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Original Research Article

Significance of Echogenic Intracardiac Foci in Predicting Fetal Aneuploidy: A Prospective Observational Study in Rural Tertiary Care Hospital

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Abstract

Background: Echogenic intracardiac foci (EIF) are described as hyperechoic regions on papillary muscles or Chordae tendineae in the fetal cardia. The incidence ranges form 4-30% in euploid and aneuploidy respectively. The positive likely-hood ratio between Trisomy 21 and EIFs is 1.4 to 1.8.

Objective: This study was done to understand the association between EIF and aneuploidy and congenital anomalies.

Methodology: Out of all the cases undergoing obstetric scan after 14 weeks of gestation in Adichunchanagiri Institute of Medical sciences, showing Echogenic Intracardiac Foci were included in our study. All the cases were examined in detail and followed up till 3 months postpartum to look for aneuploidy.

Results: 842 cases underwent scan after 14 weeks in our hospital and out of that 30 (3.5%) had isolated EIF whereas 5 had EIF associated with other soft markers. Out of this 30 cases with isolated EIF only 2 had congenital anomalies.

Conclusion: Isolated EIFs are not markers of aneuploidy and hence are not an indication for invasive testing for the same.

Keywords: Echogenic Intracardiac foci, Fetal Aneuploidy, Congenital Anamolies.

Introduction

Echogenic intracardiac foci (EIF) were first described in 1987 [1]. They are small, discrete, bright areas visualised on papillary muscles or chordae tendineae in either of the ventricles of fetal heart but most commonly in left ventricle. They are well visualised in four chamber cardiac view, but diagnosis is confirmed when visualised in 2 distinct cardiac planes.

The incidence of EIF is 4-7% in euploid foetuses and 15-30% in foetuses with trisomy 21[2]. EIF was initially thought be a marker

of aneuploidy and congenital abnormalities, however over the years the significance of EIF has been debated extensively and studies have found that isolated EIFs have positive likely-hood ratio of 1.4 to 1.8 (0.95)[3]. On the other hand few studies have found that EIF have been associated with anueploidy and congenital anamolies. ACOG has declared isolated EIF as clinically insignificant or normal variant and no advanced invasive screening is advised unless associated with additional soft markers like echogenic bowel, hypoplastic nasal bone, single umbilical artery. Since no data is available about the significance of EIFs in this part of Karnataka and we undertook this study to understand the correlation between EIFs with aneuploidy.

Materials and Methods

This prospective observational study was carried out in Department of Obstetrics and Gynecology, Adichunchanagiri Institute of Medical Sciences, Mandya. India. This study was conducted from January 2020 to January 2023.The study protocol was presented to the IEC of the Adichuchanagiri Institute of Medical Sciences, Mandya and clearance was obtained before the start of the study. Enrolling 842 cases who met the inclusion criteria

All the cases undergoing ultrasonography over 14 weeks from January 2020 to January 2023 were included in the study. A detailed Ultrasonographic examination was done. Cases showing echogenic foci in either of the ventricles in four chamber cardia view and confirmed in 2 planes were diagnosed to have Echogenic intracardiac foci. Thorough examination was done to look for any other associated soft marker or structural malformations. Cases with isolated EIFs were only included in the study. Amniocentesis and NIPT was offered in cases with advanced maternal age and other high risk factors however due to economic constraints none of them gave consent for the tests. The cases were followed up throughout pregnancy and 3 months post-delivery. Fetal outcome was analysed in terms of presence or absence of aneuploidy.

Statistical Analysis

The data was collected using a pre-formed paper sheet, entered in the MS Excel sheet and cleared. Categorical data was presented in the form of percentages and continuous data was presented in the form of means and standard deviation. Analysis of the data, binominal proportional and Chi square analysis were employed to determine the significant correlation of positive rate in hormone receptors of primary and metastatic sites. P-value less than 0.05 was regarded as a significant difference. SPSS-25 software was used for statistical analysis.

Results

842 cases underwent second trimester scan in our hospital. The demographic characters of our study group is summarised in the table below.

Table 1: Demographic characters

Parameter	Number (%)
Age	
< 20 years	5 (16.67)
20-35 years	22 (73.33%)
>35 years	3 (10%)
Gravidity	
Primi	18 (60%)
Multigravida	12 (40%)

Socioeconomic status	
LSES	17 (56.67%)
HSES	13 (43.33%)
History of previous child with aneuploidy	3 (10%)
History of congenital abnormality in family	4 (13.33)

Out of 842 cases, 35 had echogenic intracardiac foci. 30 had isolated EIFs while 5 were associated with other soft makers.

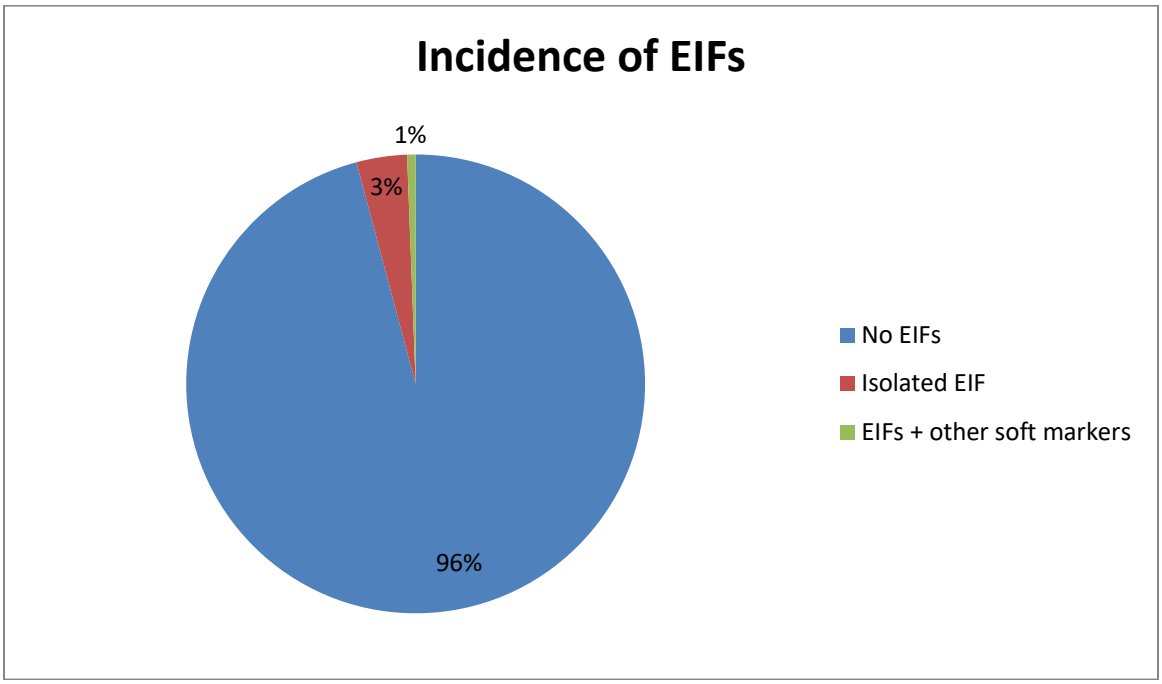

Figure 1: Incidence of Echogenic intracardiac foci in our study.

Therefore the incidence of EIFs was 4.15% while incidence of isolated EIFs was 3.5%. Majority of the cases [21 (70%)] had left ventricular EIFs whereas only 8 (30%) had right ventricular EIFs. Among the 5 cases associated with other soft markers, 2 had EIFs with choroid plexus cyst, 1 had EIFs with echogenic bowel, 1 had EIFs with choroid plexus cyst and pelviectasia and 1 had EIFs with thickened nuchal translucency and echogenic bowel.

Table 2: Table showing EIFs associated with other soft markers

Other soft markers	Number (%)
EIFs + Choroid plexus cyst	2 (40%)
EIFs + echogenic bowel	1 (20%)
EIFs + pelviectasia	1 (20%)
EIFs + Thickened NT + echogenic bowel	1(20%)
Total	5

Out of 30 cases 22 underwent Vaginal delivery and 8 underwent caesarean delivery. No babies needed NICU admission and out of 30 babies, only 2 babies had congenital abnormality. One baby had perimembranous VSD and one baby had U/L multicystic kidney. On follow up over phone the baby with Trisomy 21 had mild delay in attaining milestones whereas the baby with multicystic kidney had normal development.

Discussion

The association between isolated EIFs with aneuploidy has been analysed time and again due to the contradicting results with many studies. Goncalves *et al*[4] found that there was increased risk of fetal aneuploidy and congenital heart disease in pregnancies with EIFs. However, meta-analysis by Lorente A M R *et al*[5] concluded that isolated EIFs had low sensitivity (21.8%) for detecting Trisomy 21. The result from our study also supports this.

The incidence of isolated Echogenic intracardiac foci in our study was 3.5% which is in line with the global incidence of 4-7%[2]. Also 70% of the cases had left ventricular EIFs whereas only 30% had right ventricular EIFs this is similar to the data by Society for maternal-Fetal medicine (SMFM)[3].

In our study only 2 babies (6.67%) had congenital anamolies and the anamolies found in our study were Trisomy 21 and U/L multicystic kidney. Whereas Usta C S *et al*[6] found Aortic coarctation, VSD, Ompalocele, Ileal atresia in their study. A meta-analysis has found the likelyhood ratio of 0.95 between EIFs and Trisomy 21[3]. Overall the rate of aneuploidy in cases with EIFs is less and hence the SMFM recommends that if cfDNA or serum screen is negative no aneuploidy evaluation is needed and if no previous screening available, counselling should be done regarding NIPT; routine antenatal management should be done.

Conclusion

Isolated echogenic intracardiac foci have less likelyhood ratio and same results were found in our study. Therefore if not associated with other soft markers, there is no need for extensive aneuploidy evaluation however, adequate counselling should be done to explain the condition and decrease the apprehension.

Ethical approval

Permission for the study was obtained from the College authorities prior to commencement.

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