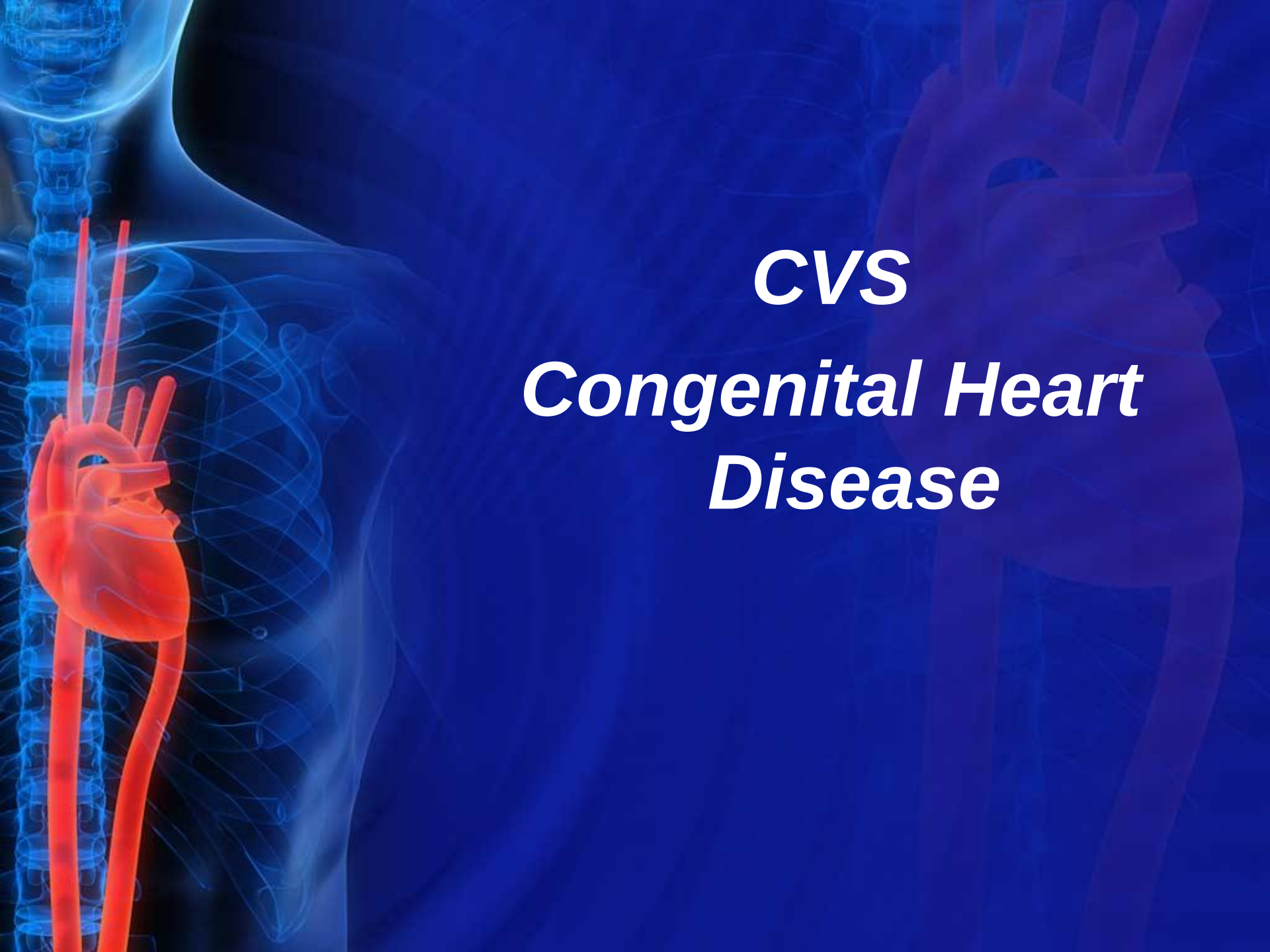


CVS

Congenital Heart Disease

Introduction

Severe anomalies are incompatible with intrauterine survival

Defects that affect individual chambers or discrete regions of the heart are often compatible with embryologic maturation and eventual live birth.

Septation defects: ***atrial septal defects (ASDs)***
ventricular septal defects (VSDs).

Unilateral obstructions: ***level of the cardiac valve***
entire cardiac chamber
e.g hypoplastic left heart syndrome.

Outflow tract anomalies: ***inappropriate routing of the great vessels from the ven- tricular mass.***

Cardiac Development

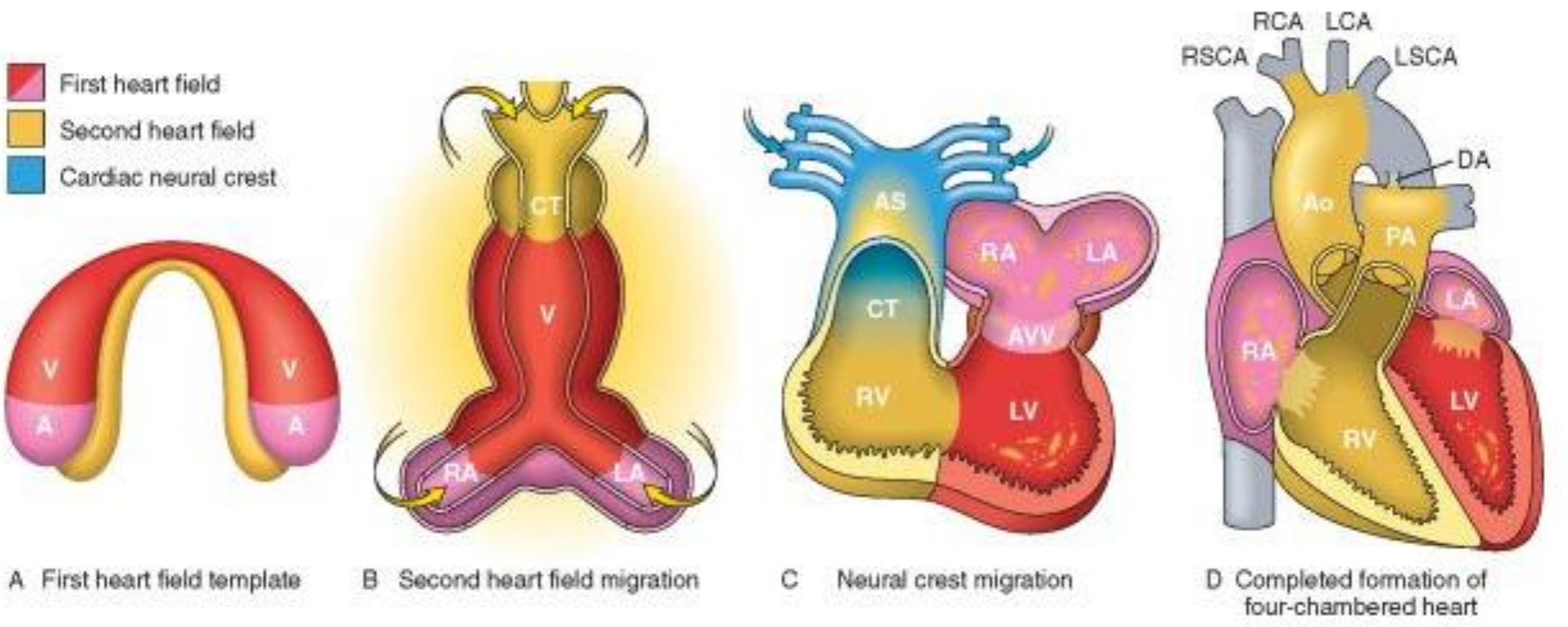
A. Day 15. First heart field (FHF) cells form a crescent shape in the anterior embryo with second heart field (SHF) cells near the FHF.

A. Day 21. SHF cells lie dorsal to the straight heart tube and begin to migrate into the anterior and posterior ends of the tube to form the RV,CT, and part of atria

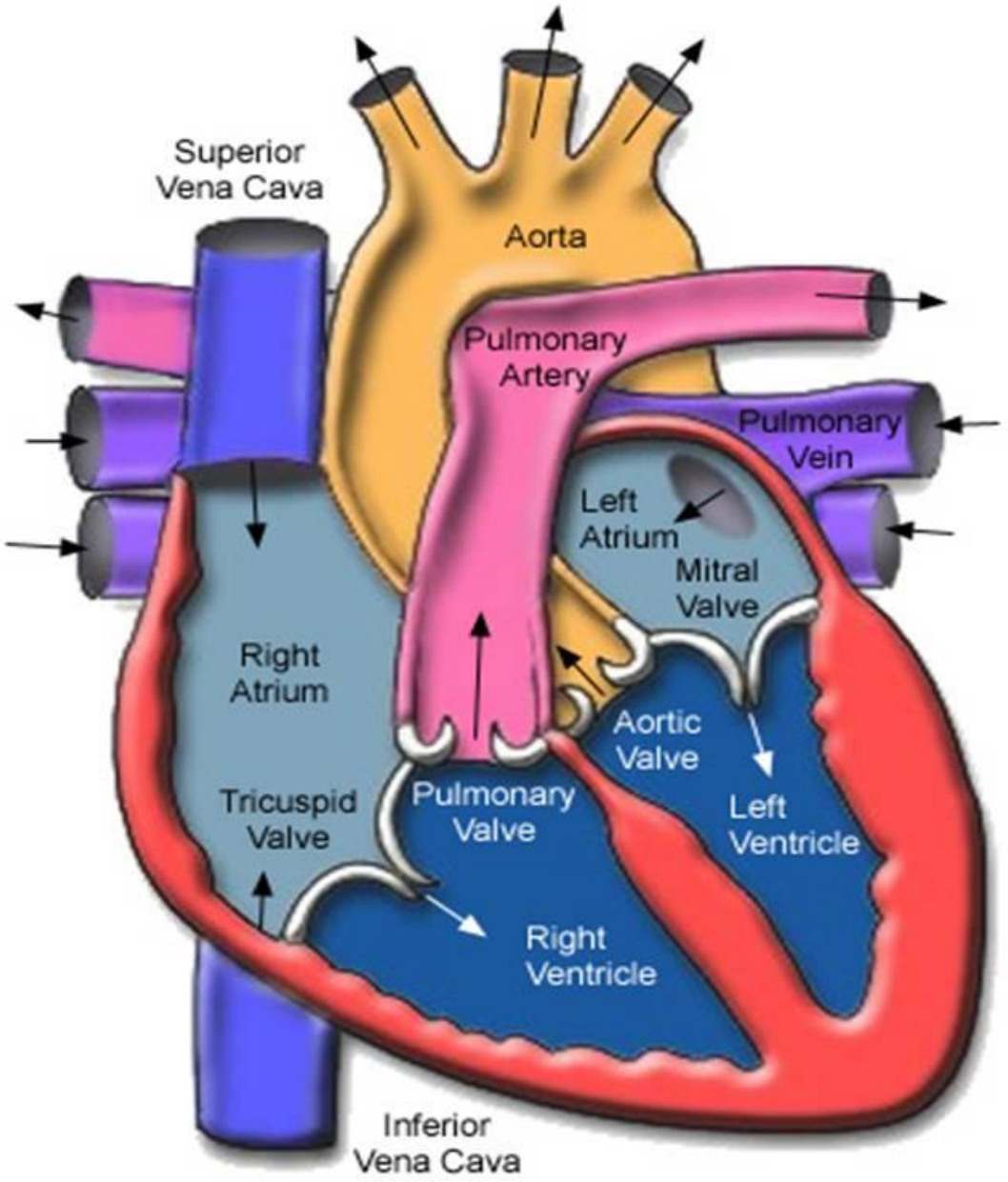
A. Day 28. Following rightward looping of the heart tube, cardiac neural crest cells also migrate into the outflow tract

Cardiac Development

***Day 50*. Septation of the ventricles, atria, atrioventricular valves (AVV)**



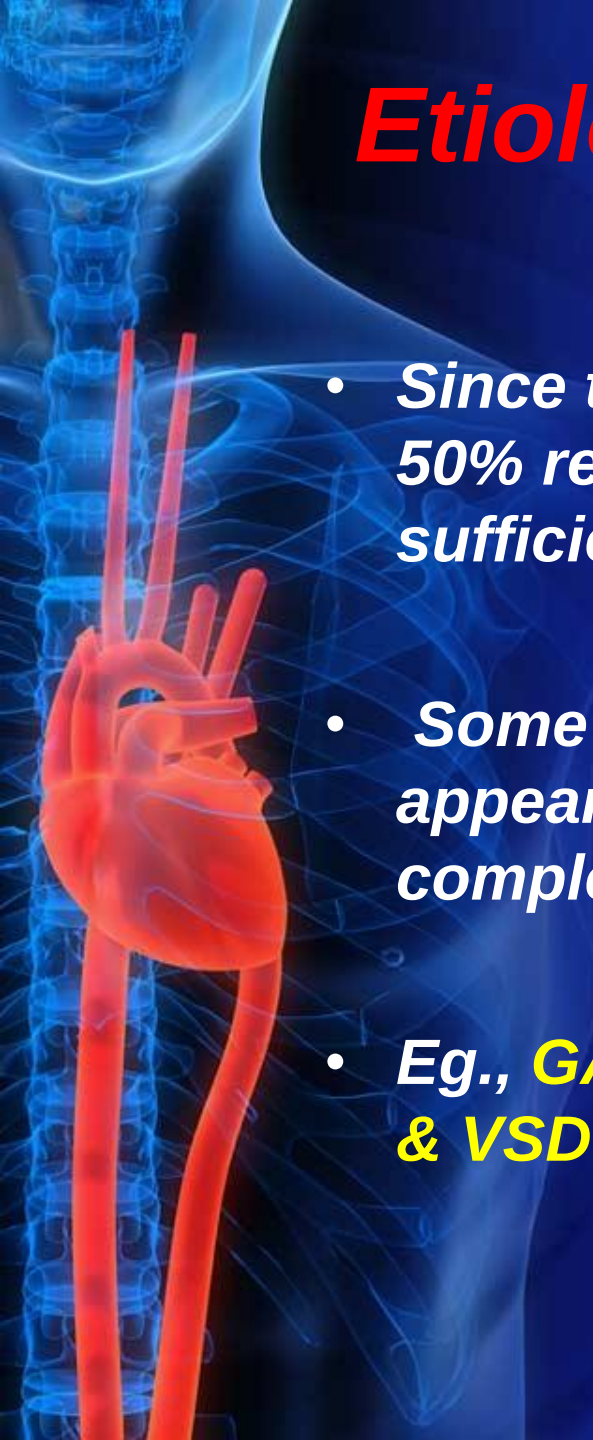
Normal Structure of Heart



Etiology and Pathogenesis

- Main known cause- sporadic genetic abnormalities,
- Single gene mutations,
- Small chromosomal deletions, and
- Additions or deletions of whole chromosomes (trisomies and monosomies).
- Mutations affect genes encoding transcription factors that are required for normal heart development.

Etiology and Pathogenesis



- ***Since the affected patients are heterozygous - 50% reduction in the activity of these factors is sufficient to derange cardiac development.***
- ***Some of the affected transcription factors appear to work together in large protein complexes***
- ***Eg., GATA4, TBX5, and NKX2-5, -mutated in ASD & VSD***

Clinical Features

- The varied structural anomalies in CHD fall primarily into three major categories:
- Malformations causing a left-to-right shunt
- Malformations causing a right-to-left shunt
- Malformations causing an obstruction.

- A shunt is an abnormal communication between chambers or blood vessels.

- Abnormal channels permit the flow of blood down pressure gradients from the left (systemic) side to the right (pulmonary) side of the circulation or vice versa.

Clinical Features

- ***Hypoxemia and cyanosis** (a dusky blueness of the skin and mucous membranes) result because of the admixture of poorly oxygenated venous blood with systemic arterial blood (called **cyanotic congenital heart disease**).*

- ***With right-to-left shunts, emboli arising in peripheral veins can bypass the lungs and directly enter the systemic circulation :paradoxical embolism***

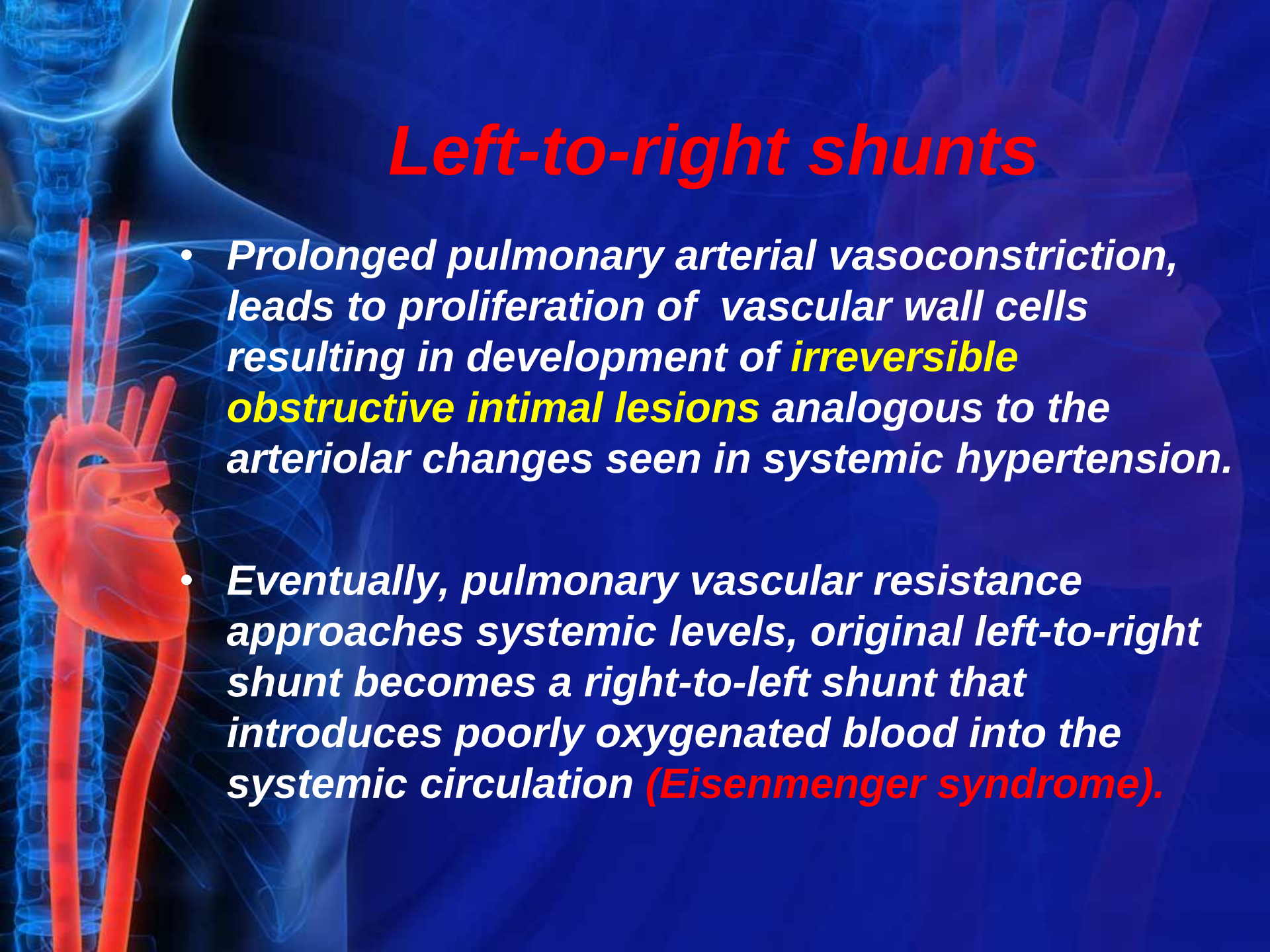
- ***Brain infarction and abscess are potential consequences.***

Clubbing of fingers

Severe, long-standing cyanosis also causes clubbing of the tips of the fingers and toes (called hypertrophic osteoarthropathy) and polycythemia.

Left-to-right shunts

- ***Increase pulmonary blood flow*, are not initially associated with cyanosis.**
- ***Raise both flow volumes and pressures* in the normally low-pressure, low-resistance pulmonary circulation, which can lead to RVH and atherosclerosis of the pulmonary vasculature.**
- **Muscular *pulmonary arteries* (<1 mm diameter) respond to increased pressure and flow by undergoing *medial hypertrophy and vasoconstriction*, which maintains relatively normal distal pulmonary capillary and venous pressures, and prevents pulmonary edema.**



Left-to-right shunts

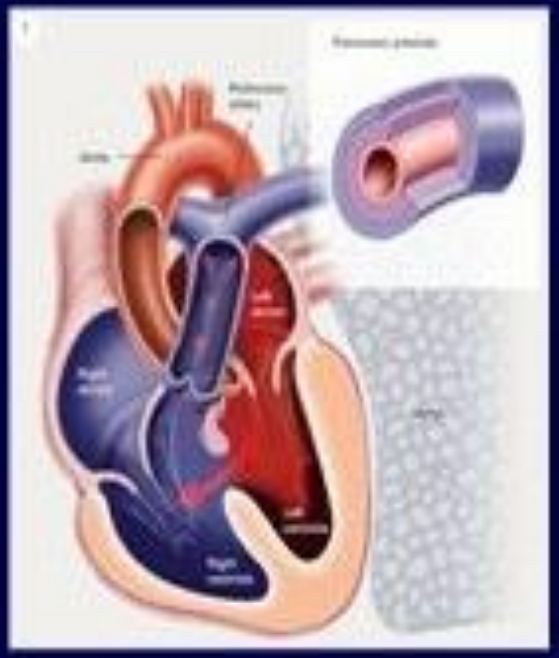
- ***Prolonged pulmonary arterial vasoconstriction, leads to proliferation of vascular wall cells resulting in development of irreversible obstructive intimal lesions analogous to the arteriolar changes seen in systemic hypertension.***

- ***Eventually, pulmonary vascular resistance approaches systemic levels, original left-to-right shunt becomes a right-to-left shunt that introduces poorly oxygenated blood into the systemic circulation (Eisenmenger syndrome).***

Left-to-right shunts

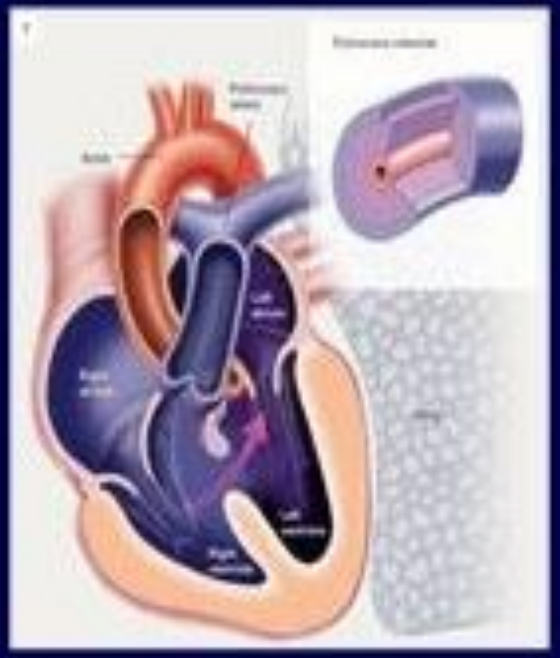
- ***Once irreversible pulmonary hypertension develops, the structural defects -irreparable.***
- ***Subsequent right heart failure eventually leads to death.***
- ***Need for early intervention, either surgical or nonsurgical, in those with left-to-right shunts.***

Evolution of Eisenmenger Syndrome



ASD, VSD, or complex defect ↑ Qp and/or PAp, with L-to-R shunting

Over time, PVR ↑ resulting in bi-directional flow



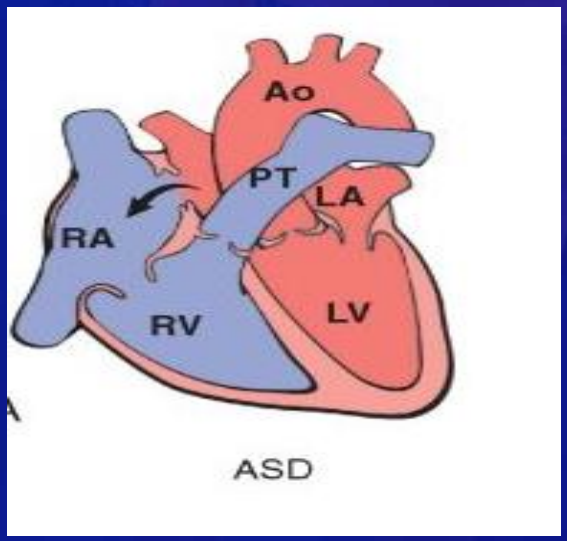
PVR ↑'s: shunt reverses: R-to-L → Eisenmenger syndrome: ↑ cyanotic

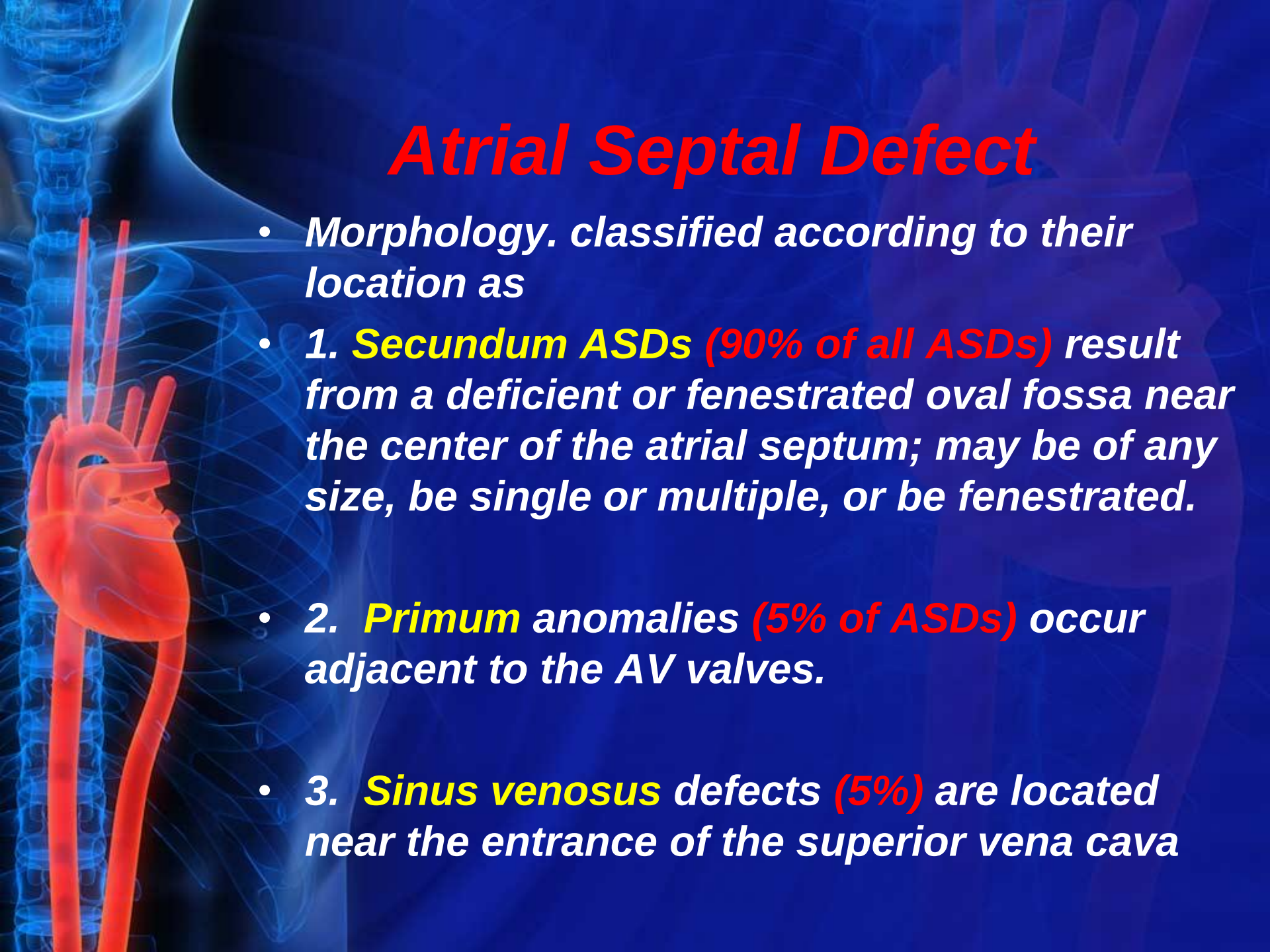
Left-to-right shunts

- ***Atrial Septal Defect***
- ***Ventricular Septal Defect***
- ***Patent Ductus Arteriosus***
- ***Complete Atrioventricular Canal Defect***

Atrial Septal Defect

- *ASD abnormal, fixed openings in the atrial septum caused by incomplete tissue formation that allows communication of blood between the left and right atria*
- *Usually asymptomatic until adulthood*
- *Result from defects in the formation of the interatrial septum*

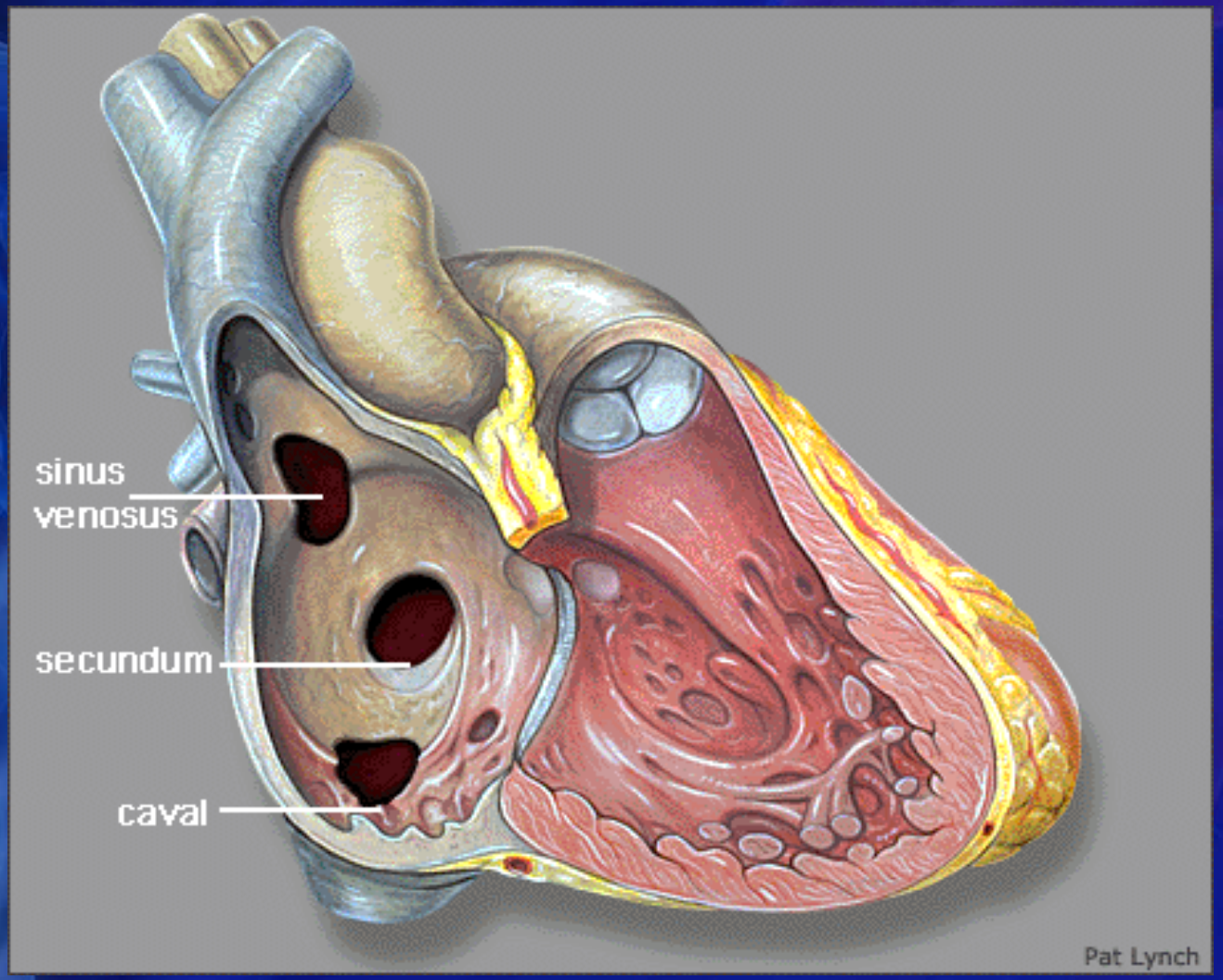




Atrial Septal Defect

- Morphology. classified according to their location as
- 1. Secundum ASDs (90% of all ASDs) result from a deficient or fenestrated oval fossa near the center of the atrial septum; may be of any size, be single or multiple, or be fenestrated.
- 2. Primum anomalies (5% of ASDs) occur adjacent to the AV valves.
- 3. Sinus venosus defects (5%) are located near the entrance of the superior vena cava

Atrial Septal Defect



Atrial Septal Defect

- ***Clinical Features***

- ***ASDs result in a left-to-right shunt, largely because pulmonary vascular resistance is less than systemic vascular resistance***

- ***Because the compliance (distensibility) of the right ventricle is much greater than that of the left. Pulmonary blood flow may be two to four times normal.***

Atrial Septal Defect

- *A murmur is often present as a result of excessive flow through the pulmonary valve.*
- *Usually do not become symptomatic before age 30*
- *Surgical or catheter-based closure of an ASD reverses the hemodynamic abnormalities and prevents complications.*
- *Mortality is low. Long-term survival is comparable to that of the normal population.*

Ventricular Septal Defect

- ***MC** form of CHD. **Most VSDs associated with TOF** only 20% to 30% are isolated.*

***Morphology**.*

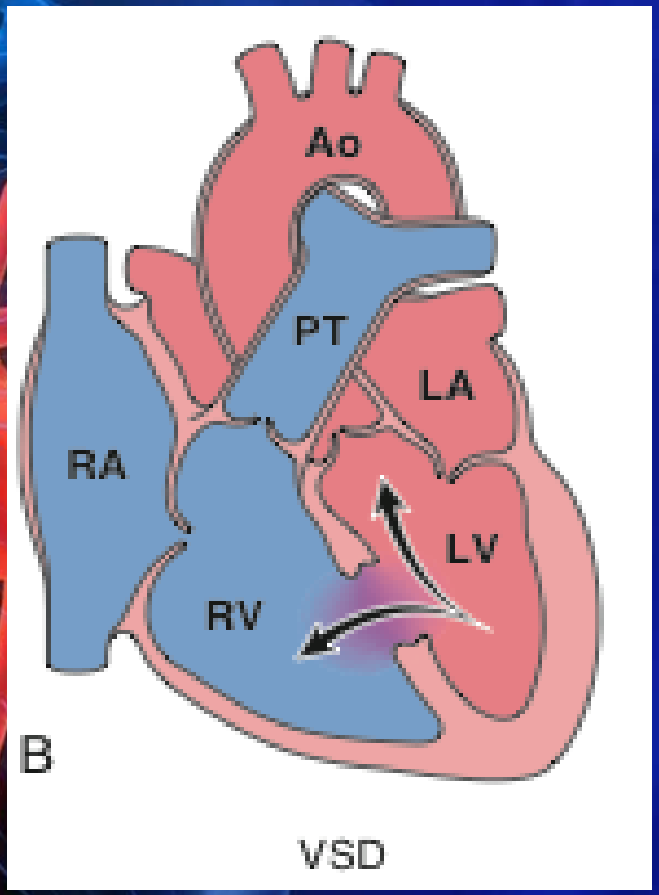
- *VSDs are classified according to their size and location.*

- *About **90%** involve the region of the membranous interventricular septum (**membranous VSD**)*

- *Remainder lie below the pulmonary valve (**infundibular VSD**) or within the muscular septum.*

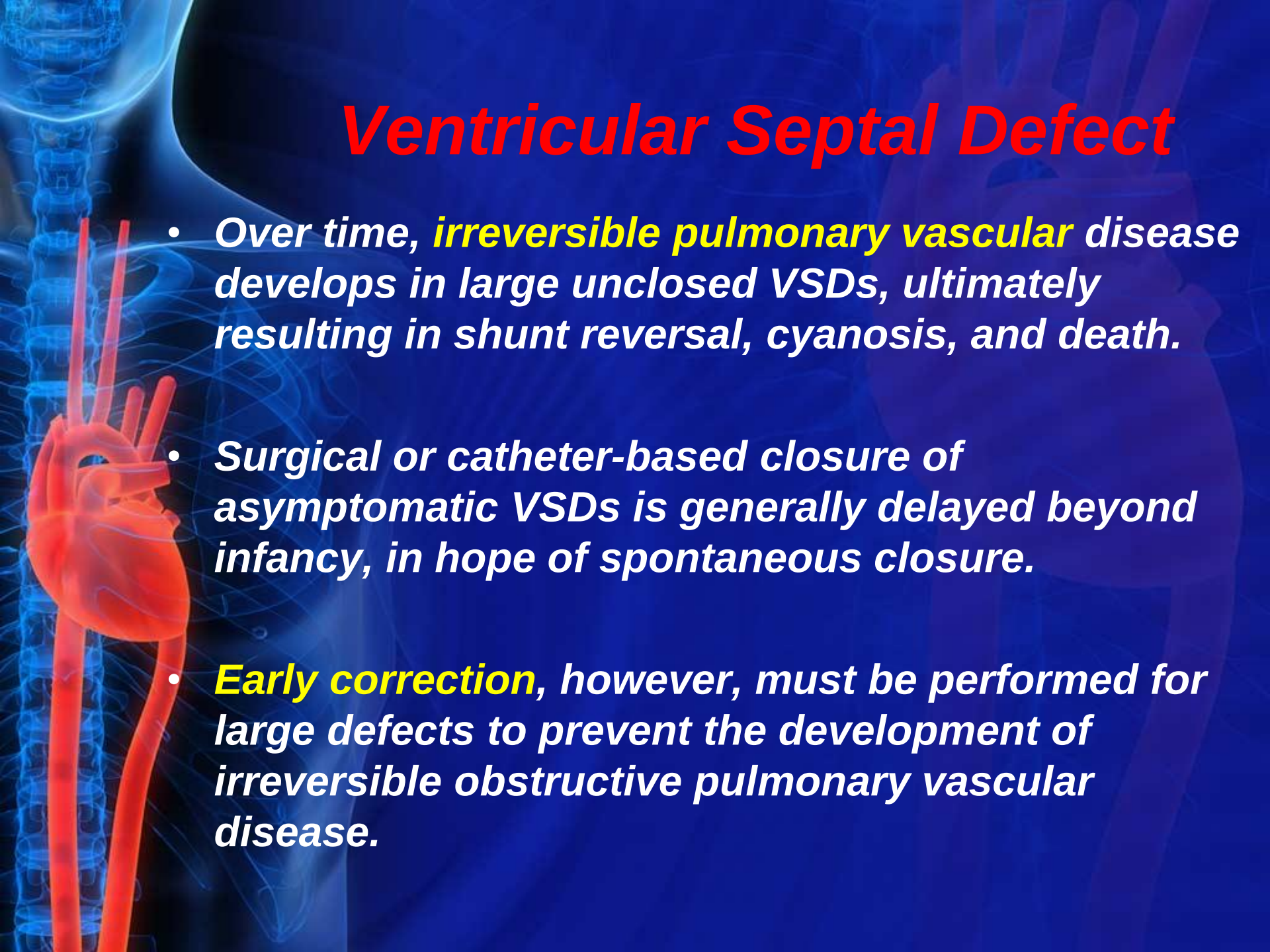
- *Most are single, those in the muscular septum may be multiple (so-called **“Swiss-cheese”** septum).*

Ventricular Septal Defect



Ventricular Septal Defect

- ***Clinical Features***
- ***Most VSDs that clinically manifest in the pediatric age group are associated with other congenital cardiac anomalies such as Tetralogy of Fallot.***
- ***Only 20% to 30% are isolated, usually in adults***
- ***Large VSDs cause difficulties virtually from birth.***
- ***Approximately 50% of small muscular VSDs close spontaneously.***
- ***Right ventricular hypertrophy and pulmonary hypertension***



Ventricular Septal Defect

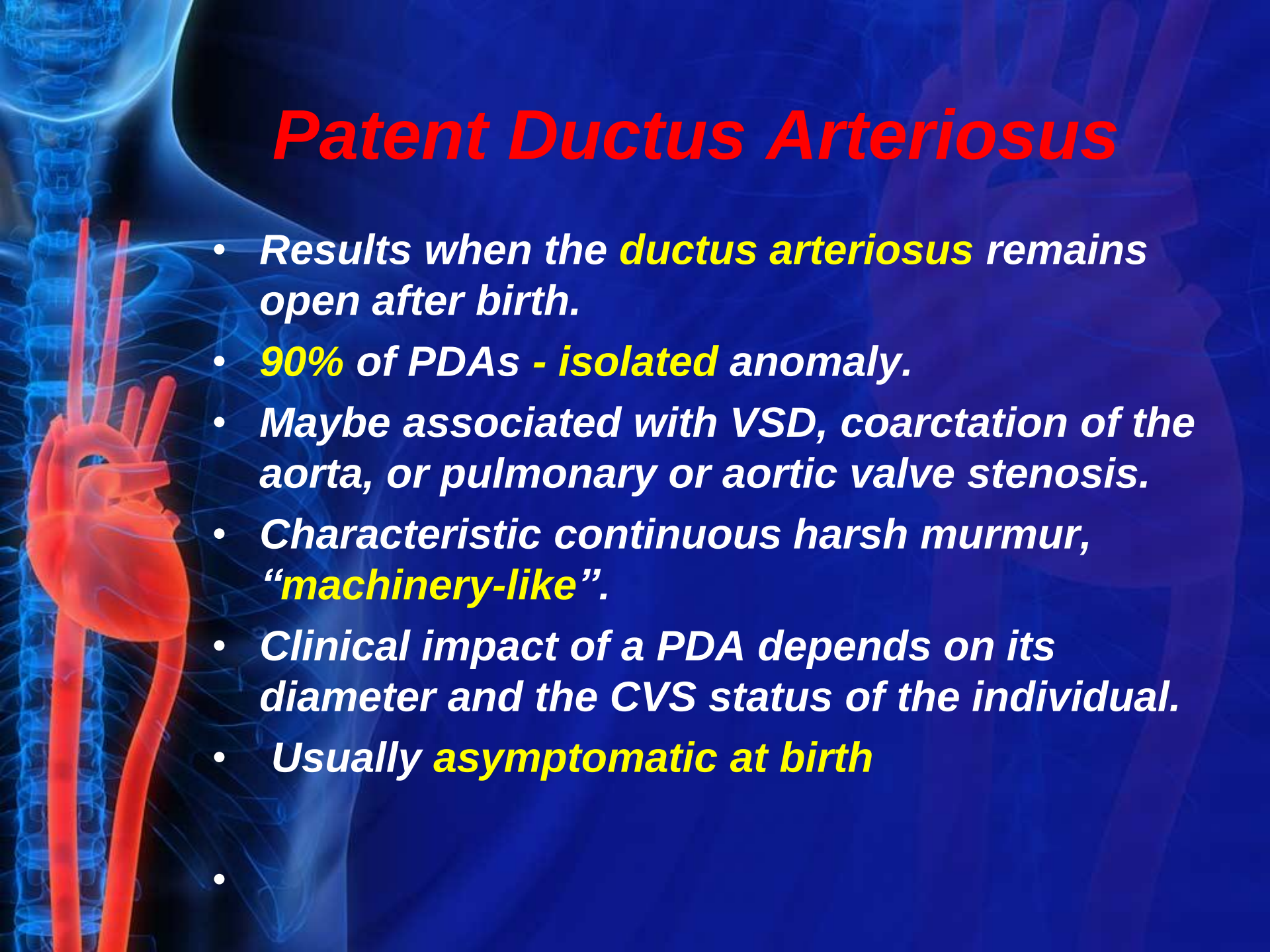
- Over time, irreversible pulmonary vascular disease develops in large unclosed VSDs, ultimately resulting in shunt reversal, cyanosis, and death.
- Surgical or catheter-based closure of asymptomatic VSDs is generally delayed beyond infancy, in hope of spontaneous closure.
- Early correction, however, must be performed for large defects to prevent the development of irreversible obstructive pulmonary vascular disease.

Patent Ductus Arteriosus

Ductus arteriosus arises from the pulmonary artery and joins the aorta just distal to the origin of the left subclavian artery.

During intrauterine life, it permits blood flow from the pulmonary artery to the aorta, thereby bypassing the Non-oxygenated lungs.

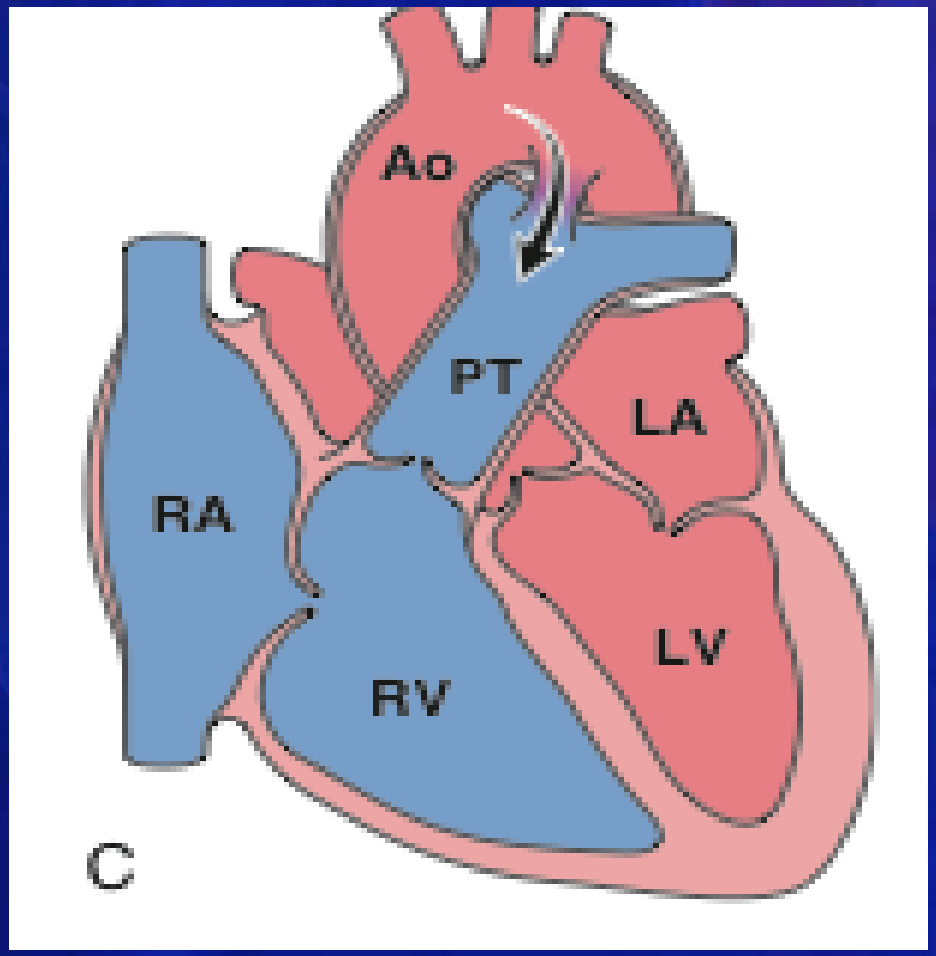
Shortly after birth in healthy term infants, the ductus constricts and is functionally closed after 1 to 2 days; in response to increased arterial oxygenation, decreased pulmonary vascular resistance, and declining local levels of prostaglandin E2



Patent Ductus Arteriosus

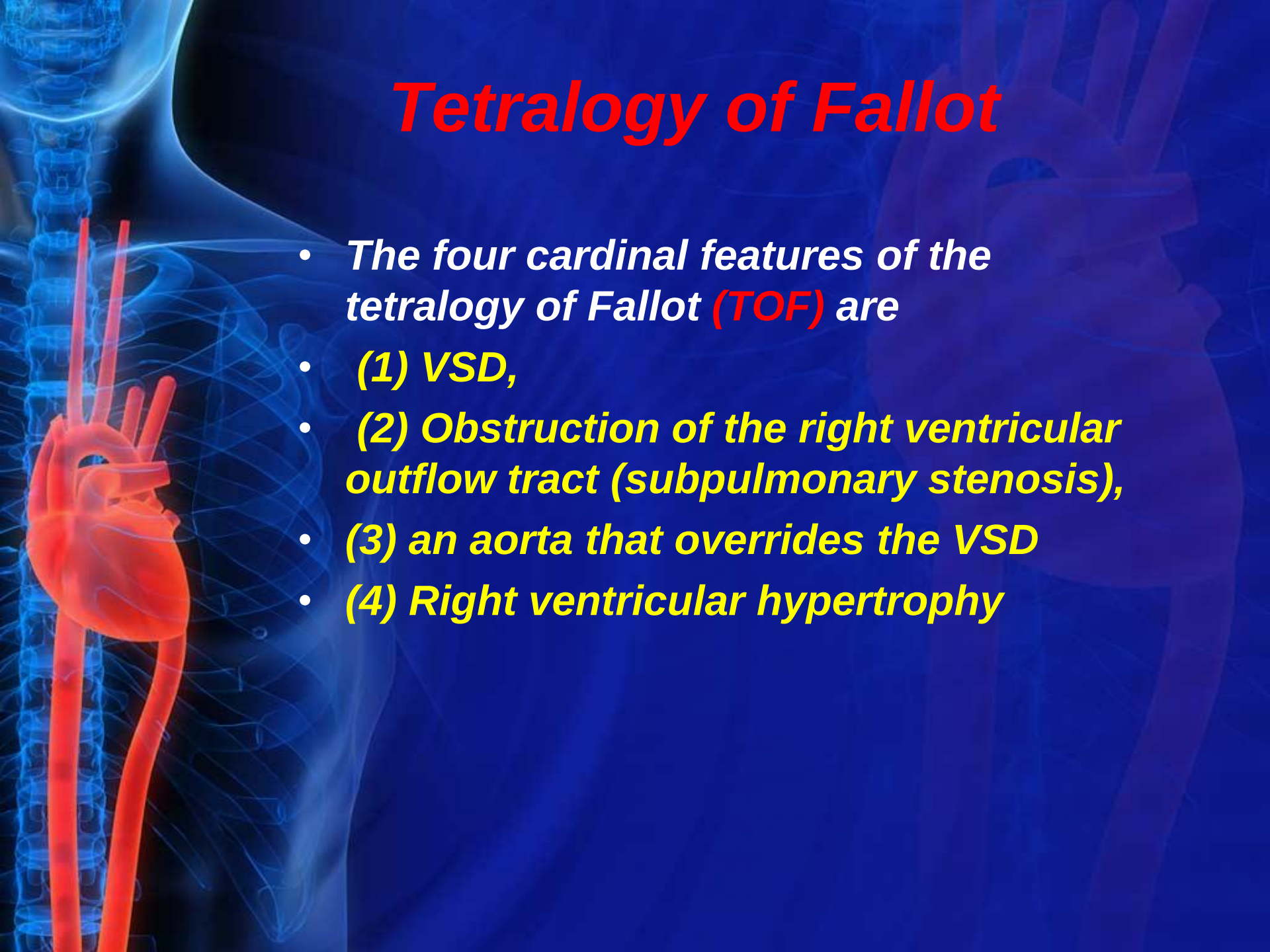
- Results when the ductus arteriosus remains open after birth.
- 90% of PDAs - isolated anomaly.
- Maybe associated with VSD, coarctation of the aorta, or pulmonary or aortic valve stenosis.
- Characteristic continuous harsh murmur, "machinery-like".
- Clinical impact of a PDA depends on its diameter and the CVS status of the individual.
- Usually asymptomatic at birth
-

Patent Ductus Arteriosus



Right to left shunts

- *Tetrology of Fallots*
- *Transposition of the great arteries*
- *Persistent truncus arteriosus*
- *Tricuspid atresia*
- *Total anomalous pulmonary venous connection.*



Tetralogy of Fallot

- The four cardinal features of the tetralogy of Fallot (TOF) are
- (1) VSD,
- (2) Obstruction of the right ventricular outflow tract (subpulmonary stenosis),
- (3) an aorta that overrides the VSD
- (4) Right ventricular hypertrophy

Tetralogy of Fallot

- All features result embryologically from ant-sup displacement of infundibular septum.

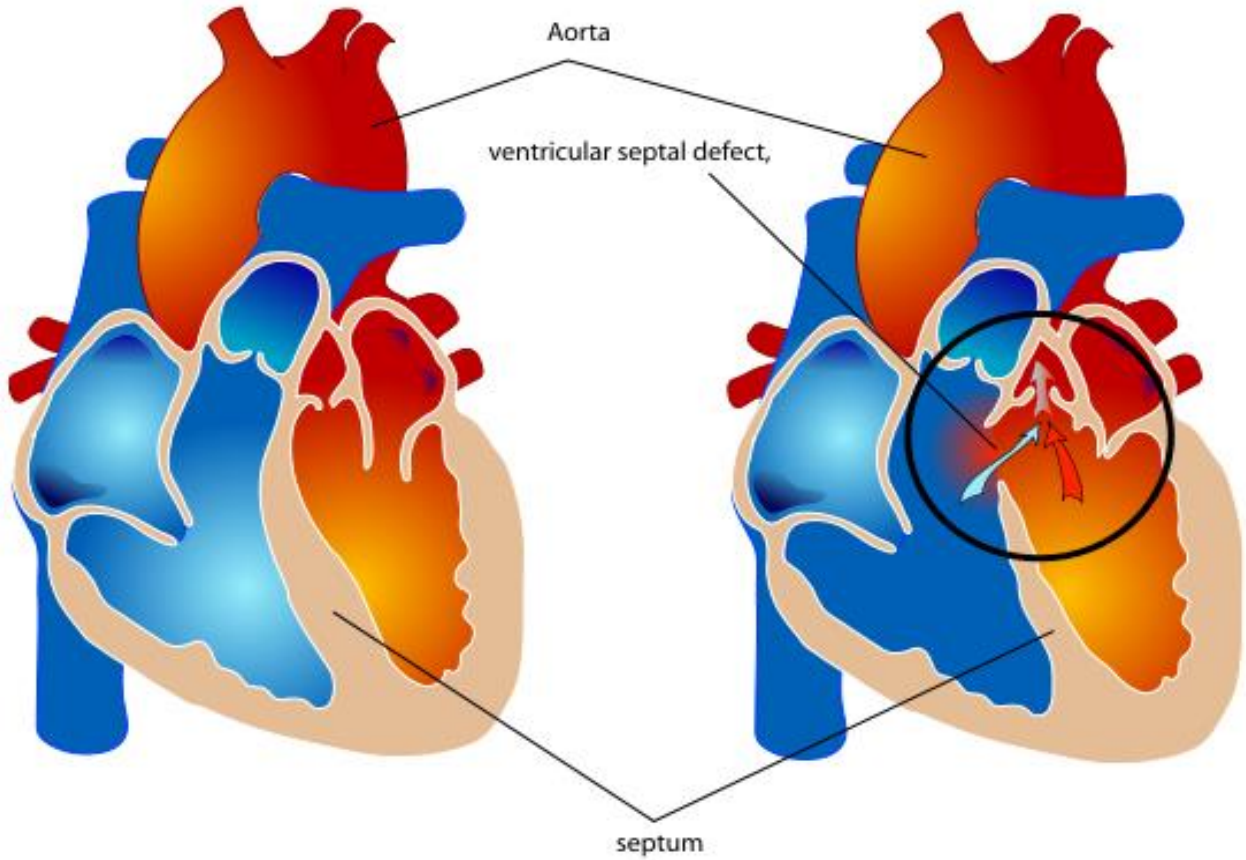
Morphology

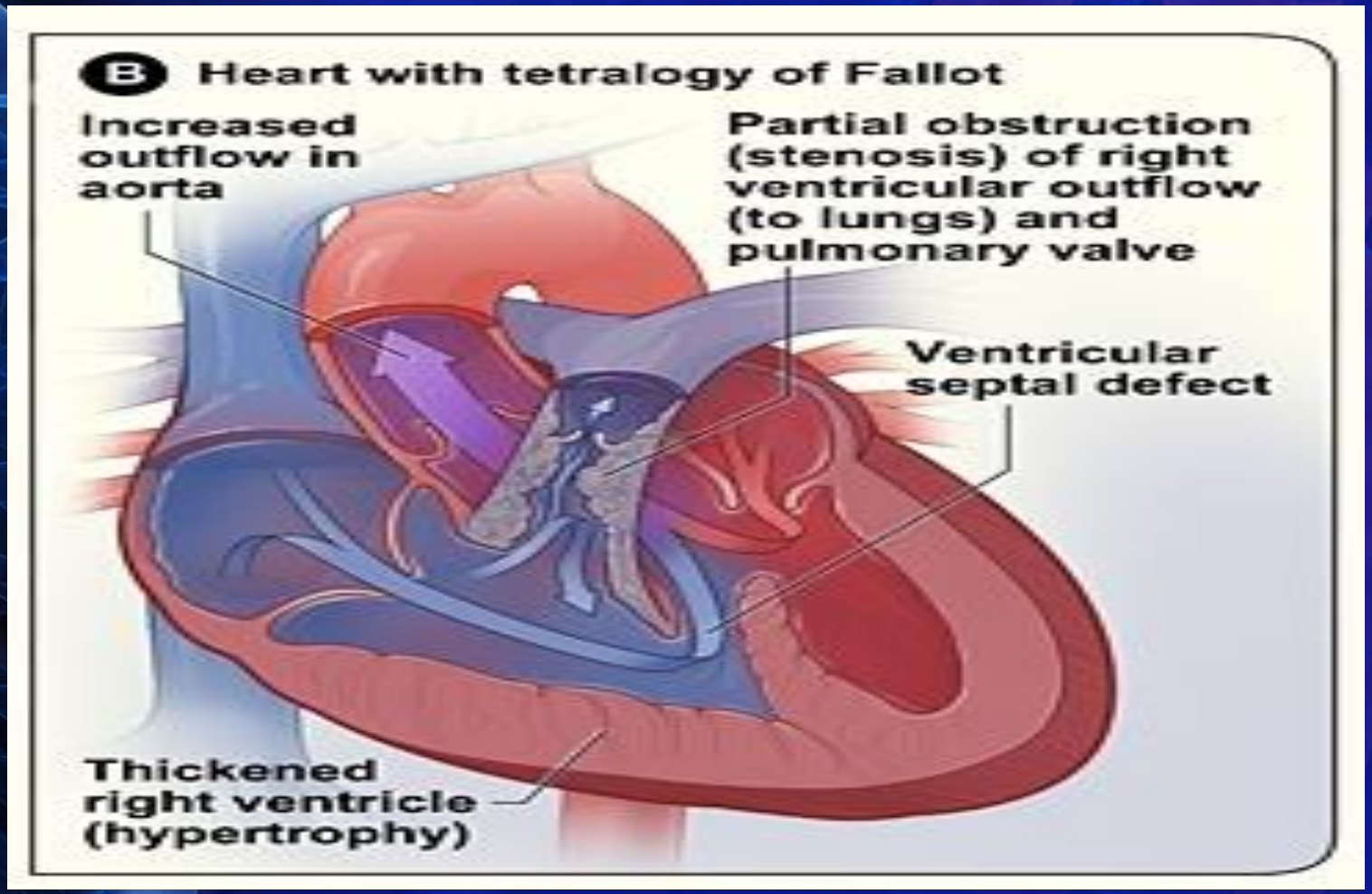
- The heart is often enlarged and may be "boot-shaped" due to right ventricular hypertrophy, particularly of the apical region.

- The VSD is usually large.

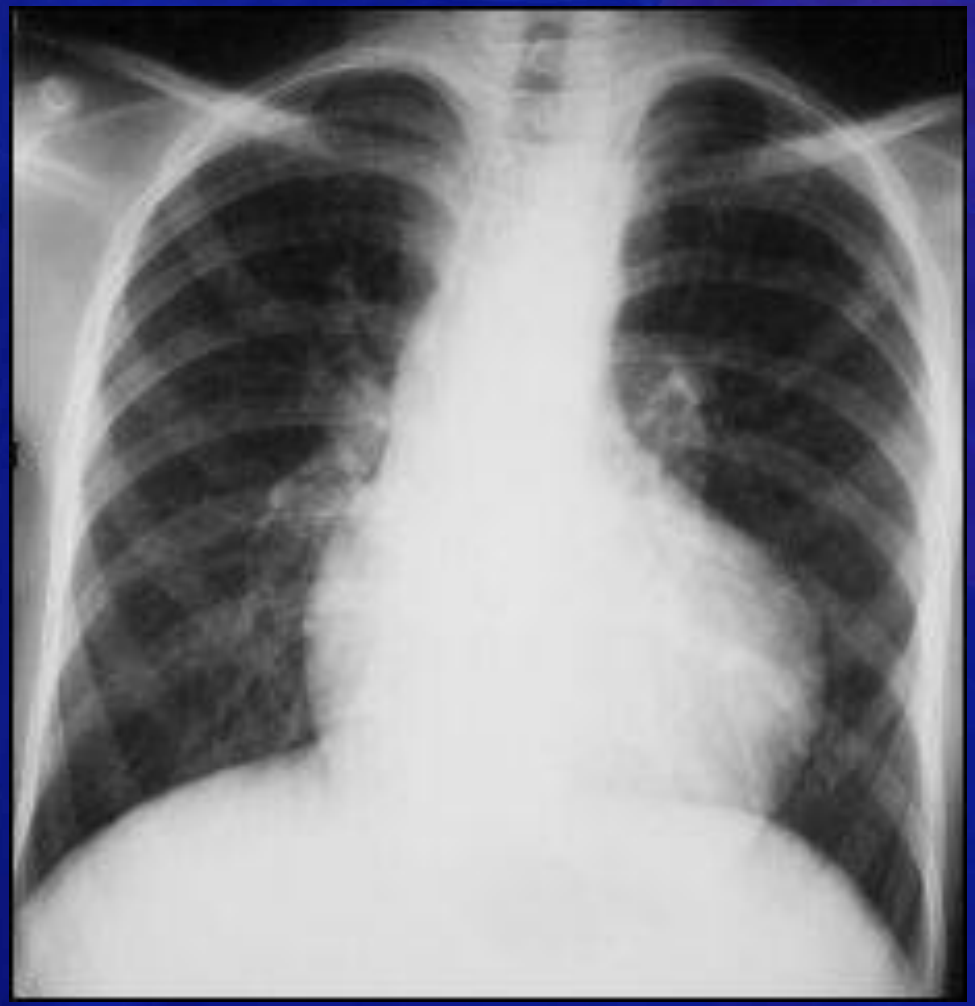
- The obstruction to right ventricular outflow is most often due to narrowing of the infundibulum (subpulmonic stenosis) but can be accompanied by pulmonary valvular stenosis.

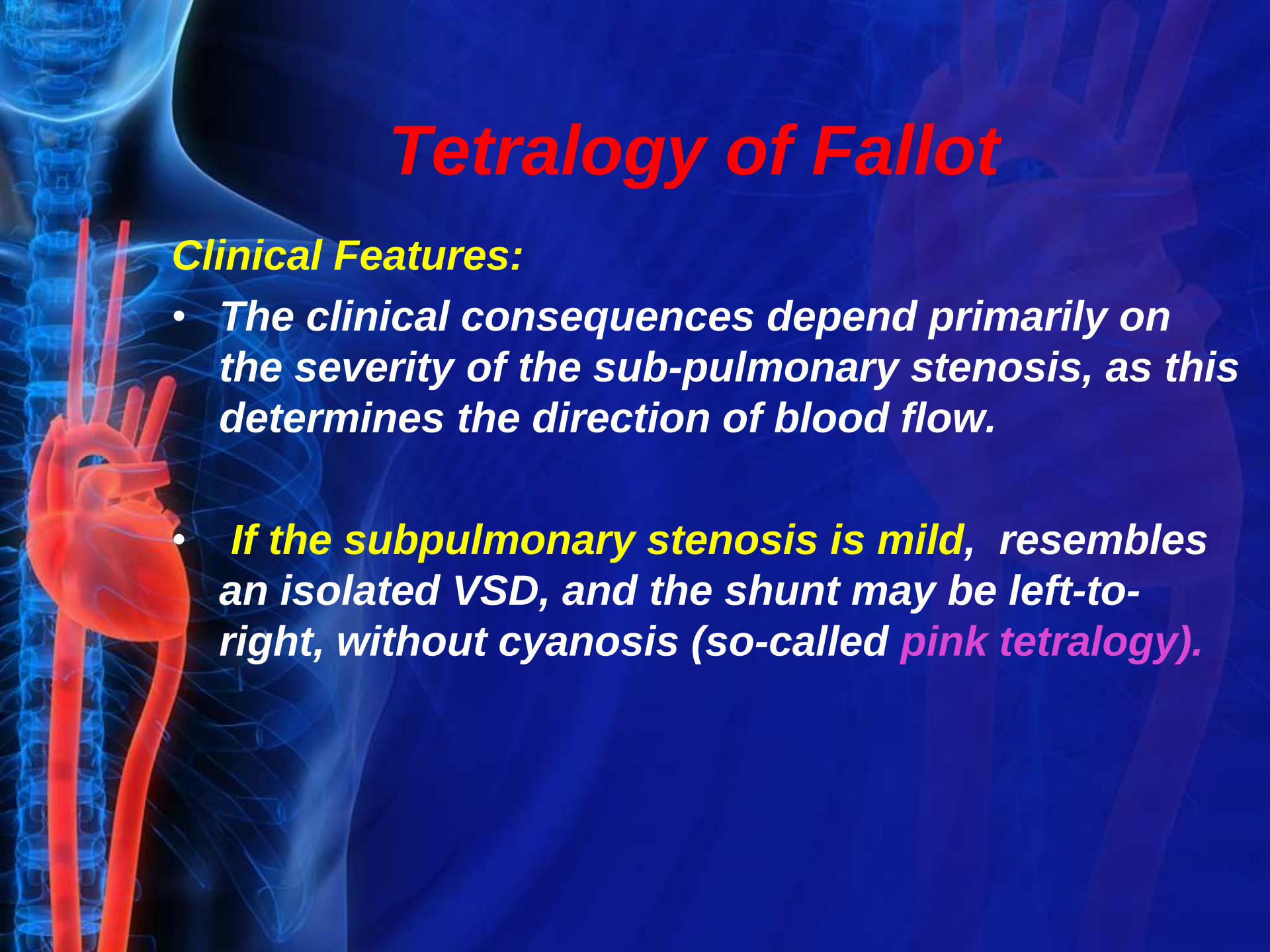
Overriding Aorta




B Heart with tetralogy of Fallot
Increased outflow in aorta
Partial obstruction (stenosis) of right ventricular outflow (to lungs) and pulmonary valve
Ventricular septal defect
Thickened right ventricle (hypertrophy)

TOF “boot-shaped” Heart





Tetralogy of Fallot

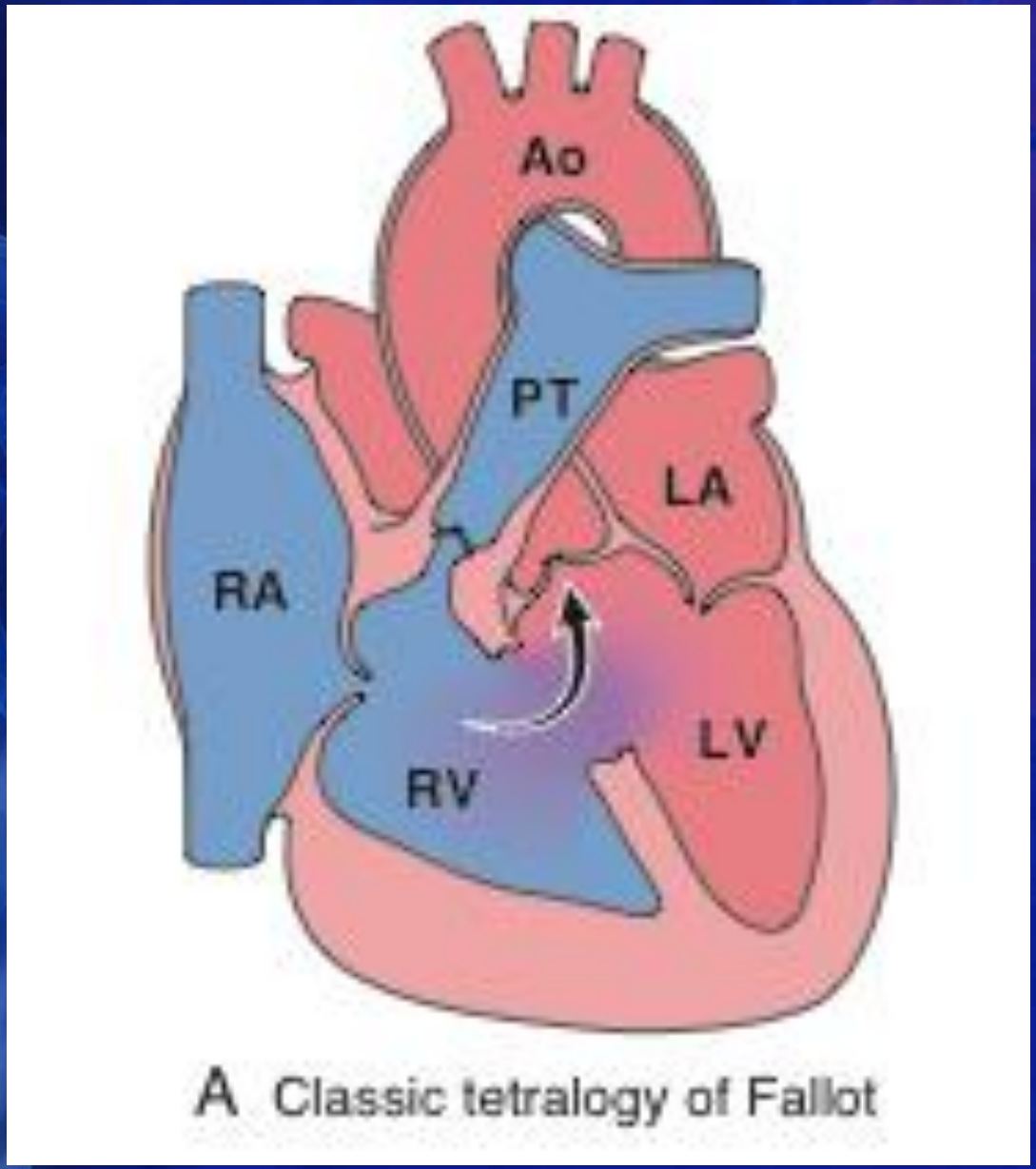
Clinical Features:

- ***The clinical consequences depend primarily on the severity of the sub-pulmonary stenosis, as this determines the direction of blood flow.***

- ***If the subpulmonary stenosis is mild, resembles an isolated VSD, and the shunt may be left-to-right, without cyanosis (so-called pink tetralogy).***

Tetralogy of Fallot

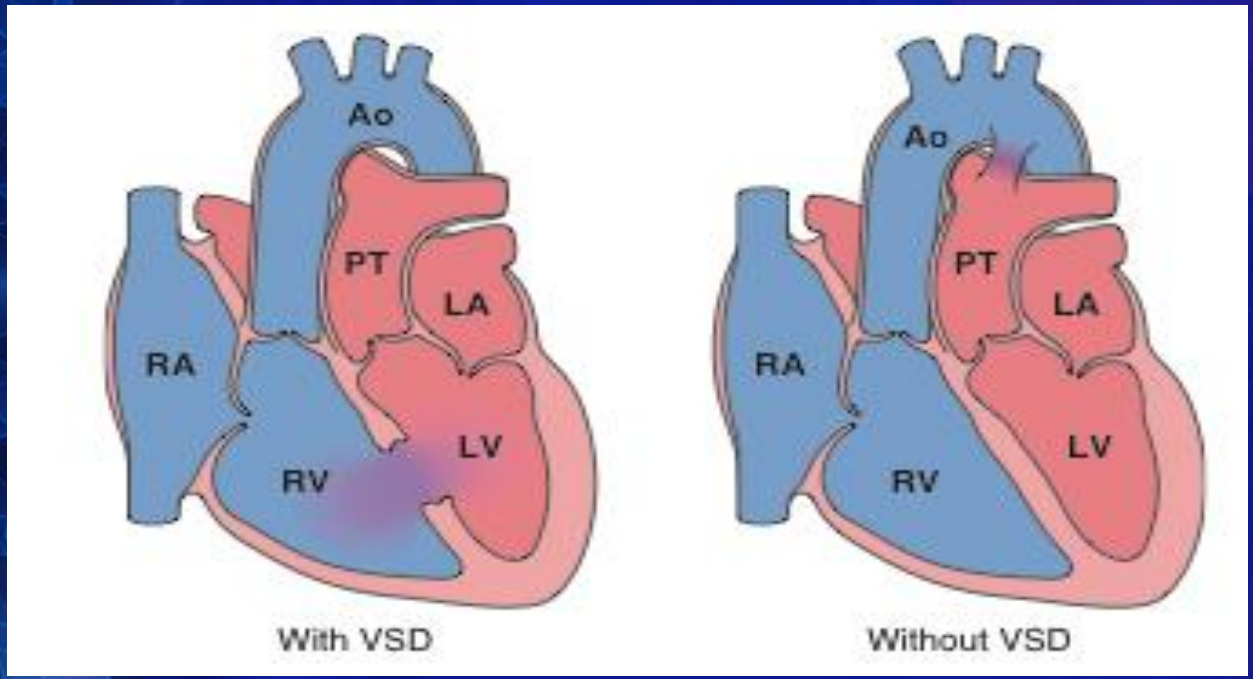
- As right-sided pressures approach or exceed left-sided pressures, right-to-left shunting develops, producing cyanosis (classic TOF).
- With increasingly severe subpulmonic stenosis, the pulmonary arteries become progressively smaller and thinner walled (hypoplastic), and the aorta grows progressively larger in diameter.
- As the child grows and the heart increases in size, the pulmonic orifice does not expand proportionally, making the obstruction progressively worse.



A Classic tetralogy of Fallot

Transposition of the Great Arteries

- ***TGA produces ventriculo-arterial discordance: the aorta arises from the right ventricle, and lies anterior and to the right of the pulmonary artery, which emanates from the left ventricle***



Transposition of the Great Arteries

- *The embryologic defect in complete TGA stems from abnormal formation of the truncal and aortopulmonary septa.*
- *Result in separation of the systemic and pulmonary circulations, a condition incompatible with postnatal life unless a shunt exists for adequate mixing of blood.*

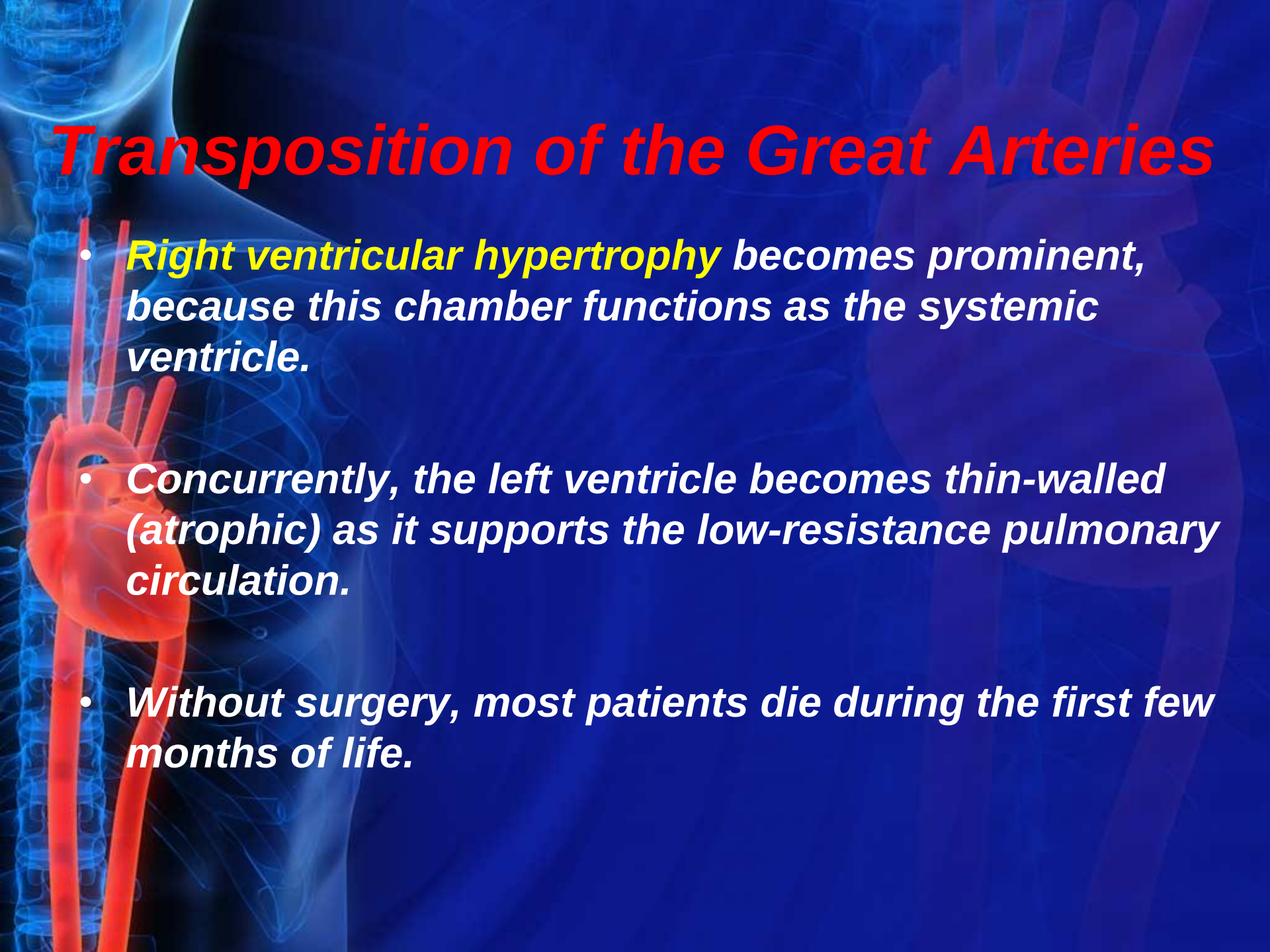
Transposition of the Great Arteries

- *The outlook for infants with TGA depends on the degree of "mixing" of the blood, the magnitude of the tissue hypoxia, and the ability of right ventricle to maintain the systemic circulation.*

- *Patients with TGA and a VSD (~35%) may have a stable shunt.*

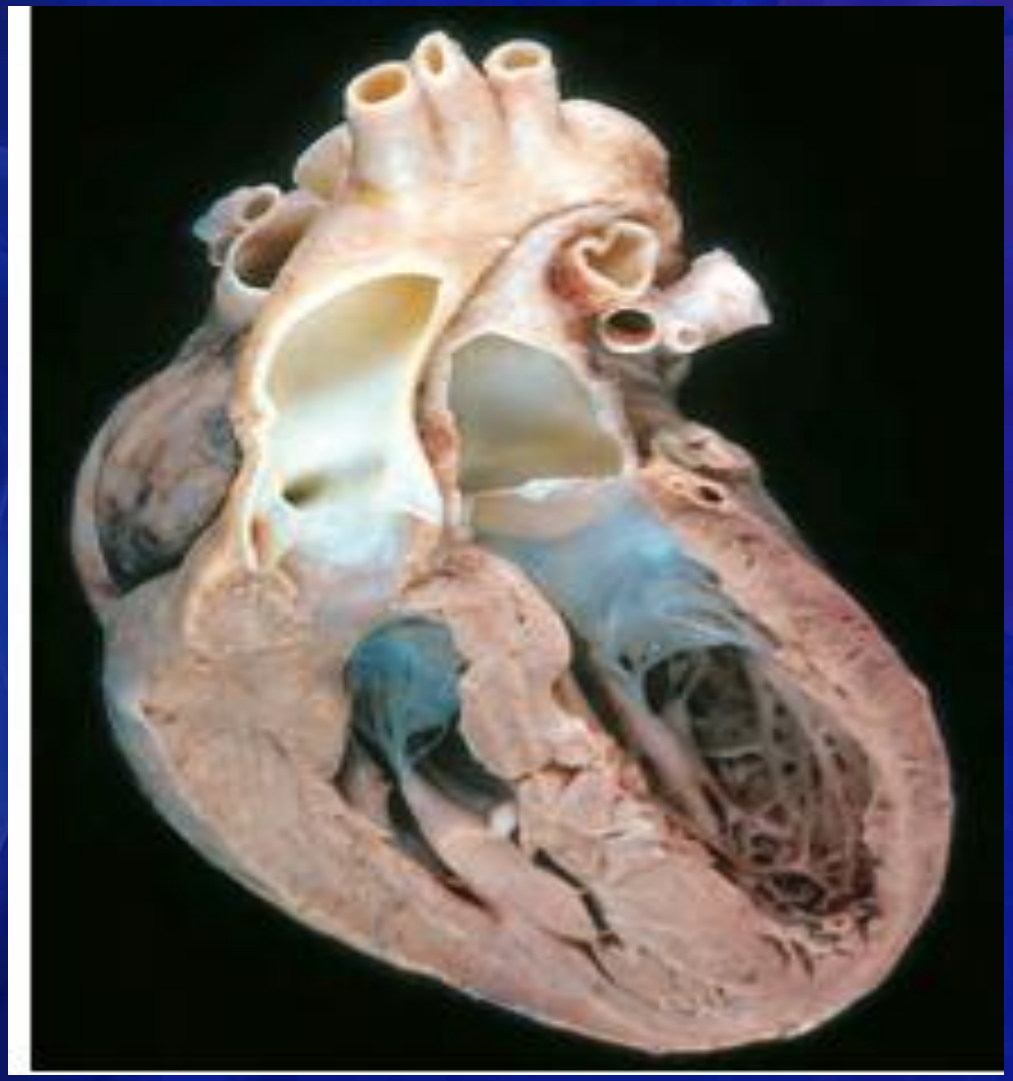
- *Those with only a patent foramen ovale or ductus arteriosus (~65%), have unstable shunts that tend to close and therefore require immediate intervention*

Transposition of the Great Arteries



- ***Right ventricular hypertrophy** becomes prominent, because this chamber functions as the systemic ventricle.*
- *Concurrently, the left ventricle becomes thin-walled (atrophic) as it supports the low-resistance pulmonary circulation.*
- *Without surgery, most patients die during the first few months of life.*

Transposition of the Great Arteries



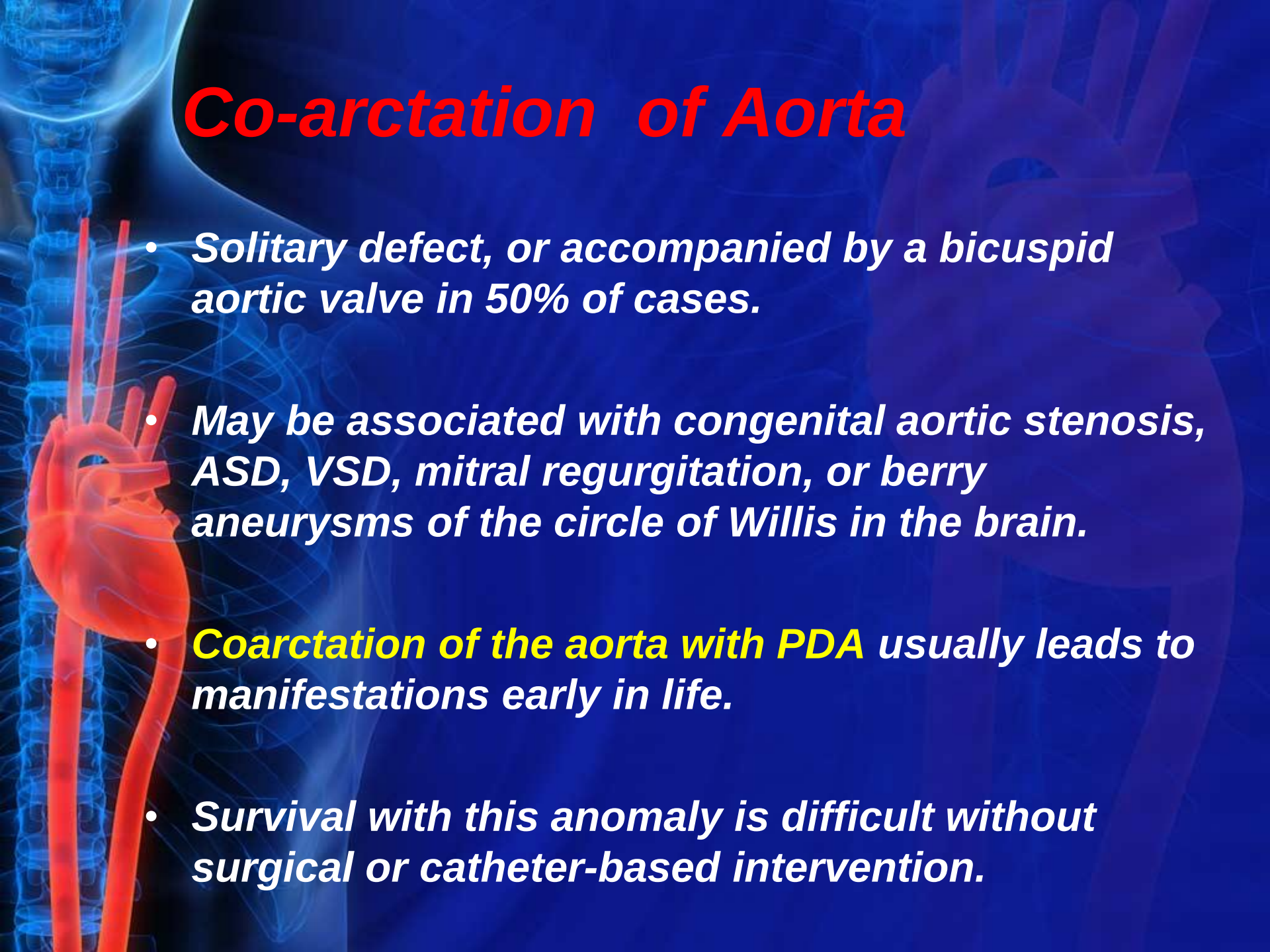
Coarctation of Aorta

- Coarctation (narrowing, constriction) of the aorta ranks high in frequency
- Males : Females 2:1, although females with Turner syndrome frequently have a coarctation

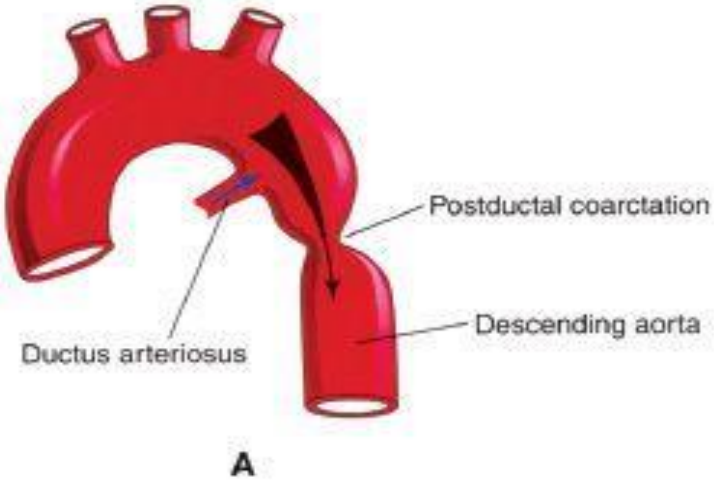
- Two classic forms
- An "infantile" form with tubular hypoplasia of the aortic arch proximal to a patent ductus arteriosus.

- An "adult" form in which there is a discrete ridgelike infolding of the aorta, just opposite the closed ductus arteriosus (ligamentum arteriosum) distal to the arch vessels

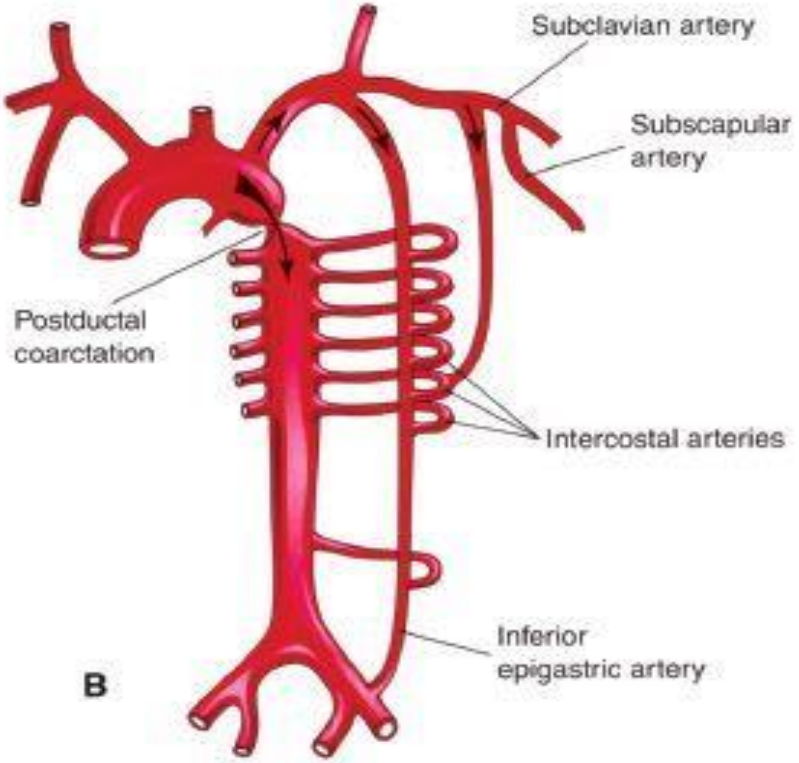
Co-arctation of Aorta



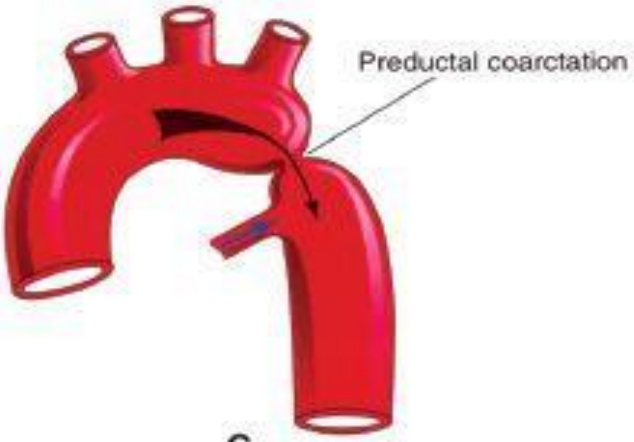
- ***Solitary defect, or accompanied by a bicuspid aortic valve in 50% of cases.***
- ***May be associated with congenital aortic stenosis, ASD, VSD, mitral regurgitation, or berry aneurysms of the circle of Willis in the brain.***
- ***Coarctation of the aorta with PDA usually leads to manifestations early in life.***
- ***Survival with this anomaly is difficult without surgical or catheter-based intervention.***


Postductal coarctation
Ductus arteriosus
Descending aorta

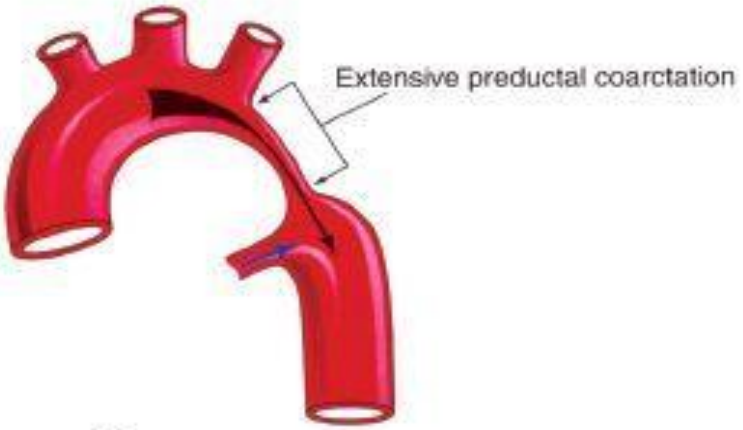
A


Subclavian artery
Subscapular artery
Postductal coarctation
Intercostal arteries
Inferior epigastric artery

B


Preductal coarctation

C


Extensive preductal coarctation

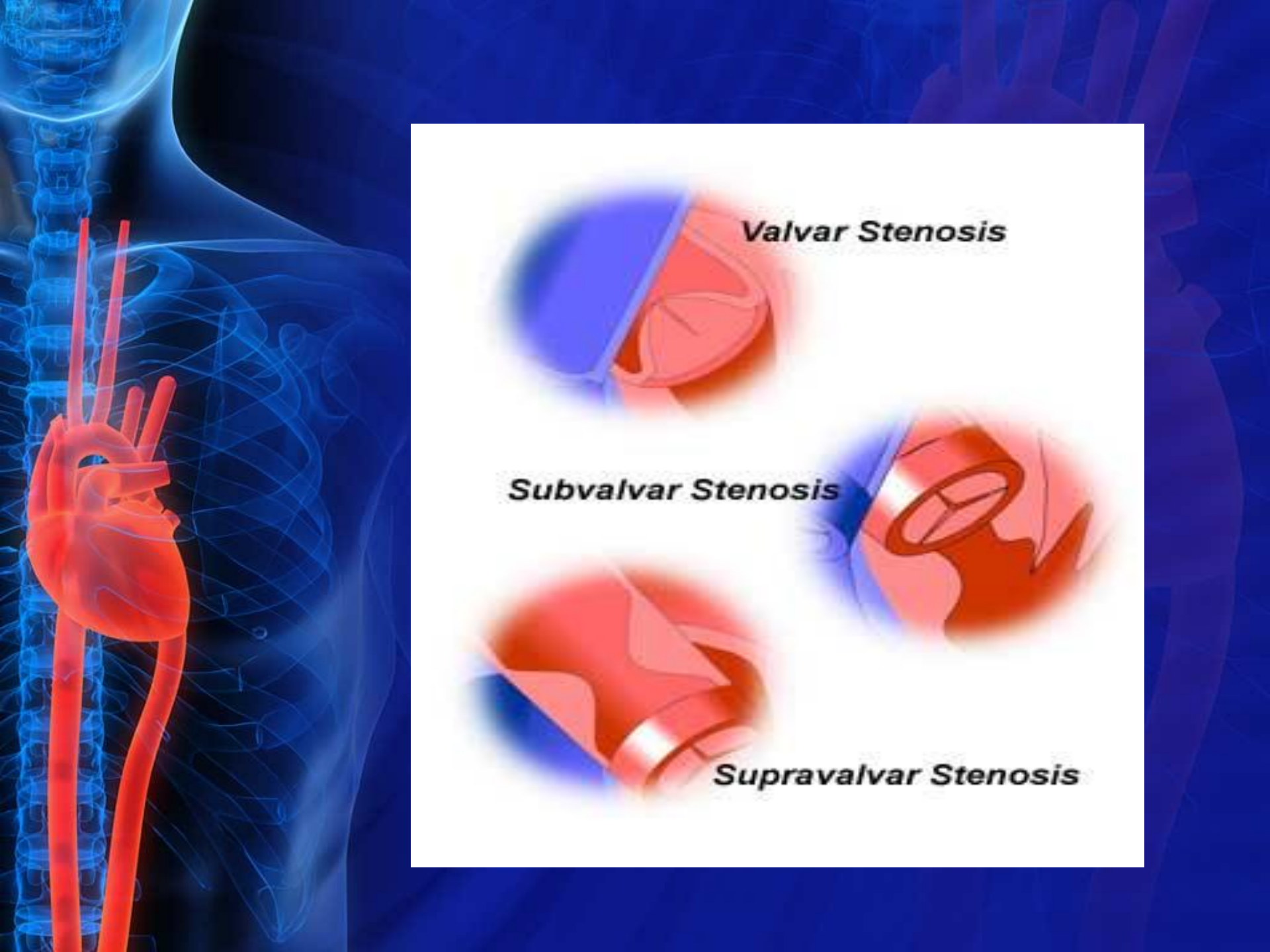
D

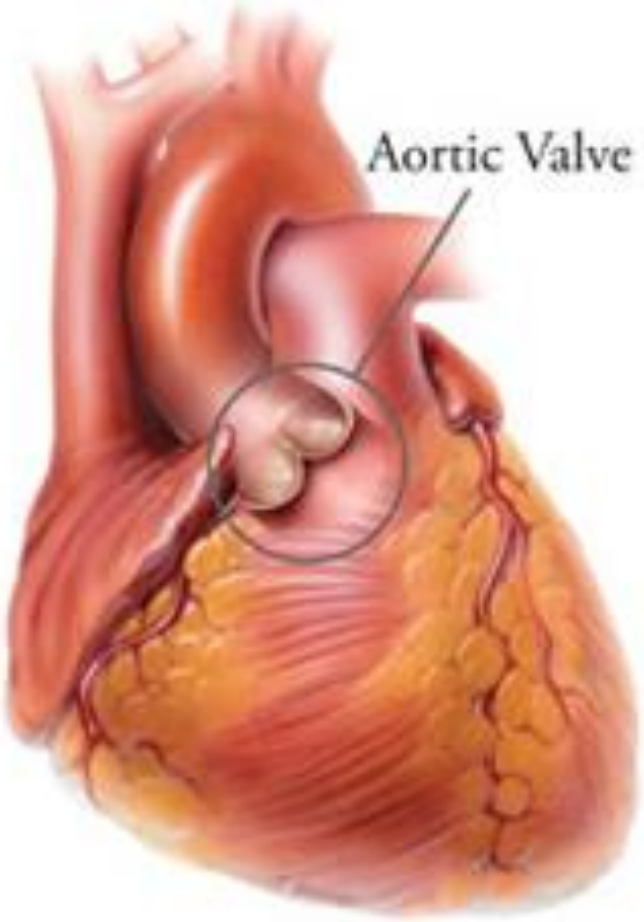
Co-arctation of Aorta

- In such cases, the delivery of unsaturated blood through the PDA produces cyanosis localized to the lower half of the body.
- Coarctation of the aorta without a PDA, unless it is very severe.
- Most children are asymptomatic, and the disease may go unrecognized until well into adult life.
- Typically there is hypertension in the upper extremities; in contrast, there are weak pulses and hypotension in the lower extremities, associated with manifestations of arterial insufficiency

Aortic Stenosis and Atresia

- Congenital narrowing and obstruction of the aortic valve can occur at three locations:
  - valvular
  - subvalvular
  - supravalvular
- Valvular aortic stenosis the cusps may be hypoplastic (small), dysplastic (thickened, nodular), or abnormal in number (usually acommissural or unicommissural).
- Isolated lesion in 80% of cases.


Valvar Stenosis
Subvalvar Stenosis
Supravalvar Stenosis



NORMAL AORTIC VAVLE

Open

Closed

AORTIC VALVE STENOSIS

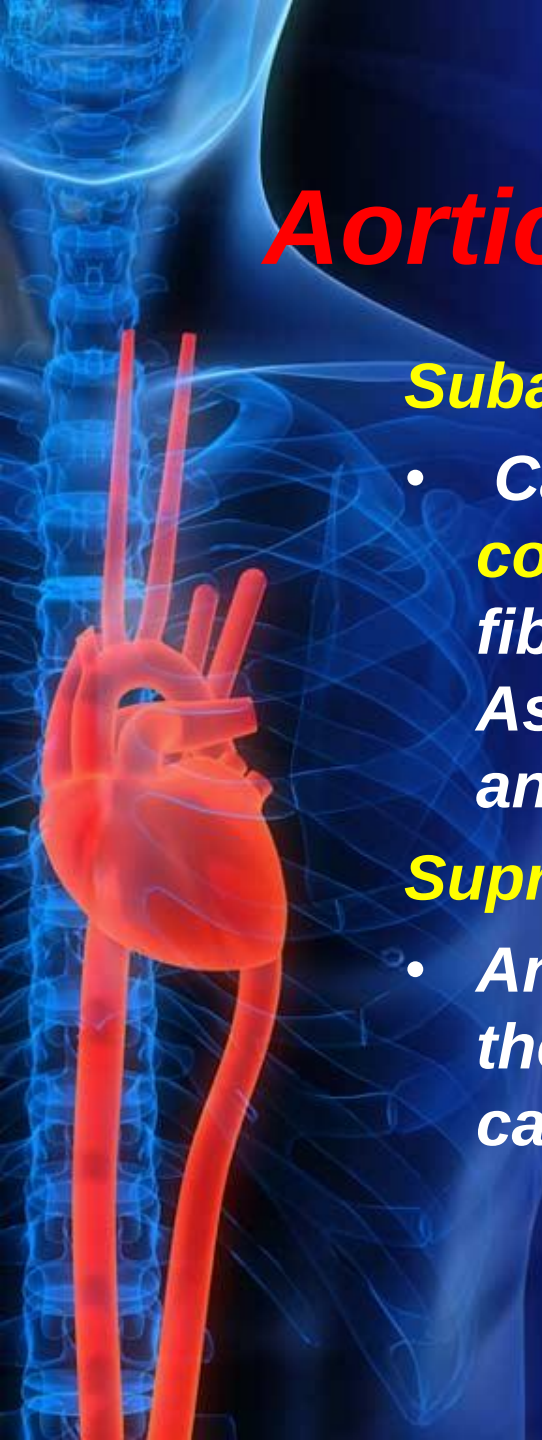
Open

Closed

Aortic Stenosis and Atresia

- *In severe congenital aortic stenosis or atresia, obstruction of the left ventricular outflow tract leads to underdevelopment (hypoplasia) of the left ventricle ascending aorta sometimes accompanied by dense, porcelain-like left ventricular endocardial fibroelastosis.*

- *The ductus must be open to allow blood flow to the aorta and coronary arteries. This constellation of findings, called the hypoplastic left heart syndrome, is nearly always fatal in the first week of life*



Aortic Stenosis and Atresia

Subaortic stenosis

- ***Caused by a thickened ring (discrete type) or collar (tunnel type) of dense endocardial fibrous tissue below the level of the cusps. Associated with a prominent systolic murmur and sometimes a thrill.***

Supravalvular aortic stenosis

- ***An inherited form of aortic dysplasia in which the ascending aortic wall is greatly thickened, causing luminal constriction.***


Thank You