Diagnosis of Transposition of the Great Arteries in the Fetus

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Introduction

Transposition of the great arteries (TGA) was first described by Mathew Baillie in 1797 [1]. The term transposition was first used by Farre in 1814, in which “trans” means “across” and “position” means “placement.” Transposition means that the aorta and pulmonary artery are placed across the ventricular septum. A few decades ago, Van Praagh et al suggested that the aberrations in conotruncal development could result in a continuum of malpositions of the great arteries [2]. TGA is simply one subtype of these malpositions. Other subcategories of these malpositions include double-outlet right ventricle, double-outlet left ventricle, and anatomically corrected malposition [3].

The reported prevalence of congenital heart disease (CHD) varies between four and 10 per 1000 live births [4–6]. Due to advances in fetal ultrasound resolution and techniques, obstetricians have the increasing ability to detect fetal CHD prenatally, thereby prompting timely referral for adequate perinatal management. Based on

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a review of the literature, the antenatal diagnosis and transfer of neonates with CHD could improve both short-term and long-term outcomes, especially in those with cyanotic congenital heart diseases [6,7].

In a population-based review, cardiac defects account for almost half of the neonatal deaths attributed to congenital malformations [8]. Tetralogy of Fallot and TGA are the most common cyanotic heart defects, each constituting approximately 10% of fetal congenital heart defects [9–13]. However, in most fetuses, TGA remains undiagnosed before birth. According to a population-based review [14], TGA is diagnosed prenatally in only 17% of affected neonates. In contrast, approximately one-half of cases of tetralogy of Fallot can be correctly diagnosed in utero. We conducted a comprehensive review for the diagnosis of TGA in the fetus because of this low detection rate.

Terminology

TGA as well as other CHD are best diagnosed using the segmental approach of the fetal heart [15–17]. To enhance understanding of the terminology used in the segmental approach for the diagnosis of TGA, the following terms should be clarified in advance [2,3,16,18–20].

Atrioventricular (A-V) discordance: The morphologic right atrium (RA) is erroneously connected to the morphologic left ventricle (LV); and the morphologic left atrium (LA) is incorrectly drained to the morphologic right ventricle (RV).

Ventriculoarterial (V-A) discordance: The pulmonary artery (PA) arises from a morphologic LV, and the aorta (Ao) arises from a morphologic RV.

Complete transposition of the great arteries (TGA): Indicates the conditions of ventriculocardiac discordance. The Ao arises from a morphologic RV, and the PA arises from a morphologic LV. The A-V connection is correct.

Congenitally corrected transposition of great arteries (ccTGA): Refers to the condition of A-V discordance plus V-A discordance. In brief, the RA enters the LV, which gives rise to the PA. The LA connects the RV, which gives rise to the Ao. Thus, the circulation becomes physiologically corrected although double errors occur.

d-loop: Denotes the normal rightward (dextro, d ) loop of the embryonic cardiac tube. The inflow portion of the RV is to the right of the morphologic LV.

l-loop: Denotes the cardiac tube bending leftward (levo, l ) during embryogenesis. The inflow portion of the morphologic RV is to the left of the morphologic LV.

Detecting transposition of the great arteries in the fetus

According to the policy of the Bureau of Health Promotion in the Department of Health of Taiwan, general practitioners should perform routine ultrasound to evaluate fetal growth at 20 weeks' gestation. The diagnosis of TGA can be made by carefully and appropriately evaluating the anatomic locations of cardiac chambers and the connections between the atria, ventricles, and great arteries at this gestational age with high-resolution ultrasound. However, it must be emphasized that diagnostic accuracy is only half based on the literature [21–23]. The gold standard and the additional signs of ultrasound in the diagnosis of fetal TGA are discussed in the following paragraphs.

The localization of cardiac chambers and great arteries

The first step in the diagnosis of fetal TGA is the localization of the cardiac chambers and their connections with the great arteries [22]. This is best done by the segmental approach as described in the literature [15,24–26]. These cardiac structures can be identified on the basis of their specific morphologic features. Anatomically, the differentiation of the atrial chamber is based on the morphologic aspect of the atrial appendages. The atrial appendages are the earlike extensions of the atria. Typically, the RA appendage is triangular in shape, whereas the LA appendage is fingerlike [15]. However, differentiation of atrial appendages is difficult during the antenatal period. In addition, systemic and pulmonary venous connections may provide important clues to define situs of the atria in the fetus. The supradiaphragmatic portion of the inferior vena cava (IVC) provides a reliable landmark to identify the anatomic RA (because of the rule of venoatrial concordance) [15], and the drainages of four pulmonary veins usually define the location of anatomic LA (Fig. 1A and B).

Certain features that could help differentiate the right and left ventricles are the texture and distribution of internal trabeculae. The trabeculae are coarse in the RV, but thin and delicate in the LV (Fig. 1B and 1C). Moreover, the papillary muscles of the RV are attached to both the interventricular septum and the lateral wall, yet the two papillary muscles of the LV are attached only to the lateral wall of myocardium [15,27,28] (Fig. 1C). Nevertheless, these anatomic features may not be apparent in the fetus. The insertion of the tricuspid valve to the ventricular septum is lower than that of the mitral valve, and the location of the moderator band in the apical area identifies the location of the RV (Fig. 1B). Both characteristics are valuable in defining the morphologic RV.

The major characteristic of the PA is that the three branches (right and left PA, ductus arteriosus) immediately emerge from the main PA when it arises from the RV (Fig. 1D). In contrast, the Ao does not branch into the ascending portion except for the coronary arteries. The aortic arch can be seen by positioning the transducer to the left parasagittal plane. In the view, the Ao is seen to arise from the middle of the fetal thorax with acute curvature (usually described as having a ‘candy cane’ appearance). Three branches from the transverse arch can be identified as the innominate, the left common carotid, and the left subclavian arteries (Fig. 1E). The ductal arch can be seen by sliding the transducer to the left from the aortic arch (with a slight tilt of the transducer). In the normal fetus, the ductal arch arises from the anterior thorax with a wide curvature (usually described as having a ‘hockey stick’ appearance). There is no branch in the
Transverse portion of the ductal arch, which can be used to differentiate the aortic arch (Fig. 1F).

‘Gold standard’ of the diagnosis of fetal TGA

The diagnosis standard of fetal TGA is the demonstration of V-A discordance. The Ao arises from the morphologic RV while the PA arises from the morphologic LV [18] (Fig. 2). The diagnosis of V-A discordance can be achieved by rotating the transducer from the four-chamber view to the left ventricular outflow tract view and the right ventricular outflow tract view subsequently. Sometimes these two views can be obtained simultaneously in the same plane. These two views essentially establish the diagnosis of fetal TGA.

After demonstrating V-A discordance, the A-V connection should be inspected to differentiate complete transposition (d-TGA) from congenitally corrected transposition (cCTGA). If V-A discordance combined with a correct A-V connection is noted, the diagnosis is complete transposition of the great arteries (d-loop TGA, or d-TGA). In contrast,
when V-A discordance plus A-V discordance (Fig. 3) is demonstrated, the diagnosis is changed to congenitally corrected transposition of the great arteries (ccTGA; l-loop TGA). These two entities should be differentiated because the clinical manifestation, outcome, and surgical options are totally different.

The additional signs of the diagnosis of fetal TGA

Several additional scanning planes and signs of the diagnosis of fetal TGA have been described in the literature [21,22,29–33]. These additional views, although not essential to the prenatal diagnosis, may help to identify TGA or assist diagnosis in certain conditions such as unfavorable fetal position or diagnostic dilemma between similar pathologies. The identification of fetal TGA may improve perinatal outcome with well-timed transport of the mother to a tertiary center for neonatal cardiovascular assistance [34,35].

1. **Three-vessel and trachea view**: The three-vessel or three-vessel-trachea view is a transverse view of the fetal upper mediastinum [32,36,37]. In this plane, the main PA, Ao, and superior vena cava (plus the cross section of trachea in front of the spine) are consequently demonstrated from the left side (Fig. 4). However, in the fetus with TGA, this anatomic sequence is altered and usually demonstrates a solitary transverse Ao in the same plane. The PA and superior vena cava sometimes may be seen on the same side of the transverse Ao (Fig. 5).

2. **The arch view**: In the fetus with TGA, the aortic arch may be shifted to the anterior fetal thorax, with a ductal arch in its posterior aspect. Both arches may demonstrate a parallel configuration without a normal spiraling relationship (Fig. 6).

3. **The outflow tract view**: In the normal fetus, the great arteries emerging from the bilateral ventricles show a ‘crossover’ relationship. However, in the fetus with TGA, the great arteries are usually aligned in the parallel course, which allows visualization of both great arteries from ventricles in a single scanning plane (Fig. 4). However, not all fetuses with TGA have this parallel relationship of the great arteries.

4. **The ‘big-eye frog’ view** [21]: Using four-dimensional spatiotemporal image correlation technique, the en-face view of the fetal A-V valves could be seen. In this view, the PA is at the 1-2 o’clock direction (left anterior side) of the Ao. However, the anatomic relationship is altered in the fetuses with d-TGA. In those fetuses with TGA, the PA deviates posteriorly to the directions of 3 or 4-5 o’clock direction of Ao (i.e., PA parallel or left posterior to the Ao) (Fig. 7).
Cardiac anomalies associated with TGA

Complete transposition of the great arteries (d-TGA)
Approximately one-half of the fetuses with TGA have associated ventricular septal defect (VSD). The location of VSD could be anywhere, but is most commonly found at the part of the outlet portion with an anterior or posterior malalignment [38]. Anterior malalignment of the outlet septum may result in aortic root stenosis and widening of the pulmonary tract. With a great degree of pulmonary valve overriding the ventricular septum (Fig. 8), TGA may merge into the continuum of the double outlet of the right ventricle. In contrast, when there is posterior malalignment of the outlet septum, pulmonary stenosis with overriding Ao may be seen. In this condition, it may be difficult to distinguish tetralogy of Fallot from TGA [1,7,16,18,39].

Left ventricular outflow tract obstruction (LVOTO) is common in d-TGA due to the posterior malalignment of the outlet septum, resulting in a small PA in comparison to the Ao (Fig. 9 A and B). In contrast, right ventricular outflow tract obstruction (RVOTO) with aortic stenosis is less common in d-TGA. Premature closure of the foramen ovale is relatively common in the fetuses with TGA, probably due
to highly oxygenated blood streaming through the foramen ovale [40] (Fig. 10). When both VSD and the foramen ovale are closed or nearly closed, the affected neonates may require intensive care (including extracorporeal membrane oxygenation) immediately after birth, and early surgery. Complete transposition of the great arteries may often combine with atrial isomerism, common A-V canal, and double outlet of RV.

Congenitally corrected transposition of the great arteries (ccTGA)

Complete transposition of the great arteries is often an isolated lesion. In contrast, 90% of fetuses with ccTGA will have other cardiac malformations. Approximately 70% of affected fetuses have VSD, mostly in the perimembranous portion. LVOTO and tricuspid stenosis occur in 40% and 30% of fetuses with TGA, respectively. Bradycardia and complete A-V block were occasionally noted.

Genetic counseling of TGA diagnosed in the fetus

The morphogenesis of the fetal heart concerns complex developmental changes in the first few weeks of embryonic life. Many chromosomal and genetic factors may contribute to the development of congenital cardiac defects. Based on a population-based case-control study, the prevalence of chromosome aberrations in fetuses with CHD is approximately 13% [41]. Most of these aberrations are trisomies 21, 18, and 13. However, the incidence of chromosome anomaly in fetuses with TGA is 0.9% [42], which is identical to that of the normal population. DiGeorge syndrome results from the microdeletion of chromosome 22q11, which occurs in 1 per 4000 live births [43]. Phenotypic manifestations include cardiac defects, characteristic facies, and thymic hypoplasia. This condition reportedly is associated with tetralogy of Fallot or other conotruncal malformations. However, it rarely occurs in the fetus with TGA. Extracardiac defects usually are associated with other congenital cardiac defects, but the incidence in TGA is low. Approximately 9% of neonates with TGA are reported to have associated extracardiac malformations, and no specific type of malformation is described [42].

References


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