

CONGENITAL HEART DISEASE

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- Congenital heart disease is the abnormality of the heart present from birth. It is the most common and important form of heart disease in the early years of life and is present in about 0.5% of newborn children. The incidence is higher in premature infants. The cause of congenital heart disease is unknown in majority of cases. It is attributed to multifactorial inheritance involving genetic and environmental influences. Other factors like rubella infection to the mother during pregnancy, drugs taken by the mother and heavy alcohol drinking by the mother have all been implicated in causing in utero injury resulting in congenital malformations of the heart.

- **Classification.** Congenital anomalies of the heart may be either shunts (left-to-right or right-to-left) or defects causing obstructions to flow. However, complex anomalies involving combinations of shunts and obstructions are also often present.

I. Malpositions of the Heart

Dextrocardia is the condition when the apex of the heart points to the right side of the chest. It may be accompanied by situs inversus so that all other organs of the body are also transposed in similar way and thus heart is in normal position in relation to them. However, isolated dextrocardia is associated with major anomalies of the heart such as transposition of the atria in relation to ventricles or transposition of the great arteries.

II. Shunts (Cyanotic congenital heart disease)

- A shunt may be left-to-right side or right-to-left side of the circulation.

Classification of Congenital Heart Diseases

Malpositions of the Heart

II. Shunts (Cyanotic congenital heart disease)

A. Left-to-right shunts

(Acyanotic or late cyanotic group)

1. Ventricular septal defect (VSD) (25-30%)
2. Atrial septal defect (ASD) (10-15%)
3. Patent ductus arteriosus (PDA) (10-20%)

B. Right-to-left shunts (Cyanotic group)

1. Tetralogy of Fallot (6-15%)
2. Transposition of great arteries (4-10%)
3. Persistent truncus arteriosus (2%)
4. Tricuspid atresia and stenosis (1%)

III. Obstructions

(Obstructive Congenital Heart Disease)

1. Coarctation of aorta (5-7%)
2. Aortic stenosis and atresia (4-6%)
3. Pulmonary stenosis and atresia (5-7%)

Left-to-Right Shunts

(Acyanotic or Late Cyanotic Group)

In conditions where there is shunting of blood from left-to-right side of the heart there is volume overload on the right heart producing pulmonary hypertension and right ventricular hypertrophy. At a later stage, the pressure on the right side is higher than on the left side creating late cyanotic heart disease. The important conditions included in this category are described below:

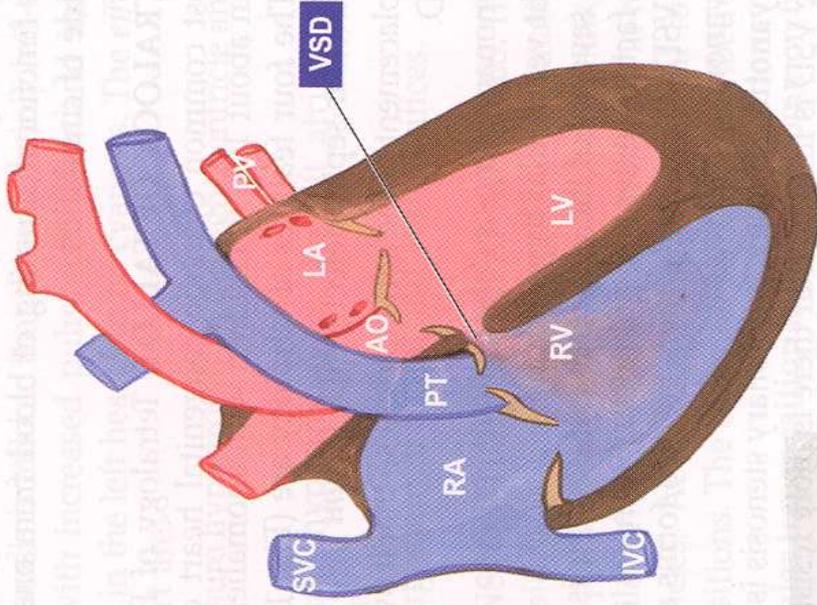
- 1. Ventricular Septal Defect (VSD).** VSD is the most common congenital anomaly of the heart and comprises about 30% of all congenital heart diseases. The condition is recognized early in life. The smaller defects often close spontaneously, while larger defects remain patent and produce significant effect.

Depending upon the location of the defect, VSD may be of the following types:

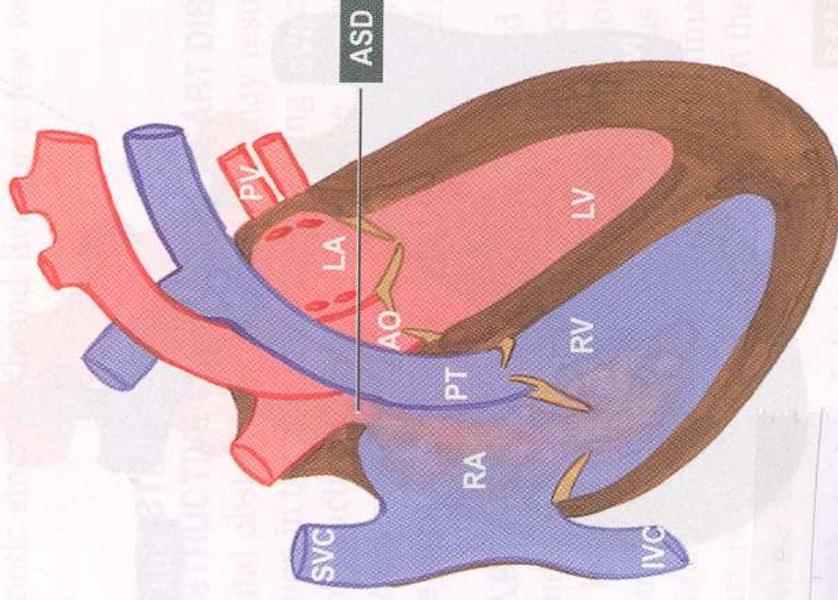
1. In 90% of cases, the defect involves membranous septum and is very close to the bundle of His.
2. The remaining 10% cases have VSD immediately below the pulmonary valve (subpulmonic), below the aortic valve (subaortic), or exist in the form of multiple defects in the muscular septum.

The effect of VSD are produced due to left-to-right shunt at the ventricular level, increased pulmonary flow and increased volume in the left side of the heart. These effects are:

- i. Volume hypertrophy of the right ventricle.
- ii. Enlargement and haemodynamic changes in the tricuspid and pulmonary valves.
- iii. Endocardial hypertrophy of the right ventricle.
- iv. Pressure hypertrophy of the right atrium.
- v. Volume hypertrophy of the left atrium and left ventricle.
- vi. Enlargement and haemodynamic changes in the mitral and aortic valves.



Ventricular septal defect, a schematic representation (LA = Left atrium; LV = Left ventricle; AO = Aorta; PV = Pulmonary valve; PT = Pulmonary trunk; RA = Right atrium; RV = Right ventricle; SVC = Superior vena cava; IVC = Inferior vena cava).



Atrial septal defect fossa ovalis type, a schematic representation (LA = Left atrium; LV = Left ventricle; PV = Pulmonary vein; AO = Aorta; PT = Pulmonary trunk; RA = Right atrium; RV = Right ventricle; SVC = Superior vena cava; IVC = Inferior vena cava).

2. Atrial Septal Defect (ASD). Isolated ASD comprises about 10% of congenital heart diseases. The condition remains unnoticed in infancy and childhood till pulmonary hypertension is induced causing late cyanotic heart disease and right-sided heart failure.

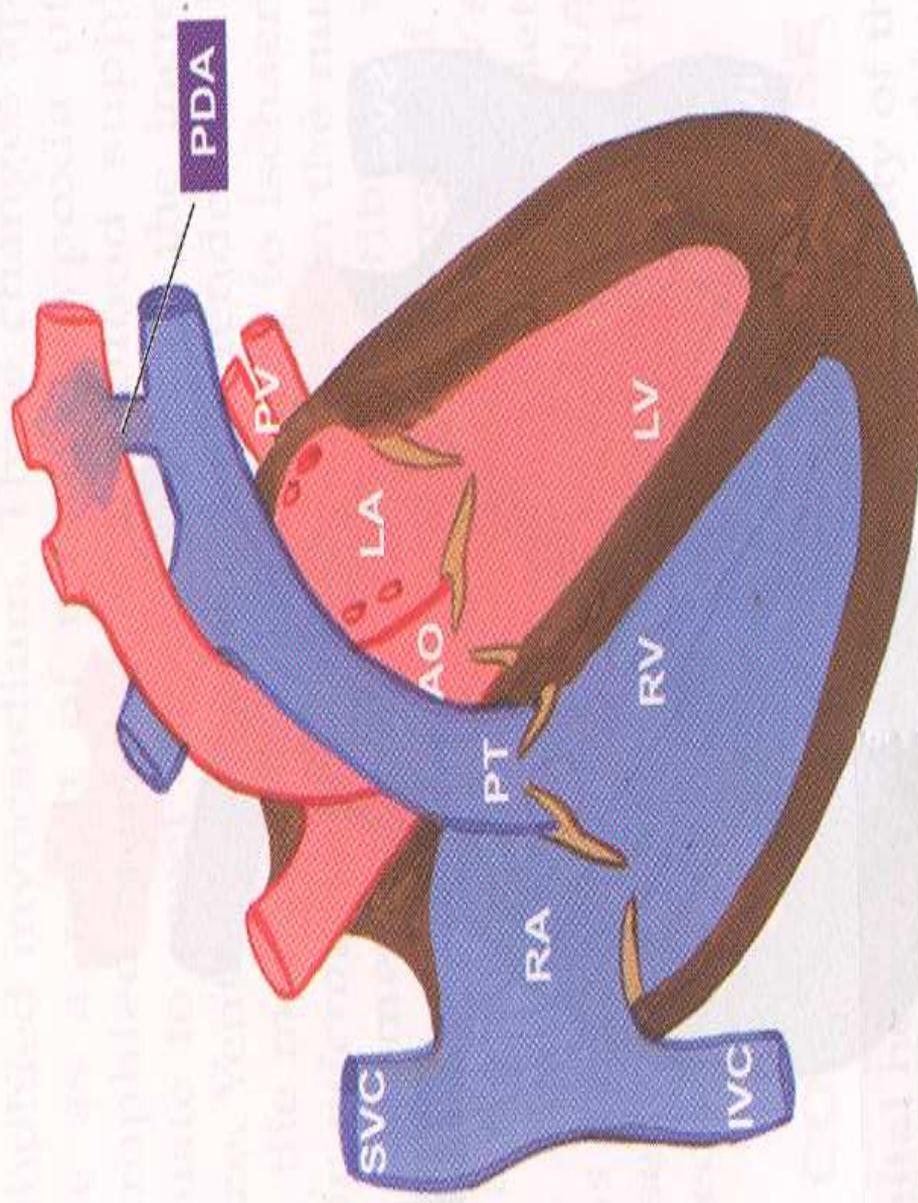
Depending upon the location of the defect, there are 3 types of ASD:

- i. Fossa ovalis type or ostium secundum type** is the most common form comprising about 90% cases of ASD. The defect is situated in the region of the fossa ovalis.
- ii. Ostium primum type** comprises about 5% cases of ASD. The defect lies low in the interatrial septum adjacent to atrioventricular valves. There may be cleft in the aortic leaflet of the mitral valve producing mitral insufficiency.
- iii. Sinus venosus type** accounts for about 5% cases of ASD. The defects is located high in the interatrial septum near the entry of the superior vena cava.

The effect of ASD are produced due to left-to-right. Shunt at the atrial level with increased pulmonary flow. These effects are:

- i. Volume hypertrophy of the right atrium and right ventricle.
- ii. Enlargement and haemodynamic changes of tricuspid and pulmonary valves.
- iii. Focal or diffuse endocardial hypertrophy of the right atrium and right ventricle.
- iv. Volume atrophy of the left atrium and left ventricle.
- v. Small-sized mitral and aortic orifices.

3. Patent ductus arteriosus (PDA). The ductus arteriosus is a normal vascular connection between the aorta and the bifurcation of the pulmonary artery. Normally, the ductus closes functionally within the first or second day of life. Its persistence after 3 months of age is considered abnormal. The cause for patency of ductus arteriosus is not known but possibly it is due to continued synthesis of PGE₂ after birth which keeps it patent as evidenced by association of PDA with respiratory distress syndrome in infants and pharmacologic closure of PDA with administration of indomethacin to suppress PGE₂ synthesis. PDA constitutes about 10% of congenital malformations of the heart and great vessels. In about 90% of cases, it occurs as an isolated defect, while in the remaining cases it may be associated with other anomalies like VSD, coarctation of aorta and pulmonary or aortic stenosis. A patent ductus may be upto 2 cm in length and upto 1 cm in diameter.



Patent ductus arteriosus, a schematic representation (LA = Left atrium; LV = Left ventricle; PT = Pulmonary trunk; PV = Pulmonary vein, AO = Aorta; RA = Right atrium; RV = Right ventricle; SVC = Superior vena cava; IVC = Inferior vena cava).

The effects of PDA on heart occur due to left-to-right shunt at the level of ductus resulting in increased pulmonary flow and increased volume in the left heart. These effects are as follows:

- i. Volume hypertrophy of the left atrium and left ventricle.
- ii. Enlargement and haemodynamic changes of the mitral and pulmonary valves.
- iii. Enlargement of the ascending aorta.

B. Right-to-Left Shunts (Cyanotic Group)

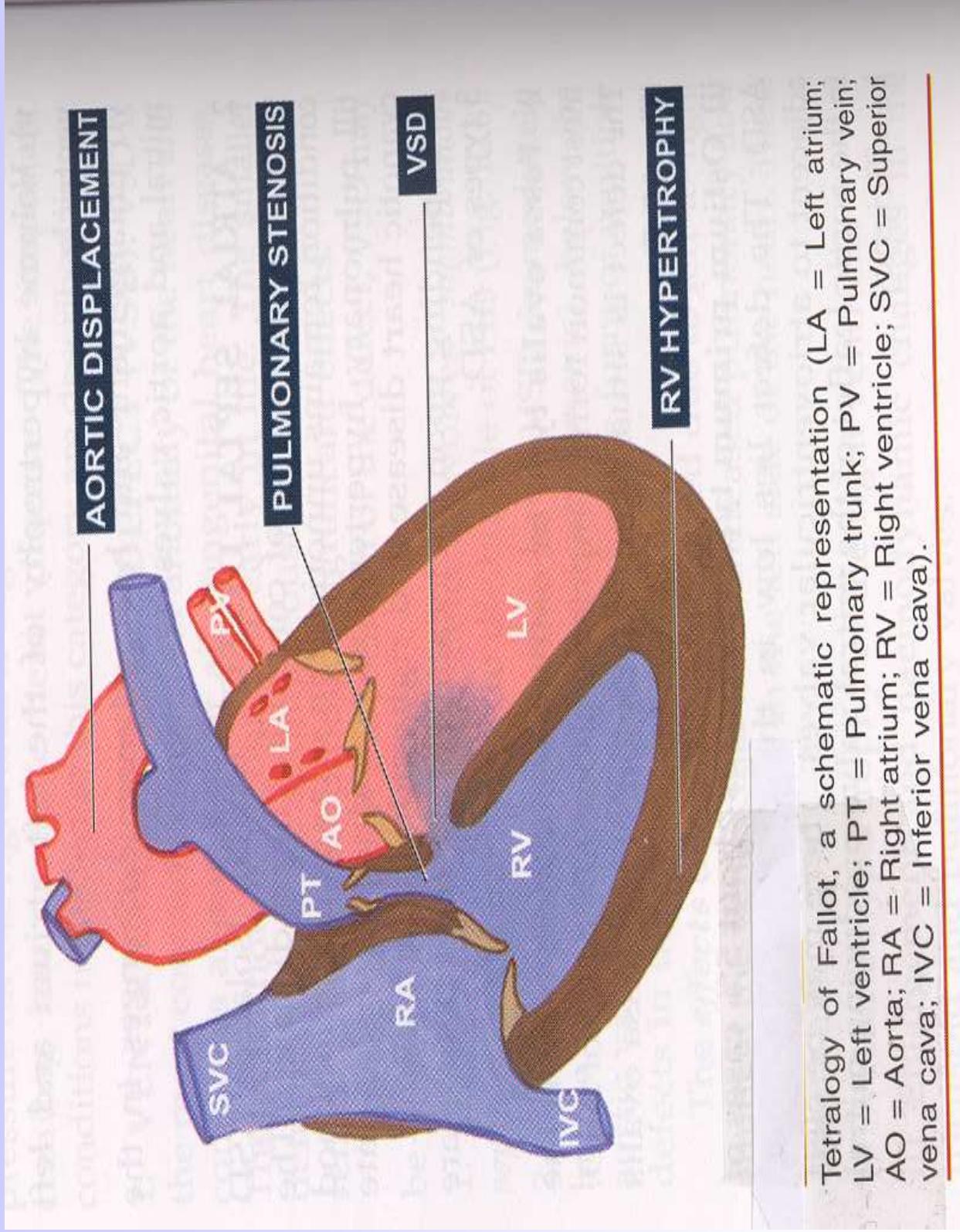
In conditions where there is shunting of blood from right side to the left side of the heart, there is entry of poorly-oxygenated blood into systemic circulation resulting in early cyanosis. The examples described below are not pure shunts but are combinations of shunts with obstructions but are described here since there is functional shunting of blood from one to the other side of circulation.

1. **Tetralogy of Fallot.** Tetralogy of Fallot is the most common cyanotic congenital heart disease, found in about 10% of children with anomalies of the heart. The four features of tetralogy are:
 - i. Ventricular septal defect (VSD) ('shunt').
 - ii. Displacement of the aorta to right so that it overrides the VSD.
 - iii. Pulmonary stenosis ('obstruction').
 - iv. Right ventricular hypertrophy.

The severity of the clinical manifestations is related to two factors: extent of pulmonary stenosis and the size of VSD. Accordingly, there are two forms of tetralogy: cyanotic and acyanotic.

In **cyanotic tetralogy**, pulmonary stenosis is greater and the VSD is mild so that there is more resistance to the outflow of blood from right ventricle resulting in right-to-left shunt at the ventricular level and cyanosis. The effects on the heart are:

- i. Pressure hypertrophy of the right atrium and right ventricle.
- ii. Smaller and abnormal tricuspid valve.
- iii. Smaller left atrium and left ventricle.
- iv. Enlarged aortic orifice.



Tetralogy of Fallot, a schematic representation (LA = Left atrium; LV = Left ventricle; PT = Pulmonary trunk; PV = Pulmonary vein; AO = Aorta; RA = Right atrium; RV = Right ventricle; SVC = Superior vena cava; IVC = Inferior vena cava).

In **acyanotic tetralogy**, the VSD is larger and pulmonary stenosis is mild so that there is mainly left-to-right shunt with increased pulmonary flow and increased volume in the left heart but no cyanosis. The effects on the heart are:

- i. Pressure hypertrophy of the right ventricle and right atrium.
- ii. Volume hypertrophy of the left atrium and left ventricle.
- iii. Enlargement of mitral and aortic orifices.

- 2. Transposition of great arteries.** The term transposition is used for complex malformations as regards position of the aorta, pulmonary trunk, atrio-ventricular orifices and the position of atria in relation to ventricles. Accordingly, there are several forms of transpositions. The common ones are described below.
- i) Regular transposition** is the most common type. In this, the aorta which is normally situated to the right and posterior with respect to the pulmonary trunk, is instead displaced anteriorly and to right. In regular complete transposition, the aorta emerges from the right ventricle and the pulmonary trunk from the left ventricle so that there is cyanosis from birth.
 - ii) Corrected transposition** is an uncommon anomaly. There is complete transposition of the great arteries with aorta arising from the right ventricle and the pulmonary trunk from the left ventricle, as well as transposition of the great veins so that the pulmonary veins enter the right atrium and the systemic veins drain into the left atrium. This results in a physiologically corrected circulation.

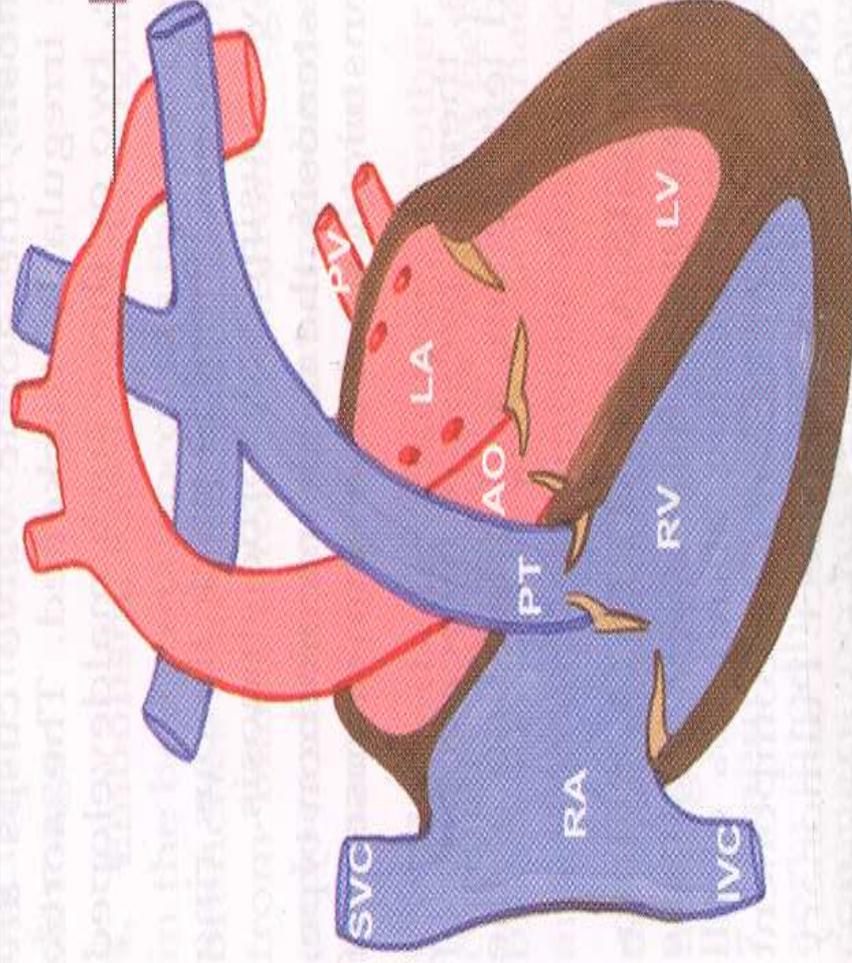
- 3. Persistent truncus arteriosus.** Persistent truncus arteriosus is a rare anomaly in which the arch that normally separates the aorta from the pulmonary artery fails to develop. This results in a single large common vessels receiving blood from the right as well as left ventricle. The orifice may have 3 to 6 cusps. There is often an associated VSD. There is left-to-right shunt and frequently early systemic cyanosis. The prognosis is generally poor.
- 4. Tricuspid atresia and stenosis.** Tricuspid atresia and stenosis are rare anomalies. There is often associated pulmonary stenosis or pulmonary atresia. In tricuspid atresia, there is absence of tricuspid orifice and instead there is a dimple in the floor of the right atrium. In tricuspid stenosis, the tricuspid ring is small and the valve cusps are malformed. In both the conditions, there is often an interatrial defect through which right-to-left shunt of blood takes place. Children are cyanotic since birth and live for a few weeks or months.

III. Obstructions (Obstructive Congenital Heart Disease)

Congenital obstruction to blood flow may result from obstruction in the aorta due to narrowing (coarctation of aorta), obstruction to outflow from the left ventricle (aortic stenosis and atresia) and obstruction to outflow from the right ventricle (pulmonary stenosis and atresia).

1. **Coarctation of Aorta.** The word 'coarctation' means contracted or compressed. Coarctation of aorta is localized narrowing in any part of aorta, but the constriction is more often just distal to ductus arteriosus (postductal or adult), or occasionally proximal to the ductus arteriosus (preductal or infantile type) in the region of transverse aorta.
 - i) **In postductal or adult type,** the obstruction is just distal to the point of entry of ductus arteriosus which is often closed. In the stenotic segment the aorta is drawn in as if a suture has been tied around it. The aorta is dilated on either side of the constriction. The condition is recognized in adulthood, characterized by hypertension in the upper extremities, weak pulses and low blood pressure in the lower extremities and effects of arterial insufficiency such as claudication and coldness. In time, there is development of collateral circulation between pre-stenotic and post-stenotic arterial branches so that intercostal arteries are enlarged and palpable and may produce erosions on the inner surface of the ribs.

ADULT COARCTATION



Postductal or adult type coarctation of the aorta, a schematic representation (LA = Left atrium; LV = Left ventricle; PT = Pulmonary trunk; PV = Pulmonary vein; AO = Aorta; RA = Right atrium; RV = Right ventricle; SVC = Superior vena cava; IVC = Inferior vena cava).

ii) In preductal or infantile type, the manifestations are produced early in life. The narrowing is proximal to the ductus arteriosus which usually remains patent. The narrowing is generally gradual and involves larger segment of the proximal aorta. There is often associated interatrial septal defect. Preductal coarctation results in right ventricular hypertrophy while the left ventricle is small. Cyanosis develops in the lower half of the body while the upper half remains unaffected since it is supplied by vessels originating proximal to the coarctation. Children with this defect have poor prognosis.

2. Aortic stenosis and atresia. The most common congenital anomaly of the aorta is bicuspid aortic valve which does not have much functional significance but predisposes it to calcification. Congenital aortic atresia is rare and incompatible with survival. Aortic stenosis may be acquired (e.g. in rheumatic heart disease, calcific aortic stenosis) or congenital. Congenital aortic stenosis may be of three types: valvular, subvalvular and supra-valvular.

- i) In **valvular stenosis**, the aortic valve cusps are malformed and are irregularly thickened. The aortic valve may have one, two or three such maldeveloped cusps.
- ii) In **subvalvular stenosis**, there is thick fibrous ring under the aortic valve causing subaortic stenosis.
- iii) In **supra-valvular stenosis**, the most uncommon type, there is fibrous constriction above the sinuses of valsalva.

In all these cases, there is pressure hypertrophy of the left ventricle and left atrium, and dilatation of the aortic root.

3. **Pulmonary stenosis and atresia.** **Pulmonary stenosis** is the commonest form of obstructive congenital heart disease comprising about 7% of all congenital heart diseases. It may occur as a component of tetralogy of Fallot or as an isolated defect. Pulmonary stenosis is caused by fusion of cusps of the pulmonary valve forming a diaphragm-like obstruction to the outflow of blood from the right ventricle and dilatation of the pulmonary trunk.

In **pulmonary atresia**, there is no communication between the right ventricle and lungs so that the blood bypasses the right ventricle through an interatrial septal defect. It then enters the lungs via patent ductus arteriosus.