

CASE REPORT

# Prenatal diagnosis of complete transposition of the great arteries at 12 weeks of gestation in a fetus with normal nuchal translucency: a case report

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## ABSTRACT

**Introduction:** Complete transposition of the great arteries (TGA) is a common cardiac malformation with atrioventricular concordance and ventriculoarterial discordance with an incidence of 20-30 per 100,000 cases. While prenatal diagnosis of TGA remains challenging, especially in the first trimester ultrasound scan, advances in ultrasound equipment and sonographer training have resulted in an increased detection rate (from 12.5% to 72.5%) in the last decades.

**Case Presentation:** We present the case of a 31-year-old Caucasian primigravida with no medical or family history of congenital anomalies, who attended our unit for the routine first trimester ultrasound examination. The initial scan revealed a singleton live fetus with a gestational age of 12 weeks and a normal nuchal translucency, nasal bone, flow pattern in the ductus veno-

sus and no regurgitation in the tricuspid valve of the fetal heart. While the four-chamber view of the heart appeared normal, careful examination of the outflow tracts failed to show the crossing of the pulmonary artery with the aorta. The parallel course of the great arteries confirmed the diagnosis of complete transposition of the great arteries.

**Conclusion:** Examination of the two outlet echocardiographic views during the 11 - 13+6 ultrasound scan by obstetric sonographers allows for early detection of TGA. The presence of TGA warrants a thorough anomaly scan and genetic counselling as TGA is associated in 10% of the cases with other noncardiac malformations. Finally, antenatal detection of TGA results in better clinical status before surgery and improved postoperative outcome of the neonate.

## KEY WORDS

Transposition of the great arteries; TGA; X-sign; cardiac defect; parallel course

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## Introduction

Dextro-transposition of the great arteries (d-TGA) characterized by situs solitus, atrioventricular concordance and ventriculoarterial discordance, is associated with early and severe neonatal central cyanosis, requiring urgent diagnosis and appropriate treatment [1]. It represents 5-7% of all congenital cardiac malformations, corresponding to an incidence of 20-30 per 100.000 cases and a 1.5:1 to 3.2:1 male preponderance [1-5]. In 10% of the cases d-TGA may be associated with other non-cardiac malformations [2], while the karyotype in most cases is normal [6]. Prenatal diagnosis of transposition of TGA remains a great challenge in fetal medicine, due to the difficulties associated with the evaluation and correct identification of great arteries and their origin [7-11]. The diagnosis of TGA during the first trimester of pregnancy is an even greater challenge. Studies regarding prenatal screening for congenital heart malformations show a sensitivity of identifying d-TGA around 3-17% [1,7,12]. This detection rate according to some studies has increased the last decades from 12.5% to 72.5% [13]. The diagnosis of d-TGA is possible during the 11-13+6 weeks scan, but as expected detecting d-TGA at that time is more difficult than in the second trimester and most cases are missed [14]. The relationship between increased nuchal translucency (NT) and major cardiac defects has been established and an early fetal echocardiography in fetuses with increased NT is suggested [15-16]. However, the effectiveness of detailed examination of the fetal heart as a routine, in fetuses considered low-risk after the NT examination and first trimester sonographic markers – nasal bone (NB), ductus venosus (DV) and tricuspid regurgitation (TR) – remains unclear [17-19]. We present a case of TGA diagnosed at 12 weeks of gestation during the routine 11-13+6 weeks scan, in a patient with normal NT, and normal DV and TR.

## Case description

A 31-year-old Caucasian woman in her first pregnancy, with an unremarkable medical and family history for congenital malformations or genetic disorders, attended our unit for the routine 11-13+6 ultrasound examination, for screening of fetal chromosomal abnormalities. The examination in our unit is performed according to the guidelines of the fetal medicine foundation (FMF) with measurement of the NT and the other first trimester so-



**Figure 1.** Normal 4 chamber view of the fetal heart

nographic markers suggested by FMF for the screening of chromosomal defects (NB, DV, TR, facial angle - FA) in combination with maternal serum biochemistry (PAPP-A and free $\beta$ -hCG). All examiners are accredited by the FMF for all the above examinations. In our unit as part of a multicenter study we perform an extended morpho-genetic ultrasound protocol during the 11-13+6 weeks scan for the detection of structural abnormalities. Ultrasound examinations are performed with a GE 730 PRO and PHILIPS HD 11 ultrasound machine with abdominal transducer. The initial ultrasound examination showed a singleton live pregnancy with a fetal heart rate (FHR) of 162 bpm and a crown rump length (CRL) of 53.6 mm, corresponding to gestational age (GA) of 12 weeks, which was in agreement with the GA calculated by the last menstrual period of the woman (LMP). Further ultrasound assessment of the fetus showed a normal NT for the GA (NT=1.9 mm), normal NB, normal flow pattern in the DV and no regurgitation in the tricuspid valve of the fetal heart. According to our protocol we proceeded to further assessment of the fetal anatomy. For the assessment of the fetal thorax the protocol of our study is as follows: transverse planes (transverse cardiac sweep): a. situs evaluation, b. area one quarter to one third of the chest and angle  $45\pm 15^\circ$  from the antero-posterior midline (subjective appreciation, measured only if seemed abnormal), c. atrio-ventricular valve offsetting in four chambers view and tricuspid valve (TV) flow assessment using pulsed Doppler, d. aorta arising from the left ventricle and pulmonary trunk arising from the anteriorly placed right ventricle and crossing to the fetal left side over the ascending aorta, e. color-flow investiga-



**Figure 2.** The parallel course of the great arteries

tion of four-chamber view, outflows emergence - 'X' sign (the crossing of the main pulmonary artery with the aorta and being equal in size), and three vessel view - 'V' sign (the connection of the aortic arch and ductus arteriosus), f. ductus venosus (DV) flow assessment using pulsed Doppler.

The examination revealed a normal four chamber view (Figure 1) and three vessel view of the fetal heart. However, careful examination of the outflow tracts failed to show the crossing of the pulmonary artery with the aorta (X-sign). The fetus was examined by a specialist in fetal echocardiography who confirmed the "parallel course" of the great arteries (Figure 2), raising the possibility of congenital heart disease affecting the origin of great arteries, including transposition of the great arteries. The pulmonary artery coming out from the left ventricle is depicted in Figure 3.

The fetus was reassessed at 14<sup>+</sup> weeks with an ultrasound examination which confirmed the diagnosis. The couple had extensive counseling by specialists in fetal medicine, fetal echocardiography, neonatal cardiology and pediatric cardiac surgery, from the tertiary neonatal unit that our unit is affiliated with. They were informed about the follow up they should have during pregnancy, the possibilities and the prognosis of the neonatal outcome. They also decided to proceed to examination of the fetal karyotype at 16 weeks of GA, in order to exclude particularly microdeletions of 22q11. The amniocentesis showed a normal male karyotype (46XY). However, the parents decided to proceed with termination of the pregnancy at 18 weeks of gestation, due to socio-economic reasons. The postmortem examination showed



**Figure 3.** The pulmonary artery coming out from the left ventricle

a complete transposition of great arteries without any other obvious cardiac or extracardiac abnormalities.

## Discussion

D-transposition of the great arteries is one of the most common cyanotic congenital heart defects in the neonatal period, representing 5-7% of all congenital heart diseases, corresponding to 20-30/100000 live births. In 10% of cases TGA is associated with other noncardiac malformations [2,20]. Antenatal diagnosis of TGA results in better clinical status before surgery and improved postoperative outcome, compared to those diagnosed postnatally [21]. Early management includes intravenous administration of prostaglandin E1 in order to maintain the patency of the arterial duct [22]. When prostaglandin infusion proves insufficient, balloon atrial septostomy (known as the Rashkind procedure) is performed to ensure proper oxygenation and to allow for more time before the corrective operation is performed [23]. The treatment of choice is the arterial switch operation which has shown survival rates of 88% at both 10 and 15 years of age [24].

The diagnosis of D-TGA was infrequently recognized by obstetric sonographers in the era of the four-chamber view [12, 25]. There is a rise in the detection rate from 20% with the increased number of routine antenatal scans and a policy of careful training for two outlet echocardiographic views [12, 25]. Regions of Paris achieved a detection rate of 72% between 1995 and 2000 [13].

Early detection of D-TGA allows for more time for genetic counselling and fetal karyotyping. Even though d-TGA is rarely associated with genetic syndromes, it has been sporadically associated with trisomy 8, trisomy 18, VACTERL syndrome, CHARGE syndrome, tuberous sclerosis, deletion of the long arm of chromosome 11 and the short arm of chromosome 18, Turner syndrome, Noonan syndrome, Williams syndrome and Marfan syndrome [26-32]. Early detection also facilitates planning of the

delivery in a tertiary hospital with a neonatal intensive care unit and a pediatric cardiac surgery department, hence improving neonatal outcomes. However, antenatal detection of congenital heart defects has been also associated with an increased probability for termination of pregnancy decisions [33]. In any case, every abnormal early fetal echocardiogram should be followed by a re-evaluation scan by a fetal cardiology expert in mid-gestation to corroborate the initial diagnosis. ■

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## CITATION

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