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Early fetal echocardiography: indications, abnormal findings and comparison with postnatal results

Zamora M, Comas M, Serra L, Donoso L, Lesmes C, Mellado F
CORPORACIO SANITARIA PARC TAULI, SABADELL, Spain

Objective

Due to technical advances in ultrasonography, fetal echocardiography has become feasible in the late first and early second trimesters. Early fetal echocardiography (EFE) is recommended in high-risk patients for both maternal and fetal indications, with the intention of examining the fetal heart in greater detail than in the standard screening ultrasound. Our main objective was to describe the indications and findings of EFE performed in our centre and secondly, to determine the diagnostic efficacy of this technique.

Methods

This retrospective study included all EFE carried out between 2016 and 2018 in our centre. Examinations were performed at 13-16 weeks of gestation using transabdominal and/or transvaginal ultrasonography in patients with high or medium-risk of congenital heart defect (CHD). Most of them also underwent a regular echocardiographic exam in the second trimester. In cases of anomalies, follow-up was carried out by postnatal echocardiography or by autopsy report in cases of pregnancy termination. Cases lost to follow up were excluded from this analysis.

Results

A total of 148 patients were included. The main referral indication to perform EFE was an increased nucal translucency (31.1%) followed by pre-gestational diabetes (23%), morbid obesity (12.8%), reverse ductus venosus (10.8%), previous child with CHD (6.8%), positive Anti-Ro (5.4%), family history of CHD (5.4%) and teratogens (3.4%) The prevalence of abnormal findings was 5.4% (8/148). EFE was normal in 129 cases (87.2%) and in 11 patients (7.4%) it was considered incomplete. Pathologic findings in this early echocardiographic exam were: suspected ventricular septal defect (4 cases), mitral insufficiency, DV agenesis, tricuspid atresia and left heart hypoplasia (1 case each). Of these 8 pathologic results, 5 (62.5%) were confirmed in the postnatal study. Of the 129 normal EFE, 3 (2.4%) had a definitive result of CHD: one auricular septal defect, one ventricular septal defect and a case of Aorta coarctation. According to these results, sensitivity and specificity of EFE were 62.5% and 97.9%, respectively.

Conclusion

EFE appears to be a reasonable screening tool for CHD in a selected risk population. However, due to the natural history of CHD, where some pathologies can only visualized in later stages of pregnancy, EFE should always be followed by echocardiography at mid gestation.

Table 1. Characteristics of study population

Descriptive analysis		Mean (SD)
Maternal age (years)		33 (5)
Time of early echocardiography (weeks)		14,5 (1)
Time second-term echocardiography (weeks)		21,6 (2,3)
Time postnatal echocardiography (days)		32 (29)
Body Mass Index (BMI) (kg/m2)		28,30 +/- 7,75
Descriptive analysis		**N (%)**
Indication of early echocardiography	Pathologic NT	46 (31%)
	Pre-gestational diabetes	34 (23%)
	Morbid obesity	19 (13%)
	Pathologic DV	16 (101%)
	Previous child with CHD	10 (7%)
	Familiar history of CHD	8 (5%)
	Ro antibodies	8 (5%)
	Drugs	5 (3%)
	Other	2 (1%)
Result of early echocardiography	Normal	129 (87%)
	Pathologic	8 (5%)
	Incomplete	11 (7%)
Result of 2nd T echocardiography	Normal	117 (92%)
	Pathologic	10 (8%)
Results postnatal study	Normal	32 (80%)
	Pathologic	8 (20%)
Nuchal translucency (NT)	Normal	102 (69%)
	95th-99th centile	7 (5%)
	>99th centile	39 (26%)
Ductus venosus (DV)	Normal	128 (86%)
	Reverse	20 (13%)
Other major abnormalities		13 (9%)
Genetic disorders		4 (3%)