Tetralogy of Fallot with absent pulmonary valve: main differences with classic Fallot are crucial for an accurate prenatal diagnosis and counselling

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DESCRIPTION

Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease. Classic features are a large perimembranous ventricular septal defect; aorta overriding the interventricular septum; right ventricular hypertrophy and right ventricular outflow tract obstruction (RVOTO) at subvalvar, valvar or supravalvar level and hypoplastic pulmonary arteries (PA), with variable severity.

A different and very rare form of TOF (<3% of total cases) occurs with absent/dysplastic pulmonary valve (AVP) leaflets, often with a hypoplastic ring causing RVOTO. In this rare condition, prenatal images are pathognomonic, with pulmonary valve regurgitation, enlargement of pulmonary trunk and branches and usually absent duc tus arteriosus.

Prenatal diagnosis can differentiate both conditions, which is of utmost importance for counselling, as it permits scheduling childbirth in a tertiary centre with the support of paediatric cardiology and/or cardiothoracic surgery in the case of AVP.

Prenatal management significantly improves the chances of survival and long-term results. Nevertheless, these cases remain highly difficult to predict, given the various degrees of involvement of the lung parenchyma, from mild bronchomalacia to severe dysplasia.

We present a case of TOF with AVP, diagnosed at 26 weeks’ gestation with typical features of TOF, severe pulmonary valve regurgitation (video 1 and figure 1), significant dilatation of the pulmonary trunk and branches and absent duc tus arteriosus. PA's dimensions remained stable during pregnancy. Investigation of 22q11 deletion was refused by the mother. Pregnancy was uneventful. Delivery occurred in a tertiary centre at 38 weeks gestation. Immediate severe respiratory distress with oxygen refractory hypoxemia resulted in invasive ventilation at 20 min of life. Transthoracic echocardiogram showed exclusive right to left shunt in the absence of RVOTO. Pulmonary venous return anomalies were excluded. Ventilatory parameters were optimised and inhaled nitric oxide was initiated. After starting inotropic support with temporary improvement, there was an inexorable decline. Extracorporeal membrane oxygenation (ECMO) was considered but refused by a multidisciplinary team since the patient was very unstable, needed transport to another hospital and ECMO would not preclude a surgical correction.

This marked instability was probably due to a severe parenchymal lung disorder.

As parents, we felt that we were given the clear notion that there were no prenatal echocardiographic markers that could anticipate or predict neonatal evolution being aware that this outcome would be possible.

Learning points

► Tetralogy of Fallot (TOF) with absent pulmonary valve is a rare variant of TOF, easy to diagnose, but hard to predict the outcome, with an overall poorer prognosis.
► There are currently no known definitive fetal markers of disease severity, therefore referral for programmed delivery at a tertiary centre with multidisciplinary neonatal support should be the standard of care for all cases.
► Even in the setting of the best neonatal conditions, parents should understand the risk of morbidity and mortality and the unpredictable evolution of each case.

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