Transposition of the great arteries

Synonyms: TGA, complete transposition of the great arteries, d-TGA, simple transposition, ventriculo-arterial discordance.

The aorta and pulmonary artery are transposed so that the aorta arises from the morphological right ventricle and the pulmonary artery arises from the morphological left ventricle. In the most common form called complete transposition, the atria are normal in position (situs solitus of atria), there is atrioventricular concordance (right atrium connected to right ventricle and left atrium connected to left ventricle), d loop of the ventricles (right ventricle on the right and left ventricle on the left), ventriculo-arterial discordance (aorta arising from the right ventricle and pulmonary artery from the left ventricle) and the aortic valve is located to the right of pulmonary valve. This is dextro-TGA(D-TGA). There is a rarer levo-TGA (L-TGA).

The systemic venous blood returning to the right atrium is pumped back via the right ventricle and aorta into the body while the pulmonary venous blood returning to the left atrium is pumped back via the left ventricle and pulmonary artery into the lungs. This results in the systemic and pulmonary circulations becoming parallel instead of the normal in-series circulation. Survival of the infant depends on mixing of blood which can only happen if there are intercirculatory shunts such as an atrial septal defect (ASD), ventricular septal defect (VSD) or patent ductus artery (PDA).

The aetiology is unknown but familial patterns of recurrence suggest a polygenic cause and research has identified a susceptibility locus on chromosome 3p14.3 near WNT5A. Data support a causal role for WMT5A.[1] The recurrence risk in first-degree relatives of patients with transposition of the great arteries (TGA) is low compared to other conotruncal defects.[2]

Transposition is often associated with other heart defects - eg, VSD, left ventricular outflow obstruction, ASD, PDA. The presence or absence of associated cardiac anomalies determines the presentation and management.

Transposition of the great arteries epidemiology[3]

- Transposition is the most common cyanotic congenital heart lesion presenting in the neonate. It constitutes 3% of all congenital heart disease (CHD) and 20% of all neonatal cyanotic CHD.
- The overall annual incidence is 1 in 4,000 live births.
- It is more common in males than in females, with a ratio of about 3:1.
- Maternal factors associated with an increased risk include rubella or other viral illness during pregnancy, alcoholism, maternal age over 40 and diabetes.
- Transposition is rarely associated with syndromes or extracardiac malformations.

Pathology

There are three pathological or anatomical types described which determine the clinical presentation:

1. TGA with intact ventricular septum.
2. TGA with VSD.
3. TGA with VSD and pulmonary stenosis (PS).

Transposition of the great arteries symptoms (presentation) [4]

Asymptomatic cases surviving to adulthood are reported.[5]

Symptoms

- Infants with TGA with intact septum usually present with cyanosis within the first week of life which may be obvious in some soon after birth. They may otherwise be well and asymptomatic initially although with time they become tachypnoeic and develop respiratory distress. If they are not treated they develop metabolic acidosis and become severely ill.

- Those with a large VSD may not be diagnosed until several weeks of age. They usually present with symptoms of congestive heart failure (tachypnoea, tachycardia, sweating and poor feeding) between 4-8 weeks of life. Cyanosis is often minimal.
- If there is a VSD and pulmonary stenosis, the presentation can vary depending on the severity of PS. If the PS is severe, the presentation is similar to that of an infant with Fallot's tetralogy. However, those with poor mixing of blood can present early with cyanosis similar to TGA with intact septum and some with mild PS may present late with heart failure.

Signs

- Babies with TGA with intact septum are usually very cyanosed but without distress until severe hypoxaemia and acidosis develop. The S2 is single and loud and there are no audible murmurs.
- In patients with a VSD there may be a systolic murmur, which increases in intensity as the pulmonary vascular resistance falls.
- An ejection systolic murmur is usually present in those with PS.

Differential diagnosis

- Non-cardiac causes of a severely ill neonate - eg, infection, respiratory problems (such as infant respiratory distress syndrome, meconium aspiration, pneumothorax, pneumonia, congenital diaphragmatic hernia).
- Other causes of CHD, especially:
    - Pulmonary atresia.
    - Fallot's tetralogy.
    - Total anomalous pulmonary venous connection.
    - Tricuspid atresia.
    - Truncus arteriosus.

Diagnosis in fetal life

- Antenatal diagnosis of TGA results in better clinical status before surgery and improved postoperative outcome.[6]
- The prenatal detection rate of TGA on antenatal ultrasound has improved with inclusion of additional outlet views but still remains below 50%.[7]
- Children diagnosed antenatally have improved cognitive skills when compared with those diagnosed postnatally in whom pre-operative acidosis and profound hypoxaemia are more common.[8]
- Once fetal diagnosis has been made, a multidisciplinary team approach ensuring that the delivery is carried out in a unit with ability to provide immediate and corrective management is important.

Investigations

- Pulse oximetry done on day 1 of life is likely to show low saturations suggesting the possibility of cyanotic heart disease. Pulse oximetry screening has been shown to minimise the risk of discharging infants with transposition.[9]
- CXR may appear normal in newborns with TGA and intact ventricular septum but may demonstrate the classic 'egg on a string' appearance (heart is slightly enlarged and appears like an egg lying on its side, narrow vascular pedicle because aorta and pulmonary artery lie one in front of the other and the thymic shadow involutes rapidly) and increased vascular lung markings.
- With an associated VSD, the CXR usually shows cardiomegaly with increased pulmonary arterial vascular markings.
- ECG in a neonate with TGA and intact septum may be normal with the usual right ventricular dominance seen at this age. After a few days right ventricular hypertrophy (RVH) is usually present and additionally right atrial hypertrophy may be present in some. In those with a large VSD biventricular hypertrophy may be seen.
- Echocardiography (two-dimensional and colour Doppler) usually provides all the anatomical and functional information needed for diagnosis and management of these babies.

- With advances in echocardiography cardiac catheterisation is usually not needed for diagnostic purposes but may be necessary to ascertain the origins of the coronary arteries.[10] MRI has no routine role.

Transposition of the great arteries treatment and management

Careful preoperative management is required to minimise the time spent in a hypoxic state.[11] Timed delivery and careful measurement of tissue oxygenation is suggested.

- Once cyanotic heart disease is suspected in a neonate treatment to maintain ductal patency should be started immediately in the form of prostaglandin infusion. Delay in commencing prostaglandins while waiting for echocardiographic confirmation of diagnosis is unacceptable.[12]

- Prompt transfer to a cardiac centre should be arranged, especially for the severely acidotic and cyanosed neonate because most will require urgent atrial septostomy. Not all babies need to be ventilated for transfer. Those who are clinically stable on a prostaglandin infusion rate of less than 15 micrograms/kg/minute may be transported safely without mechanical ventilation.[13]

- Balloon atrial septostomy (BAS) which was developed by Rashkind nearly half a century ago remains an important part of the management of babies with TGA with intact ventricular septum.[14] It improves mixing of blood at the atrial level by creating or enhancing an atrial communication. BAS may be associated with vascular trauma, atrial arrhythmias, atrial perforation and tamponade.

Surgical

- The definitive corrective procedure is the **arterial switch operation**, which has replaced the previous procedures (Mustard or Senning) focused on achieving a physiological rather than an anatomical correction of circulation in TGA.[15]

- Most full-term neonates with uncomplicated TGA can undergo an arterial switch operation (ASO) as a single operation, with minimal mortality. It has been traditional to wait for several days before performing the procedure but increasingly the procedure is performed at an earlier age. A recent study supports 3 days of age as the ideal time for an ASO.[16]
- Data confirming that ASO produces excellent long-term results in TGA with low mortality and morbidity and confirming it as the procedure of choice are emerging.[17]
- The alternative is **Rastelli procedure** which is indicated in patients presenting with D-TGA, a large VSD, and pulmonary stenosis.[3] During this procedure, the VSD is closed using a baffle so oxygenated blood from the left ventricle is directed into the aorta. A conduit is then placed from the right ventricle to the pulmonary artery to shunt deoxygenated blood into the pulmonary artery.

Complications and prognosis[18]

- Survival into adulthood after ASO is common with a 15-20-year survival rate of 90%, and 95% in some studies.[3] However, this is not always trouble free and several long-term consequences are recognised. These include:
  - Neopulmonary stenosis.
  - Neoaortic regurgitation.
  - Neoaortic root dilatation.
  - Coronary artery disease.
- Anywhere between 2% to 8% of patients may require intervention, including balloon angioplasty, transcatheter stenting or surgical patch arterioplasty.
- Obstructed coronary arteries are present in 5% to 7% of survivors and remain the most common cause of morbidity and mortality after ASO. The incidence of myocardial ischaemia, infarction and death is highest in the first three months after ASO.

- The incidence of sudden cardiac death in repaired TGA patients is reported to be between 0.3% to 0.8%. This is thought to be related to primary arrhythmia, myocardial ischaemia or myocardial infarction and most occur one to five years after ASO.
- There is a high incidence frequency of neurodevelopmental (ND) abnormalities in these patients. All TGA patients should have ND evaluation ideally in early childhood.[19]
- Low gestational age and a high pre-operative lactate are the most important predictors of poor developmental outcome.[6]
- Echocardiography follow-up after five years reports changes in morphology and function, particularly in the right ventricle; globular form and decreased systolic function.[20]

Further reading

- Pizula J, Devera J, Ng TMH, et al; Outcome of Pregnancy in Women With D-Transposition of the Great Arteries: A Systematic Review. J Am Heart Assoc. 2022 Dec 6;11(23):e026862. doi: 10.1161/JAHA.122.026862. Epub 2022 Nov 29.
- Silversides CK, Roche SL; Congenitally Corrected Transposition of the Great Arteries: Untangling the Mechanisms of Right Ventricular Dysfunction. JACC Cardiovasc Imaging. 2022 Apr;15(4):575-577. doi: 10.1016/j.jcmg.2021.12.002. Epub 2022 Feb 16.

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