

Indication Specific Diagnostic Yield of Fetal Echocardiography

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Objectives: To document the indication specific diagnostic yield of fetal echocardiography, when performed by established referral indications.

Methods: Retrospective study based on referral indications and documented findings of fetal echocardiograms performed over 6 years (1997-2013) in an academic referral center for fetal cardiology. Referral indications have been classified as either supported by literature (Ind+) or not (Ind-). Indication specific diagnostic yields and Odds Ratios (O.R) for any abnormality, fetal congenital heart disease (fCHD) and critical fCHD when fetal echocardiography was applied in Ind+ and Ind- pregnancies have been estimated.

Results: 1782 out of 1847 fetuses (1804 pregnancies) were included, having complete referral data. 915 (51.3%) corresponded to Ind+ cases (50.4% fetal, 36.9% familial, 31% maternal origin of referral indication). Most of Ind- cases (52%) had no referral indication (family or physician wish), the remaining not established indications (including previous abortion, reduced fetal heart imaging, echogenic foci, borderline NT <3.5mm, etc).

Overall, the incidence of any abnormality, fCHD and critical fCHD was 35.7%, 27.6% and 1.9%, respectively. The majority of fCHD were ventricular septal defects (38.6%) followed by (5-10% each) aortic coarctation, atrial septal defects and (<5% each) valvular stenosis, arch abnormalities and more complex forms of CHD.

Ind+ compared to Ind- referrals were associated with significantly increased risk (Chi-square $p < 0.001$) for any abnormality (46.1% vs 24.5%, O.R: 2.6, 2.1-3.2), for fCHD (35.8% vs 18.9%, OR: 2.3, 1.9-2.9) and for critical fCHD (3.2% vs 0.6%, O.R:5.6, 2.6-14.5).

Diagnostic yield (%) for fCHD / critical CHD were highest (>50%) in Ind+ referrals for polyhydramnion (71 / 14) abnormal heart configuration in anomaly scan (56/ 8), chromosomal abnormalities (62 /1) , followed (25-50%) by referrals for fetal malformations (39/3) increased NT (36/1.8), history of maternal CHD (45 /4.5). Lowest yields (<25%), were documented in referrals for IVF (21 / 1.3), maternal diabetes (18/1.9), teratogen drug exposure (18/ 0), monochorionic twins (16/0).

Conclusions: Fetal echocardiography performed for established indications is associated with significantly increased diagnostic yield for fetal CHD, which is referral indication specific. However, critical fetal CHD cases might still escape antenatal diagnosis in the absence of indication for fetal echocardiogram.