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Radiology Section

Significance of Echogenic Cardiac Nodule in Fetus as Soft Marker

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ABSTRACT

Introduction: The purpose of this study was to determine the pathological significance of echogenic cardiac nodule in the heart of fetuses with no other sonographic abnormalities and in the absence of other risk factors for chromosomal abnormality. A common ultrasound finding in relation with fetal abnormality was identified as echogenic foci. The association of this soft marker abnormality was less known.

Aim: The aim of this study was to document the outcome of fetuses having single or multiple echogenic cardiac foci with no maternal risk factors for chromosomal abnormality in Central Maharashtra.

Materials and Methods: The location and number of echogenic cardiac nodule on fetal scans were recorded prospectively for fetuses seen at the Tertiary Hospital, Kumbhari, Maharashtra, India, between May to December 2016. A total of 71 fetuses were identified with single or multiple echogenic cardiac nodules. This represents 17.2% of the total number of fetuses scanned (n=413).

Results: The most frequent finding was a single echogenic cardiac nodule (n=62, 87.3%), but multiple foci were also observed in 9 (12.6%). The most common findings were isolated echogenic foci, 64 (90.1%), in the left ventricle. An echogenic cardiac nodule in the right ventricle occurred in 2 cases (2.8%). Associated abnormalities were diagnosed in 5 fetuses, each one of bilateral club foot (1.4%), Left PUJ obstruction (1.4%), microcephaly (1.4%), Rhizomelic dwarfism (1.4%), Sacro coccygeal teratoma (1.4%). Postnataly in two patients Trisomy 21 (2.8%) was identified. The majority of fetuses which had echogenic cardiac nodule were normal.

Conclusion: We emphasize that single ecogenic cardiac focus is very common finding in our study. If it is not associated with other structural abnormalities, need to perform invasive genetic diagnostic tests may not be warranted.

Keywords: Echogenic cardiac foci, Left ventricle, Trisomy 21, Ultrasonography

INTRODUCTION

An increase in the prenatal detection of structural findings associated with anomalies was possible due to technological advances in ultrasonography. Recently, adjusting the overall maternal age related risk for aneuploidy, subtle ultrasound findings have been accepted as potential genetic markers. Echogenic Intracardiac Focus (EIF), Echogenic Bowel (EB), Choroid Plexus Cyst (CPC), shortened long bones (femur and humerus), and Renal Pelvicaliectasis (RP) are few most common markers for aneuploidy. Rate of chromosomal abnormality is considered to be high in advanced maternal age (i.e., >35 years old) but if we consider this single criteria for screening then 80% of fetal aneuploidy will be missed [1]. The relationship between advanced maternal age and fetal aneuploidy was established by Hook in 1981 [2]. Antenatal ultrasound examination of the fetus frequently includes imaging of the fetal heart, and detection of congenital heart disease by this method is now well established [3]. Single or multiple echogenic cardiac foci is common finding during obstetric sonography [4]. Echogenic cardiac foci are more common in the left ventricle than right ventricle [5-7], the reported incidence of echogenic foci in left ventricle ranges from 0.45 [5] to 22% [6].

There is diverse opinion regarding significance and management of ultrasound “soft markers” in low risk populations and its association with aneuploidy, particularly EIF [8-9].

The finding of a single echogenic focus in the left ventricle is not of clinical significance [4]; but few studies do not support this it. One of 26 fetuses with an echogenic focus in the left ventricle, reported by Schechter and colleagues

[10], had trisomy 21, and a pathological study of three fetuses demonstrated mineralization within a papillary muscle, which was suggested as the cause of the echogenic focus [11]. At autopsy, calcification of papillary muscles has been observed more frequently in trisomies 21 and 14 than in normal fetuses [12] and this, in conjunction with echocardiogaphic studies, has raised the possibility that echogenic foci may be a marker of fetal karyotypic abnormality. But this view is controversial as there is high incidence of echogenic cardiac focus in otherwise low risk pregnancies. Fetuses with major chromosomal abnormalities can be recognized by detailed fetal ultrasound scanning [13]. Considering high incidence of echogenic cardiac foci in fetus and risk involved in karyotypic procedure, it is controversial to consider isolated soft marker like echogenic cardiac focus as a significant karyotypic abnormality in low risk pregnancies.

MATERIALS AND METHODS

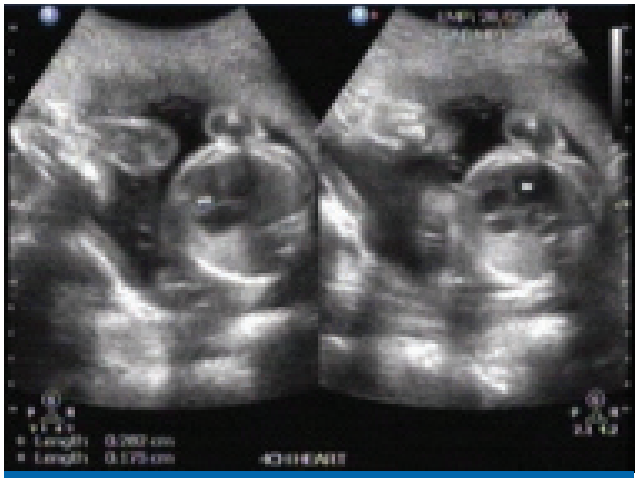
The location and number of echogenic cardiac nodule on fetal scans were recorded prospectively for fetuses seen at the Tertiary Hospital, Kumbhari, Maharashtra, India, between May to December 2016. The informed consent and Institutional ethical clearance was obtained prior to beginning of study. The study consisted of 71 cases in which echogenic foci were identified but in which there were no maternal risk factors for fetal abnormality, such as raised maternal age (>35 years), abnormal serum screening or a history of genetic disorders. In addition, fetuses with other soft markers such as nuchal oedema, choroid plexus cysts, echogenic bowel were excluded from the study population. In view of the association of congenital heart defects with karyotypic abnormalities, any fetus with a cardiac abnormality was also excluded. The study group, therefore, consisted of fetuses with finding of single or multiple echogenic foci. All had an obstetric ultrasound scan in addition to a detailed echocardiogram. The variables were analyzed included age, gestational age at ultrasound screening, echogenic nodule size, location and the presence or absence of anomalies. The total number of fetal anatomy ultrasounds performed during the study period was assessed and also the data was extracted from the patient's records as well as from ultrasound database. Sonographic examination for the detection of associated anomalies was performed between 15 and 36 weeks of gestation [Table/Fig-1,2].

STATISTICAL ANALYSIS

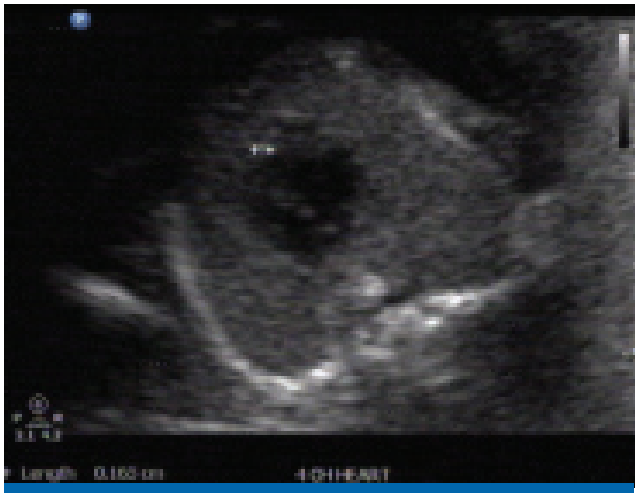
Descriptive statistics such as mean, SD and percentage was used to present the data. Data analysis was performed by using software SPSS v16.0

RESULT

A total of 71 fetuses ultrasound screening during the study period. The median gestational age of the 71 fetuses was 21 weeks (range 15-36 weeks). The median maternal age was 23 years with standard deviation (SD) 4.0. The average echogenic nodule size was 1.91. Majority of mothers belongs to age group 20-24 years (42.3%) followed by 25-29 years (25.4%). Mean maternal age was 23±4.0. [Table/Fig-3]. Majority of echogenic cardiac nodule was single i.e., 62 (87.3%) [Table/Fig-4]. The location of echogenic foci are shown in [Table/Fig-5]. The most common findings were isolated echogenic foci, 64 (90.1%), in the left ventricle. Approximately 3% of fetuses had 'golf balls' in the right ventricle, either in combination with 'golf balls' in



[Table/Fig-1]: Echogenic cardiac nodule in the left ventricle.



[Table/Fig-2]: Echogenic cardiac nodule in the right ventricle.

Maternal Age (years)	Frequency	Percentage (%)
< 20	16	22.5
20 – 24	30	42.2
24 – 29	18	25.4
>= 30	7	9.9
Total	71	100.0

[Table/Fig-3]: Maternal age distribution.

Number	Frequency	Percentage (%)
One Echogenic Cardiac Nodule	62	87.3
Two Echogenic Cardiac Nodule	9	12.7
Total	71	100.0

[Table/Fig-4]: Distribution of number of echogenic cardiac nodule.

Location	Frequency	Percentage (%)
Both Ventricles	4	5.6
Left Ventricle	64	90.1
Right Ventricle	2	2.8
Near Tricuspid Valve	1	1.4
Total	71	100.0

[Table/Fig-5]: Distribution of location of echogenic cardiac nodule detected by antenatal ultrasound scanning in fetuses.

the left ventricle. Echogenic foci were not associated with any impairment of cardiac function. From the above table, it was observed that 5 (7.04%) patients were found with other sonographic markers associated anomalies [Table/Fig-6].

Associated Anomalies	Frequency	Percentage (%)
Bilateral Club Foot	1	1.4
Left Puj Obstruction	1	1.4
Microcephaly	1	1.4
Rhizomelic Dwarfism	1	1.4
Sacro Coccygeal Teratoma	1	1.4
No Associated Anomalies	66	92.9
Total	71	100.0

[Table/Fig-6]: Distribution of associated anomalies of echogenic cardiac nodule.

DISCUSSION

The present study validates that echogenic foci may be single or multiple and found in either or both ventricles of the fetal heart. The most frequent finding in this series was a single focus in the left ventricle. This is consistent with previous reports [4-7,10,14]. However, the number of foci observed in a single fetus in this series is in the excess of that reported previously. The overall incidence of echogenic foci in this study (17.2%) overestimates the incidence in the general obstetric population, because the referral reason for many fetuses was the presence of echogenic foci on routine obstetric ultrasound scans. There was strong relationship between this ultrasound finding and aneuploidy, reported in the pathological study of fetal hearts [12].

In this series of isolated echogenic foci, two fetuses with chromosomal abnormalities was identified. A postnatal diagnosis of trisomy 21 was made in two fetuses that had a single echogenic focus in each ventricle. Ecogenic foci appear to be a marker of a number of karyotypic abnormalities rather than being specific to trisomy 21, which is also supported by the work of Sepulveda W and colleagues [15] and Twining P [16], who have described echogenic foci in association with trisomy 13, trisomy 18 and monosomy X. Moreover, this study also suggests that, in discriminating between genetically normal and abnormal fetuses, the number and location of echogenic foci are not helpful.

Echogenic foci are not of any significance as we did not observe functional cardiac abnormality like atrioventricular valve obstruction, outflow tract obstruction or impairment of cardiac function in any of our cases. The relationship of echogenic foci to structural cardiac abnormalitites has not yet been evaluated, but one fetus in this study had a small ventricular septal defect and there is a report of transposition of the great arteries in an affected fetus in another series [5].

Shipp TD et al., [17] reported 30.4% prevalence of EIF in Asian patients. There, however, were only 46 (489 total patients) patients in the Asian cohort of their study. Whereas, in another study, the incidence was reported as 7.3% in other low risk groups [18]. Study done by Rebarber A et al., reports 14.8% prevalence of EIF in patients of Japanese ancestry and also they concluded that, there is an increased prevalence of EIF in Asians of Japanese origin [19].

Nyberg DA et al., studied 186 fetuses with trisomy 21 and 8728 controls and found EIF as the most common marker (7.1%) [20].

LIMITATIONS

Limitations of the present study include a small sample size may not be truly representative of the general population. Further studies with larger sample size should be conducted in order to have clearer picture about echogenic cardiac foci.

CONCLUSION

In 83% of these cases, we found a single echogenic focus within the left ventricle, which allows us to conclude that this is the most frequent location inside the heart. In view of the incidence of karyotypic abnormalities in this study, the policy we have adopted is for detailed anomaly scanning of all fetuses with echogenic foci, regardless of their number or location. The option of karyotyping the fetus is discussed with parents, who are given 7% risk of genetic abnormality. The risk of a karyotype abnormality can now be more accurately weighed against the risks of invasive procedures, such as amniocentesis or fetal blood sampling, which are used to obtain such information.

We conclude that echogenic cardiac focus is most commonly seen soft marker in central Maharashtra during fetal ultrasound scanning, and the left ventricle being the most frequent location. Single echogenic cardiac nodule (87.3%) was more

common than multiple.

Therefore, we emphasize that single ecogenic cardiac focus is very common finding in this study. If it is not associated with other structural abnormalities, need to perform invasive genetic diagnosis may not be warranted.

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