

● Original paper

THE FOLLOW UP OF 114 FETUSES AND NEWBORNS (WITHOUT CHROMOSOMAL ABERRATIONS) WITH ECHOGENIC INTRACARDIAC FOCUS DETECTED IN PRENATAL USG



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Abstract

Introduction: The majority of research regarding echogenic intracardiac focus (EIF) concentrates on its weak correlation with the occurrence of Down syndrome. The aim of our research was to approach this problem from a wider perspective and to find out, if the prenatal diagnosis of EIF is connected with the occurrence of other abnormalities of prenatal and postnatal period.

Materials & Methods: The data of 114 patients with prenatally diagnosed EIF were analyzed retrospectively. No fetal or neonatal chromosomal abnormalities were included.

Results: In 13/114 (11,4%) fetuses cardiological abnormalities other than EIF were diagnosed: 8/114 (7%) cases of congenital heart defects and 7/114 (6,1%) cases of tricuspid valve regurgitation. Extracardiac malformations were diagnosed in 11/114 (8,8%) of fetuses. In 7/114 (6,1%) of the cases the abnormal volume of amniotic fluid was diagnosed. In 4/114 (3,5%) of pregnancies the premature rupture of membranes (PROM) occurred. Six, 6/114 (5,3%) of pregnancies were at risk of intrauterine asphyxia in perinatal period. 12/114 (10,5%) newborns were delivered before 37th week of gestation, stillbirth occurred in 1/114 (0,9%) case. Most newborns (86/114; 75,4%) birth weight >3000g. In 19/114 (16,7%) of newborns birth weight was 2500g-3000g. In 9/114 (7,9%) of newborns birth weight was <2500g

Conclusions: Fetuses with EIF without chromosomal aberrations may present heart defects which are hard to diagnose in basic obstetrical USG scan. Therefore, those patients should be directed to prenatal cardiology facilities for evaluation of the fetal heart.

Prenatal EIF in fetuses without chromosomal aberrations may indicate low birth weight (<2500g) in the future. Further research of this matter is needed.

Key words: echogenic intracardiac focus; EIF; Bright Spot; fetal echocardiography; prenatal cardiology; prenatal diagnostic; ultrasonography

INTRODUCTION

The clinical significance of detecting the echogenic intracardiac focus (EIF) in prenatal USG scan is not clear. Most of the studies so far investigated its correlation with Down syndrome. However, only a few publications describe its connection with other abnormalities occurring in fetuses and newborns. The aim of this research was to evaluate the medical condition of fetuses and newborns with EIF detected in prenatal USG examination, excluding the cases with chromosomal aberrations.

MATERIALS

The studied material consisted of the medical documentation of 147 patients of the Department of Prenatal Cardiology of Polish Mother's Memorial Hospital, who were diagnosed with echogenic intracardiac focus in the years 2000-2013 (photo 1). 32 patients without postnatal follow up and 1 case of triplet pregnancy were excluded. Fetuses and newborns with chromosomal aberrations were not included in the studied group. The data of the remaining 114 patients were retrospectively analyzed in search for the following abnormalities:

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Fetal period:

- normal/abnormal anatomy and function of fetal heart
- extracardiac malformations
- volume of amniotic fluid

Postnatal period (newborns):

- preterm birth (<37 hbd)
- birth weight <3000g and <2500g
- abnormalities in perinatal period
- signs and symptoms of infection (gastroenterocolitis and congenital pneumonia)

RESULTS

The mean age of pregnant patients was 29,9 years (SD 5,7).

The mean fetal age at time of diagnosis in the reference medical facility was:

- according to the last menstrual period: 25,7 weeks (SD 4,4)
- according to biometric measurement : 26,1 weeks (SD 4,6)

In 13/114 (11,4%) fetuses cardiac defects other than EIF were detected. In 8/114 (7%) cases the USG scan performed in the fetal cardiology department revealed: stenosis of the aortic valve (which evolved form mild to critical defect during the course of pregnancy); tetralogy of Fallot; ventricular septum defect; 2 cases of disproportion of heart ventricles; pericardial effusion; heart muscle hypertrophy; heart muscle hypertrophy with pericardial effusion (table 1). Moreover, in 7/114 (6,1%) fetuses functional regurgitation of tricuspid valve was detected.

Extracardiac abnormalities were detected in 11/114 (8,8%) fetuses: 3 cases of pyelectasis; hydrothorax; hyperechogenic bowel; increased thickness of the nuchal fold; multiple defects: esophageal atresia, hypertelorism and pyelectasis; multiple defects: duodenal atresia and annular pancreas; lymphoid cyst in thoracic cavity and cyst (probably bronchogenic) in mediastinum (table 2).

In 6,1% of cases abnormal volume of amniotic fluid was detected (2 x anhydramnion, 3 x oligohydramnion and 2 x polihydramnion).

The analysis of the perinatal follow up revealed, that premature rupture of membranes (PROM) occurred in 4/114 (3,5%) pregnancies. In 6/114 (5,3%) of cases the risk of intrauterine fetal demise was present.

Miscarriage occurred in 1/114 (0,9%) of the pregnancies. The remaining 113/114 (99,1%) pregnancies ended with live birth.

Majority of newborns (102/114; 89,5%) were delivered on or after 37th week of pregnancy, 12/114 of newborns (10,5%) were born before 37th week (chart 1.) .Majority of newborns (86/ 114) had a birth weight > 3000g. In 19/114 (16,7%) newborns birth weight was 2500g- 3000g. In 9/114 (7,9%) newborns birth weight was <2500g (chart 2.) .

Clinical signs and symptoms of infection (gastroenterocolitis and/or congenital pneumonia) were observed in 2/114 (1,8%) newborns.

Photo 1. 4 chamber view with bilater EIF at 19th week of gestation; at 26th week of gestation mild aortic stenosis; at 36week - critical aortis stenosis; baloon valvuloplasty after delivery with good results

DISCUSSION

Echogenic intracardiac focus is an ultrasound finding of density close to the bone tissue. It visualizes calcification



Photo 1. 4 chamber view with bilater EIF at 19th week of gestation; at 26th week of gestation mild aortic stenosis; at 36week - critical aortis stenosis; baloon valvuloplasty after delivery with good result



Video file can be found here: <http://echoplodu.fetalecho.pl/en/articles/show/the-follow-up-of-114-fetuses-and-newborns-without-chromosomal-aberrations-with-echogenic-intracardiac-focus-detected-in-prenatal-usg,224.html>

Nr	Gestational age (based on LMP and on biometry)	Cardiac diagnosis
1	24 / 24	Benign aortic stenosis at 26 wks and critical at 36 wks
2	28,4 / 23,6	Tetralogy of Fallot
3	31,6 / 29,3	Ventricular Septal Defect
4	31,4 / 30,4	Disproportion
5	26,3/ 26,3	Disproportion
6	25 / 25,6	Pericardial effusion
7	27,4 / 27,3	Myocardial hypertrophy, pericardial effusion
8	29,1 / 28,1	Myocardial hypertrophy
	Mean 27,9 (SD 2,8)/ 27 (SD 4,6)	

Table 1. Structural and functional cardiac anomalies in analysed group of fetuses with EIF (bright spot)

in heart papillary muscle. The most common localization of EIF is the left ventricle. In the majority of cases it is singular, however multiple EIF have also been described^{1,2}. In normal pregnancies it occurs with the frequency of 4-5%, significantly more often in the Asian population^{2,3}. In USG scan EIF may be mistaken with the ultrasound signal reflecting from the septomarginal trabecula, ventricular endocardium or annulus of tricuspid valve⁴. EIF may be detected in the 2nd trimester routine obstetrical ultrasound examination or in fetal echocardiography. Prenatally detected EIF is considered to be a soft marker

of Down syndrome and indicates the need for very careful evaluation of fetal morphology in search for other markers of aneuploidy. If other ultrasound markers are absent and results of mother's serum analysis are within normal range, the relative risk of Down syndrome is low. According to this, prenatal detection of isolated EIF is not an indication for evaluating fetal karyotype and amniocentesis^{5,6,7,8}. Some researchers claim, that EIF does not correlate with increased risk of Down syndrome at all and informing mothers about this finding is a source of unnecessary

anxiety^{9,10,11,12}. However, it is worth noticing, that almost all the research concerning EIF focuses on its correlation with Down syndrome and only a few publications describe its coincidence with other abnormalities occurring in fetuses and newborns. Considering this, our study is innovative and based on the largest study group both in Polish and English literature.

Frequency of congenital heart defects (CHD) is estimated at 1% (according to data from the Poznan Polish Registry of Congenital Malformations in the years 2005-2006:

0.71%)¹³. In the studied group the frequency of CHD was seven times greater (7%). The problem of increased risk of CHD development in fetuses with EIF was previously mentioned only once by A. Carriço et al.¹⁴. The alarming finding in our material is the fact, that basic heart evaluation performed during obstetrical USG scan did not detect any of the 8 cases of CHD accompanying the EIF. In our facility one of the fetuses with EIF was diagnosed with "mild stenosis of aortic valve" with V max up to 120 cm/sec, which in a few weeks presented increased flow gradient through the aortic valve (up to 3,5 m/sec). The newborn needed percutaneous balloon valvuloplasty of the aortic valve immediately after birth (with satisfying effect).

According to data from Polish Central Statistical Office in 2011 the frequency of preterm birth in Poland was 6,7%. It was 10,5% in the studied group. Those differences are close to statistical significance (Chi-square test: p= 0,1).

The low birth weight (<2500g, including only singleton pregnancies) occurred in 4,6% of Polish newborns in 2011. In the

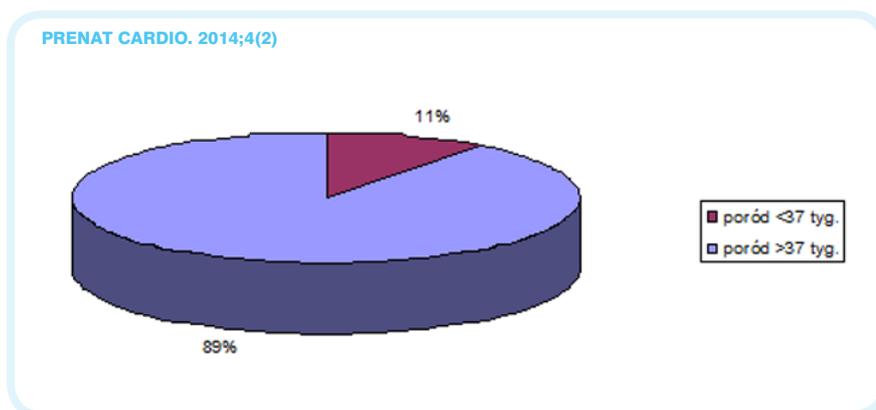


Fig. 1. Time of delivery (red < 37th week of gestation, blue > 37th week of gestation)

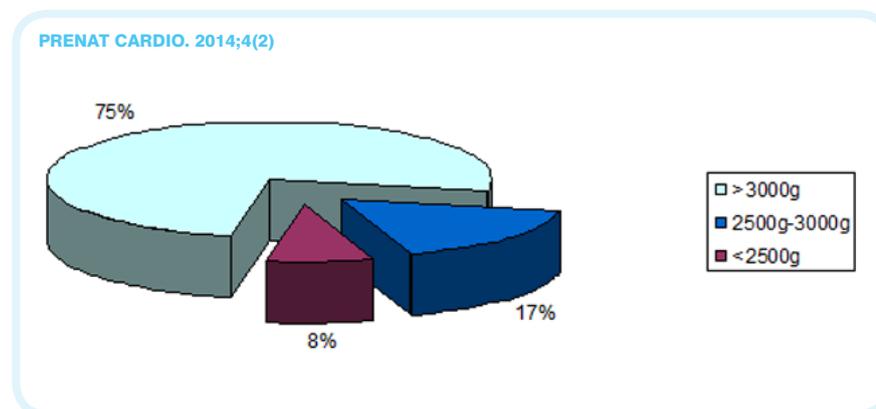


Fig. 2. Birth weight of newborns with EIF

Nr	Gestational age based on LMP / biometry	Prenatal diagnosis
1	27 / 28	<i>Pyelectasis</i>
2	21,4 / 21,2	<i>Pyelectasis</i>
3	35,2/ 36,4	<i>Pyelectasis</i>
4	25,2 / 25,6	<i>Hydrothorax (unilateral)</i>
5	20,5 / 18,6	<i>Hyperechogenic bowel</i>
6	33,3 / 34,1	<i>Increased thickness of nuchal fold</i>
7	27,4 / brak danych	<i>Multiple anomalies: esophageal atresia, hipotelorysm, pyelectasis</i>
8	24,3 / 24,4	<i>Duodenal atresia (anular pancreas)</i>
9	30,3 / 31,5	<i>Chest lymphatic cyst</i>
10	36,6 / 35,6	<i>Bronchogenic cyst in mediastinum</i>
	Mean 28,1 (SD 5,6)/ 28,4 (SD 6,4)	

Table 1. Extracardiac anomalies in fetuses with EIF (brigh spot)

studied group it was 7,5%. The differences were close to statistical significance (Chi-square test: $p = 0,09$).

The important finding in the studied group is the large number of cases, in which at least one of the analyzed abnormalities was present (31,6%-43,9%). If the 3000g was the lower limit of normal birth weight, than in 50/114 (43,9%) of cases at least one abnormality was observed. For the 2500g as the lower limit of normal birth weight, at least one abnormality occurred in 36/114 (31,6%) of the cases.

The previously mentioned findings may indicate, that prenatal detection of echogenic intracardiac focus is not without meaning and may be significant for the wellbeing of fetuses and newborns.

However, we must be aware of the fact, that the studied group was composed of the patients of the Department of Prenatal Cardiology in Polish Mother's Memorial Hospital. Patients are directed to this facility due to increased risk of fetal defects and/or abnormal findings in routine screening ultrasound examinations.

According to that, they are not a random sample of the general population, which may put to question the comparison of received results to epidemiological data. Therefore, the statistical analysis used in our study is a guide only. Creating a control group from the patients of our facility would be very difficult or even impossible.

Some studies negate the coincidence of EIF with pre and postnatal abnormalities^{15,16,17}. However, most of them focused on searching for correlation with genetic syndromes and were based on smaller groups of patients.

The results of our research seem to point out two new aspects, which were not described in literature before: the possibility of developing congenital heart defect in fetuses

with EIF during the course of gestation and a risk of low birth weight in this group. Does the EIF indicate fetal infection? We leave this question for future research to answer.

CONCLUSION

Fetuses with EIF without chromosomal aberrations may present heart defects which are hard to diagnose in basic obstetrical USG scan. Therefore, those patients should be directed to prenatal cardiology facilities for evaluation of the fetal heart.

Prenatal EIF in fetuses without chromosomal aberrations may indicate low birth weight (<2500g) in the future. Further research of this matter is needed.

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Authors and the division of work:

M. Respondek-Liberska: the idea of the research, collecting data, final version of the article, choice of photos for the article

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