*Leading Article*

# Congenital heart disease: When to act and what to do?

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Congenital heart disease has an incidence of 8 per 1000 live births[1]and contributes significantly to childhood morbidity and mortality. Therefore, basic understanding on pathophysiology of different lesions and management options is vital for a practicing paediatrician.

**Anatomical diagnosis or physiological diagnosis**

For a physician the most important aspect is physiological diagnosis as numerous anatomical diagnoses will lead to only few physiological types and medical management is based on physiological effects.

Congenital heart disease can be categorized into two basic types, acyanotic and cyanotic. Acyanotic lesions can be further categorized into shunts and obstructions whereas cyanotic lesions can be divided into Tetralogy of Fallot (TOF) physiology, Transposition of Great Arteries (TGA) physiology and admixture physiology.

**Shunt lesions**

Shunts can be further divided into atrial septal defect (ASD), ventricular septal defect (VSD) and patent ductus arteriosus (PDA). Even though these are three anatomical lesions, they are better considered as physiologies because there are many other anatomical entities which have similar physiological effects.

In ASD there is left to right shunt before the tricuspid valve and if the shunt is significant (Qp:Qs >1.5-2) it can lead to volume overload of right atrium and ventricle. Because of high flow of blood through the tricuspid valve there will be a mid diastolic murmur in the tricuspid area. As the pressure differences between the left and right atria are low, the shunt is relatively less, there is no pressure transmission to pulmonary circulation and damage to pulmonary vasculature is less. They are usually not symptomatic and are mostly detected on careful clinical examination. Ostium secundum ASD, sinus venosus ASD, ostium primum ASD and partial anomalous pulmonary venous drainage fall into this physiology.

VSD is a shunt which occurs after tricuspid valve but before semilunar valves. This is a high pressure shunt as left ventricular pressure is significantly higher than the right ventricular pressure. The shunt occurs during systole and leads to volume overloading of left atrium and left ventricle. In addition to pansystolic murmur due to shunt across the VSD, if the shunt is significant, there will be a mid diastolic murmur in mitral area due to high flow through the mitral valve. All types of ventricular septal defects, univentricular heart without pulmonary stenosis and double outlet right ventricle with subaortic VSD, TOF with minimal pulmonary stenosis are some anatomical entities which fall under this physiology.

PDA is also a post tricuspid shunt but the shunt occurs after the semilunar valves as well. Therefore, shunting occurs during systole as well as during diastole because aortic pressure is much higher than pulmonary artery pressure during the whole cardiac cycle. This leads to a significant shunt with volume overloading of left atrium and left ventricle. There is a continuous murmur in the left subclavicular area and a mid diastolic murmur in the mitral area if the shunt is significant. Aorto-pulmonary window also falls in to this physiology.

**Obstructive lesions**

Obstructive lesions can be divided into left sided obstructions and right sided obstructions. Left sided obstructions will compromise systemic perfusion and right sided obstructions will compromise pulmonary blood flow. Common right sided obstructive lesions are pulmonary stenosis or atresia and common left sided obstructions are aortic stenosis and coarctation of aorta. If the obstruction is severe, distal perfusion is usually maintained through the PDA which is called duct dependent systemic or pulmonary

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circulation depending on the compromised system. Therefore, in these children maintaining the patency of the ductus arteriosus is important for survival. If the PDA closes in a patient with duct dependent pulmonary circulation the patient will become severely cyanosed and will eventually become acidotic due to hypoxia. If it is a duct dependent systemic circulation the patient will maintain saturation but will eventually become acidotic due to poor perfusion. Heart failure in obstructive lesions is very rare and is associated with critical stenosis. It is a medical emergency and after initial stabilization the patient should be immediately subjected to either transcatheter or surgical correction.

**Cyanotic Lesions**

Cyanotic lesions can be divided into three basic physiologies; TOF, TGA and admixture.

**TOF physiology**

In this, the most important determinant is the degree of obstruction to pulmonary blood flow. If it is severe, there will be less blood flow into pulmonary circulation resulting in lesser volume of oxygenated blood entering into left atrium. As there is a non restrictive VSD, depending on the degree of pulmonary obstruction, a volume of deoxygenated blood from right ventricle will flow through the VSD into left ventricle and systemic circulation. Ultimate saturation will be determined by the proportions of deoxygenated and oxygenated blood that flows into left ventricle and then into systemic circulation. This in turn is determined by the degree of right ventricular outflow tract (RVOT) obstruction as this is the deciding factor for pulmonary blood flow. It is very important to understand this simple principle as management of TOF physiology and its complications like hypercyanotic spells are based on this. Other than classical TOF, common AV canal defect with pulmonary stenosis, double outlet right ventricle with subaortic VSD and pulmonary stenosis are some of the anatomical entities that fall under this physiology.

**TGA physiology**

In TGA, oxygenated blood flows back into lungs and deoxygenated blood flows back into the body creating two parallel circuits. Saturation depends on amount of mixing at the level of atria, ventricles or great arteries. There may be no murmur unless there is associated pulmonary stenosis or VSD. Only in TGA physiology is there evidence of high pulmonary blood flow along with deep cyanosis. Classical transposition of great arteries and double outlet right ventricle with subpulmonic VSD fall into this category.

**Admixture physiology**

Complete mixing of pulmonary and systemic venous return at venous level, cardiac chamber or great artery level, before it is pumped into systemic and pulmonary circulations is called admixture physiology. Best example is tricuspid atresia where systemic venous return flows into left atrium as tricuspid valve is atretic and gets completely mixed with oxygenated pulmonary venous return in left atrium and then in left ventricle before it is pumped into pulmonary artery and aorta. Therefore, in admixture lesions the aortic and pulmonary artery saturations are equal and as we can assume the saturations in pulmonary venous blood (98%) and systemic venous blood (about 70%) we can calculate the pulmonary and systemic flow ratio (Qp:Qs). If aortic saturation is 85% Qp:Qs is 1:1 and if it is 94% the Qp:Qs is 4:1. This indicates that in a patient with admixture physiology, if the aortic saturation is high there is high pulmonary blood flow which is an important factor which adversely affects the outcome if he is suitable only for univentricular repair later. Lesions that fall into this category include total anomalous pulmonary venous drainage, tricuspid atresia, mitral atresia and pulmonary atresia with duct or collateral dependent pulmonary circulation.

## Management of common conditions

**Management of shunt lesions**

Lesions with a significant left to right shunt (Qp:Qs >1.5-2) should be surgically corrected as they can lead to pulmonary vascular damage and irreversible pulmonary hypertension later in life.

Management of ASD depends on the type of defect and its anatomical features. Small secundum ASDs are likely to close spontaneously. Most of the secundum ASDs can be closed using a device in the cardiac catheterization laboratory (transcatheter closure). Few of secundum defects, all of sinus venosus and ostium primum ASDs are not suitable for closure using a device and need surgical correction. Closure is done when the child is around four years of age as there is no added advantage by closing it before. These patients usually remain asymptomatic and rarely need anti-failure medication. If a child with ASD is symptomatic it is important that we exclude associated problems like mitral stenosis, partial anomalous pulmonary venous

drainage and other less obvious shunts like aorto-pulmonary window before sending them for surgical closure.

Management of VSD mainly depends on the size and its location in the interventricular septum. Most of the children with either moderate to large VSDs are symptomatic and need anti-failure treatment until they go for closure of the defect. Mainstay of treatment is surgical closure. Very few patients with multiple muscular VSDs or very large VSDs will need pulmonary artery banding as a palliative procedure. Transcatheter occlusion of perimembranous VSD is associated with complete heart block in approximately 5% of patients[2]. Therefore, the procedure is not recommended in children especially those weighing less than 10 kg[3].

Small or restrictive VSDs will not need any intervention unless they develop a complication. Aortic valve can prolapse through perimembranous or subpulmonic VSDs. The child may be asymptomatic and the VSD may appear small as it gets restricted by prolapsing aortic valve. However, these children need surgical closure of VSD as it can progress to severe aortic regurgitation needing aortic valve replacement in later life. The other complication which warrants surgical closure in a small VSD is repeated episodes of endocarditis.

Perimembranous and muscular VSDs can close spontaneously and therefore can be managed medically if the child is relatively asymptomatic. Subpulmonic VSDs and inlet VSDs are unlikely to close spontaneously. Therefore, they need surgical closure if they are symptomatic or if there is evidence of volume overload of left atrium and left ventricle.

PDA is the worst out of the three shunts as a high pressure left to right shunt continues during systole as well as during diastole. Therefore, these patients are more symptomatic and develop pulmonary hypertension early. Majority of PDAs are suitable for transcatheter closure with either a coil or a device. Very few newborns or infants less than 3-4 kg who are ventilator dependent may need surgical closure of the defect.

**Heart failure in shunt lesions**

Diagnosis and management of heart failure is important in shunt lesions. Tachycardia, tachypnoea and hepatomegaly are features of heart failure but all these three will be seen in a child with respiratory tract infection or bronchiolitis (apparent hepatomegaly due to liver being pushed down). Chest x-ray will help to differentiate, which will show cardiomegaly if there is a shunt lesion or hyper-expanded lung fields if there is bronchiolitis. Heart failure in a shunt lesion should be managed using diuretics alone (frusemide with spironolactone) or a diuretic with an afterload reducing agent (frusemide with captopril).

**Management of obstructive lesions**

Aortic stenosis is commonly associated with bicuspid aortic valve and treatment of choice is balloon dilatation. However, the procedure is done only if the child is symptomatic or if there is significant left ventricular hypertrophy as there is approximately a 20% chance of developing significant aortic valve regurgitation after balloon dilatation[4].

Coarctation of aorta is one of the congenital heart diseases which can be confidently diagnosed during neonatal examination. Absent or low volume femoral pulse compared to brachial pulse should raise the suspicion and the patient should be referred for further evaluation to a paediatric cardiology centre. However, if there is a large ductus feeding the lower limbs, the pulse volume will be good but there will be desaturation in lower limbs compared to right upper limb which is called differential cyanosis. Treatment of choice is surgical correction as there is a significant incidence of re-coarctation and aneurysm formation after balloon dilatation[5]. However, balloon dilatation can be used as a bridging therapy in those who have left ventricular failure or those who have other risk factors for surgery.

Isolated valvar pulmonary stenosis, which is the commonest right side obstructive lesion, is usually dealt with by transcatheter balloon dilatation. The procedure is done only when the obstruction is moderate to severe. Patients with subvalvar or supravalvar obstructions need surgical correction of the lesion.

**Management of cyanotic lesions**

Management of cyanotic lesions depend on two basic facts; whether the patient is suitable for biventricular or univentricular repair and whether there is low pulmonary blood flow or high pulmonary blood flow.

**Management of patients suitable for biventricular repair**

**Tetralogy of Fallot**

This is the commonest cyanotic heart disease and management depends on the degree of cyanosis which in turn is determined by the degree of RVOT obstruction. When the obstruction is severe they become more cyanosed due to shunting of deoxygenated blood from right to left through the VSD. If they have well developed branch pulmonary arteries total correction is done around one year of age. If the branch pulmonary arteries are smaller in size or if they have significant cyanosis or hypercyanotic spells a shunt is created from innominate artery to pulmonary artery (Blalock Taussig Shunt) to augment growth of pulmonary arteries and to improve oxygen saturation. Patients with Tetralogy of Fallot do not go into heart failure (except in rare variants of Tetralogy of Fallot) and their major issue is hypercyanotic spells.

**Management of hypercyanotic spells**

What really causes a hypercyanotic spell is poorly understood and there are many theories proposed, including the infundibular spasm theory. However, the popular infundibular spasm theory fails to explain why it occurs even in pulmonary atresia. Hypercyanotic spells are not limited to Tetralogy of Fallot and can occur in any condition where there is a common pump head (single ventricle or two ventricles with a large VSD) pumping into both systemic and pulmonary circulations in combination with restricted pulmonary blood flow. Therefore, it can occur in univentricular heart with pulmonary stenosis, any condition with pulmonary atresia with duct dependent pulmonary circulation and even in tricuspid atresia with pulmonary stenosis.

Hypercyanotic spells should be managed as a medical emergency and the main aim of management is to increase pulmonary blood flow. This can be achieved by keeping them in the knee-chest position and by giving a fluid bolus of 10-20ml/kg. 100% oxygen should be administered and morphine 0.1-0.2mg/kg IV/IM can be used to reduce distress and hyperpnoea. Propranolol can be given intravenously, 0.05–0.1 mg/kg slow bolus over 10 minutes, but patient should be closely monitored for bradycardia. Acidosis can be corrected with sodium bicarbonate. If the patient continues to spell he may need general anaesthesia and ventilation or an emergency surgical intervention.

A single episode of a definite hypercyanotic spell is an indication for early surgical repair either total correction or Blalock Taussig shunt.

**Transposition of great arteries**

This is the second commonest cyanotic heart disease. If there is significant cyanosis at diagnosis, transcatheter balloon atrial septostomy is done to increase mixing and thereby oxygen saturation. Arterial switch operation is done within 2-3weeks of life, before left ventricular muscle mass regresses, if there is no associated VSD or PDA. If diagnosed late and left ventricle has already regressed, atrial switch operation is done where blood is redirected in to systemic and pulmonary circulations at atrial level.

**Total anomalous pulmonary venous drainage**

These patients usually have clinical features of a large ASD with cyanosis and features of heart failure. If there is obstruction to the pulmonary venous drainage leading to severe pulmonary hypertension, it becomes a medical emergency and patient should undergo immediate surgical correction.

**Management of patients suitable for univentricular repair**

**Complex cyanotic heart diseases**

There is no definition for a complex cyanotic heart disease. Generally, there is a combination of shunts and obstructive lesions and most of them are suitable only for univentricular repair. During univentricular repair systemic venous return is directly diverted into the pulmonary circulation usually in two stages.

First stage of univentricular repair is the bidirectional Glenn Shunt where superior vena cava is connected to right pulmonary artery. This is done preferably between 6 months to 2 years of age as blood flow to upper body is higher compared to lower body at this age. As the child grows this ratio reverses and lower body gets a larger proportion compared to upper body. Therefore, connection of inferior vena cava to pulmonary artery (Fontan completion or total cavo-pulmonary communication) is done preferably between 2-5years of age. Both the SVC and IVC are anastomosed to right pulmonary artery in an end to side manner and therefore blood flows into both right and left lungs.

After completion of univentricular repair, systemic venous return should flow passively through pulmonary circulation. Therefore these patients need

preparation prior to univentricular repair so that they will have good size pulmonary arteries to accommodate systemic venous return and low pulmonary vascular resistance (PVR) and low pulmonary artery pressure (preferably PA mean pressure less than 15mmHg) for blood to flow through pulmonary vascular tree. Patients with high pulmonary blood flow (e.g. univentricular heart without pulmonary stenosis) will have good size pulmonary arteries but PA pressure and PVR will rise with time. Therefore, they need pulmonary artery banding to prevent rise in pulmonary vascular resistance and pulmonary artery pressure. In the other extreme, where the patient has low pulmonary blood flow (e.g. univentricular heart with critical pulmonary stenosis or pulmonary atresia) he will have low PVR and low PA pressure but the pulmonary arteries will be too small to accommodate systemic venous return. Therefore he needs aorta to pulmonary artery shunt (e.g. Blalock-Taussig Shunt) to augment growth of pulmonary arteries. Those who have a balanced shunt, in whom the pulmonary stenosis is adequate to prevent development of pulmonary hypertension but the flow is enough for growth of branch pulmonary arteries, can be closely followed up and can directly go for bidirectional Glenn shunt.

**Conclusion**

Proper management of congenital heart disease is important in bringing down morbidity and mortality associated with it. Even for simple lesions, the decision on whether to operate or not depends on anatomy, haemodynamics, presence of heart failure, risk of development of pulmonary hypertension, risk of endocarditis, risk of cerebral thromboembolism, failure to thrive, recurrent lower respiratory tract infections, recurrent hospitalizations, effect on rest of the family and socio-economic factors. All these need to be weighed against morbidity and mortality risk of intervention or surgery. Timing of intervention or surgery is also important as this affects overall outcome. Medical management of symptoms of heart failure or cyanotic spells is only a bridging therapy until definitive intervention or surgery is done or for patients who are decided for conservative management.

**References**


1. Hoffman JI. Incidence of congenital heart disease: I. Postnatal incidence. *Pediatr Cardiol* 1995; **16** (3):103-13.

2. Carminati M, et al. Transcatheter closure of congenital ventricular septal defects: results of the European Registry. *Eur Heart J* 2007; **28**(19): 2361-8.

3. Sullivan ID. Transcatheter closure of perimembranous ventricular septal defect: is the risk of heart block too high a price? *Heart* 2007; **93**(3): 284-6.

4. Jindal RC, et al. Long-term results of balloon aortic valvulotomy for congenital aortic stenosis in children and adolescents. *J Heart Valve Dis* 2000; **9**(5): 623-8.

5. Cowley CG, et al. Long-term, randomized comparison of balloon angioplasty and surgery for native coarctation of the aorta in childhood. *Circulation* 2005; **111**(25): 3453-6.