

Truncus arteriosus

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Abstract

Truncus arteriosus (TA) is an uncommon congenital cardiovascular anomaly, characterized by a single arterial trunk arising from the heart by means of a single semilunar valve (i.e. truncal valve). Pulmonary arteries originate from the common arterial trunk distal to the coronary arteries and proximal to the first brachiocephalic branch of the aortic arch. TA typically overrides a large outlet ventricular septal defect (VSD). Intracardiac anatomy usually reveals situs solitus and atrioventricular (AV) concordance. Prevalence ranges from 0.03 to 0.056 per 1,000 live births. No striking sex difference in frequency is observed although most series contain more males than females. Neonates with TA present with clinical features of congestive heart failure depending on the high volume of pulmonary blood flow and the presence or absence of truncal valve insufficiency. Symptoms of failure manifest as falls in pulmonary resistance and increases in pulmonary overcirculation. Tachypnea, tachycardia, excessive sweating, poor feeding may be the first signs to appear. In the last 10 to 15 years there have been clinically significant improvements in treatment with early repair and now TA is ideally repaired in the neonatal period with low morbidity and mortality (5%) in selected series. Currently surgical management consists of complete repair with closure of VSD. Etiology of TA remains unknown. In experimental animal models, TA has been linked to abnormal development of cells from the neural crest that are normally located in the outflow region of the developing heart; this is thought to be an important etiologic factor in some cases of human TA also. Prenatal detection of truncus arteriosus by ultrasound is documented.

Keywords

Common arterial trunk, cardiovascular anomaly, ventricular septal defect, congestive heart failure, echocardiography, band 22q11 deletion

Disease name and synonyms

Truncus arteriosus (TA)

Common arterial trunk

Common aortico-pulmonary trunk

Excluded disease

Sepsis in neonatal period

Definition and diagnostic criteria

TA is an uncommon congenital cardiovascular anomaly, characterized by a single arterial trunk arising from the heart by means of a single semilunar valve (*i.e.* truncal valve). Pulmonary arteries originate from the common arterial trunk distal to the coronary arteries and proximal to the first brachiocephalic branch of the aortic arch. TA typically overrides a large outlet ventricular septal defect (VSD). Pulmonary arteries may arise from the common trunk in one of several patterns which are often used to classify subtypes of TA.

The earliest classification was developed by Collet and Edwards in 1948 (1). The commonly-used classification proposed by Van Praagh in 1965 (2), includes four primary types TA (see table 1)

Table 1: The four primary types of truncus arteriosus

Types	Description
Type 1	It is characterized by the origin of a partially separate main pulmonary trunk from the lateral aspect of the common trunk because of the presence of incomplete aortico-pulmonary septum.
Type 2	The pulmonary arteries originate separately from the common trunk.
Type 3	It is characterized by absence of one pulmonary artery from the ascending aorta. The other pulmonary artery can originate from the ductus arteriosus or directly from descending aorta (major aortopulmonary collateral artery or MAPCA).
Type 4	It is defined not by the origin of the pulmonary arterial branch but rather by the coexistence of an interrupted aortic arch or aortic hypoplasia or preductal aortic coarctation. In these cases a well-documented correlation with Di George syndrome is observed (3).

Intracardiac anatomy usually reveals situs solitus and atrioventricular (AV) concordance. Two balanced ventricles are generally present and are separated by a large VSD. Nevertheless, TA has been described in cases with asplenia (4), tricuspid atresia and double inlet ventricle with hypoplastic left ventricle (5, 6). A very rare form is TA with discordant AV connection (7).

The truncal valve may be:

- tricuspid in 61% of cases
- quadricuspid in 31% of cases
- bicuspid in 8% of cases (Van Praagh).

It may be stenotic or incompetent or both and the leaflets are usually thickened.

Intrinsic stenosis, hypoplasia of one or both pulmonary arterial branch(es) may be present which may influence management and outcome. Proximal coronary arteries are abnormal in many patients, with a single coronary artery and an intramural course as the most important variations.

Differential diagnosis

In cases of TA and large pulmonary blood flow, differential diagnosis includes the other congenital heart diseases causing early heart failure with mild or absent cyanosis:

- [VSD](#),
- [Patent ductus arteriosus](#) (PDA),
- Aorto-pulmonary window,
- AV fistulas,
- Pulmonary artery with VSD,
- Total anomalous pulmonary venous connection, etc.

However, thanks to increasing experience, echocardiography allows diagnosis of TA to be established.

Frequency

TA is an uncommon cardiac anomaly with a prevalence ranging from 0,03 (8) to 0,056 (9) per 1,000 live births.

TA represents 0,7% (10) to 1,4% (8) of all congenital heart diseases.

Prevalence of TA is thought to be higher in pregnancies complicated by maternal diabetes (11). Among aborted fetuses and stillborn infants with cardiovascular anomalies, TA represents almost 5% of defects.

No striking sex difference in frequency is observed although most series refer more males than females.

Clinical description

Neonates with TA present with clinical features of congestive heart failure depending on the high volume of pulmonary blood flow and the presence or absence of truncal valve insufficiency.

Symptoms and signs of congestive heart failure are probably more common findings than cyanosis in patients presenting early in life. Symptoms of failure manifest as falls in pulmonary resistance and increases in pulmonary overcirculation. Tachypnea, tachycardia, excessive sweating, poor feeding may be the first signs to appear. Pulse pressure is usually increased and the heart is hyperperactive; a left precordial bulge and a systolic thrill may be noted; the first heart sound is normal, a loud ejection click is present, the second heart is loud and single.

Continuous murmur is rare (suggestive of pulmonary artery stenosis), then pulmonary atresia with VSD or PDA or MAPCAS must be excluded.

ECG is generally non specific; combined ventricular hypertrophy and left atrial enlargement are often seen. Chest X-rays show cardiomegaly, increased pulmonary blood flow and an abnormally high origin of the left pulmonary artery.

Management and treatment

High mortality rate is noted in patients with untreated TA in the neonatal period or early infancy. About 65% of patients who were treated only medically did not survive beyond 6 months of life and more than 90% died before 1 year of age (8). The first corrective operation was performed in 1967 by Mc Goon. In the last 10 to 15 years there have been clinically significant improvements with early repair and now TA is ideally repaired in the neonatal period with low morbidity and mortality (5%) in selected series (12).

Currently surgical management consists of complete repair with closure of VSD, committing the common arterial trunk to the left ventricle and reconstruction of the right ventricular outflow tract.

In patients with both branch pulmonary arteries arising from the common trunk, right outflow tract reconstruction can be obtained by placing a valved conduit: a cryo-preserved valved aortic or pulmonary allograft or a synthetic tube graft or autologous flap.

In patients with one pulmonary artery arising from the common trunk and one underside of the aortic arch, the pulmonary arteries are disconnected separately and anastomosed together and then to the conduit or anastomosed to the conduit.

Operative factors significantly associated with poorer survival over time were operative weight of 2,5 kg or less and truncal valve replacement (12).

Coexisting anomalies are repaired when appropriate with cardiopulmonary bypass and sometimes deep hypothermic arrest.

Published reports have documented mortality rates ranging from 4 to 5% with mean age at repair as low as 11 days, in selected series in absence of associated anomalies (12).

The physiological basis for improved outcomes with earlier repair is the avoidance of damaging sequelae of pulmonary overcirculation and heart failure.

Improved operative outcomes have also been achieved with:

- 1) aggressive truncal valve repair versus replacement in the presence of truncal valve dysfunction,
- 2) right ventricular outflow tract reconstructive techniques that are patient-specific.

In a recent series of 60 patients by Brown (13) right ventricle – pulmonary artery continuity has been established by aortic homograft, porcine valve conduit, direct anastomosis and glutaraldehyde-treated bovine jugular vein valved conduit (Contegra). In his paper Breymann (14) reported no signs of conduit or

valve degeneration during a follow-up up to 27 months.

Associated cardiac lesions, like severe truncal valve regurgitation, interrupted aortic arch or coronary arteries anomalies, were risk factors for death after TA repair, with only 71% of survival (13). In addition, heightened awareness of anomalies of coronary artery ostial location, angle of take off and degree of patency can result in avoidance of inadvertent injury to the coronary artery and associated myocardial insult.

Follow-up

Freedom from reoperation for right ventricle outflow tract obstruction was 64% at 7 years in cases without risk factor and 36% at 10 years in patients surviving with risk factors (13).

Although late mortality among patients undergoing early repair is minimal, it can be due to the problems related to right ventricular outflow tract reconstruction (conduit replacement, revision or dilation).

The reported actual survival rate among all hospital survivors was 90% at 5 years, 85% at 10 years and 83% at 15 years (15). Significant independent risk factor for poorer long-term survival was truncal valve insufficiency before repair (16).

The 10-year freedom from reoperation and survival, respectively, was 54% and 71%, respectively, as reported by Monro (17).

Etiology

Like most forms of congenital heart disease, the causes of TA are not known.

In experimental animal models, TA has been linked to abnormal development of cells from the neural crest that are normally located in the outflow region of the developing heart; this is thought to be an important etiologic factor in some cases of human TA also. Similarly to other congenital cardiac anomalies of conotruncal region, patients with TA have frequently associated (approximately 35-40%) microdeletion within chromosome band 22q11.2 which is thought to affect migration or development of cardiac neural crest cells. Patients with anomalies of branch pulmonary arteries may have a higher incidence of association with band 22q11 deletion (18). Nevertheless, a causal association has not been established. Some reports found increased incidence of TA in children of mothers with significant diabetes mellitus during pregnancy.

In animal models, certain teratogens (retinoic acid, bisdiamine...) have been found to predispose patients to TA, but no evidence in humans has been reported. [Di George syndrome](#) (also called velocardiofacial syndrome, often included together as variations

of CATCH 22 syndrome) is present in approximately 30-35% of patients with TA; most of these patients have deletions in band 22q11 (3).

The majority of non cardiac anomalies in patients with TA are those found in association with CATCH 22 syndrome such as velopharyngeal insufficiency, cleft palate and thymic and parathyroid dysfunction.

Diagnostic methods

Most neonates with uncomplicated TA may be referred to surgery relying on the clinical and echocardiographic examination. In many papers the classic echocardiographic features of TA have been widely documented (19, 20, 21).

Using subcostal coronal and parasternal long-axis images is the best way to visualize the single arterial trunk arising from the ventricles, with variable override the ventricular septum.

Morphology of the truncal valve and origin of proximal coronary arteries are best observed from the parasternal short axis view.

Doppler color imaging from these windows is critical to evaluate pulmonary arteries and the regurgitation or stenosis of the truncal valve.

A hemodynamic study is indicated in cases with associated anomalies and when it is necessary to have more details (or for specific anatomic indications) such as:

- Ventricular anatomy, if echocardiography-features suggest atrioventricular septal defect or undeveloped right or left ventricle.
- Pulmonary artery anatomy: when only one pulmonary artery is visualized.
- Coronary artery anomalies: distribution and origin.
- A hemodynamic study is also indicated in older infants or children where pulmonary vascular obstructive disease is being considered.

Genetic counseling

Since band 22q11 deletion is present in approximately one third of patients with TA, genetic counseling can be offered for some of these patients.

Antenatal diagnosis

Prenatal detection of truncus arteriosus by ultrasound is documented (21,22). On screening obstetric ultrasound, four-chamber and great vessel views are sufficient to detect cardiac anomalies. In such an event the parents should be referred for fetal echocardiography which allows more accurate definition of the anatomy of TA (23).

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