Case **report**

TRANSPOSITION OF GREAT ARTERIES (D-TGA) IN THE FIRST TRIMESTER - A CASE REPORT

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Abstract

D-type transposition of the great arteries (d-TGA) is a critical heart defect lesion, that should be diagnosed prenatally, as early postnatal management greatly relies on this information. Recently, in Poland more heart defects of this type are diagnosed prenatally. However, there is a lack of data regarding the diagnosis of d-TGA at the time of nuchal translucency measurement at 11-13+6 weeks of pregnancy. We present a case of d-TGA that was detected and properly diagnosed during the first trimester scan. The diagnostic plane that enabled the diagnosis was the three vessel-trachea view (3VT) presenting one, wide vessel instead of a typical V-sign.

Key words: *d-TGA, ultrasound, first trimester*

INTRODUCTION

Transposition of the great arteries type d first introduced by Matthew Baillie in 1808[1], is one of the most common congenital heart defects. In Poland, according to the National Registry of Fetal Cardiac Pathology (ORPKP), d-TGA was the fourth most common anomaly after Hypoplastic left heart syndrome (HLHS), Tetralogy of Fallot (TOF), atrioventricular septal defect (AVSD) with a prevalence of 6.9%[2]. d-TGA is a structural heart defect with an atrio-ventricular concordance and ventriculo-arterial discordance. In 30-40% of cases it can be associated with ventricular septal defect, left ventricular outflow tract stenosis or coronary artery anomalies [3]. The prenatal diagnosis of d-TGA remains as one of the most difficult due to the fact that the four chamber view (4CV) is normal in most cases. According to EUROCAT, d-TGA was diagnosed prenatally in 41% of cases, compared to 72% of HLHS in 2007-2011 [4]. The data gathered by Polish Mother's Memorial Hospital- Research Institute (ICZMP) shows that in Poland the average gestational age on detection of d-TGA in years 1997-2012 has decreased from 36 weeks (2007) to 30 weeks (2012)[5]. However, there is a lack of data regarding the diagnosis of d-TGA in the first trimester of pregnancy.

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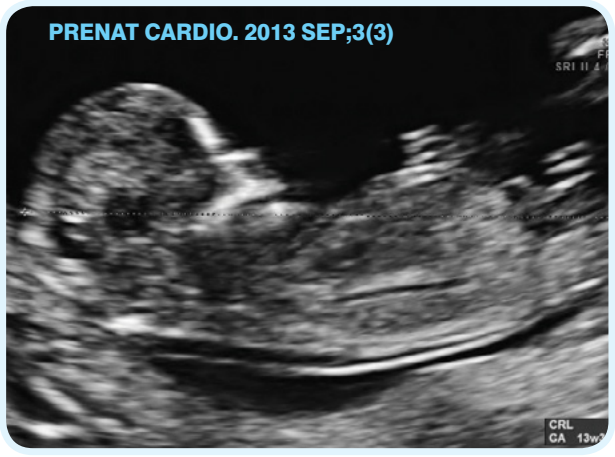
CASE REPORT

A 27-year-old patient G-1 P-0, came for the routine ultrasound scan at 11-13+6 weeks. The gestational age, based on LMP, was 12 weeks and 6 days. The patient and her husband were not consanguineous. The patient's husband was 28 years old. The measurement of the fetus based on crown-rump length (CRL) was 72.2 mm which corresponds to 13 weeks and 3 days (photo 1). During the scan, markers of aneuploidy were examined according to the Fetal Medicine Foundation (FMF) guidelines and included: nuchal translucency (NT- photo 2) thickness, nasal bones ossification (NB), ductus venosus (DV-photo 3) flow and tricuspid flow (TV-photo 4). The scan protocol included early anatomy evaluation



Fot. 1. First trimester scan.CRL 72 mm

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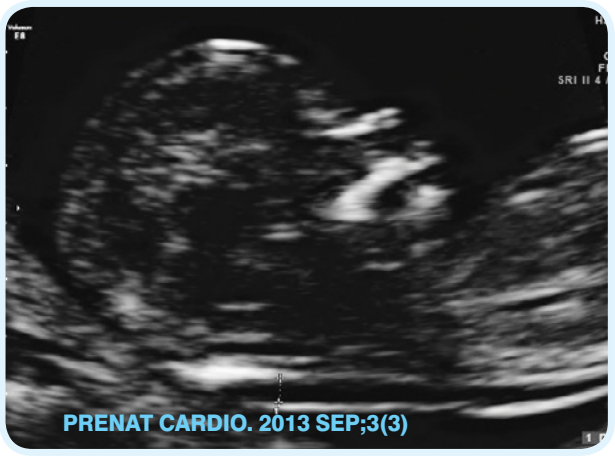
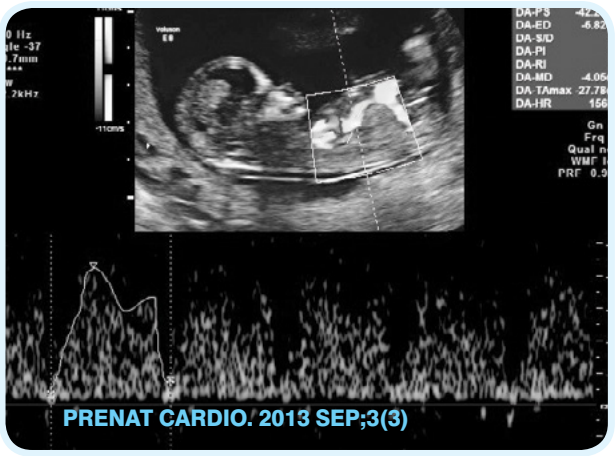


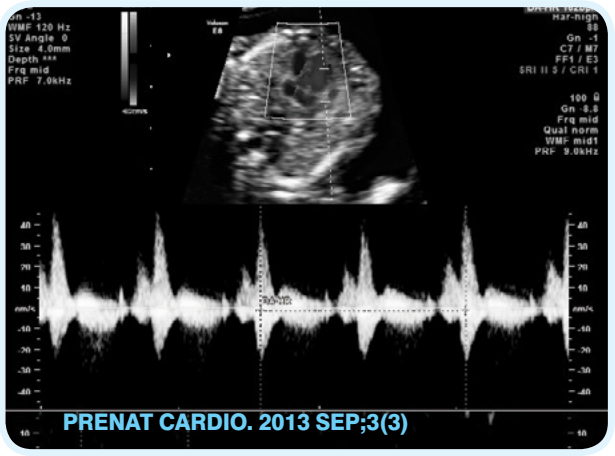
Fot. 2. Nuchal translucency (NT) and nasal bone (NB) in fetus with d-TGA

covering, according to the ISUOG guidelines[6]. The heart



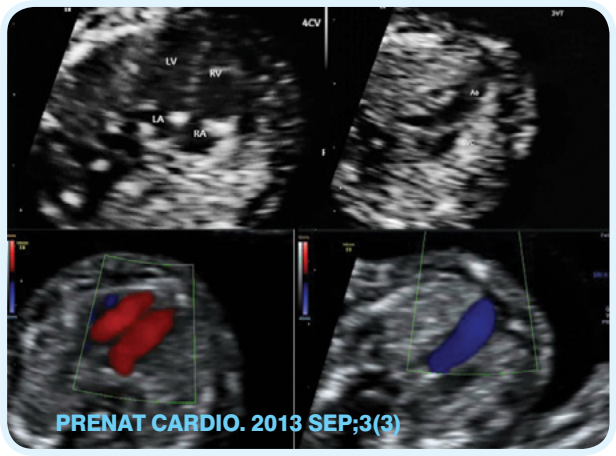
Fot.3. Ductus venosus flow in fetus with d-TGA

examination included evaluation of the four chamber view



Fot. 4. Tricuspid valve blood flow in fetus with d-TGA

(4CV) -inflow to the ventricles and the confluence of the ductus arteriosus and the aortic isthmus at the level of the three vessel and trachea view in color mapping (3VT). The ultrasound scan did not reveal any positive markers of aneuploidy: NT= 2.1 mm, TV- normal; DV-normal



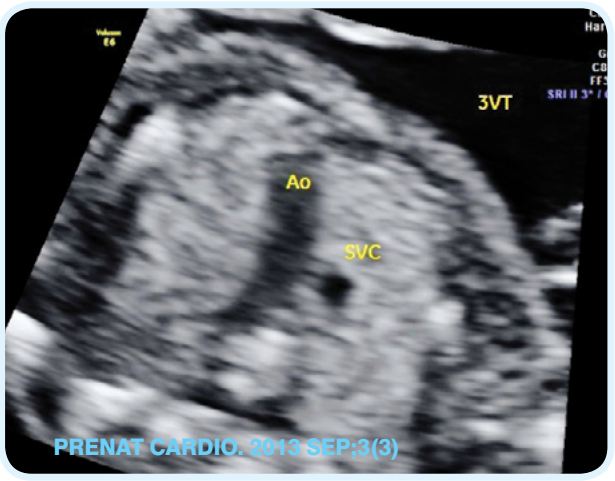
Fot. 5. Fetal heart with d-TGA at the level of the 4 chamber view and 3 vessels & trachea view (3VT) with color Doppler. At the level of 3VT view two vessels are seen - aorta and superior vena cava. Pulmonary artery is not visible because it is lower and back towards the aorta.

a- wave, NB-present, FHR- 162 bpm. Background risk of aneuploidy resulting from the age of the mother was 1:862 (trisomy 21); 1:2199 (trisomy 18); 1:6872 (trisomy 13). When the markers of aneuploidy were included, the new, adjusted risk of chromosomal aberrations was: T21- 1:17237; T18 <1:20000; T13 <1:20000.

The inflow at the level of the four chamber view in Color Doppler presented two stripes of equal size. At the level of the three vessel-trachea view 3VT the classical V-sign, that demonstrates from left to right the main pulmonary trunk continuous with DA that joins the aortic arch, could not be visualized. At this level only one, wide arterial vessel (Ao) was seen, a typical sign of d-TGA (photo 5). Other structural anomalies were not found. The Patient was referred to the Ultrasound Unit of the Head of Gynecology and Obstetrics in Krakow to verify the diagnosis. In the next ultrasound scan at 17 weeks+5 days an isolated d-TGA was confirmed. (photos: 6,7,8,9). The data concerning the prenatal scan was introduced to the National Registry of Fetal Cardiac Pathology (ORPKP-19860210). The patient was informed extensively about the management of pregnancy in case of d-TGA. The parents decided to continue with the pregnancy. At this time of pregnancy the patient was consulted by a cardiothorasic surgeon regarding the type of surgical treatment, prognosis and the potential need for urgent septostomy, within hours after delivery.

DISCUSSION

The fetal echo performed routinely during the second trimester of pregnancy enables the diagnosis of the majority of heart defects. However, the relationship between increased nuchal translucency in the first trimester and the risk of aneuploidy and congenital heart disease (CHD)[7-9] initiated the attempts to evaluate the heart at this point in pregnancy[10-12]. Most of the structural heart defects develop early, during the first weeks of embryonic life and present similar abnormal pictures in either the first, second

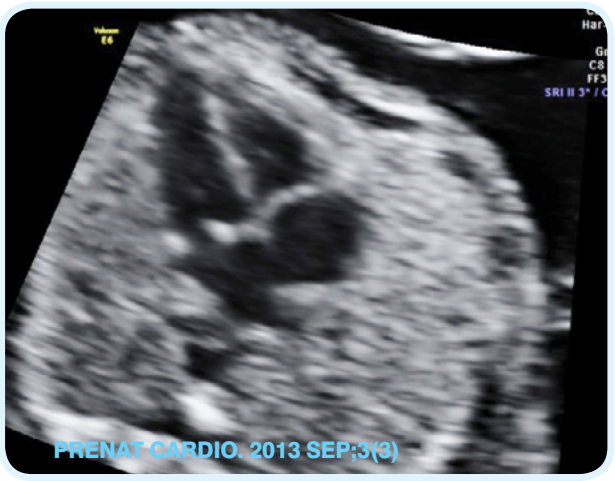


Fot. 6. 3VT in 17,5 wks of pregnancy in fetus with d-TGA. Single big vessel-aorta. The similar scan as obtained in 1st trimester.

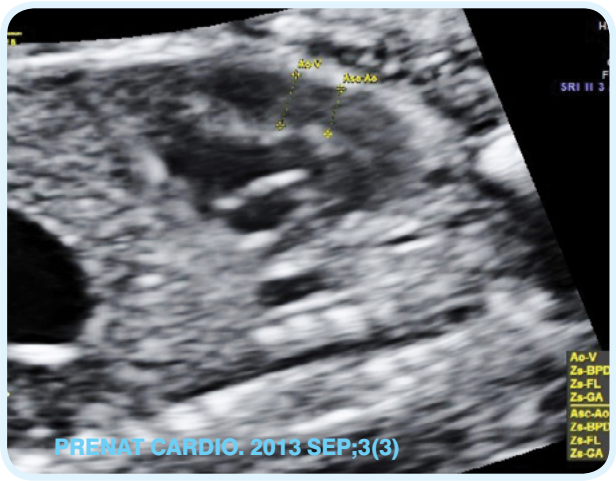
or third trimester of pregnancy, and include: d-TGA, AVSD, double outlet right ventricle (DORV), common arterial trunk (CAT), and tricuspid atresia –(TA)[3]. Some of the cardiac anomalies that continue to evolve, such as HLHS on the basis of critical aortic stenosis, cardiomyopathies or cardiac tumors, develop later, during the second trimester of pregnancy.

The examination of the heart in the first trimester includes the evaluation of the upper abdomen, four chamber view and transverse sections of the mediastinum. The normal view in these planes, in most cases, excludes severe heart defects. Smreck et al. visualized the above-mentioned sections in 45% of cases in the 11th week and in 90% at 13 weeks+6days[14]. The visualization of the heart at this point in pregnancy is dependent on several factors, such as the equipment, the experience of the examiner, the fetal position and the echogenicity of the abdominal tissue.

So far, there is a number of reports that present the detection rate of CHDs in the first trimester (Table 1). The diagnosis of d-TGA is often made during the second trimester scan. There is a handful of reports regarding the diagnosis of this particular anomaly in the first trimester[10,30]



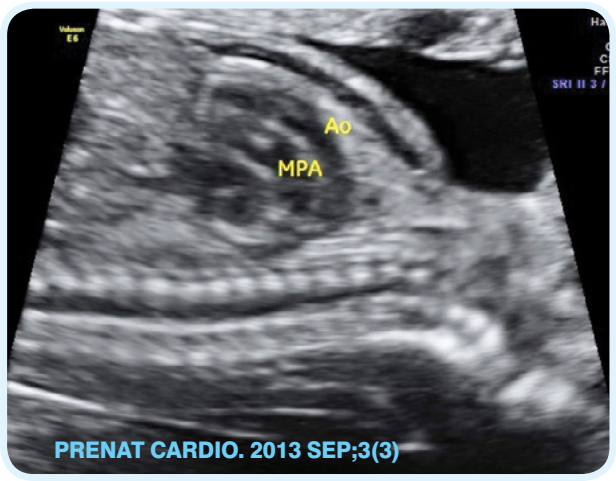
Fot 7 4Ch view in fetus at 17,5 wks of gestation.Normal view



Fot. 8. Longitudinal scan d-TGA. Z-score for the vessel from right ventricle - aorta, 17,5 wks of gestation

which may suggest that the defects of the outflow tracts are more difficult to diagnose at this point in pregnancy[10,25,31]. A misdiagnosis of two cases of d-TGA during the first trimester scan in the study of McAuliffe FM et al. could be attributed to the lack of clear visualization of great artery relationship in one case, and false negative image suggesting a normal crossing of the vessels in Color Doppler that turned out to be aortic and ductal arches in an oblique section through the fetal chest, rather than the proximal great arteries.[32]. Volpe et al. presented four cases of correctly diagnosed d-TGA at 11-14 weeks. However, the authors did not describe the methodology and the most important sonographic views that could aid in the diagnosis of this anomaly at this point in pregnancy[29].

In order to confirm the diagnosis of d-TGA, the outflow tracts, the left ventricular outflow tract particularly, should be visualized. The most characteristic sonographic sign is the parallel view of the great arteries. Nevertheless, its oblique view, with the use of the rotational technique, is not always easy to visualize. So far, many sonographic signs that would aid in the diagnosis of d-TGA have been proposed [33-35]. However, the technique of the evaluation of the mediastinum at the level of the three



Fot. 9. d-TGA in the longitudinal scan. Parallel vessels. The anterior vessel proximal to the head diverged into cephalad vessels

Authors	Gestational age	Number of fetuses detected before 14 wks	Type of heart defect	Autopsy
DeVore et al. (1987)16	14	1	VSD, PS	100
Gembruch et al. (1990)17	11	1	AVSD, PS, całkowity blok serca	100
Bronshtein et al. (1990)18	13-14	2	TOF	100
Bronshtein et al. (1991)19	12-16	10	AVSD, VSD, TOF, HLHS, SA, SV	80
Gembruch et al. (1993)20	11-16	5	AVSD, PS, SV, MGA	100
Achiron et al. (1994)21	10-12	8	ectopia cordis, AVSD, anomalia Uhl, TAC,TOF, tachykardia	63
Carvalho et al. (1998)22	12	1	AVSD, IAA	100
Baschat et al. (1999)23	11-14	4	Blok serca, AVSD, DORV, MGA, PS, LI	75
Zosmer et al. (1999)24	13-17	24	CoA, TA, TGA, AVSD, TOF, PA, HLHS, bradykardia, LI DILV, RI, PS,	NS
Simpson et al. (2000)25	12-15	13	PA, TV dysplasia, PA:IVS, CoA, AVSD, TOF, HLHS, isomerism, bradycardia	46
Haak et al. (2002)10	11-14	10	AVSD, VSD, DORV, HLHS	89
Huggon et al. (2002)26	10-14	60	AVSD, HLV, VSD, ebstein anomaly, TA, PA, LI	45
Comas Gabriel et al. (2002)27	12-17	38	AVSD, VSD, HLHS, DORV, TA, PA, TAC, TOF, CoA, ectopia cordis, SV, MA, TGA	45
Smrcek et al.(2006)28	11-14	40	AVSD, TOF, APVS, CoA, PS, AS, TAC, PA, VSD	65
Axt-Fliedner et al. (2008)29	11-14	78	AVSD, VSD, HLHS, TOF, APVSD, AS, PS, CoA, TAC, TGA, DORV, ysplazja zastawki trójdzielnej, jednokomorowe serce, ectopia cordis, zespół heterotaksji	68
Volpe et al. (2012)30	11-14	36	AVSD, TGA, TOF, TOF+LSVC, CoA, IAA, AS, PS, TR, VSD, VSD+LSVC, RAA, Dextrocardia	NS

Table 1. Congenital heart defects detected in the 1 and early second trimester of pregnancy based on the literature

vessel view (3VV) and three-vessel and trachea view (3VT) proposed by Vinals et al. simplifies and standardizes the evaluation of d-TGA, which is very important especially at the screening level. The geometry of the anomaly is constant at every stage of pregnancy, presenting normal 4CV and abnormal relation of the great arteries. The aorta leaves the right ventricle, with its valve located anteriorly to the pulmonary valve. The ascending aorta and its arch is located above the pulmonary arch that forms the left ventricular outflow tract. At the level of 3VV in d-TGA the abnormal arrangement of the vessels, with the aorta anterior to the pulmonary artery is seen. Under normal conditions the linear relation of the vessels in this plane is visualized; the main pulmonary trunk most anteriorly and to the left and SVC most posteriorly to the right and Ao in-between. At the level of the 3VT view, in d-TGA only two vessels, (Ao and SVC) instead of three can be seen, due to the interior location of the pulmonary trunk. That is why the typical V-sign at this plane cannot be visualized. (photo 10). Vinals et al. with the use of these planes in a grey scale demonstrated d-TGA in different trimesters, the earliest at 13 weeks, with the same ultrasound picture at every stage of pregnancy[30]. It should be underlined that the view of a single vessel at the level of 3VT is not synonymous with d-TGA. It can present similar pictures in other anomalies such as TOF or CAT. Therefore, the evaluation of the fetal heart in different planes is essential in order to confirm the diagnosis.

In the Polish National Registry for Fetal Cardiac Pathology in 2012, d-TGA took the 4th place on the list of the most common heart defects after HLHS, TOF and AVSD, and accounted for 6.9% of all registered cardiac malformations[2]. Although in recent years there was a tendency for a better prenatal detection of d –TGA at an earlier gestational age, it remains a diagnostic challenge. Due to this fact, the section of Echocardiography and Prenatal Cardiology of the Polish Ultrasound Society funded an award of dr. A. Respondek for the obstetrician who is the first to detect d-TGA anomaly in a current year [36]. In 2012, the award was given to the obstetrician who diagnosed another type of transposition- type L TGA. So far, there is a lack of information regarding the implications of d-TGA detection in the first trimester. The detection of this anomaly in the first trimester does not change the management regarding the follow-up during pregnancy. Late third trimester is the most critical period, when the hemodynamic changes may occur as a result of a premature closure of ductus arteriosus (DA) or restrictive patent foramen ovale (FO)[37-39]. The restriction at the level of foramen ovale, hypermobile septum of FO[39] or reverse diastolic flow in ductus arteriosus are the findings that may predict the need for an early, urgent balloon atrioseptostomy (the Rashkind procedure). In the recent analysis of 55 newborns with d-TGA, 23 required the Rashkind procedure during 48h after delivery, while arterial switch (the Jatene procedure) was performed, on average, 11 days after delivery[5].

The transposition of great arteries is a ductal-dependent anomaly that should be detected prenatally, as the early cardiosurgical treatment relies on the prenatal diagnosis. Furthermore, prenatal detection of d-TGA is associated with decreased neonatal mortality and morbidity, when compared to the postnatal diagnosis.[40] More extensive studies are necessary to draw conclusions regarding the role of early d-TGA detection in the first trimester of pregnancy.

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