Case **Report**

THE 1ST AND 2ND TRIMESTER SCAN OF THE FETAL HEART IN THE CASE OF INTERRUPTED AORTIC ARCH - CASE REPORT

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Abstract

Interrupted aortic arch is a rare and very difficult to diagnose congenital heart defect, which is divided into three types according to the place of interruption. The most common is type B, where the interruption is between the left carotid artery and left subclavian artery. This case report shows the description of the anomaly during the first and second trimester scan and review of the literature regarding interrupted aortic arch.

Key words: *fetus, ultrasound, interrupted aortic arch*

INTRODUCTION

An interrupted aortic arch (IAA) is a rare, accounting in about 1% of cases congenital heart defects but very serious anomaly, in which in the neonatal period a part of circulation depends on ductus arteriosus.

This anomaly is characterized by complete discontinuity between two adjacent segments of the aortic arch.

According to the classification proposed in 50[ties] of XX century there are three morphological types depended on the place of interruption – type A, B and C. In type A the interruption is distal to the left subclavian artery and it is present in 30-43% of cases. In the most frequent occurring type B (53-65% of cases) the interruption is between the left carotid and left subclavian arteries; and in type C, interruption is between the innominate artery and the left carotid artery. Type C is the least common, accounting for 4-5% of cases[1-6]. This case report shows description of the anomaly during the first and second trimester scan and review of the literature.

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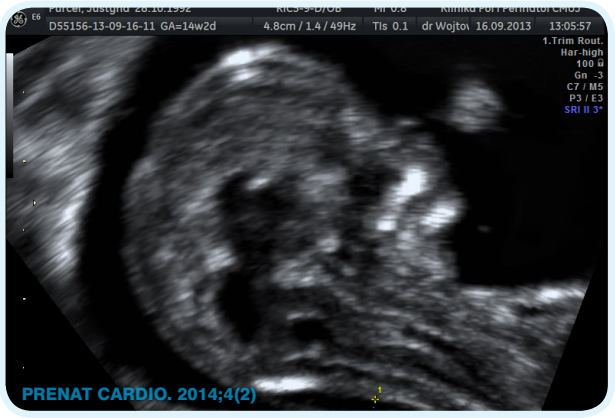

Photo 1. Fetal profile in the 1st trimester scan and nuchal translucency

CASE

In 21-year old primigravida at 14+2 weeks of gestation (according to last menstrual period) ultrasound scan showed the biometry of fetus as at 12+2 weeks, what was consistent with earlier measurements and history of irregular menses. Nuchal translucency was 2.1mm (fot.1), there was hypoplastic nasal bone, tricuspid regurgitation and radial aplasia on the left side as well as single umbilical artery was present. Heart anatomy was taken in to consideration. The four chambers view was abnormal with smaller left ventricle and left atrium than right ventricle and right atrium (fot.2). Color Doppler showed in lateral position two ventricular septal defects (VSD) and in three vessels view in the upper mediastinum V sign with blood flow from the heart but with smaller ascending aorta than pulmonary artery (fot.3). PAPP-A test showed that the patient was in high risk of aneuploidy. The patient was offered amniocentesis, but the karyotype was normal. The

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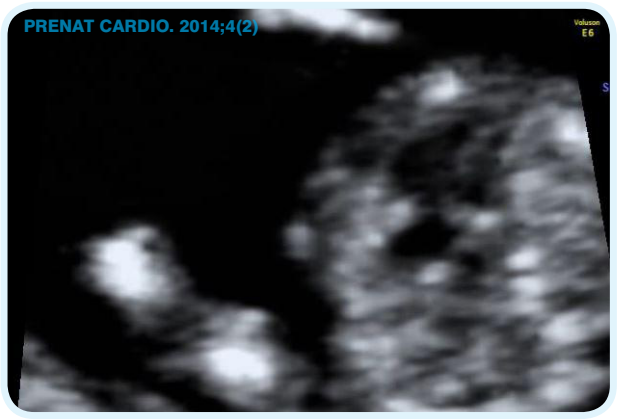

Photo 2. Four chambers view in the 1st trimester

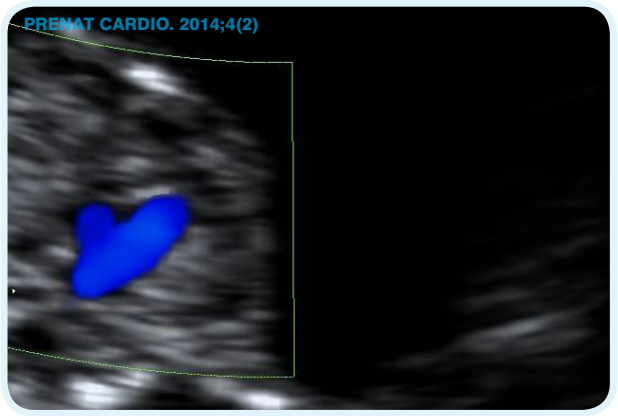

Photo 3. Three vessels view in the 1st trimester

second scan at 21 weeks of gestation confirmed single umbilical artery, radial aplasia on the left and abnormal view of the heart. The fetal growth was measured as at 20 weeks of gestation. In the four chambers view there was still discrepancy of the chambers with the domination of the right ventricle (fot.4). Atrioventricular connections were concordant (MV=2,5mm Zscore -7,16; TV=6,4mm Zscore 0,63). There was trivial tricuspid regurgitation. Posterior ventricular septal defect was found. The valve of the foramen ovale was in the left atrium as well as inferior pulmonary veins drained to the left atrium. Ventriculoarterial connections were concordant, but narrow left outflow tract paid attention (LVOT 2,6mm Zscore -1,43) with normal right ventricular outflow tract (RVOT 4,6mm Zscore 2,08). The position of the three vessels in the upper mediastinum was normal, but the ascending aorta smaller (2,2mm Zscore -3,73) with blood flow from the LV (fot.5). In this scan as well as in the sagittal view aortic arch was not able to obtain. Ascending aorta went straight towards the head with carotid branches giving Y sign (fot.6). In the sagittal view the blood flow in the distal part of the aortic arch was from ductus arteriosus. The thymus was absent. Nuchal fold was normal (3,29mm) but profile of the fetus with shorter mandibula and nasal bone (4,5mm) paid attention. The diagnosis of interruption of the aortic arch and microdeletion 22q11.2 was made. FISH was proposed, but the patient did not give consent to it. At the time of collecting data to this case report it was 36 weeks of gestation and the fetus biometry was about 3 weeks smaller .

DISCUSSION

Imaging of the aortic arch in the fetus could be challenging for the sonographers and these days in our country is not included in the guidelines of the Polish Society of Gynecology for screening fetal anomaly scan [1]. Prenatal detection of IAA has been reported only a few times and usually as case reports. Even in the most experienced tertiary care centers detection rate is among 11 and 43% [7]. However, the rate of prenatal diagnosis is increasing, what is associated with improved image quality, growing experience of the sonographers and increased awareness of the cardiac defects. In our case abnormal four chambers view and three vessels view was detect at 12 weeks of gestation, but the precise diagnosis was not able at this stage. The diagnosis of VSD and asymmetry of the great arteries was made, what could indicate coarctation or interruption of the aorta. In PubMed there is only one case series including one case of the diagnosis of IAA made in the 1st trimester based on STIC technique [8]. In our case the diagnosis was made in 2nd trimester as in case series of Volpe et al. [5], where the mean time of detection was 24 weeks.

The diagnosis of IAA is difficult, because we can see

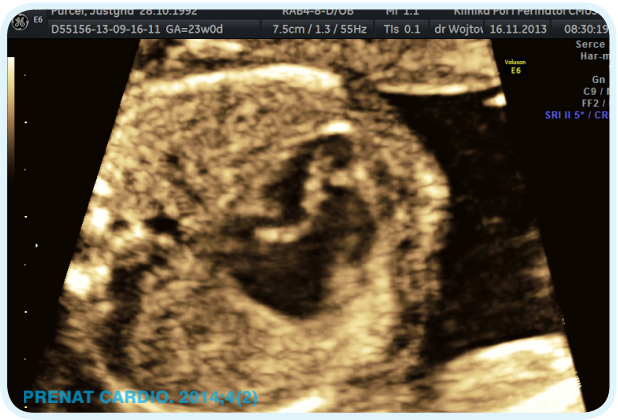

Photo 4. Four chambers view in the 2nd trimester scan

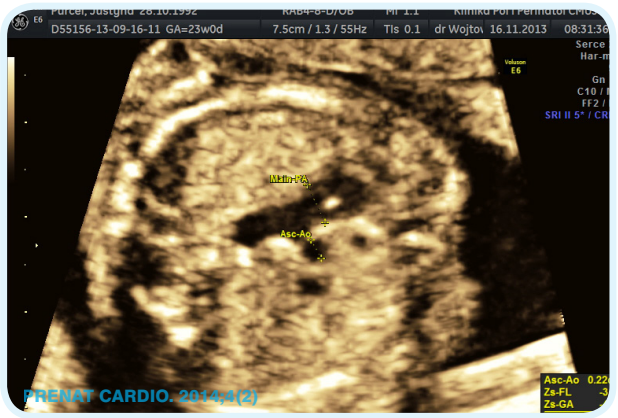

Photo 5. Three vessels view in the upper mediastinum

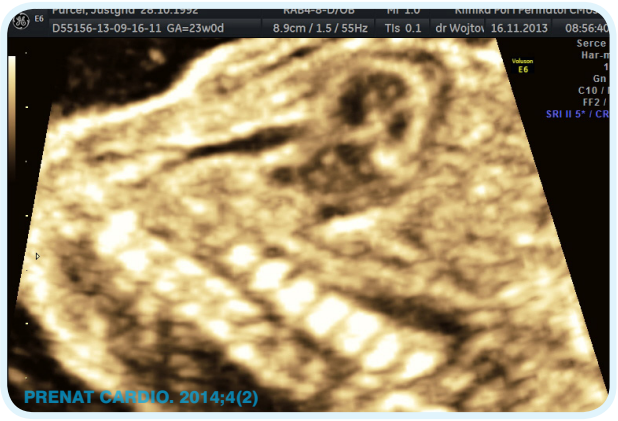

Photo 6. Ascending aorta with y sign

four chambers and correct connections between chambers and great vessels. Discrepancy with the domination of the right part of heart, which is often present in left outflow tract anomalies or in coarctation of the aorta is not always present in IAA. It could be explained by VSD, through it blood flows to LV and the ventricle grows normally. In our case the discrepancy between chambers was seen in the 1st trimester as well as VSD. Vogel et al [7] recognized lower ratio of the aortic valve to pulmonary valve (0,54+/-0,11) and ascendens aorta to pulmonary trunk (0,50+/-0,09) in the absence of the discrepancy between chambers. Universal presence of VSD in case of IAA type B was emphasized. Such VSD was described as posterior malalignment VSD. Significant discrepancy between pulmonary valve and aortic valve is not pathognomonic to IAA, but it should be considered as an indication of IAA or severe coarctation of the aorta [7]. Słodki et al. [9,10] showed that the ratio of the great vessels width in the upper mediastinum may be helpful to make a diagnosis of the coractation of the aorta and IAA. They demonstrated the meaningful discrepancy between PA/Ao diameters in typ A and B of the interrupted aortic arch (2,1+/-0,09 vs 2,9+/- 0,2; p=0,0007 respectively). In our case the discrepancy was significant amounted 2.

The difficulty in prenatal diagnosis of type of the interruption may be caused by the similarity of the ductus arteriosus arch and aortic arch, as well as that left subclavian artery arises from proximal part of the descending aorta, a little distal to ductus arteriosus arch, what can be interpreted as carotid vessels arising from aorta. A frequent sign in type B of IAA is that ascending aorta comes straight towards the head and does not turn where the first vessel arises from aorta which is innominate artery. Sonographers may see Y sign, with aorta divided in to two vessels as we can see on the Fot.6.

It should be kept in mind that in case of the detection of one anomaly precisely fetal anatomy should be checked, because another anomaly can be detect as in our case. There is a strong association between IAA type B and microdeletion 22q11.2 syndromes [11]. IAA type B is the most frequent cardiac defect in Di George syndrome, which is characterized by hypoplasia/aplasia of the thymus in connection with cardiac defects (over 90% of cases), especially conotruncal malformations [5,7,12,13]. 43 % of patients with Di George syndrome have type B of IAA, and 68% of all with IAA have Di George syndrome [14]. Postnatal case series showed that type A of IAA was rarely associated with del 22q11.2 [15], whereas 50-80% of cases with type B and other extracardiac anomalies [16,17].Interrupted aortic arch was also present in fetuses with such syndromes as CHARGE, Klippel-Feil, Turner, VACTERL and so on [17-20].

In our case abnormal heart anatomy was described in the 1st trimester and the karyotype was normal. Patient did not consent to another amniocentesis to perform FISH. However, taking in to consideration that pathology of the forearm, thymus aplasia, abnormal fetal profile and intrauterine growth retardation were present we can suspect the microdeletion 22q11.2. The answer will be after delivery. The question is, if we have anomalies in 1st trimester scan, especially cardiac defects, aside from karyotype FISH should be performed.

In the summary it should be state that this rare anomaly is recognizable in prenatal scan, especially in the 2nd trimester thanks to systematically evaluation the heart anatomy.

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Conflict of interest: The authors declare no conflict of interest

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