A 28-year-old woman was referred to an outside center at 20 weeks’ gestation for fetal echocardiogram, which revealed the diagnosis of tetralogy of Fallot with absent pulmonary valve. There were an anterior malalignment-type ventricular septal defect, overriding aorta, and severe pulmonary insufficiency in the setting of severely dysplastic, rudimentary pulmonary valve leaflets. The patient presented to our center at 31 weeks for follow-up evaluation and delivery management planning. Fetal echocardiogram confirmed the diagnosis. The main and branch pulmonary arteries were markedly dilated (Figure 1A and 1B and Movies I and II in the online-only Data Supplement), and there was no ductus arteriosus. The cardiac silhouette was shifted leftward with a cardiac axis of 112°, deviating significantly from a normal fetal cardiac axis of 45° (Figure 1C). The left lung appeared small and diffusely hyperechoic relative to the right lung, suggesting atelectasis or consolidation.

At 35 weeks’ gestation, a fetal magnetic resonance imaging (MRI) study was obtained to assess the lungs and airway to help predict whether respiratory compromise would be present at birth. The MRI demonstrated significant leftward shift of the mediastinum and posterior and leftward deviation of the heart with severely dilated pulmonary arteries. Aeration of the lungs was markedly asymmetrical (Figure 2A). The right lung, particularly the middle and lower lobes, was overinflated with increased T2 signal, suggesting trapped fetal lung fluid, consistent with congenital lobar emphysema. The left lung was small and underinflated, with a lung volume of 5 mL compared with a right lung volume of 93 mL (Figure 2B). Only a small segment of the intrathoracic trachea could be identified, suggesting bronchial compression by the dilated pulmonary arteries (Figure 2C).

Given the severity of the lung pathology seen on fetal MRI, a plan of care was created that included scheduled cesarean section at 38 weeks with a specialty team comprising a neonatologist, a cardiologist, and a cardiac intensivist in the delivery room to initiate care at the time of cord clamping. The cardiac surgeon was available on standby in the cardiac intensive care unit with the extracorporeal membrane oxygenation pump primed and ready if needed. At birth, there was profound hypoxia, necessitating immediate intubation and ventilation with 100% oxygen. The baby was transferred directly from the delivery room to the cardiac intensive care unit, where chest x-ray confirmed the presence of severe right lung overexpansion and left lung collapse or hypoplasia (Figure 3).

During the first 3 hours of life, a variety of specialized ventilatory strategies were attempted with the goal of minimizing air trapping and volutrauma. The right lung continued

Figure 1. A, Fetal echocardiogram image demonstrating massively dilated main and branch pulmonary arteries. Arrow indicates the rudimentary pulmonary valve. LPA indicates left pulmonary artery; and RPA, right pulmonary artery. B, Sagittal fetal echocardiogram image demonstrating a cross section of the massively dilated right pulmonary artery. Ao indicates aorta. C, Axial echocardiographic view of the fetal chest demonstrating marked leftward deviation of the cardiac axis in the setting of right lung overexpansion and left lung hypoplasia vs compressive volume loss. The angle demonstrates a cardiac axis of 112°. Note that the left lung appears more echo-bright compared with the right lung. LL indicates left lung; LV, left ventricle; RL, right lung; and RV, right ventricle. *The fetal spine.
to become more hyperexpanded, resulting in compression of the heart, a dramatic shift in cardiac axis to the point of near-inversion of the apex (Figure 4A and 4B), and clinical evidence of diminished cardiac output. The patient became more acidotic and difficult to ventilate effectively despite maximal support. The decision was made to initiate venoarterial extracorporeal membrane oxygenation support before further decompensation and cardiovascular collapse. Once hemodynamic stabilization was achieved on extracorporeal membrane oxygenation, cardiac angiography confirmed the presence of massively dilated branch pulmonary arteries with distal branch hypoplasia (Figure 5). The patient was taken to surgery at 2 days of age for full repair with ventricular septal defect closure and replacement of the main and proximal branch pulmonary arteries with a bifurcating right ventricular to pulmonary artery conduit. She was decannulated from extracorporeal membrane oxygenation in the operating room with stable hemodynamics. The postoperative course was complicated by marked tracheomalacia and chronic lung disease, but the patient was eventually discharged home at 7 months of age.

Discussion

Tetralogy of Fallot with absent pulmonary valve is a rare congenital heart defect that, even in the modern era with improved surgical outcomes, remains associated with significant perinatal morbidity and mortality, particularly in those born with severe respiratory compromise. A recent review of case series from the past 2 decades reports survival beyond infancy ranging from 14% to 50%. Although fetal echocardiography has become increasingly sophisticated in recent years, it remains difficult to reliably predict whether a patient will be born with significant compromise and whether the patient will subsequently have a poor outcome. Multiple studies have attempted to identify fetal predictors of postnatal respiratory distress and early mortality but have been limited by small study populations. To date, fetal echocardiographic measurements have failed to predict clinical presentation or outcomes.

Postnatal MRI and computed tomography imaging have previously been reported to identify bronchial compression and congenital lobar emphysema in these patients. Neonates born with tetralogy of Fallot with absent pulmonary valve and severe respiratory compromise were found to have overexpansion of the affected pulmonary lobe resulting from airway narrowing with a resulting ball-valve effect and trapping of fetal lung fluid.

This report is, to the best of our knowledge, the first to document the use of MRI to evaluate a fetus with tetralogy of
Fallot with absent pulmonary valve to assess anatomic airway and lung findings suggestive of postnatal compromise. Being able to anticipate this patient’s postnatal clinical presentation allowed adequate preparation by the delivery room, intensive care unit, and surgical teams and facilitated a multidisciplinary plan of care that included preparation for respiratory failure at birth. This advanced knowledge allowed the team to intervene with appropriate ventilatory management, a smooth transition to extracorporeal membrane oxygenation, and early surgery despite the rapid deterioration in clinical status that occurred. This report suggests that fetal MRI may serve as a useful adjunctive clinical tool for predicting clinical presentation at birth in patients with this defect.

Figure 4. A and B. Consecutive subcostal echocardiographic images obtained within the first and third hours of life, respectively, demonstrating a profound leftward shift of the cardiac silhouette and near inversion of the heart with increasing overinflation of the right lung. I indicates inferior; L, leftward; LV, left ventricle; R, rightward; RV, right ventricle; and S, superior.

Figure 5. X-ray angiographic image confirming the presence of severely dilated main and branch pulmonary arteries (right pulmonary artery [RPA] is larger than the left pulmonary artery [LPA]). Arrow indicates the dysplastic pulmonary valve.

References


Disclosures

None.
Clinical Utility of Fetal Magnetic Resonance Imaging in Tetralogy of Fallot With Absent Pulmonary Valve
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