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**Fetal Echocardiography as a "genetic screening" tool?**

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**Introduction:**
Amniocentesis is routinely recommended in pregnancies with increased risk for fetal karyotype abnormalities (fKA), as assessed by maternal age combined with biochemical and sonographic indexes. Fetal Echocardiography (fEcho), performed upon established indications (EI) allows for fetal congenital heart disease (fCHD) detection, often independently associated with fKA. We evaluated whether the detection of fCHD increases the risk for fCA in pregnancies with and without EI for fEcho.
**Methods:**
Retrospective study of 2,202 fEcho records (years 2008-16) from a tertiary referral centre, regarding 1) referral indications, classified as established (EI) when based on recent recommendations (AHA 2014) or non-established (NEI), in remaining cases 2) diagnosis of fCHD (excluding ventricular or great artery disproportion, inflow valve regurgitation, pericardial fluid, PDA constriction, arrhythmias), 3) results of fetal karyotype (prior or following fEcho).
The prevalence of abnormal karyotype in the presence/absence of fCHD was compared, in subgroups of EI(+) and EI(-) fEcho cases (Pearson Chi-square test, Odds Ratios-O.R)
**Results:**
Included were 258 studies with complete referral data, fEcho diagnoses and available fetal karyotype results.

| Indication-based fECHO | fCHD | Abnormal karyotype | Normal Karyotype |
|---|---|---|---|
| yes | yes | 18 | 81 |
| | no | 13 | 80 |
| no | yes | 1 | 24 |
| | no | 1 | 40 |

The prevalence of fetal karyotype abnormalities, in presence vs absence of fCHD respectively, was: A) 15% vs 10.6% in the whole group (O.R: 1.5, 95% C.I:0.7-3.1, p=0.282, n.s)
B) 18.2% vs 14% for EI(+) cases (O.R: 1.4, 95% C.I:0.6-2.9, p=0.429, n.s),
C) 4% vs 2.4%, for EI(-) cases (O.R:1.6, 95%C.I:0.1-27, p=0.72, n.s).
Cases having established indications for fEcho (EI+) had a significant association with the presence of fKA (16.6%) compared to cases where fECHO was “routinely” performed (3%), O.R: 6.3, 95% C.I:1.4-7.3, p=0.005)
**Conclusions:**
The detection of fCHD during fetal echocardiography is associated with an increased trend (1.5 fold) for fetal karyotype abnormalities. However, the strongest predictor (6-fold increased risk) is the presence of an established indication for fetal echocardiography itself. Although fetal echocardiography should not be used as a means of fetal karyotype “screening”, still careful obstetric history talking might identify pregnancies that could benefit from both fetal echocardiography and fetal karyotyping.