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IMAGES IN PEDIATRIC CARDIOLOGY

Prenatal Diagnosis of a Patient With Tetralogy of Fallot and an Absent Pulmonary Valve

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A 34-week-gestation fetus of a gravid 3 mother 29 years of age was referred to our hospital due to an abnormal vascular image on obstetric ultrasonographic examination. The family history of congenital heart disease was negative.

The mother had been using valproate due to epilepsy. Fetal echocardiography showed aneurysm of the pulmonary artery and rudimentary pulmonary valve and pulmonary regurgitation (Figs. 1, 2, movie 1). Tetralogy of Fallot and an absent pulmonary valve was diagnosed (Figs. 3, 4, 5).

At birth, the patient was symptomatic with severe respiratory failure. Postnatal echocardiography confirmed the diagnosis of dextroversion due to right lung atelectasia in addition to tetralogy of Fallot and an absent pulmonary valve. The aneurysm of the pulmonary artery was measured as 3.5 cm. Clinical examination showed no extra cardiac malformations or dysmorphic findings.

Genetic testing for 22q11.2 deletion was not possible. The chest tomography showed a right lung compression due to an aneurysmatically dilated pulmonary artery. The patient's condition deteriorated while on the ventilator due to bilateral pneumothorax. He died on the day 6 after the delivery due to severe respiratory distress while awaiting cardiac surgery.

Tetralogy of Fallot and an absent pulmonary valve are a rare congenital malformation characterized by an absent or rudimentary valve, typically both stenotic and regurgitant,

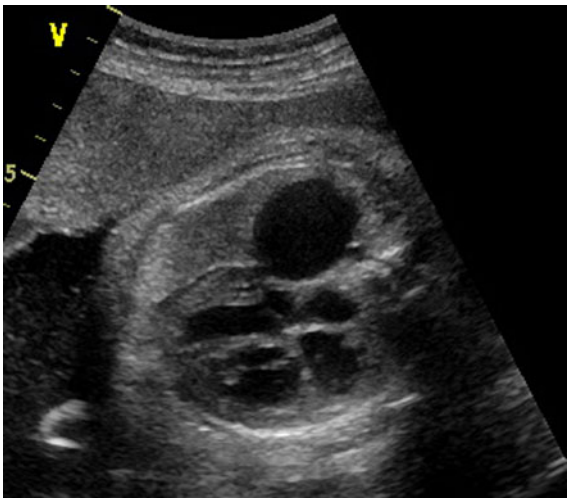

Fig. 1 Four-chamber views of fetal echocardiography showing a dilated vascular image behind the left atrium

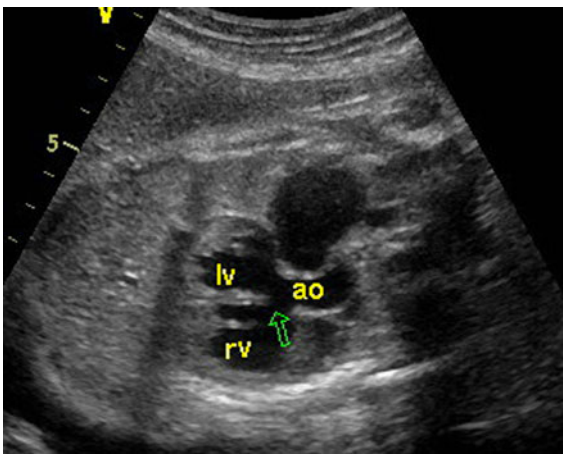

Fig. 2 Fetal echocardiography showing overriding of the aorta, a subaortic ventricular septal defect, and an aneurysmatically dilated pulmonary artery

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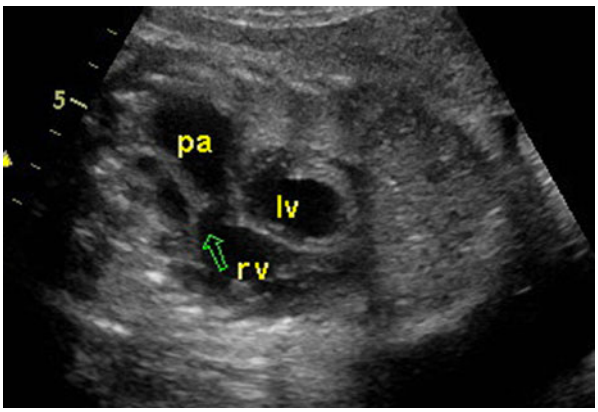

Fig. 3 Rudimentary pulmonary valve and aneurysmatically dilated pulmonary artery

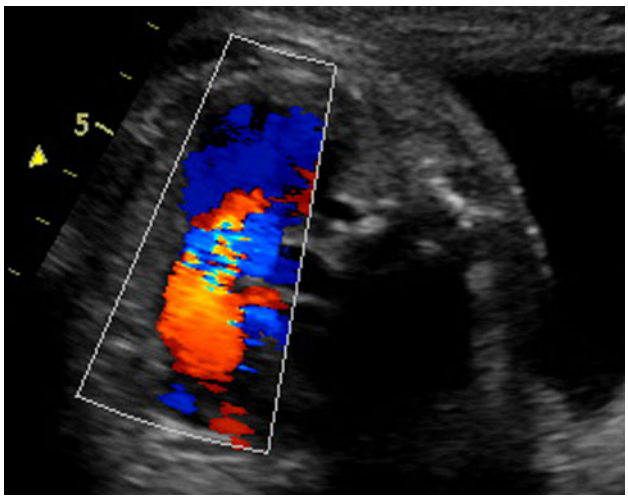

Fig. 4 Color-coded fetal echocardiography showing severe pulmonary regurgitation

together with aneurysmally dilated pulmonary arteries and a malaligned outlet ventricular septal defect [1]. The majority of the patients show respiratory symptoms of inspiratory and expiratory stridor, dyspnea caused by lobar collapse, and at times lobar emphysema due to compression of the bronchial tree by the grossly dilated proximal pulmonary arteries during the neonatal period and infancy. Prenatal diagnosis and early symptoms after birth have been correlated with a poor prognosis [3]. Severity of the defect, tracheobronchial and esophageal compression, and bronchomalacia due to compression of dilated branch pulmonary arteries are the poor signs of the outcome.

Agenesis or early closure of the ductus arteriosus plays an important role in pathogenesis [4]. Findings show 22q11.2 deletion in 75% of the patients with an absent pulmonary valve [2].

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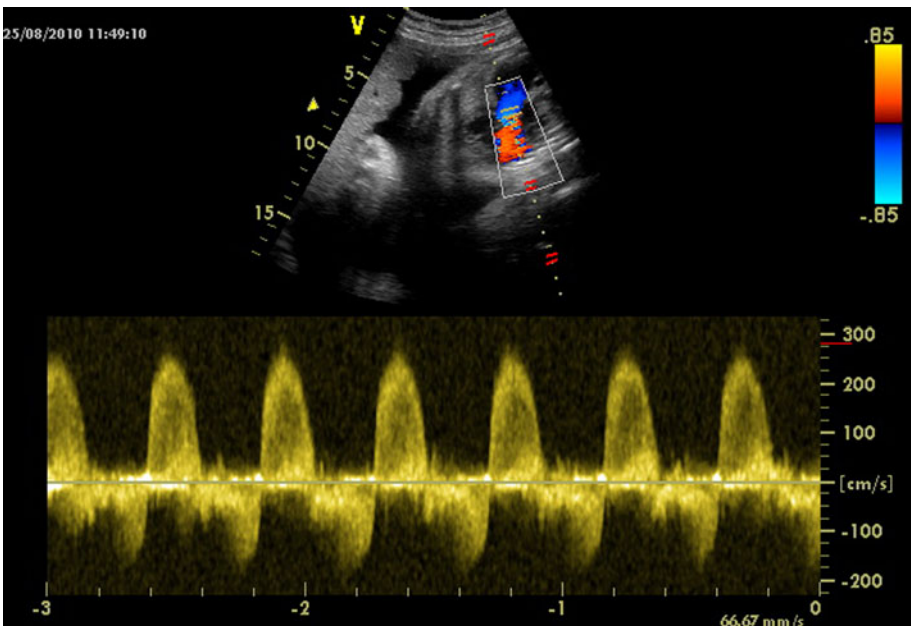

Fig. 5 Pulse-wave Doppler echocardiography showing a bidirectional flow pattern over the rudimentary pulmonary valve