



EP-08.80 Prenatal risk stratification and counselling considerations of different variants of fetal Tetralogy of Fallot



Camilla Fazi, Lucia Pasquini, Olga Patey

Introduction

TOF is the most common cyanotic CHD (~1:3,000 live births, 5–7% of all CHD). While prenatal diagnosis is established, translating echocardiographic findings into structured risk stratification and variant-specific counselling remains incompletely defined.

Methods

A narrative review of the available literature was conducted for each TOF variant. Fetal echocardiographic parameters with established prognostic value were identified and analysed, including pulmonary valve z-scores, right ventricular outflow tract obstruction severity, ductal flow patterns, and branch pulmonary artery dimensions. Associated cardiac and extracardiac anomalies were reviewed, and prevalence data for genetic syndromes and chromosomal abnormalities across variants were synthesised to inform a structured counselling framework.

Conclusions

- This review presents the first structured framework linking prenatal echocardiographic risk profiles to individualised counselling for all three major TOF variants.
- By combining systematic parameter selection, anomaly characterisation, and outcome prediction, it offers clinicians a practical tool to improve both antenatal surveillance and parental communication in families facing a TOF diagnosis.

Result Table

Classic TOF (TOF-PS)	TOF with pulmonary atresia	TOF with absent pulmonary valve
Key echocardiographic parameters		
• Pulmonary valve Z-score • PA diameter and growth • Ductal flow direction • RVOTO severity • VSD dimension / restrictiveness • Aortic arch sidedness • Extracardiac anomalies	• PA branches confluence and size • Ductal morphology and flow • MAPCAs identification • Native PA size and growth • Aortic arch sidedness • Fetal MRI if no native PAs	• RV size and function • LV function • Biventricular dysfunction • Main PA & PA branches diameters • Bronchomalacia risk • DA (usually absent) • Extracardiac anomalies
Counselling implications		
• Pink vs ductus-dependent • PGE1 if ductus-dependent • Repair at 3–6 months • Reintervention up to 40% • Mortality <2–3% • 22q11 ~15%, T21 ~5% • ExCM ~1/3 non-syndromic • Recurrence risk ~3% • Lifelong ACHD follow-up • Neurodevelopmental f/u	• DA vs MAPCAs-dependent • Multi-staged surgery ~100% • Conduit reintervention ~100% • Biventricular repair in ~64–79% • 10-yr survival ~80% • 40-yr survival ~50% • 22q11 ~40–50% • ExCM 34–60%	• Hydrops/IUFD ~7–15% • Respiratory distress ~50% • Survival 38% (with respiratory compromise) • Survival ~95% (w/o respiratory compromise) • Conduit reintervention ~100% • 22q11 ~45% • ExCM ~76% • ECMO availability required