Conotruncal Anomalies

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Abstract: In the chapter, the 2D, color Doppler and 4D features of major conotruncal abnormalities will be described. In particular, the echocardiographic views on which the various lesions are detected will be described. In addition, the role of color Doppler in the recognition of valve stenosis or insufficiency will be illustrated. Finally, the diagnostic role of 4D echocardiography will be described, only in those cases in which it has additional clinical value. Videos of major diagnostic features are also provided, to facilitate the understanding of the text.

Key Words: 4D Ultrasound, Fetal Echocardiography, Conotruncal Anomalies.

TETRALOGY OF FALLOT (TOF)

Anatomy. Tetralogy of Fallot (TOF) is a cardiac malformation characterized by: 1) a malalignment subaortic VSD, 2) an aorta overriding the defect, 2) an obstruction of the right outflow tract, mainly involving the infundibular part, of varying degree; 4) a consequent hypertrophy of the right ventricle which becomes manifest only after birth. In the fetus, the pulmonary stenosis is often late onset and the main diagnostic features become then the malalignment VSD with the overriding aorta. The TOF spectrum comprises a number of variants and subtypes among which TOF with absent pulmonary valve and pulmonary atresia with VSD, which will be addressed separately.

Ultrasound diagnosis. As for most conotruncal anomalies, the 4-chamber view is unremarkable, unless anomalies of the atrioventricular plane are associated, which is rather uncommon. The classic form of TOF is diagnosed on the outflows' views, where the malalignment VSD with the overriding aorta can be seen (Fig. 1 - video 1).

Figure 1: Tetralogy of Fallot. On the left outflow tract view, it is possible to detect the malalignment ventricular septal defect (arrow). See also video 1 (Ao: ascending aorta LV: left ventricle; RV: right ventricle)

On the right outflow tract view, the smaller pulmonary artery can be detected. As mentioned above, the infundibular stenosis is not a constant finding in the 2nd trimester, but a significant stenosis, sometimes even progressing to atresia, can develop in the 3rd trimester [1]. This is why serial ultrasound monitoring is important in order to demonstrate antegrade flow through the pulmonary artery late in gestation to exclude the potential need for prostaglandin therapy after birth. Color Doppler can be used to demonstrate flow through the VSD toward the aorta (Fig. 2 - video 2) from both ventricles and flow through the smaller pulmonary outflow tract.

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Of note that in utero it is unusual to detect any significant acceleration of blood across the right outflow tract, even in the presence of an obvious reduction of the vessel size. Spectral Doppler is of limited value, as is 4D echocardiography [2]. The latter may be of help if anomalies of the aortic arch are associated.

From what has been said above, in utero TOF cannot be differentiated from a simple malalignment VSD, if evident pulmonary stenosis is absent. Therefore, in the absence of pulmonary outflow obstruction, it is always necessary to consider the possible evolution from malalignment VSD to TOF. Another important concept is that the degree of aortic overriding is variable, and cannot be fully appreciated in the fetus. Therefore, when the aorta emerges about 50% from the right ventricle, differentiation of TOF from a Fallot-like double-outlet right ventricle can be difficult, the two being distinguished only by the degree of aortic overriding. With regard to the differential diagnosis with other conotruncal anomalies, it should be considered that common arterial trunk (CAT) and pulmonary atresia with ventricular septal defect (PAVSD) share with TOF the presence of a malalignment VSD. Hence, if aortic overriding is found, the right outflow tract should be evaluated: if a small pulmonary artery is connected to the right ventricle, the diagnosis is TOF; if this is atretic, it is PAVSD; if the pulmonary artery originates from the single emerging vessel, it is CAT.

Prognosis and survival. The association with extracardiac anomalies is frequent, in particular gastrointestinal and thoracic ones (esophageal and duodenal atresia, and diaphragmatic hernia), even independently of chromosomal anomalies. The aneuploidy risk is high (up to 20% in fetal case series), with equal distribution between trisomies 21 and 18,4,15 [3-5] Consequently, karyotyping is mandatory. There is a lesser association with microdeletion 22q11, except in the variant with absence of the pulmonary valve, where the association is about 25%. [6]. Also the risk of association with non-chromosomal syndromes is relatively high [7].

As for the obstetric management, it is safer to plan the delivery in tertiary referral centers in order to warrant optimal multidisciplinary management of possible associated malformations, notwithstanding the fact that the shunt is significant enough to warrant oxygenation in classic TOF.

The overall prognosis will depend on several factors, including karyotype, associated extracardiac malformations, and cardiac anatomy. This last factor is extremely important as the anatomy of the defect may vary significantly. With regard to survival, case series of patients with isolated TOF report long-term survival rates as high as 80-90%. Hence, if no unfavorable prognostic factors are found in utero, and, above all, after birth, TOF is an easily correctable heart defect, with excellent survival and good quality of life [5].

**ABSENT PULMONARY VALVE SYNDROME (APVS)**

Definition. The absent pulmonary valve syndrome refers to a rare congenital anomaly, namely a severely hypoplastic pulmonary valve with anular stenosis, aneurysmal dilatation of the main pulmonary artery and branches. This anomaly may occur as an isolated lesion, or, more commonly be associated with TOF and ductal agenesis. In the former, the interventricular septum is intact, the pulmonary arteries are less dilated, and the ductus arteriosus is present.

Ultrasound diagnosis. Unlike most conotruncal malformations, APVS can be suspected on the 4-chamber view. The two key features are an evident cardiomegaly and an abnormal cardiac axis [6]. On this view, on some occasions,
the pulmonary trunk may be so dilated to become visible. On the left outflow view, the malalignment VSD with an
overriding aorta can be seen. Sweeping further cephalad from this view, a severe dilatation of the pulmonary trunk
and branches comes into view (Fig. 3).

**Figure 3:** Tetralogy of Fallot with absent pulmonary valve. On color Doppler, the right outflow tract view demonstrates the
severe dilatation of the main pulmonary trunk and branches, due to the steno-insufficiency of the severely hypoplastic pulmonary
valve. Aliasing due to high velocity is also evident on color Doppler. (LPA: left pulmonary artery; RPA: right pulmonary artery;
RV: right ventricle)

Color Doppler is used to detect the stenosis and insufficiency of the rudimentary pulmonary valve (Fig. 3). We may
apply 4D echocardiography to demonstrate effectively the degree of pulmonary artery dilatation (Fig. 4 - [video 3])
and to characterize the severely dysplastic pulmonary valve [2].

**Figure 4:** Tetralogy of Fallot with absent pulmonary valve. 4D-echocardiography clearly demonstrates the severity of the
dilatation of the pulmonary main artery and branches. See also [video 3]. (LPA: left pulmonary artery; RPA: right pulmonary
artery; RV: right ventricle)

Prognosis and survival. The risk of association with chromosomal anomalies is extremely high for APVS-TOF, with
25% association rate with the microdeletion 22q11 [6, 8]. On the contrary, the variant with an intact ventricular
septum is rarely associated with aneuploidies. Therefore, karyotyping including FISH analysis for the DGCR on
chromosome 22 is mandatory in all cases of APVS-TOF [7]. Delivery should take place in a tertiary referral center
in order to ensure adequate neonatal management, which may require resuscitation and, in some cases, tracheotomy.

Survival is lower than that reported for TOF, due to the frequent association with the microdeletion 22q11. In
addition, severe respiratory distress due to compression of the bronchial tree by massively dilated pulmonary arteries
and tracheomalacia may lead to demise after birth.

**PULMONARY ATRESIA WITH VENTRICULAR SEPTAL DEFECT (PAVSD)**

Anatomy. PAVSD represents an extreme form of TOF. There is a malalignment VSD with an overriding aorta, while
the right ventricular outflow tract is, in most cases, similar to that of TOF, with the muscular outlet septum
being anteriorly displaced. In most cases, the muscular outlet septum fuses directly with the parietal musculature of the right ventricle, obliterating the ventriculopulmonary junction. The anatomy of the hypoplastic pulmonary vessel is variable. In the most frequent arrangement, the right and left pulmonary arteries are confluent (communicating with each other) and supplied by the ductus arteriosus. Alternatively, the central pulmonary arteries may be confluent and coexist with Major Aorto-Pulmonary Collateral Arteries (MAPCAs). The third pattern of arterial supply is complete absence of the central pulmonary arteries, the lungs being directly supplied by multiple MAPCAs.

Ultrasound diagnosis. The 4-chamber view is usually normal; in some cases, minor leftward rotation of the cardiac axis and/or cardiomegaly can be appreciated. The malalignment VSD is best visualized on the left outflow tract view (Fig. 5 - video 4), on which sometimes, as in the case shown, also the hypoplastic pulmonary artery is displayed.

Figure 5: Pulmonary Atresia + Ventricular Septal Defect. On the left outflow tract view, it is possible to demonstrate the malalignment ventricular septal defect with the overriding aorta. It is also shown, just behind the aorta, the small, atretic pulmonary artery (arrow) - See also video 4. (Ao: ascending aorta; LV: left ventricle; Pa: main pulmonary artery; RV: right ventricle)

When the right and left pulmonary arteries are present, they are commonly smaller than normal and confluent, with the characteristic appearance of a ‘flying seagull’, and their size is usually dependent on the source of arterial supply. The pulmonary vascular bed may be supplied with blood flow from a ductus arteriosus, from MAPCAs, or from a combination of both. Color Doppler can be used to confirm overriding of the aorta (see Fig. 2 and video 2), and to demonstrate the retrograde blood flow across the ductus arteriosus. The use of color Doppler or power Doppler is also important to demonstrate the presence of MAPCAs branching off the descending thoracic aorta. Spectral Doppler has a limited diagnostic role to play in PAVSD. 4D echocardiography has recently been demonstrated to be very helpful in the definition of the vascularization pattern of the pulmonary arteries [2, 9]. In particular, the use of B-flow imaging and/or the inversion mode can demonstrate the confluent pulmonary arteries and the MAPCAs better than 2D ultrasound. As for the the differential diagnosis, it should be underlined that differentiating PAVSD from CAT can be challenging. CAT types II and III and PAVSD share the reduced dimensions of the pulmonary branches and the prevalence of the aortic vessel. When doubts arise, the following anatomic details should be sought to make the final diagnosis: the aortic/truncal valve is always dysplastic (from two to five cusps) and typically stenotic and/or insufficient in CAT, not in PAVSD; the direction of flow within the arterial duct is reversed in PAVSD, anterograde in CAT.

Prognosis and survival. The risk of association with chromosomal anomalies is high for PAVSD, which shows a 29% association rate with the 22q11 microdeletion and, to a much lower extent with trisomies 13 and 18 [7]. Therefore, karyotyping with FISH analysis for the DGCR on chromosome 22 is mandatory. Delivery should take place in a tertiary referral center to allow proper neonatal management, being the pulmonary circulation ductus-dependent.

The main prognostic factors for PAVSD are represented by the anatomy of the pulmonary arteries and by the sources of the pulmonary blood supply. The most favorable arrangement, from a surgical point of view, is that in which the two pulmonary branches are confluent and are supplied by the arterial duct. On the contrary, the complete absence of the central pulmonary arteries, with the lungs being directly supplied by multiple MAPCAs, represents the worst scenario, being associated with a significantly worse prognosis; also, it is the most difficult to treat postnatally.
COMMON ARTERIAL TRUNK (CAT)

Anatomy. Common arterial trunk (CAT) is characterized by a single arterial trunk arising from the base of the heart, which supplies the systemic, coronary, and pulmonary blood flow. CAT results from a septation failure during development of the ventricular outlets and the proximal arterial segment of the heart tube. The earliest classification, developed by Collett and Edwards [10], includes four different anatomical subtypes with respect to the origin of the pulmonary arteries: in type I, a short main pulmonary trunk arising from the common arterial trunk which gives rise to right and left pulmonary arteries (48-68% of cases); In types II and III, the pulmonary trunk is absent and the two pulmonary branches arise close to one another (29-48% of cases - type II) or distant one from the other (6-10% of cases - type III). Type IV is currently defined as PAVSD.

Ultrasound diagnosis. In CAT, the 4-chamber view is usually unremarkable, being a slight increase of the cardiac axis noted in a minority of cases. On the left outflow tract view, the constant malalignment VSD and the usually abnormal truncal valve are seen (fig.6). Sometimes, the truncus may straddle one ventricle, especially if there is dominance of one ventricle [11]. Visualization of the pulmonary arteries is essential to distinguish CAT from PAVSD and to identify the CAT subtype (fig.6).

![Figure 6: Common arterial trunk, type I.](image)

In the case of PAVSD, the hypoplastic pulmonary arteries are supplied by the ductus arteriosus and/or by the major aorto-pulmonary collateral arteries (MAPCAs). As previously mentioned, in type I CAT, the main pulmonary trunk arises from the posterolateral aspect of the common trunk and bifurcates into two pulmonary arteries (fig.6). In types II and III, the pulmonary arteries arise separately. The truncal valve is often dysplastic, and may be regurgitant or stenotic. Color Doppler may be used and may help to evaluate the ventriculo-arterial connection (fig.7), checking for possible steno-insufficiency of the truncal valve, and to trace the course and connection of the pulmonary trunk/arteries.

![Figure 7: Common arterial trunk, type I.](image)
The use of 4D echocardiography is extremely useful in the identification of the small pulmonary branches in type II/III CAT (fig.8) and in the characterization of the pulmonary trunk anatomy in PAVSD (especially with inversion mode and B-flow) [2].

Figure 8: Common arterial trunk type II (both pulmonary arteries branching off at the same distance from the valve). On 4D-echocardiography, it is possible to demonstrate the two pulmonary branches directly departing from the arterial trunk (arrowheads). This diagnosis is extremely challenging on two-dimensional echocardiography and is much simpler and straightforward using 4D-echocardiography. (aa: aortic arch; LV: left ventricle; RV: right ventricle; T: truncus arteriosus)

Prognosis and survival. CAT can be associated with cardiac and extracardiac abnormalities in a significant percentage of cases, and some of these may have an impact on management and outcome. Associated cardiac defects, which occur in 20-30% of cases, include absence of ductus arteriosus (50% of cases), interruption of the aortic arch or right aortic arch, and atrioventricular valve atresia [11]. The risk of association with chromosomal anomalies is significant, in the range of 5-10%, but what is extremely high is the risk of association with the microdeletion 22q11, which is present in 30-40% of the cases both in prenatal and in postnatal series [11]. Therefore, karyotyping with FISH analysis for the DGCR is mandatory. Delivery should take place in a tertiary referral center, so that the neonate can be transferred to a pediatric cardiology unit to confirm the diagnosis and receive an adequate management.

The final prognosis depends on on the presence of extracardiac and chromosomal anomalies and of unfavorable cardiac anatomy (e.g. severe truncal valve regurgitation, IAA, and straddling with ventricular hypoplasia). Immunodeficiency in case of an underlying DiGeorge syndrome is also an issue.

DOUBLE-OUTLET RIGHT VENTRICLE (DORV)

Anatomy. DORV encloses a wide range of lesions characterized by a double right ventriculo-arterial connection that may have completely different hemodynamic characteristics. The main feature of DORV is that both great vessels arise for more than 50% from the same ventricle and that the spatial relationship of the two arteries may vary extensively. They can show a normal relationship, with the aorta posterior and the pulmonary artery anterior and to the left, or they can be malposed, with the aorta arising anteriorly, behind the sternum, and the pulmonary trunk posterior above the VSD. The VSD position and commitment is also variable: non-committed, sub-pulmonary, subaortic, doubly committed. Besides, there may be an obstruction of the pulmonary outflow or, much less frequently, the aortic outflow, due to pulmonary stenosis/atroresia or aortic coarctation. The most frequent variants are the Fallot type (subaortic VSD, great vessels in normal spatial relationship, and pulmonary artery obstruction), the Taussig-Bing type (subpulmonary VSD and malposed great arteries), and the type with subaortic VSD but without pulmonary stenosis. In many cases, other major cardiac defects such as ventricular hypoplasia (almost always due to straddling and overriding of one of the two atrioventricular valves), aortic coarctation, AVSD, and cardioplenic syndromes can be associated. Ultrasound diagnosis. It is important to underline that, unless severe anomalies of the atrioventricular junction are associated, the 4-chamber view is unremarkable, as in most conotruncal anomalies. The double right ventriculo-arterial connection is detected on the outflow tract views: the crossover is missing and the great vessels run parallel and arise from the anterior ventricle in normal relationship or malposed (Fig. 9 - video 5).
Figure 9: Double Outlet Right Ventricle (DORV). On the outflow tracts’ view, it is possible to demonstrate the absence of the crossover, with the two great arteries coursing parallel one to the other. The arteries are also malposed, with the aorta anterior and the stenotic pulmonary artery posterior (arrow). The small sub-pulmonary ventricular septal defect is also evident. See also video 5 (Ao: ascending aorta; LV: left ventricle; Pa: main pulmonary artery; RV: right ventricle)

The use of color Doppler may facilitate the assessment of the spatial relationship of the great vessels (Fig. 10).

Figure 10: Double Outlet Right Ventricle (DORV). On the right outflow tract view, using color Doppler it is possible to demonstrate again the absence of the crossover and the two parallel vessels. In this case, there was no malposition, and the aorta arises normally from the central part of the heart. (Ao: ascending aorta; Pa: main pulmonary artery; RV: right ventricle)

Noteworthy, the diagnosis of outflow obstruction in DORV is based on comparison of the vessel size rather than on increased transvalvular velocity. Very important is the definition of the spatial relationship of the great vessels because it identifies the type of surgical approach required, and therefore the prognosis. Also, DORV may change through the course of pregnancy: the degree of pulmonary outflow obstruction can worsen significantly in the 3rd trimester, and, if one of the atrioventricular valves show straddling or overriding, ventricular hypoplasia can develop. 4D echocardiography may be used to confirm the double ventriculo-arterial connection and the absence of the crossover [2], but these diagnoses can be effectively made on 2D also.

Prognosis and survival. From the prognostic standpoint, if DORV is not associated with extracardiac or chromosomal anomalies, the major negative prognostic feature is the presence of other cardiac defects (AVSD, cardiosplenic syndromes, aortic coarctation, and straddling/overriding of the atrioventricular valves with consequent hypoplasia of the underlying ventricle). The general prognosis is very poor if DORV is associated with chromosomal anomalies or syndromic conditions. The risk of association with chromosomal anomalies is very high, in the range of 12-45%, with a prevalence of trisomies 18, 13, and, to a lesser extent, 22q, 11 microdeletion and trisomy 21 [3-5]. Hence, karyotyping is mandatory, especially if extra-cardiac anomalies are found in association. The delivery should take place in a tertiary referral center, so that the neonate can be transferred to a pediatric cardiology unit to confirm the diagnosis and to receive adequate management.

Due to the strong association with aneuploidy and extracardiac anomalies, the overall survival of fetal DORV is 46-50%, if terminations of pregnancy are excluded. However, it is shown that the overall surgical mortality rate for biventricular repair is as low as 13%, with an 86% 10-year survival rate. The mortality is much higher if other major cardiac lesions are associated.
COMPLETE TRANSPOSITION OF THE GREAT ARTERIES (TGA)

Anatomy. Complete transposition of the great arteries (TGA) is determined by the presence of a discordant ventriculo-arterial connection, with the aorta arising from the right ventricle and the pulmonary artery connected to the left ventricle. The pulmonary and systemic circulations function in parallel, rather than in series. This malformation is caused by abnormal formation of the aorto-pulmonary septum. TGA can occur as an isolate anomaly (simple TGA) or be associated with a VSD and/or pulmonary outflow obstruction; less commonly with aortic arch anomalies. It is very rare to find atrioventricular valve abnormalities, including straddling and overriding, associated with TGA. Significant anomalies of the coronary pattern, which are not diagnosable in utero, are associated in two-thirds of the cases. Ultrasound diagnosis. The 4-chamber view is unremarkable, as for most conotruncal anomalies, unless major anomalies of the atrioventricular junction are associated. The diagnosis is made on the outflow tract views, where there is no crossover, with the two arteries following a parallel course. The aorta arises from the anterior right ventricle, and the pulmonary artery is connected posteriorly with the left ventricle (fig 11). In order to detect possible (valvular) obstructions of the pulmonary artery and of the aorta (arch coarctation/interruption), it is important to compare the size of the two vessels. On longitudinal views, the anteriorized connection of the ascending aorta gives the aortic arch a wider angle curvature similar to a “hockey club” and not to the classic “umbrella handle” of the normal arch. In case of TGA with an intact ventricular septum, particular attention should be paid in late gestation to signs possibly indicative of a restrictive foramen ovale, such as consistent bulging into the left atrium and/or thickening, or limited movement of a small foramen ovale flap - ref.12).

Figure 11: Transposition of the Great Arteries (TGA). The left outflow tract view demonstrates also in TGA the absence of the crossover, as in DORV. In this case, however, each vessel is connected with the wrong ventricle: the aorta, anterior, arises from the right ventricle, the pulmonary artery, posterior, from the left ventricle. The type of vessel is identified by showing the two pulmonary branches (arrows) in the pulmonary artery (Ao: ascending aorta; LV: left ventricle; Pa: main pulmonary artery; RV: right ventricle)

Color Doppler may contribute to confirm the ventriculo-arterial discordance, (Fig. 12 - video 6), to recognise small VSDs not evident on greyscale ultrasound, and to detect possible pulmonary/aortic outflow obstruction. 4D echocardiography has been shown to be useful in the assessment of the spatial relationship of the great arteries, deriving the risk of association with coronary abnormalities (video 7) [13].

Figure 12: Transposition of the Great Arteries (TGA). Color Doppler contributes to the assessment of the ventriculo-arterial connection: each vessel is clearly emerging from a different ventricle. See also video 6 and video 7. (Ao: ascending aorta; LV: left ventricle; Pa: main pulmonary artery; RV: right ventricle)
It can also demonstrate very nicely the parallel course of the great vessels (Fig. 13).

![Figure 13: Transposition of the Great Arteries (TGA). 4D-echocardiography nicely demonstrates the absence of crossover and the ventriculo-arterial discordance. In this case, inversion mode rendering was used. (Ao: ascending aorta; LV: left ventricle; Pa: main pulmonary artery; RV: right ventricle)](image)

As to the differential diagnosis, TGA can be distinguished from DORV paying attention to the ventriculo-arterial connection. Prognosis and survival. From the cardiological standpoint, the worst prognostic indicator is the occurrence of significant right outflow obstruction, for this represents a contraindication to the classic arterial switch operation. On the other hand, TGA seems to protect from aneuploidy, like cTGA and cardiosplenic syndromes; and is also exceptionally rare the association with other syndromic conditions. Hence, karyotyping can be avoided, unless clear markers of aneuploidy are associated. Serial follow-up scans are indicated, especially in case of TGA with intact ventricular septum, in order to recognize a restrictive foramen ovale, if present. If this is the case, it is necessary to organise the delivery so that the neonate is transferred to the interventional catheterization room within 30 minutes at the latest, in order to perform a life-saving Rashkind atrioseptostomy. As a consequence, delivery should be organized in a tertiary referral center, alerting the cardiac hemodynamist of the imminent delivery [14]. If there is significant shunting across a VSD, together with a non-restrictive foramen ovale, the neonate may be transferred to the pediatric cardiology unit in the first hours of life if early neonatal management is properly planned, this CHD has a long-term survival rate greater than 90% in good functional conditions. What makes the difference is certainly prenatal diagnosis, being responsible for a 7% reduction in surgical mortality [14]. Patients undergoing palliative surgical procedures (Mustard or Senning) have lower survival rates (80%).

**CORRECTED TRANSPOSITION OF THE GREAT ARTERIES (cTGA)**

Anatomy. cTGA is defined by the association of atrio-ventricular and ventriculo-arterial discordance, with the double discordance functionally correcting the circulation. The left atrium is connected via a tricuspid valve to a morphologically right ventricle from which the aorta emerges and, viceversa, the right atrium is connected via the mitral valve to a morphologically left ventricle from which the pulmonary artery emerges. The discordance between cardiac situs and position is very often associated, with dextrocardia in situs solitus and levocardia in situs inversus. Frequently associated are other major cardiac lesions, including VSDs, abnormalities of the left-sided tricuspid valve, pulmonary outflow obstruction and rhythm disturbances.

Ultrasound diagnosis. Fundamental for the diagnosis of cTGA is the 4-chamber view, on which the atroioventricular discordance is evident (Fig. 14): the morphologically right ventricle is positioned to the left and is connected to the left atrium; conversely, the left ventricle, forming the apex of the heart, is positioned to the right and is connected to the right atrium.
Figure 14: Congenitally corrected transposition of the great arteries (cTGA). On the apical 4-chamber view, it is possible to detect the atrio-ventricular discordance. Note the lower insertion of the left-sided abnormal tricuspid valve (arrow) and the fact that the apex of the heart is made by the ventricle positioned on the right side (morphological left ventricle). (LA: left atrium; mLV: morphological left ventricle; mRV: morphological right ventricle; RA: right atrium)

Other features that may increase the confidence of the diagnosis are the different attachments of the chordae tendinae of the two atrioventricular valves: the left-sided tricuspid valve papillary muscles attach to the ventricular apex, whereas the right-sided mitral valve papillary muscles attach to the lateral free wall (Fig. 15).

Figure 15: Congenitally corrected transposition of the great arteries (cTGA). On the transverse 4-chamber view, it is possible to detect the differential attachments of the chordae tendinae. The left-sided tricuspid ones attach onto the apex of the ventricle (arrowheads), whereas the right-sided mitral ones attach on the lateral myocardial wall (arrow). This is an additional feature supporting the diagnosis of atrio-ventricular discordance. (LA: left atrium; mLV: morphological left ventricle; mRV: morphological right ventricle; RA: right atrium)

Assessment of the outflows demonstrates the pulmonary artery arising from the right-sided left ventricle and the aorta emerging from the left-sided right ventricle. Recognition of dextrocardia in situs solitus should always prompt assessment of central connections to rule out a cTGA. Anomalies of the tricuspid valve, VSDs, pulmonary and aortic outflow obstruction, and complete heart block are frequently associated [15]. The heart block is progressive, very often appearing only in the 3rd trimester or after birth, and is due to the abnormal position of the conduction tissue. Color Doppler may be helpful to distinguish the spatial relationship of the great arteries and in the detection of the VSD. To quantify the transvalvular gradient in the case of pulmonary stenosis it is possible to use spectral Doppler. 4D echocardiography can surely demonstrate the inverted position of the ventricles by inversion-mode or B-flow renderings (Fig. 16, video 8).
Figure 16: Congenitally corrected transposition of the great arteries (cTGA). 4D-Echocardiography confirms both the atrio-ventricular discordance and the differential attachments of the chordae tendinae, with the left-sided tricuspid ones attaching onto the apex of the ventricle (arrowheads) and the right-sided mitral ones attaching on the lateral myocardial wall (arrow). See also video 8, (da: descending aorta; LA: left atrium; mLV: morphological left ventricle; mRV: morphological right ventricle; RA: right atrium)

Prognosis and survival. Ventricular hypoplasia and outflow atresia represent ominous prognostic signs. Scant is the association with other malformations, with minor renal anomalie having been reported in association with cTGA [15]. The risk of association with chromosomal and non-chromosomal syndromic conditions is virtually non-existent, for cTGA seems to protect from aneuploidy, together with TGA and cardiosplenic syndromes. Therefore, fetal karyotyping is not recommended. It is advisable for delivery to take place in a tertiary referral center, especially if there is evident outflow obstruction, major atrioventricular anomalies or congenital heart block with low cardiac output. Postnatal management of cTGA is controversial. Major surgery can be avoided if there are no outflow obstructions or significant valve abnormalities. The real problem is the long term survival. The frequency and severity of the associated cardiac lesions and the conduction system abnormalities represent the most important determinants of survival and mortality. The main indicators of a poor prognosis are a severe insufficiency of the left-sided tricuspid valve and impaired systolic function of the right (systemic) ventricle. By 45 years of age, 67% of individuals with associated lesions showed heart failure, in comparison with 25% of individuals with isolated cTGA [16].

REFERENCES


