septum. Importantly, the increased pulmonary blood flow secondary to the chronic regurgitation can potentially cause massively dilated pulmonary arteries, which in turn compress nearby bronchioles. This particular phenomenon is worse when APVS occurs with tetralogy of Fallot, as there is no PDA to allow pulmonary blood runoff. During fetal life, the airway obstruction is tolerated as oxygenation is completed by the placenta. However, at birth, newborns who have significant bronchial compression due to dilated pulmonary arteries are at high risk for respiratory decompensation. Adequate fetal evaluation is crucial to anticipate the clinical presentation at birth for optimal delivery planning and preparation for airway management with mechanical ventilation, extracorporeal membrane oxygenation, or early surgical repair. More recently, fetal magnetic resonance imaging has been used as an adjunctive modality to perform pulmonary volume assessment, where increased and asymmetric lung volumes would infer significant bronchial compression and profound distress at birth. In 2013, Chelliah et al. first assessed APVS with this technique and demonstrated its utility and benefit to anticipate postnatal compromise and to prepare with a full multidisciplinary plan of care.

Most cases of APVS occur with biventricular physiology, specifically with an adequate right ventricle and normal tricuspid valve annulus. However, the rudimentary pulmonary valve can occur with concurrent tricuspid atresia or stenosis with right ventricular hypoplasia. This single-ventricle physiology is exceedingly rare and only sporadically documented in literature. A 2013 retrospective single-center study reported an incidence of 8% (n = 1) for APVS with tricuspid atresia, right ventricular dysplasia, and a restricted duct. A case report in 2018 described a patient with prenatally diagnosed APVS along with tricuspid atresia, a VSD, and dilated pulmonary arteries, initially palliated with main pulmonary artery banding and plication of the aneurysmal dilated central pulmonary artery and later completed with a Fontan procedure at 16 months of age. Surgical options include single-ventricle palliation, but heart transplantation should always be a consideration. In a 2014 retrospective review by Szwast et al., the presence of single-ventricle anatomy, particularly with tricuspid atresia, was strongly correlated with heart transplantation (P = .003). These patients were noted to have markedly abnormal coronary arteries causing ischemic changes and thus compromising left ventricular function. Furthermore, echocardiography demonstrated exaggerated leftward bowing of the ventricular septum, subsequently obstructing the left ventricular outflow tract and decreasing cardiac output. The constellation of these anatomic anomalies ultimately predisposed patients with APVS with tricuspid atresia to heart transplantation.

This particular case report highlights the first identified and documented occurrence of APVS with DORV and right-dominant complete atioventricular canal. The anatomy entails single-ventricle palliation secondary to the hypoplastic left ventricle. The very unusual combination of cardiac defects has special hemodynamic considerations, though survival is feasible.
Figure 3  (A) Apical view of unbalanced right dominant atrioventricular (AV) canal with hypoplastic left ventricle. (B) Parasternal short-axis view of massively dilated main (Z score = 6.1) and branch pulmonary arteries (left pulmonary artery Z score = 8.6, right pulmonary artery Z score = 7.5). MPA, Main pulmonary artery.

Figure 4  (A) Apical view of dysplastic pulmonary valve (PV) and free regurgitation. (B) Right anterior oblique view of flow turbulence across the dysplastic pulmonary valve and no anterior deviation of conal septum. PR, Pulmonary regurgitation.

Figure 5  High parasternal view in color-compare mode depicting the 3.5-mm central shunt anastomosis to the pulmonary artery. The arteries are markedly dilated.
The outcome of prenatally diagnosed APVS depends on the underlying lesion and physiology. Given the improvement in prenatal detection as well as enhanced preparation at birth, the prognosis has drastically improved with 80% postnatal survival.6

CONCLUSION

APVS in the setting of DORV and right-dominant atrioventricular canal defect is a rare constellation of cardiac defects that has never previously been reported. Its occurrence highlights the feasibility of palliation initially with a systemic shunt and later conversion to superior and inferior cavopulmonary anastomosis. Fetal and postnatal echocardiography strongly assists in the diagnosis as well as for predicting the clinical presentation in the immediate postnatal period.

SUPPLEMENTARY DATA

Supplementary data related to this article can be found at https://doi.org/10.1016/j.case.2018.12.001.

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