Total Anomalous Pulmonary Venous Return with No Connection

Valerie DeMatteo, RDCS, Michael D. Quartermain, MD, Michael DiLorenzo, MD, Michael Liu, MD, Craig Tinker, RDCS, and Meryl S. Cohen, MD, *Philadelphia, Pennsylvania*

INTRODUCTION

Total anomalous pulmonary venous return with no connection, also known as common pulmonary vein atresia, presents immediately after birth in the neonate with severe, life-threatening cyanosis. Echocardiography is helpful in diagnosing specific characteristics of this disease after birth. Patients with this disease are often in respiratory failure and are frequently placed on high-frequency oscillator ventilation. Moreover, this diagnosis can mimic pulmonary hypertension of the newborn and lead to a management strategy that would be ineffective. Because of limited acoustic windows, other imaging modalities may be required to confirm the diagnosis. Management options for total anomalous pulmonary venous return with no connection are quite limited because there is significant damage to the pulmonary vascular bed, and the cyanosis is often irreversible despite ventilation strategies. Extracorporeal membrane oxygenation can support the circulation for a limited period of time, but often surgery to connect the pulmonary venous confluence to the left atrium is unsuccessful and does not reverse the lung disease. For these reasons, the majority of patients with this rare disorder succumb to this disease. This case report highlights the features of this congenital heart defect seen on echocardiography and also describes the limitations and potential need for other imaging modalities in the diagnosis and management of these patients.

CASE PRESENTATION

The patient was a preterm infant born at 32 weeks' gestation who was transferred from an outside hospital for respiratory failure. His prenatal history was limited as the mother had limited prenatal care with no reported fetal ultrasound examinations. His perinatal history was remarkable for persistent late decelerations necessitating early cesarean section. On delivery, the neonate was apneic despite stimulation and positive pressure ventilation. He was intubated and given surfactant. His oxygen saturation remained in the 50% to 65% range on 100% fraction of inspired oxygen. His chest x-ray showed diffuse whiteout of both lungs. His blood gases showed mixed severe respiratory and metabolic acidosis. Prostaglandin infusion at a rate of 0.01 μg/kg/min was initiated because of the concern for cyanosis. Blood cultures were drawn, and antibiotics were started. Inhaled nitric oxide was also started, and the patient was subsequently transferred to our institution. On arrival, dopamine was initiated for hypotension, and his ventilation strategy was changed to high-frequency oscillation.

Echocardiography was performed shortly upon presentation to the cardiac intensive care unit. The pulmonary veins were not well visualized, though there was a suggestion of a small pulmonary venous confluence behind the left atrium (Figure 1, Video 1). A blind-ended vertical vein was suspected, but its connection back to the heart could not be identified. Thus, no flow was seen from the pulmonary venous confluence to either the left atrium or to the systemic venous system by two-dimensional or color Doppler. The echocardiogram showed exclusive right-to-left shunting across the atrial septum with a very small left atrium, as is typical of patients with totally anomalous pulmonary venous connection (Figure 2, Video 2). The right ventricle was markedly dilated, with marked septal bowing into the left ventricle and moderate systolic dysfunction, suggesting that there was severe pulmonary hypertension. A large patent ductus arteriosus was seen with almost exclusive right-to-left shunting also suggesting pulmonary hypertension with high pulmonary vascular resistance (Figure 3, Video 3). The transverse arch demonstrated retrograde diastolic flow (Figure 4, Video 4). There was no aortic insufficiency or evidence of coarctation of the aorta as the cause of the abnormal aortic flow pattern. With retrograde diastolic flow in the aorta seen on echocardiography, head ultrasound was performed to rule out a vein of Galen defect. (A vein of Galen is a cerebrovascular malformation of the arteries, which sometimes appears on echocardiography as retrograde diastolic flow in the descending aorta.) With all the right-to-left atrial shunting and the diminutive left atrium visualized on echocardiography, there was a high level of suspicion for severe pulmonary venous obstruction (Table 1). All these findings also provided evidence of severe pulmonary hypertension, which would be seen in severely obstructed total anomalous pulmonary venous return. The clinical management of the child was then adjusted to discontinue prostaglandins because of the concern for potential pulmonary venous obstruction. Head ultrasound showed no findings to suggest a vein of Galen defect. With the echocardiographic findings and the inability to accurately document the pulmonary veins returning to the heart, computed tomographic angiography was performed. At the same time, the cardiac catheterization laboratory was activated in case angiography was required. During computed tomographic angiography, there were difficulties timing the contrast injection appropriately because of the neonate's tachycardia and clinical instability. Thus, findings were nondiagnostic. Urgent cardiac catheterization was then performed.

From The Children's Hospital of Philadelphia, Philadelphia, Pennsylvania.

Keywords: Total anomalous pulmonary venous connection, Pulmonary vein atresia, Echocardiography

Conflicts of interest: The authors reported no actual or potential conflicts of interest relative to this document.

2468-6441

https://doi.org/10.1016/j.case.2017.11.001

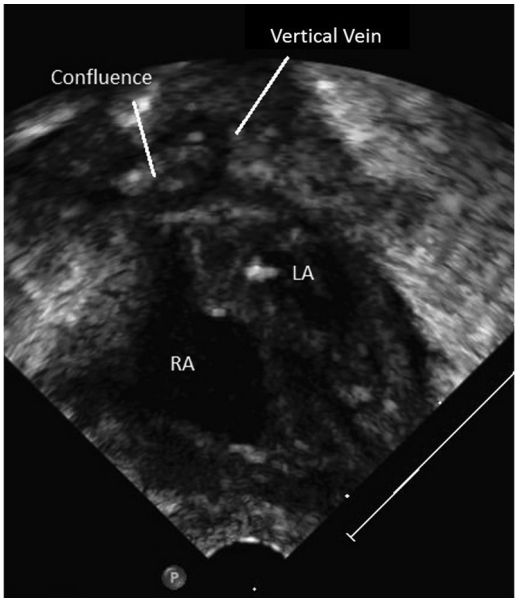

Figure 1 Subxiphoid left anterior oblique view demonstrating a small pulmonary venous confluence behind the heart. A blind-ended vertical vein is seen connected to the confluence. *LA*, Left atrium; *RA*, right atrium.

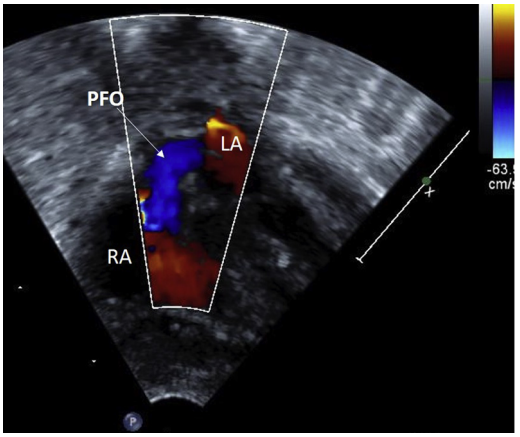

Figure 2 Subxiphoid left anterior oblique view demonstrating right-to-left flow across the atrial septum. *LA*, Left atrium; *PFO*, patent foramen ovale; *RA*, right atrium.

Limited angiography demonstrated that the pulmonary veins from each lung segment drained into a short common vein, which was atretic with no obvious egress. Instead, contrast was seen decompressing into a dense, fine network of bronchial veins to the innominate vein and below the diaphragm to other systemic veins (Figure 5, Video 5). These findings were consistent with a diagnosis of total anomalous pulmonary venous return with no connection or common pulmonary vein atresia. After a discussion with all treating physicians, it was felt that there was no viable intervention to repair this defect. Surgery was not an option for the patient because of the hypoplastic common pulmonary vein that was blind ended and did not drain back into the heart. The patient was brought back to the cardiac intensive care unit, where he expired shortly thereafter at around 12 hours of life.

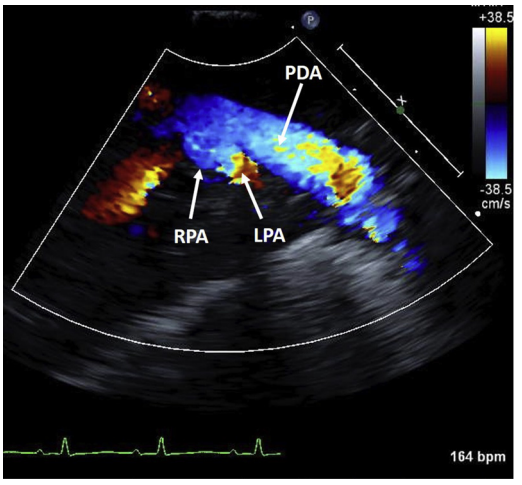

Figure 3 High parasternal view (ductal view) using color Doppler demonstrating all right-to-left flow across the patent ductus arteriosus (PDA). *LPA*, Left pulmonary artery; *RPA*, right pulmonary artery.

DISCUSSION

Total anomalous pulmonary venous return with no connection is a rare congenital heart disease first reported by Lucas *et al.*[1] in 1962 using the term *common pulmonary vein atresia.* Newborns with this disease are extremely ill and present immediately after birth with severe pulmonary hypertension and poor cardiac output due to the lack of adequate blood flow into the left side of the heart. In a recent review article, Perez *et al.*[2] reported that newborns with this disease usually expire within 48 hours of birth. This critically ill patient had limited prenatal care, with no warning of this disease. However, even with fetal ultrasound prenatal detection, pulmonary venous anomalies remain challenging in the fetus because there is limited pulmonary venous flow in utero. The majority of patients with this type of congenital heart disease are usually not diagnosed until after birth. A recent study describing the current status of prenatal detection rates across the United States for a wide range of congenital heart defects found total anomalous pulmonary venous connection to be the lowest detected among all congenital heart defects at approximately 9%.[3] In some cases, right ventricular dilation with an abnormal pulmonary venous confluence behind the atria can sometimes be identified prenatally and lead to a diagnosis, but the pulmonary veins are difficult to visualize on fetal echocardiography. Limited fetal diagnosis leads to an even higher responsibility to make the diagnosis after birth.

Total anomalous pulmonary venous return with no connection is characterized by lack of direct connection of a common chamber (confluence) of the pulmonary veins to the heart, portal system, or systemic venous system.[2] Echocardiography is useful in screening for this disease, although it lacks the ability to definitively diagnose the defect; in most cases of extracardiac total anomalous pulmonary venous connection, a vertical vein can be identified by echocardiography with a connection usually by way of a vertical vein to the portal system or the systemic venous system. In this case, echocardiography identified unique characteristics of total anomalous pulmonary venous connection without evidence of a patent vertical vein. Our inconclusive echocardiographic findings led to the need for additional imaging with a different modality. Cardiac catheterization