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

Rita Ataíde Silva, Duarte Martins, Ana Teixeira, Rui Anjos

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 ductus arteriosus.1 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.2 Prenatal management significantly improves the chances of survival and long-term results.3 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 ductus 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.
Death occurred at 26 hours of life. Autopsy confirmed the diagnosis and documented severe small airway respiratory involvement (figures 2 and 3).

Prenatal diagnosis of TOF with AVP can be made once we are aware of its particularities. Given the scarce literature about echocardiographic markers of pulmonary development and the incapability of evaluation of the impact of PA compression, parental counselling has to address every possibility, including the increased chances of success when the diagnosis is made antenatally. The delivery should take place in a tertiary hospital.

Although several series report good results with patients diagnosed prenatally, these patients generally have a much poorer prognosis than the classic form and the course of the disease is unpredictable. Professionals must be aware that there are no antenatal predictors of morbidity in this particular form of TOF. The high mortality is related to airflow obstruction caused by aneurysmal dilatation of the PA.

Contributors All authors had contributed to manuscript composition: AT has followed this pregnancy as pediatric cardiologist. She captured several echocardiographic images during pregnancy, including figure 1 and video 1. DM and RA were member of the multidisciplinary team that took care of the newborn infant during his short period of life. RAS gathered all information and discussed the case with pathologists, providing image integration. All authors had read and corrected the manuscript.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent for publication Parental/guardian consent obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

ORCID iDs
Rita Ataíde Silva http://orcid.org/0000-0002-2090-0984
Duarte Martins http://orcid.org/0000-0003-0662-5629

REFERENCES