

Fetal Counseling Provider Information

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Ebstein Anomaly

Definition/Anatomy

- The septal and posterior leaflets of the tricuspid valve are displaced into the right ventricle (RV) cavity, so that a portion of the RV is incorporated into the right atrium (atrialized RV), resulting in functional hypoplasia of the RV and tricuspid valve regurgitation (TR)
- Depending on the severity of the TR, the right atrium may be massively dilated. The RV free wall is often thin. Myocardial fibrosis can be present in the RV and LV free walls (which may cause ventricular dysfunction).

Incidence ^[1-3]

- 1/200,000 live births, 0.5-1% congenital heart disease
- Male:Female 1:1
- 3-7% CHD diagnosed in fetuses
- Increased incidence in maternal cigarette smoke exposure, first degree family members with CHD

Associated Problems ^[4-6]

- Chromosomal abnormalities: MYH7, NKX2.5, 10p13, 1p34, CHARGE, T21, T13, 1p36 deletion
- Extracardiac abnormalities: Pulmonary hypoplasia

Cardiac Abnormalities:

- ASD/PFO
- VSD
- Pulmonary stenosis or atresia, PPS

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- PDA
- Left heart lesions: mitral valve abnormalities, subAS, BAV, CoA
- LV non-compaction
- cc-TGA 15-50% of the time have Ebstein-like deformity of the tricuspid valve
- Accessory pathway up to 20%

Fetal Diagnosis

- More severe disease can be diagnosed as early as 11 weeks
- More subtle disease can be seen in later gestation with worsening of RA enlargement due to TR

Fetal Imaging Predictors of Postnatal Management/Outcome ^[7-10]

- GOSE ratio: RA/(RV+LV+LA)

GOSE Score	Ratio	Mortality
1-2	<1.0	8%
3 (acyanotic)	1.1-1.4	10% early, 45% late
3 (cyanotic)	1.1-1.4	100%
4	>1.5	100%

- Prenatal risk factors for perinatal mortality:
 - Pericardial effusion (2.5 increased odds)
 - diagnosis at <32 weeks GA (8.6 increased odds)
 - larger TV annulus z score
 - pulmonary insufficiency twice (3 times increased odds)
- Predictors of mortality on last fetal echo (>30 weeks)
 - Higher CTA ratio
 - Lower TR jet velocity
 - Depressed LV function

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- Absent or reversed diastolic flow in umbilical artery (~10 times increased odds)
- Umbilical vein Z score <1 (Odds 2.5)
- Overall perinatal mortality of 45% (fetal demise or neonatal death before discharge) in those diagnosed prenatally
- In liveborn patients risk factors for neonatal mortality:
 - 68% survivability in severe disease out of neonatal life
 - GA <37 weeks (OR 4.0)
 - Birth weight <3kg (OR 7.5)
 - Lower TR velocity
 - No antegrade pulmonary blood flow

Long term survival

- 20 year survival 76% in babies who survive neonatal period
- Arrhythmias
 - First degree AV block in 42% of patients
 - Atrial arrhythmias
 - Accessory pathway, up to 36% have more than one accessory pathway
 - Wide complex tachycardia
- Ventricular dysfunction

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