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THE VALUE OF THE INTRACARDIAC ECHOGENIC FOCI IN THE FETAL HEART: CURRENT UNDERSTANDING AND CLINICAL VALUES

ZNAČAJ INTRAKARDIJALNIH EHOGENIH ŽARIŠTA FETALNOG SRCA: SADAŠNJE SHVAĆANJE I KLINIČKA VRIJEDNOST

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Original paper

Key words: fetal echocardiography, echogenic intracardiac foci, ultrasonography, prenatal diagnosis

Summary. ***Objective.*** To estimate the degree of risk of the echogenic intracardiac foci (IEF) for fetal chromosomopathies and to determine its association with structural anomalies of the fetus. ***Material and methods.*** During the period of two years 190 pregnant patients had been send for fetal echocardiography. Examination had been performed by transvaginal (12–17 weeks of gestation) or transabdominal approach (18 weeks or more of gestation). ***Results.*** IEF was observed in 17 fetuses, multifocal appearance was found in 2 out of 17 fetuses. In 3 cases IEF had resolved during the 8 weeks period of time. Additional structural anomalies were detected in 11 fetuses. In 2 fetuses trisomy 21 had been confirmed. ***Conclusion.*** A single soft marker as IEF is commonly encountered during the second trimester among the fetuses with chromosomal aberation. As do many sonographic markers IEF can be resolved during the pregnancy and often can be found in normal fetuses.

Izvorni rad

Ključne riječi: fetalna ehokardiografija, ehogena intrakardijalna žarišta, ultrazvuk, prenatalna dijagnostika

Sažetak. ***Cilj rada*** je bio na vlastitom uzorku utvrditi u kojoj mjeri ultrazvučni nalaz hiperehogenih intrakardijalnih žarišta (IEF) pridonosi dijagnostici kromosomopatija i strukturalnih anomalija. ***Uzorak i metode.*** Tijekom dvije godine 190 trudnica između 12. i 39. tjedna trudnoće je primljeno radi fetalne ehokardiografije. Pregled je obavljen vaginalnom sondom od 5 MHz pri trudnoćama 12.–17. tjedna ili zavinutom abdominalnom sondom od 3,5 MHz nakon 17. tjedna trudnoće. ***Rezultati.*** IEF su nađeni u 17 fetusa, multifokalni u 2 od njih. U 3 fetusa su IEF u roku od osam tjedana nestali. U 11 fetusa su nađene dodatne strukturalne anomalije. Trisomija 21 je potvrđena u 2 fetusa. ***Zaključak.*** IEF su »meki« ultrazvučni biljezi fetalne aneuploidije, često su prolazni, a nalaze se i u eukariotičnih fetusa.

Introduction

Advanced technology as well as increased capability of the ultrasonographers has expanded the use of the ultrasound as a diagnostic tool for the evaluation of the fetal heart. Ultrasound has enabled us to examine the life fetus in detail and with existing technology it is possible to detect fetal abnormalities including the heart anomalies at the end of the first trimester of pregnancy. Modern approach to prenatal ultrasound diagnostic should include basis of fetal echocardiography in every pregnant patients.

Therefore the fetal exhocardiography in last two decades has been established as a reliable non-invasive technique for the evaluation of the normal anatomy of the fetal heart as well for the abnormal anatomy finding. With the combination of the skilled stuff and modern technology it is possible to confirm most of the structural anomalies, and, through the use of the so called »soft« or »minor« markers, achieve limited success in identifying pregnancies at increased risk for aneuploidy.[1] One of these »minor« markers is hyper echoic intracardiac foci. These soft markers are more commonly seen among fetuses with normal karyogram, although they have higher frequency in fetuses with aneuploidy.[1] Unlike structural anomalies, sonographic markers are insignificant and non-specific most frequently seen in normal fetuses, and are often transient. The most common sonographic markers of fetal aneuploidy in the second trimester are nuchal thickening, short or absent nasal bone, shortened extremities, chorioid plexus cysts, hyperechoic bowel, renal pyelectasis and echogenic intracardiac foci.

In the basic cross sectional view *(Figure 1.)* of the four chamber view it is possible to detect small, either isolated or multifocal echogenic foci so called »goofballs« or either bright papillary muscle. These structures appear near the papillary muscles and chordae tendineae. They move in synchronicity with the intraventricular valves. Since the first description in 1987 by Schechter et al, the topic still remains controversial.[2] Despite many investigations of fetal intracardiac echogenic foci, the relationships between »goofballs« and congenital heart malformations and chromosomal abnormalities still remain unclear. They might represent micro calcification of the papillary muscles. Small calcifications in the papillary muscle, particularly in the left ventricle, are common in second trimester fetuses and are present in approximately 5–10% of normal fetuses.[3,4] Withlow et al. had found in their study that 0.7% of all fetuses do have echogenic

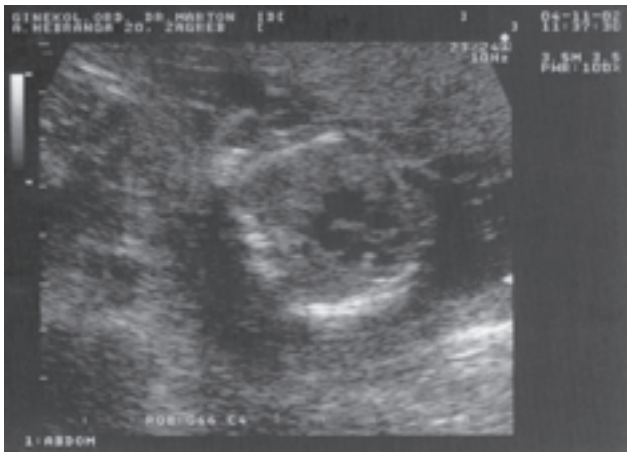

Figure 1. Apical four chamber view of the fetal heart showing the isolated echogenic focus in the left ventricle
Slika 1. Aplikalni presjek kroz fetalno srce s izoliranim hiperehogenim fokusom u lijevom ventrikulu

foci in the ventricle of the heart at the end of the first trimester, but by 18–20 weeks disappear in some fetuses but do develop in others, so that prevalence of echogenic foci is similar in the first (0.7%) and in second trimester (0.8%).[5]

Due to the substantial overlap between population of fetuses with trisomy 21 and the normal population of fetuses, these markers are not commonly part of the genetic sonogram but are used occasionally as an adjunct to other findings among the patients exposed to the high risk.[1]

Material and methods

During a 2 years period 190 pregnant women had been referred for fetal echocardiography. All ultrasound examinations had been performed on Aloka 1700 or Aloka 2000 SSD machine (Tokyo, Japan) by the transvaginal probe of 5 MHz or curved transducer with 3.5 MHz. Fetuses had been examined between 12 and 39 weeks of gestation. After the detailed structural evaluation of the fetus the fetal echocardiography had been performed either transvaginally (12–17th weeks) or by transabdominal approach (from 18th weeks on). The intracardiac echogenicity was identified from two different views: from four-chamber view and from the outlets of big vessels. The echogenicity of the »goofball« was comparable to the echogenicity of the bone. Numbers and the location of the intracardiac echogenic foci were recorded as well as additional structural anomalies if present. If the echogenic intracardiac focus was detected, detailed evaluation of the fetal anatomy was done to exclude other structural anomalies. The possibility of fetal karyotyping was discussed with parents if »minor« markers were detected as well additional structural anomalies.

Results

The intracardiac echogenic foci were detected in 17 fetuses. The mean gestational age at the time of the diagnosis was 21 weeks, ranging from 12 to 39 weeks. The mean maternal age was 36.5 years, with range 20–45. The majority of the echogenic foci were located in the left ventricle, while two isolated (single appearance) were found in the right ventricle. The multifocal echogenic »goofball« appearance had been found in two fetuses. In three fetuses echogenic foci during the 8 weeks period disappeared after the initial diagnosis had been done. Additional structural aberrations and anomalies were detected: chorioid plexus cyst, mild pyelectasis, obstructive uropathy, cardiac arrhythmia, agenesis of corpus callosum, renal bilateral agenesis, hypoplastic thorax, symmetric IUGR *(Table 1).* The findings of the intracardiac echogenic foci in combination with other sonographical markers, led us to the detection of two fetuses with chromosomal aberrations trisomy (21). The isolated presence of echogenic intracardiac foci was detected in 6 fetuses. Their biochemical tests were in normal range, so karyotyping had not been performed. In follow up these infants with normal phenotype were observed for two months and all were healthy with normal function of the heart.

Table 1. Additional structural anomalies detected in 17 fetuses with intracardiac echogenic foci
Tablica 1. Strukturalne anomalije u 17 fetusa s hiperehogenim intrakardijalnim žarištem

Structural anomalies Strukturne anomalije	Number of fetuses Broj plodova
Chorioid plexus	2
Pyelectasis	2
Obstructive uropathy	3
Cardiac arrhythmia	1
Agenesis of corpus callosum	1
Bilateral renal agenesis	1
Symmetric IUGR	1
Total – Ukupno	11

Discussion

Intracardiac echogenic foci are one of the most recent and probably the most controversial sonographic feature that has been described. Its appearance is common in fetuses during the second trimester, with reported incidence in the literature 1 out of 7 fetuses.[6,7] Like many sonographic markers, it typically resolves by the third trimester.[8] Description of the echogenic intracardiac foci is subjective, it depends on resolution of the ultrasound machine, sonographer's experience, fetal position, as exhogenic foci are best visualised when the apex of the heart is oriented toward the transducer.[9]

The significance of diffuse or right-sided echogenic foci in the fetal heart in contrast to isolated left-sided finding is not yet clear. Some investigators do suggest that right-sided or bilateral intracardiac echogenic foci had an approximately twice-greater risk of aneuploidy compared to the left-sided foci.[10] Wax and Philput reported that chromosomal aberration was more common

when echogenic intracardiac foci were diffused in both ventricles compared to isolated in one of the ventricle.[11]

Data from our study do support the benign explanation of the isolated intracardiac echogenic »goofball«. The majority (88.2%) of the fetuses had normal karyogram. According to the results from the literature and from our data, it seems to be a normal variant in the development of papillary muscles and chordae tendinae. We do share the opinion when the intracardiac foci are detected in a fetus, detailed information for the parents about the minimal increased risk for chromosomal abnormalities should be given.

The focus can be easily detected during the ultrasound evaluation of the fetal anatomy; thereafter detailed search for the conjoined structural anomalies or minor sonographical signs should be done. That includes sonographic »soft« signs for T 21 as nuchal thickening, iliac angle >90 degree, hyperechoic bowel, pyelectasis, sandal gap, sheered long bones, small ears, pericardial effusion, right-left disproportion of the heart, shortened or absent middle phalanx on the fifth digit, short or absent nasal bone. According to the data collected by Benaceraf et al. the intracardiac echogenic foci should be incorporated into sonographic index in screening program for fetuses with autosomal trisomies.[1] Approximately 12–15% of pregnant women have one or more sonographic markers identified during the routine scan at the second trimester, and it is unlikely that invasive procedures on all of these are indicated.

The risk of intracardiac echogenic foci and other soft markers are probably overestimated in studies in which the fetal karyotype is known for all patients, because sonographic findings influence patients decision making. Diffuse or large echogenic foci may have an important variable to carry a greater risk when considering amniocentesis.

The use of sonographic markers to modify the risk for T 21 is widely referred to as *»genetic sonogram«*.[6] The actual sensitivity of a genetical sonogram will depend on various factors as gestational age, markers sought, the quality of the sonographer and ultrasound machine, and indications of referral.[12] With the higher number of sonographic markers present, the risk of fetal trisomy 21 increases dramatically. Two or more markers are detected in nearly one third of fetuses with T 21 compared with less of 2% in normal fetuses.[6] In comparison, a single marker is observed in more than 11% of normal fetuses compared with 22.6% of fetuses with T 21.[6]

Structural findings also recognised, as soft sonographic markers of fetal aneuploidy, are non-specific, frequently seen in eukaryotic fetuses and often are transient. Therefore, more attention and time should be put on the education of sonographers, equipment and technique to improve the sensitivity of ultrasonography for detection of fetuses with abnormal karyotype.

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