Actual management of complete transposition of the great arteries diagnosed prenatally

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Abstract

Transposition of the great arteries (TGA) is a cyanotic congenital heart condition characterized by an atrioventricular concordance and a ventriculoarterial discordance, in which the aorta rises from the right ventricle while the pulmonary artery rises from the left ventricle. The transposition of the great arteries is commonly undiagnosed in utero, with less than half of the cases being detected and having an increased incidence in males than females. The transposition of the great arteries can sometimes be associated with a more complex genetic syndrome, such as DiGeorge syndrome, or can be an isolated condition. The transposition is not a life-threatening condition in utero due to the existence of the foramen ovale and ductus arteriosus which connect the two parallel circulation systems that are created by the condition. Adding the evaluation of the outflow tracks to the traditional four-chamber view increased the sensitivity of ultrasound screening by 2.5 times. The transposition of the great arteries can be surgically corrected after birth, with the best outcome possible if the surgery takes places in the first two weeks of life. The overall outcome for patients with transposition of the great arteries has improved in recent times due to the available surgical procedures and antenatal detection, with a good long-term survival, low morbidity and with excellent functional outcome.

Keywords: *transposition of the great arteries, ultrasonography, outflow track*

Rezumat

Transpoziția de vase mari este o boală congenitală cardiacă ce este caracterizată de o concordanță atrioventriculară, cu o discordanță ventriculo-arterială, în care aorta pornește din ventriculul drept, în timp ce artera pulmonară pornește din ventriculul stâng. Transpoziția de vase mari este în general subdiagnosticată intrauterin, cu mai puțin de jumătate din cazuri detectate, având o incidență mai mare la fețiii de sex masculin față de cei de sex feminin. Transpoziția de vase mari poate să fie unică ori asociată cu alte malformații cardiace sau să fie parte dintr-un sindrom genetic, cum ar fi sindromul DiGeorge. Transpoziția nu este o boală amenințătoare de viață intrauterin datorită existenței ductului arterial și a foramen ovale care conectează cele două circulații paralele create de boală. Adăugarea evaluării traiectului de ejecție al marilor vase la secțiunea tradițională de patru camere a crescut sensibilitatea ecografiei de screening cu peste 2,5 ori. Transpoziția de vase mari poate să fie corectată chirurgical după naștere, având cele mai bune rezultate dacă se realizează în primele două săptămâni de viață. Prognosticul pacienților cu transpoziție de vase mari s-a îmbunătățit semnificativ în ultimii ani datorită tehnicilor chirurgicale existente și detecției antenatale, cu o supraviețuire pe termen lung foarte bună, cu morbidități reduse și cu potențial funcțional excelent.

Cuvinte-cheie: *transpoziția de vase mari, ultrasonografie, traiect de ejecție*

Submission date: 22.11.2021
Acceptance date: 2.12.2021

Managementul actual al transpoziției complete de vase mari diagnosticate prenatal

Suggested citation for this article: Bohîlțea R, Tudorache D, Pariza PC, Călin AM, Dima V, Mitran M, Vlădăreanu R. Actual management of complete transposition of the great arteries diagnosed prenatally. Ginecologia.ro. 2021;34(4):14-17.

Introduction

Transposition of the great arteries (TGA) is a cyanotic congenital heart condition in which the aorta rises from the right ventricle while the pulmonary artery rises from the left ventricle. It is a disease that represent between 5% and 7% of all congenital heart conditions and about 20% of cyanotic heart disease disorders. It is characterized by an atrioventricular concordance and a ventriculoarterial discordance, which creates two parallel and closed circulation systems, a pulmonary one and a corporeal one[1-3].

The transposition of the great arteries is commonly undiagnosed *in utero*, with less than half of the cases being detected. Most of the time, transposition of the great arteries is not an isolated abnormality, usually being accompanied by ventricular septal defect, left ventricle outflow obstruction, mitral and tricuspid valvular abnormalities[1].

If not treated properly, most of the patients die within the first year of life, while the prognosis for those surgically corrected is very good, with a normal cognitive and motor development during their lifetime[3].

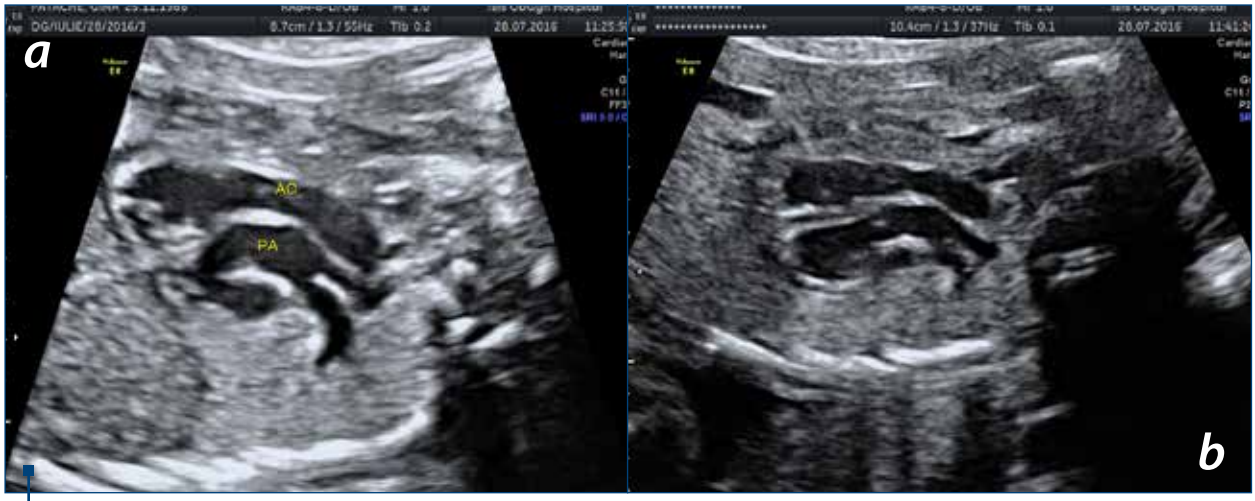

Figure 1. *Conventional two-dimensional ultrasonography showing the parallel trajectory of the aorta and the pulmonary artery with its bifurcation (**a**) and the ejection channels (**b**)*

Case report

We present the case of a 30-year-old primigesta, primipara pregnant patient who was referred for pregnancy monitoring with a singleton pregnancy that was obtained spontaneously. The patient's history showed no congenital abnormalities and no other comorbidities. During the second-trimester screening ultrasonography, the transposition of the great arteries was detected (Figures 1 and 2). Prior to that, no other structural anomalies were detected during the first-trimester ultrasonography. No associated malformations were found regarding the fetal brain, fetal abdomen, placenta or umbilical vessels.

The ultrasonography revealed four heart chambers with normal atrioventricular connections, and abnormal ventriculoarterial connections, with the aorta starting from the right ventricle and the pulmonary artery starting from the left ventricle. The parallel trajectory of the two great vessels is viewable at the base of the heart, the pulmonary veins open in the left atrium while the two venae cava open in the right atrium. The ventricular

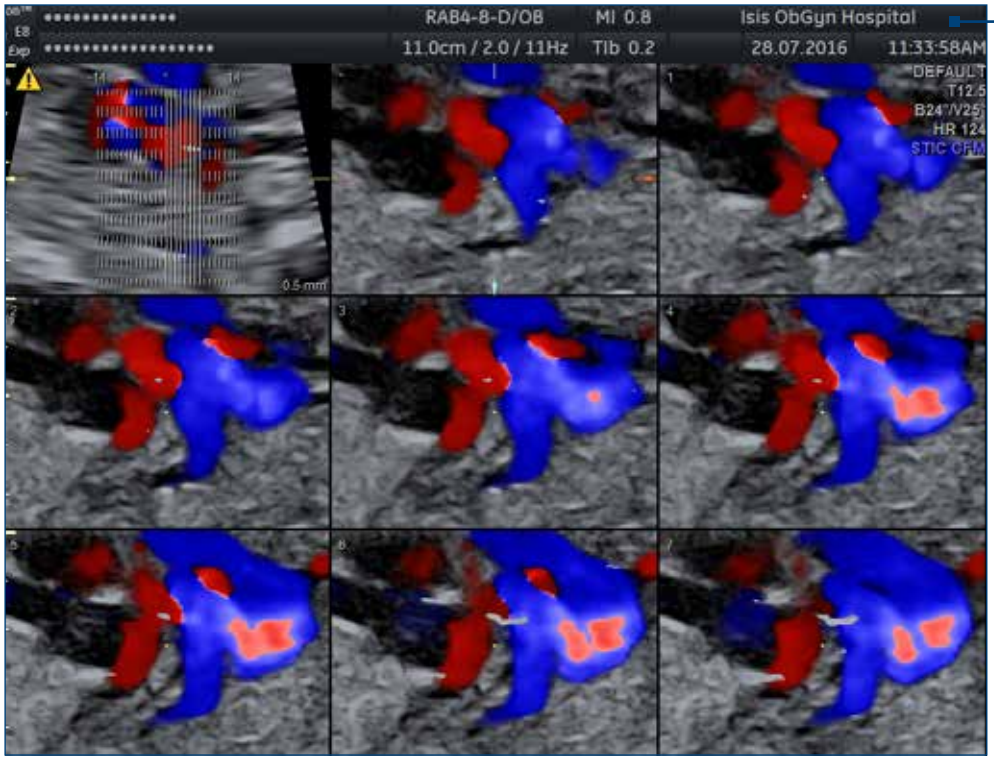

Figure 2. *Ultrasonography showing the outflow track of the two great vessels with the parallel trajectory and no crossing between them, by the spatiotemporal image correlation (STIC) with tomographic ultrasound imaging (TUI)*

septal defect that accompanies TGA in 40% of cases was absent in our case.

Amniocentesis was recommended in order to exclude a possible DiGeorge syndrome, while the patient was referred to the Târgu-Mureş Clinic for pediatric cardiovascular surgery department. The genetic test showed a normal karyotype and the fetal biometry was appropriate for the gestational age. The birth occurred without incidents at Târgu-Mureş and the transposition was surgically corrected, with a normal evolution postintervention. There was no long-term complication following the surgery, with the child being 5 years old now, having a normal development. The mother gave birth to a second child, with no anomalies.

Discussion

Transposition of the great arteries is reported to have an incidence of 5 cases in 10,000 live births, with an increased incidence in males, with a male to female ratio from 2:1 to 3:1, as reported by Grewal et al.[4] It is rarely associated with other noncardiological abnormalities, the main incriminated cause of the anomaly being hypothesized to be the abnormal evolution of the bilateral subarterial conus, with the persistence of the subaortic conus which normally resorbs, and the resorption of the pulmonary conus which normally persists, thus the conoventricular rotation cannot happen normally[1,4].

The transposition of the great arteries can sometimes be associated with a more complex genetic syndrome. It can accompany DiGeorge syndrome, although the patients only rarely have complete transposition of the great arteries, most of the cases involving only a conotruncal lesion with branching abnormality[1].

The transposition is not a life-threatening condition *in utero* due to the existence of the foramen ovale and *ductus arteriosus* which connect the two parallel circulation systems that are created by the condition. After birth, the condition is incompatible with life, as long as the connections are not maintained functionable, as there will be no oxygenated blood delivered to the body tissues. Continuous infusion of prostaglandin is required in order to maintain the *ductus arteriosus* permeable, as it allows the mixing of the two circulations[1,3].

The antenatal diagnosis of the transposition of the great arteries is mandatory in order to obtain a positive outcome of the case. Routine fetal heart screening must be performed by all the maternal-fetal specialists, including the visualization of the emergence of the great arteries, as most of the fetuses have normal ultrasonographic scans when only the four-chamber view is performed[1,2,4]. The addition of the evaluation of the outflow tracks to the four-chamber view increased the sensitivity of ultrasound screening to 69-83% from 30%, as reported by Fulton and Kane[1]. The three-vessel and trachea view, which normally includes the assessment of the position, size and trajectory of the aorta, the pulmonary artery and superior vena cava, also performed as a standard of the second-trimester prenatal screening, in most TGA cases consist in a single large vessel, which is the transverse aortic arch, with the superior vena cava to its right[2]. The prenatal follow-up should be focused on the existence of ventricular septal defect, on the possible development of pulmonary stenosis during the third trimester and on the monitoring of the premature preterm closure of the *ductus arteriosus* and/or foramen ovale.

Congenitally corrected TGA and double outlet right ventricle are the most important entities of the differential diagnosis.

The steps required to identify the existence of the transposition of the great arteries is to assess the following steps: the alignment of the aorta and the pulmonary artery, whether they cross or they are parallel, the emergence of the arteries from their ventricles, and if the pulmonary bifurcation forms a imagine like a bird's beak. Menahem et al. concluded in their study that the convexity of the aorta may be used as a marker for the transposition of the great arteries, with the convexity being to the right instead of left as in normal hearts[2,5].

Ishii et al. introduced in 2013 a new marker for the antenatal diagnosis of transposition of the great arteries with an "I-shaped" aorta being identified at 97% of the patients. Bravo-Valenzuela et. al concluded in 2019 that an easier marker is the "boomerang sign" that represents the rightward convex curvature of the right ventricle flow which normally is leftward. The study showed that the boomerang sign was identifiable from the first trimester[2,6]. Generally, the TGA can be detect at the $11\text{-}13^{+6}$ week ultrasound, but the diagnosis is missed in most cases, the enlarged nuchal translucency associated with the single great vessel on the three-vessel view section being highly suggestive and inciting for the parallel course of the two vessel search[7].

The transposition of the great arteries can be surgically corrected after birth, with the best outcome possible if the surgery takes places in the first two weeks of life. Fulton et al. reported complications in 5% to 25% of the patients, with the main postsurgical problems being represented by pulmonary artery stenosis, coronary artery stenosis or insufficiency and early coronary atherosclerotic disease[3].

The overall outcome for the patient with transposition of the great arteries has improved in recent times due to the available surgical procedures and the antenatal detection with a further correct management of the case. Patients with arterial switch operation have the best long-term survival with low morbidity and good functional outcome, with over 95% declaring no functional limitations at their follow-up[3].

The mortality at over 20 years after procedure is less than 5%, while the mortality in the first year after surgery is less than 1%. Fulton et. al reported that mortality is higher in patients with complex TGA (other cardiac anomalies present), prematurity (less than 36 weeks), low birth weight, aortic arch obstruction, and history of supraventricular tachyarrhythmias[1,3].

The neurodevelopment was reported to be in the normal range at 6 years old, with better results for

children diagnosed antenatally than in those diagnosed postnatally, the development being influenced by other genetic abnormalities present, the degree of hypoxemia in neonate period, the timing of surgery and by nutrition[3].

Conclusions

Transposition of the great arteries is a cyanotic congenital heart condition in which the aorta arises from the right ventricle while the pulmonary artery arises from the left ventricle, with two parallel circulation circuits.

Unlike most of the congenital heart disease, transposition of the great arteries is most often not associated with other conditions. The conditions that can accompany it are represented by ventricular septal defect, mitral and tricuspid valvular anomalies and left ventricle outflow obstruction.

Severely underdiagnosed in the past, the condition is now more likely to be discovered during the pregnancy due to the improvement of the ultrasonographic techniques and the possibility to visualize the trajectory of the outflow tracts of the two vessels.

The antenatal diagnosis is mandatory in order to correctly manage the pregnancy and delivery and to create all the necessary premises for a successful surgical procedure in the first two weeks of life.

Short-term and long-term outcomes are excellent, with over 97% survival rate at 20 years from surgery and over 95% of the patients declaring no functional limitations at their follow-ups. ■

Acknowledgements: No financial support declared.

Conflicts of interests: The authors declare no conflict of interests.

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