Cardiology in the Young

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Ebstein anomaly combined with unique pulmonary venous abnormality in a 9-month-old child

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Brief Report

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Abstract

Ebstein anomaly is a rare CHD known for its wide spectrum of presentation with the age of diagnosis dependent on the malformation's severity. Here, the authors describe a case of delayed diagnosis of Ebstein anomaly, secondary to lack of medical attention, which resulted in severe tricuspid regurgitation and pulmonary hypertension. Furthermore, the case was complicated by a unique pulmonary venous abnormality.

The prevalence of Ebstein anomaly was estimated to be 5.2 per 100,000 live births in the Baltimore-Washington Infant Study.[1] This rare disease has a high neonatal mortality (20–40%) with less than 50% surviving to 5 years of age.[2] Ebstein anomaly was first described in 1866 by Wilhelm Ebstein.[3,4] He based his description of the anomaly on the autopsy of a 19-year-old cyanotic man, noting that the tricuspid valve abnormality was the most significant feature. Since his initial observations, five criteria for the diagnosis of Ebstein anomaly have been established: adherence of the septal and posterior leaflets of the tricuspid valve to the underlying myocardium, a downward displacement of the tricuspid annulus, varying degrees of dilation of the "atrialised" portion of the right ventricle, tethering of the anterior leaflet without fenestrations, and dilation of the right atrioventricular junction.[5] Even with these defining characteristics, Ebstein anomaly continues to have a wide range of presentation – from the severely symptomatic neonate to the asymptomatic adult – because of the various degrees of leaflet abnormalities, right ventricle dysfunction, and coexisting cardiac abnormalities. Ventricular septal defect, pulmonary outflow obstruction, accessory conduction pathways, mitral valve prolapse, bicuspid aortic valve, and left ventricle noncompaction have been described in combination with Ebstein anomaly. However, to our knowledge, there has been no reported case in the literature of Ebstein anomaly and obstructive pulmonary venous return.

Case presentation

A 9-month-old Caucasian male presented to the emergency room after evaluation by a primary care provider and found to be hypoxic with saturations in the 50 s. The mother received no prenatal care, and the patient was born full-term at home with the assistance of a midwife. He received no vaccinations and had only been evaluated by a paediatrician three times during his life. Family reported feeding difficulties for several weeks before presentation and suboptimal weight gain.

Upon presentation to the local emergency room, the patient was found to be afebrile (97.4 F), tachypneic (RR 51), tachycardic (HR 153), and hypoxic. Initial saturation was 50%, which improved to 80% with oxygen supplementation. Physical exam showed overall pallor and cyanotic appearance, 3/6 high-frequency holosystolic regurgitant murmur best heard at the left lower sternal border, cyanosis of the nail beds, delayed capillary refill, hepatomegaly, and diffuse mottling. Labs on arrival to the emergency room showed a white blood cell count of 13.48, haematocrit of 42.4, bicarbonate of 14, creatinine of 0.2, total bilirubin of 1.9, aspartate aminotransferase of 107, alanine aminotransferase of 37, and AlkPhos of 192. A chest X-ray showed cardiomegaly without pulmonary oedema. Cardiology was consulted in the emergency room, and a limited bedside echocardiogram was performed which showed severe Ebstein anomaly, severe tricuspid regurgitation, and severely dilated right atrium and right ventricle with significant atrialisation of the right ventricle. The patient was transferred to Arkansas Children's Hospital's CVICU for further management.

Repeat echocardiogram confirmed the diagnosis and showed right ventricle pressure of 116 mmHg. Pulmonary venous course was abnormal but could not be clearly identified. However, spectral Doppler was concerning for obstruction.

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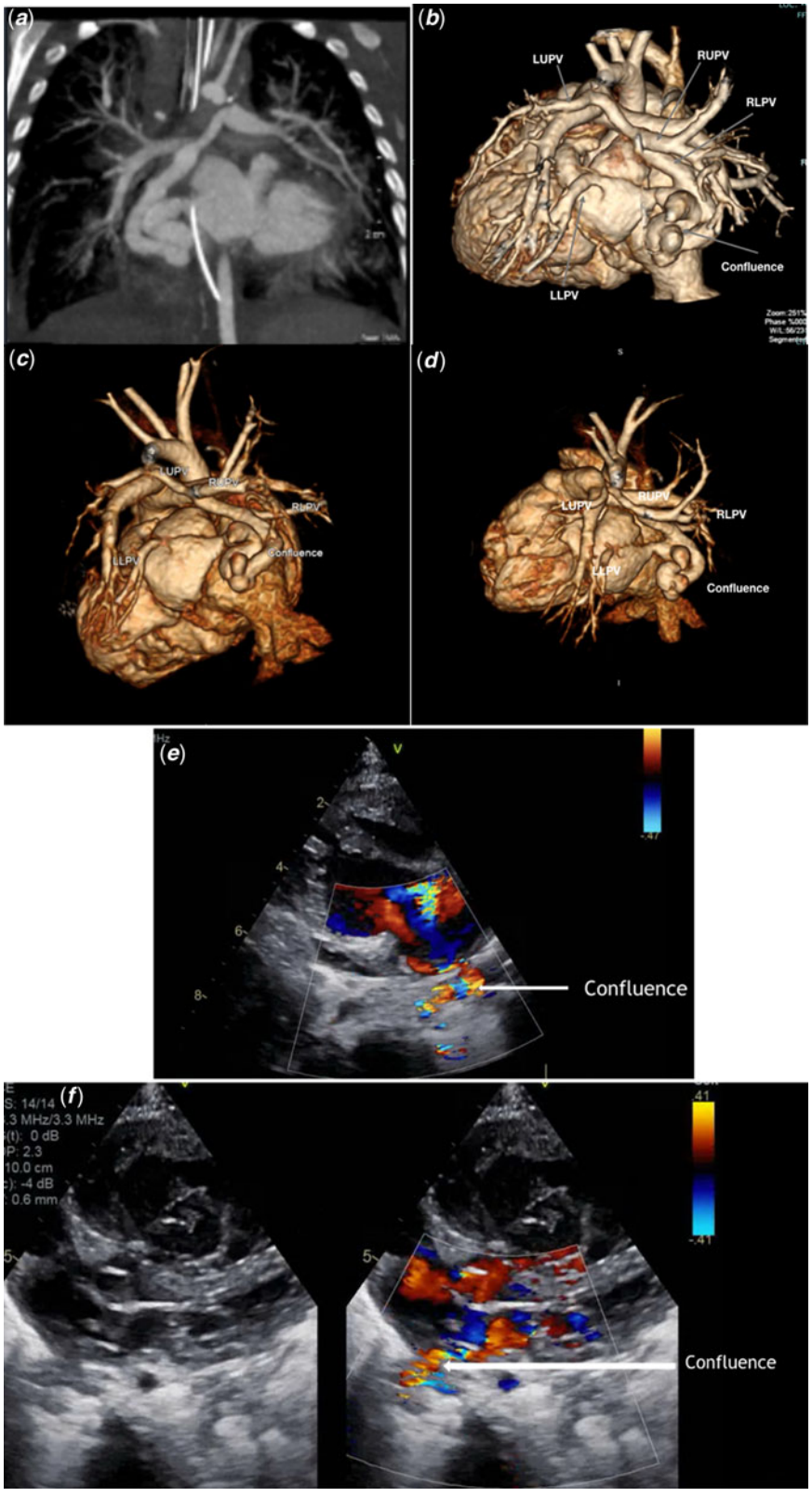

Figure 1. (*a*) Coronal view demonstrating the right upper and lower pulmonary veins as well as the left upper pulmonary veins forming a common pulmonary vein which follows a serpiginous course before entering the left atrium. Not visualised, the left lower pulmonary vein connected to the left atrium with significant focal stenosis. (*b*, *c* and *d*) 3D reconstruction of the patient's heart. (Images with removal of aortic arch and azygos vein). (*e* and *f*) Echocardiogram images demonstrating the suboptimal visualising of the unique pulmonary venous return.

He continued to have desaturations requiring initiation of nitric oxide within 24 hours of admission. Cardiac catheterisation showed abnormal pulmonary venous course returning to the left atrium; however, the procedure was challenging due to heart block, and individual assessment of the pulmonary veins could not be performed. The pulmonary artery systolic pressure was found to be equal to systemic pressure at 85 mmHg, and the diastolic pulmonary artery pressure was 36 mmHg.

Cardiac CT was preformed to further evaluate the pulmonary veins, which demonstrated obstructive pulmonary venous return.

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The right upper and lower pulmonary veins, as well as the left upper pulmonary vein formed a common pulmonary vein, which followed a long serpiginous course before entering the left atrium. A focal stenosis was noted at the junction of the common pulmonary vein and the left atrium. The left lower pulmonary vein was connected to the left atrium with focal stenosis near the junction with the left atrium. In addition, there was diffuse hypoplasia of the aorta (Fig 1).

The case was reviewed, and no surgical intervention was recommended due to the clinical state of the patient and the complexity of the pulmonary venous repair. He was made comfort care and passed soon after.

Discussion

This case is unique because of the presence of abnormally obstructive pulmonary vasculature in a patient with Ebstein anomaly, which was detected by advanced imaging modalities. The delay in the diagnosis and surgical intervention could possibly be a result of the lack of prenatal care and paediatric well-child visits. However, the coexistence of Ebstein anomaly and this particular abnormal pulmonary vasculature has never been discussed in the literature, so the overall prognosis is not known even with surgical intervention. The administration of nitric oxide in this case was prior to the definitive diagnosis of the patient's unique anatomy, and the decision to discontinue the use of nitric oxide was made once the advance imaging demonstrated the pulmonary venous return obstruction. This is of particular importance because inhaled nitric oxide in the presence of obstructive pulmonary venous return can worsen interstitial pulmonary oedema.

Many known factors contribute to the development of Ebstein anomaly in utero: psychiatric drug use during pregnancy, environmental factors, and genetic factors. However, most cases of Ebstein anomaly are known to be sporadic with rare familial cases. Several studies have identified the severity of tricuspid valve insufficiency as a risk factor for poor outcome.[6] In this case, it is possible that severe tricuspid valve insufficiency occurred later in the infancy following the progression of pulmonary venous obstruction and the development of severe pulmonary hypertension. This also might explain his late presentation with severe cyanosis. The limitations of this case report reside with the lack of medical information prior to admission to the hospital, which is unavoidable. Certainly, there is more to be learned about Ebstein anomaly and the importance of coexisting cardiac abnormalities.

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Conflicts of Interest. None.

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