ON OUTCOME OF FIVE CONSECUTIVE PREGNANCIES WITH HEART DEFECTS DETECTED IN 1ST TRIMESTER OF PREGNANCY

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Abstract

The aim of this study was an analysis of cardiac assessment in the first trimester, their outcomes, and comparison with literature data. Five cases were analysed from the year 2014. The exams were performed between 11-13 + 6 weeks. The analysis confirmed that the detection of CHD in 1 st trimester was possible and was verified by an early fetal echocardiogram. The most common symptoms of abnormal heart images in the early screening assessment were: axis, heart size and color Doppler assessment of the chambers and V sign. The outcome of pregnancies with early detection of fetal heart defects was poor. There was only one surviver.

Key words: *pulmonary valve atresia, aortic valve stenosis, fibroelastosis, 1st trimester*

The outcome of five consecutive pregnancies with heart defects detected in 1st trimester of pregnancy.

Every patient, regardless of age should have an ultrasound between 11 and 14 weeks of gestation. Screening for aneuploidy based on the measurement of NT was introduced 20 years ago[1]. The high resolution probe allows a detailed assessment of fetal anatomy, and this has caused an increase in the number of diagnoses of major defects in the first trimester of pregnancy. According to literature data, the detection of large defects in the fetus between 11-14 weeks is approx. 50% compared to 70-90% in the second trimester of pregnancy[2,3]. The incidence of heart defects was estimated at 8 / 1000 live births, but the publications analyzing autopsy studies show a significantly higher incidence of defects in fetuses up to 16% [4, 5]. The heart should also be evaluated in the cases with an increased nuchal translucency, tricuspid valve regurgitation or abnormal flow in the ductus venosus or other pathology of the fetus. The detection of fetal malformation at early stage of pregnancy enables an appropriate further diagnostic process. Performing amniocenthesis, genetic counseling and early echocardiography allows for the complete diagnosis at an early stage and lets parents make decisions regarding the pregnancy. The best time for the fetal exam is the gestational age between 18 and 22 weeks and until now most defects are still diagnosed at this time. However, many of these pathologies can be detected much earlier.

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THE AIM

The aim of this study was an analysis of cardiac assessment in the first trimester, their outcomes, and comparison with literature data.

MATERIALS AND METHODS

5 cases from the database medical center Volumed were analysed from the year 2014. Four patients were from the low-risk group and wanted to undergo prenatal screening because the exam was recommended by their obstetrician. One patient was referred for the Prenatal Screening Program because of her age.

The exams were performed between 11-13 + 6 weeks. CRL, BPD, FL and fetal heart rates were measured. The genetic markers (NT, NB, TR, DV) and fetal anatomy were assessed according to the recommendations of the Ultrasonographic Section of the Polish Gynecological Society (Table 1), the FMF and ISUOG [6, 7, 8].

In addition, cardiac evaluation was extended to four chamber view (4CHV) and three vessel view (3VV) in color

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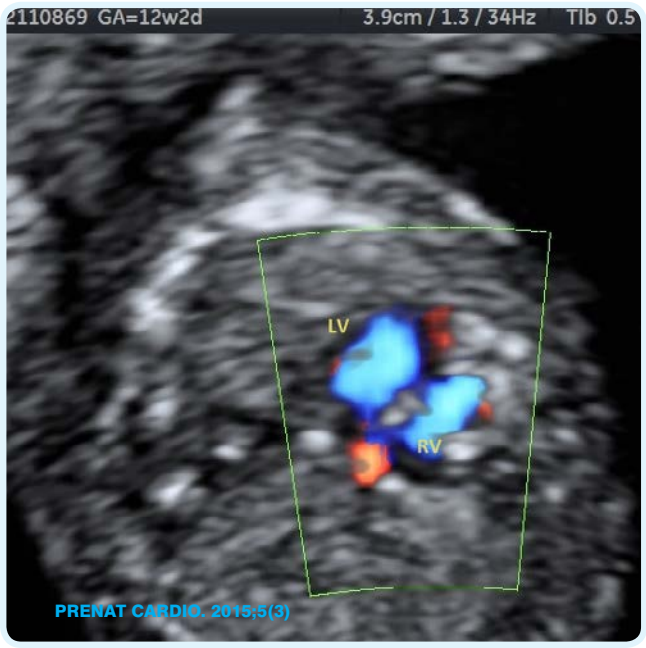
	Case 1	Case 2	Case 3	Case 4	Case 5
Medical history	Age 28 G I Low-risk group	Age 31 G I Low-risk group	Age 41 G II P II High-risk group (maternal age)	Age 33 G III PII Low-risk group	Age 34 G II PII Low-risk group
Weeks of gestation	12	12	12+4	13+1	11+6
NT(mm)	3,4	7,4	2,7 (>95%)	Normal	Normal
NB	+	Hypoplastic	Hypoplastic	+	+
TR	-	+	-	-	-
DV	PI=1,53	Reverse flow	Not assessed	Reverse flow	PI=0,9
4CH	RV<LV	LV>RV Cardiomegaly	Not assessed	Cardiomegaly	One chamber
3VV	One large vessel	Two-direction flow	Not assessed	Not assessed	One vessel
Fetal anatomy	Normal	Omphalocele Fetal oedema	Omphalocele Abnormal hand position, 2 vessels in the umbilical cord	Normal	Normal
Risk of aneuploidy	T21 1:78 T18 1:353 T13 1:336	T21 1:5 T18 1:7 T13 1:34	T21 1:9 T18 1:4 T13 1:4	T21 1:880 T18 1:1530 T13 1:2870	T21 1:2148 T18 1:2572 T13 1:3418
Genetics consultation	No (patients decision)	Yes	Yes	No (patients decision)	No (patients decision)
Amniocenthesis	No	Yes (Tris 18)	Yes (Tris 18)	No	No
Fetal echocardiography (weeks of gestation/ recognition)	14 Atresia pulmonary valve VSD Fetal edema	16 Stenosis aortic valve Hypoplasia of aortic arch Fibroelastosis of LV	15 DORV VSD	16 TOF with absent pulmonary valve	16 Complex CHD
Fetal outcome	IUD (17 wks)	IUD (31 wks)	Termination of pregnancy	Intrauterine Digoxin treatment Polyhydramion Amnioreduction	Complex CHD Normal course of pregnancy
Newborn outcome				Preterm labor (35w.g), death at 4 months	Delivery at term (40w.g. - 3950g)

Table 1. Medical data of the 5 consecutive pregnancies

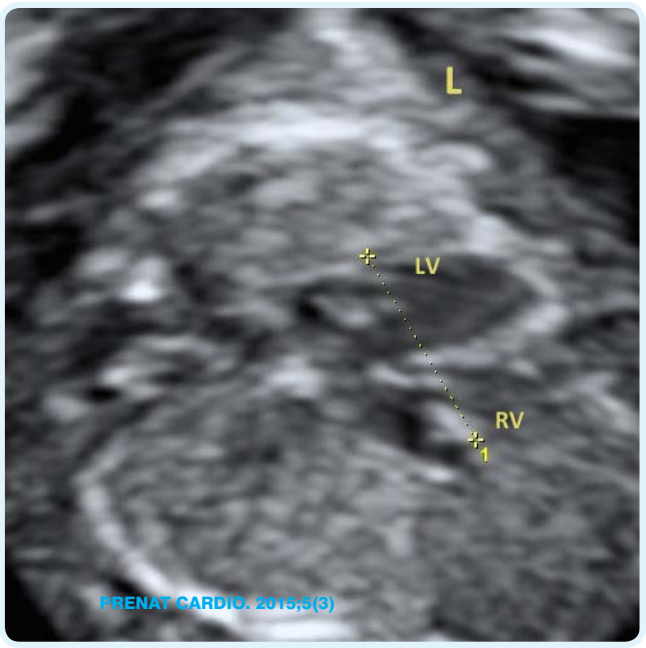
Doppler technique. After receiving the Pappa test results, risk calculation for the three most common aneuploidies was assessed. Patients received further recommendations to perform genetic counseling and early echocardiography. The medical data are presented in Table 1.

DISCUSSION

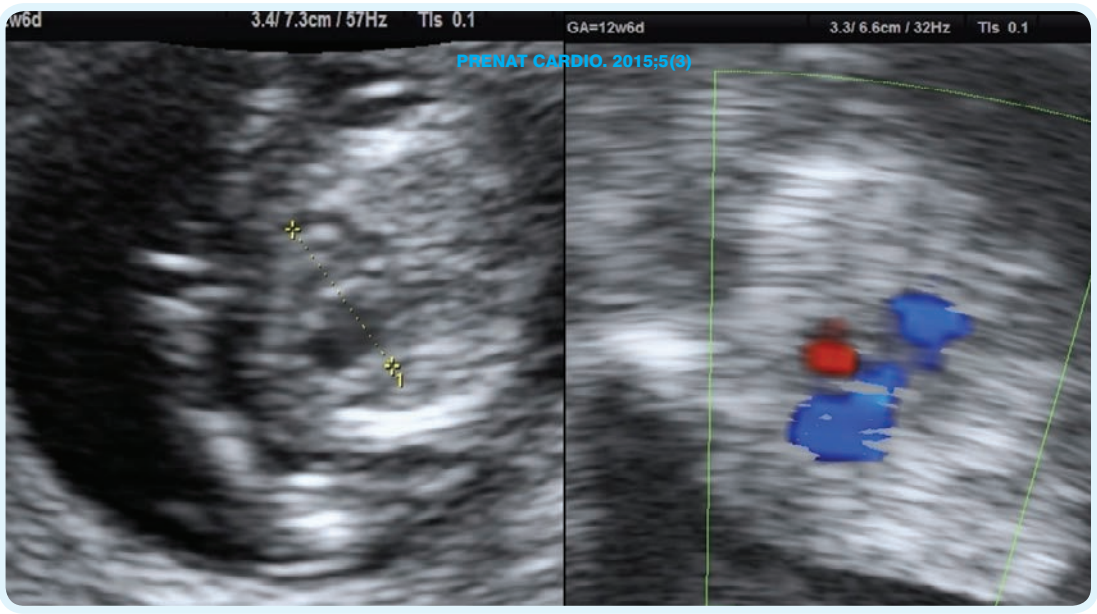
The obstetrician performing prenatal ultrasound exam in first trimester of pregnancy assesses the nuchal translucency (NT), nasal bone (NB), tricuspid valve flow, ductus venosus flow and fetal anatomy. In the case of normal karyotype, increased NT can be related to extracardiac malformations or congenital heart disease. In Moczulska et al.'s study, the NT > 3mm was associated with extracardiac fetal malformation or congenital heart disease (CHD) in 54% and 29,8%, respectively. That is why currently it is thought that an increased NT is a good marker not only for aneuploidy but also for structural anomalies of the fetus in low risk groups[9]. However, often the fetus with CHD had normal NT and only a heart assessment may allow to suspect any defects[10, 11]. According to Reddy et al. about 36% of fetuses with extracardiac anomalies had CHD, and according to Tennstedt et al., on the basis of autopsy, the frequency was 66%. That is why we should pay attention to the heart already when performing the genetic I trimester exam. The assessment of the four-chamber view can be performed already at 8 wks in 50% of fetuses. However, later at 11 wks, it is possible in 98% of fetuses. According to Hutchinson et al., the outflow track is visible in 95% of fetuses between 11 – 13+6 wks [12] Ushakov is convinced that the heart screening assessment in this period is quite easy [13]. It is



Case 1. Ventricular septal defect and smaller right ventricle in color Doppler imaging (B). RV-right ventricle, LV-left ventricle.



Case 1. Right ventricle smaller then left (A). RV-right ventricle, LV-left ventricle.

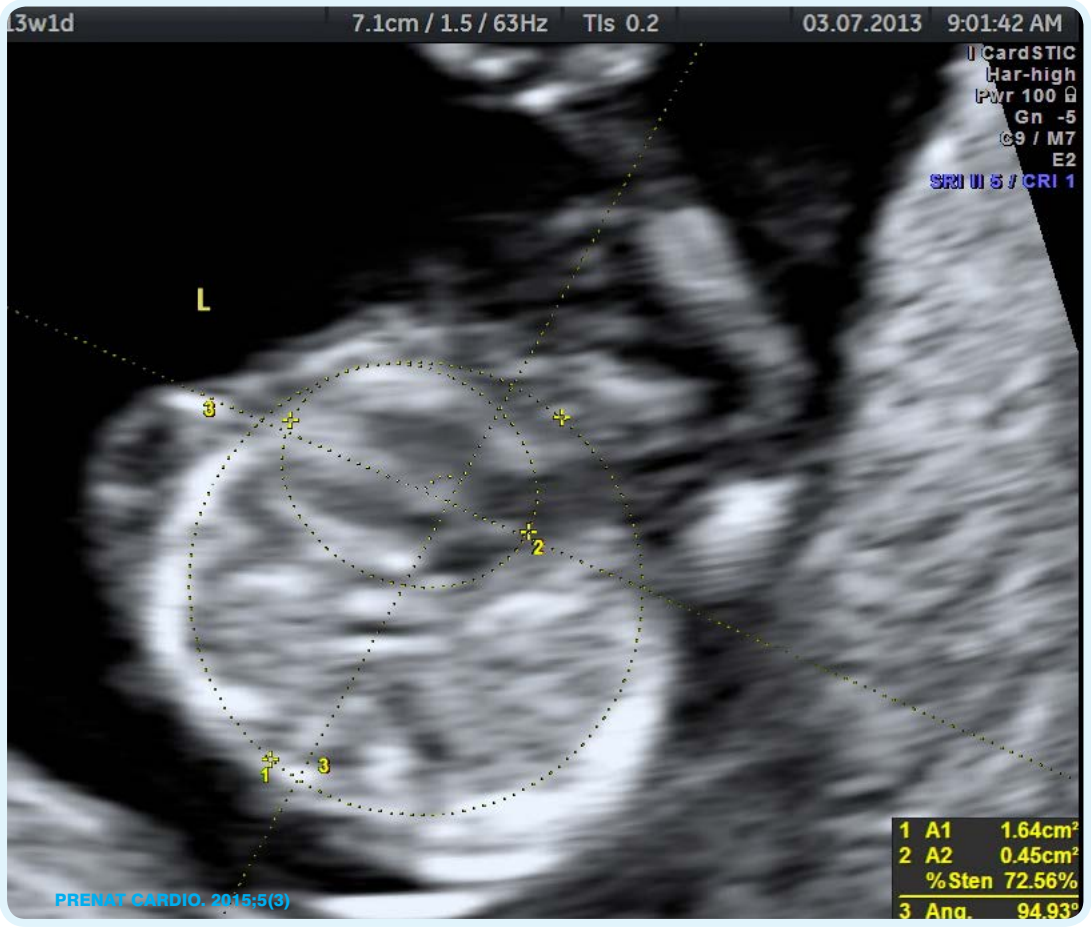


Case 2. Dilated left ventricle shown in the four- chamber view (A). Retrograde flow across the aortic isthmus shown in the three- vessel –trachea view (V-sign) (B).

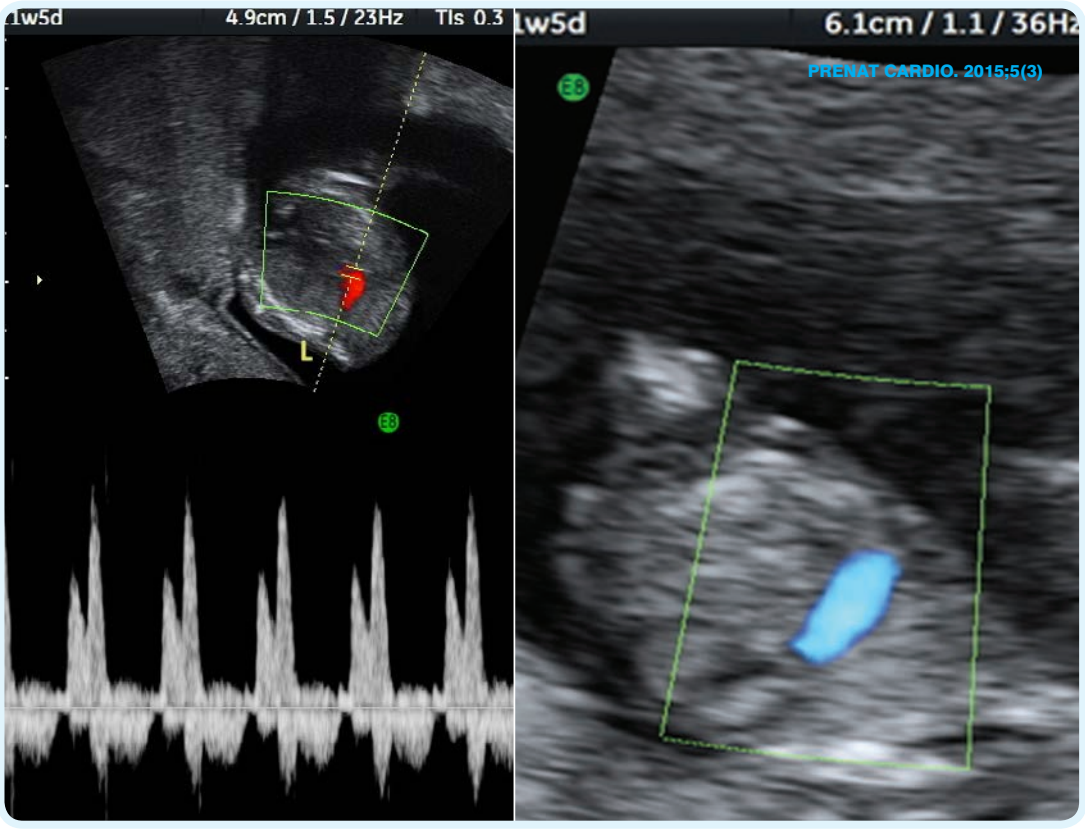
enough to use the scan to evaluate NT, move the probe above the chest of the fetus and rotate it about 90 deg. This maneuver enables the image of 4-chamber view and 3-vessel view (V-sign) in color Doppler imaging. Additionally, it is worth assessing the size and axis of the heart. An exam performed in this way enables the visibility of the main cardiac defects and referring the patient for early fetal echocardiography (EFE), which is performed between 12-16 wks.[12]. Other indications for EFE are: an increased NT and/or other genetic markers, extracardiac malformations and previous child with CHD.

The presented cases are an example of different abnormal heart imaging both in fetuses with normal and abnormal NT. In the first case no other extracardiac malformations were observed, the patient declined amniocenthesis. The EFE was performed at 14 wks and it confirmed CHD and also fetal edema, which was caused by the CHD and heart insufficiency. In the second and third case, the omphalocele was easy to recognize. In the second case, at 12 wks, an abnormal heart scan was visible, which was later confirmed in the EFE at 16 wks. In the third case, the heart assessment was not possible but EFE revealed CHD at 15 wks. In the 4th and 5th case, NT was normal and only earlier heart screening revealed heart malformation. After receiving the results of the karyotype at about 18wks, the patients had a complete medical work-up (genetic, prenatal cardiac and obstetric counseling).

The presented cases were not high-risk pregnancies, except for the age of one patient. According to Perii (2005), 89% of CHD occur in mothers from a low risk group[14]. The concomitance of heart defects and extracardiac malformations has an especially poor outcome.



Case 4. Abnormal cardiac axis-95 degrees, Ha/Ca=0,27. L-left side



Case 5. Color and pulsed Doppler across only one atrio-ventricular valve (A) and flow in only one vessel in the three- vessel –trachea view (B).

Consequently, this should be taken into consideration in prenatal counselling. Live births in this group were 35-46%, from which half died after birth in the neonatal period[15,16]. In the case of omphalocele and CHD, morbidity is even up to 100%[17,18], similar to the presented cases.

CONCLUSIONS

The detection of CHD in 1 st trimester was possible and was verified by an early fetal echocardiogram.

The most common symptoms of abnormal heart images in the early screening assessment were: axis, heart size and color Doppler assessment of the chambers and V sign.

The outcome of pregnancies with early detection of fetal heart defects was poor.

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Nawara-Baran A: concept of research, data collection and analysis, references, first draft

Radzyminska-Chrusciel B: work with manuscript, final version