

Tetralogy of Fallot and malalignment ventricular septal defect detected at early echocardiography

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Objective

To better understand the natural history of Tetralogy of Fallot (TOF) and isolated malalignment ventricular septal defect (VSD) with aortic override detected in fetal life.

Methods

All cases of TOF or isolated (without pulmonary stenosis) malalignment VSD diagnosed at early echocardiography (< 16 weeks) were successively evaluated at 21 and 34 weeks' gestation respectively. Final diagnosis was determined at birth or at postmortem examination.

Results

36 fetuses with TOF or with malalignment VSD were detected at early echocardiography between 2013 and 2018. In 14 cases, parents opted for termination of pregnancy (TOP) in the first trimester, due to other structural or chromosomal abnormalities and were excluded from the study. In 2 further cases parents opted for TOP at the mid-trimester scan. In 6 of the remaining 20 fetuses (14 cases of TOF and 6 malalignment VSD) the congenital heart disease progressed in severity in the second (n=2) and third trimester (n=4). In 4 of these cases with TOF a malalignment VSD progressed and 2 cases of TOF progressed in TOF with pulmonary atresia (PA).

Conclusion

TOF or malalignment VSD may be diagnosed at early echocardiography. In almost 40% of cases, associated anomalies were detected in the first trimester. Progression from a malalignment VSD to tetralogy of Fallot and from TOF to TOF with PA throughout pregnancy was documented in the study. Interestingly it seems to be less frequent between early and second trimester echocardiography than between second and third trimester echocardiography. However, more case series are needed to identify the correct incidence of this phenomenon. This information is also critical for appropriate patient counselling.