Echogenic Intracardiac Foci
Ashish Jain*, Bijaylaxmi Behhra

Case Scenario
A 23-year-old primigravida underwent a level 2 antenatal ultrasonography (USG) screening at 22 weeks of gestation. The USG was suggestive of an echogenic focus in the left ventricle (Figure 1). A detailed evaluation did not reveal any other fetal anomaly.

Echogenic Intracardiac Focus
An echogenic intracardiac focus (EIF) is a small structure within the fetal heart with an echogenicity similar to or greater than that of the surrounding bone, apparent during an antenatal USG. It is sometimes referred to as a “golf ball.” An EIF is usually a normal variant, observed in 5% of all fetuses during the second trimester USG.1,2 Multiple foci have been noted in 6% to 11% of fetuses.3 Biventricular foci are more frequently associated with autosomal trisomies. Studies report the presence of the foci in the left ventricle in 87.6% of both aneuploid and euploid cases, in the right ventricle in about 0% to 25% of the cases, and in both ventricles in 1.5% to 7.6% of cases.4

Etiology
The possible/suggested etiologies include
- microcalcification of the papillary muscles and
- abnormal development of the microvasculature, involving terminal branches of the coronary artery leading to early ischemic changes in the papillary muscle.5
However, an EIF could also be a normal variant of the development of the atrioventricular apparatus, in which incomplete fenestration of the papillary muscle and chordae tendineae occur during fetal development.

**Significant associations**

An EIF has been associated with chromosomal abnormalities such as trisomies 21 and 13, particularly among fetuses of women of advanced maternal age or with abnormal triple screen results. A study reports that in a group of women at high risk of fetal chromosomal abnormalities, an EIF occurred in 18% of fetuses with Down syndrome compared with 4.9% of fetuses without Down syndrome. Some of the conditions known to be associated with an EIF include trisomies 21, 13, and 18; Turner syndrome; triploidy 11; and unbalanced translocations.

A meta-analysis published in 2013 concludes that in the second trimester, an EIF is seen in 21% to 28% of fetuses with Down syndrome compared with 3% to 5% of euploid fetuses. Another study concludes that an isolated EIF in a low-risk pregnancy does not confer an increased risk of fetal aneuploidy, while a recent study advises that detection of an EIF could imply an increase in the background risk of Down syndrome by 1.8-fold.

**Antenatal diagnosis and management**

An EIF does not show/produce acoustic shadowing on antenatal ultrasound. However, during real-time ultrasound, the focus is observed to move synchronously with the valve leaflets throughout the cardiac cycle. Usually a single focus measuring about 1 to 6 mm in diameter is present near the papillary muscle and chordae tendineae. Petrikovsky et al. diagnosed 41 fetuses to have ventricular echogenic foci among the 1139 fetuses they screened. They report that follow-up echocardiographic examinations revealed that 36% of foci increased in size over time, whereas 12% decreased and 51% remained unchanged. So far, no other cardiac anomalies have been reported as co-occurring with an EIF. Most of the studies report normal fetal echocardiography in fetuses with an isolated EIF; however, some studies report an association between the cardiac anomalies namely tricuspid regurgitation, ventricular septal defect (VSD), atrial septal defect (ASD), and pericardial effusion, and an EIF. So, fetal echocardiography is not recommended in case of an isolated EIF finding. But, if an EIF is associated with diffuse echogenic areas within the heart, it clearly represents distinct pathological and clinical entities and has poor prognosis and clinical implication.

Detection of a single echogenic focus or multiple echogenic foci should be followed by prompt and thorough fetal anatomic investigations for associated anomalies and their chromosomal markers. The investigation should include an examination of the standard 4-chambered view of the heart showing the outflow tracts. According to the currently available literature, the incidental detection of an EIF in an otherwise normal fetus probably represents a normal variant of papillary muscle development. It would be of clinical significance in pregnancies with risk factors such as increased maternal age and positive triple marker test and in pregnancies where other soft markers for aneuploidy are positive. Other soft markers include increased nuchal translucency, absent nasal bone, single umbilical artery, short femur, bilateral pyelectasis (dilatation of renal pelvis), widened lateral ventricles, choroid plexus cysts, and echogenic bowel. If any of these USG markers or fetal structural anomalies are found, the couple should be counseled regarding the increased risk of chromosomal anomalies in the child.
In young women who are not at risk of fetal aneuploidy, an amniocentesis is indicated when one or more of the aforementioned USG markers are detected, or when the triple screen results are abnormal.11

Conclusion

If an EIF is detected, a clinician should perform a full genetic sonogram, including the assessment for the other sonographic markers such as in a scoring system (nuchal fold, pyelectasis, and long bone length). The location and number of foci should be determined, and the results of the sonographic analysis should be correlated with biochemical screening results. In case of an isolated finding, no further USG examinations, including echocardiography, are required. At present, a combination of these sonographic markers, maternal age, and serum screening provide most of the information to help the clinician determine the need for invasive testing.

Recommendations and Practice Points

1. An EIF should be evaluated as part of the 4-chamber cardiac review during the 16th- to 20th-week USG.
2. An isolated EIF finding with a fetal aneuploidy risk < 1/600 by maternal age (31 y) or maternal serum screen requires no further investigations (neither follow-up USG nor postnatal echocardiography).
3. An isolated EIF finding with an aneuploidy risk > 1/600 by maternal age (31 y) or maternal serum screening should be followed by fetal karyotyping.
4. When right-sided, biventricular, multiple, particularly conspicuous, or nonisolated EIF findings are detected in antenatal screens, the couple should be referred for expert review and fetal karyotyping.

References


Author Affiliations

Dr Ashish Jain, Associate Professor, Department of Neonatology, Maulana Azad Medical College, 2, Bahadur Shah Jafar Marg, New Delhi 110002; Dr Bijaylaxmi Behhra, DM Resident, Department of Neonatology, Maulana Azad Medical College and Lok Nayak Hospital, New Delhi, India