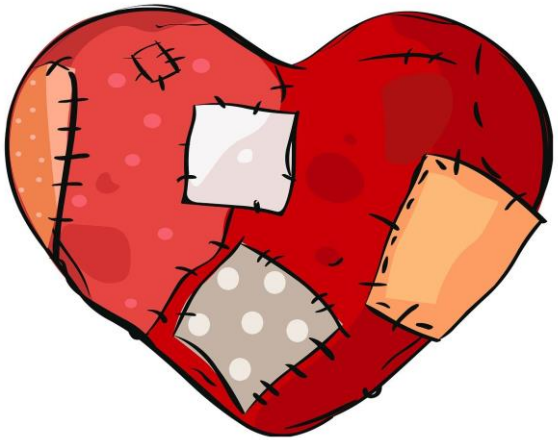
# Adult Congenital Heart Disease



Lenka Kubkova
Masaryk University and University Hospital in Brno, 2020

# Purpose of the Lecture

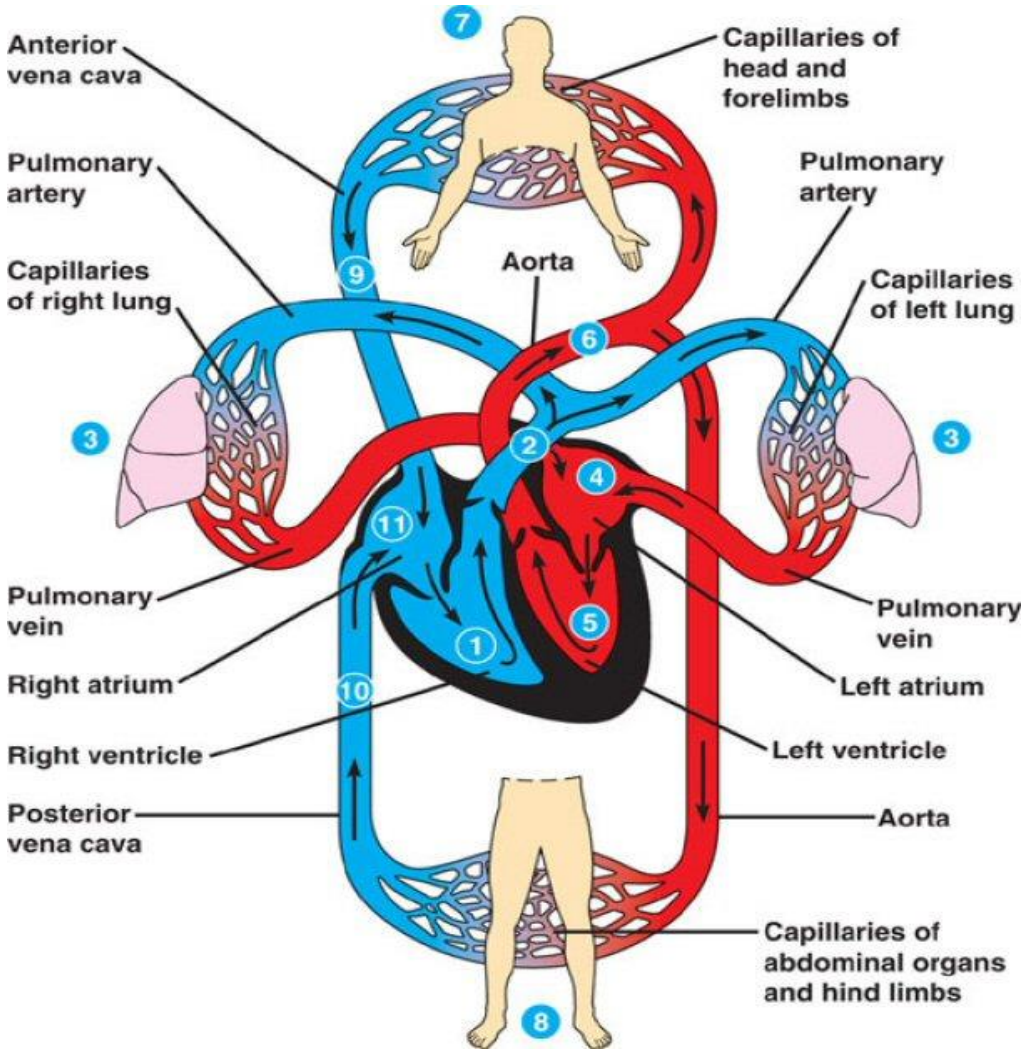
to **remember** you and **explain**:

- basics from blood circulation and hemodynaemics
- basics from anatomy and pathophysiology of the most frequent CHDs - **CHD = Congenital Heart Disease**

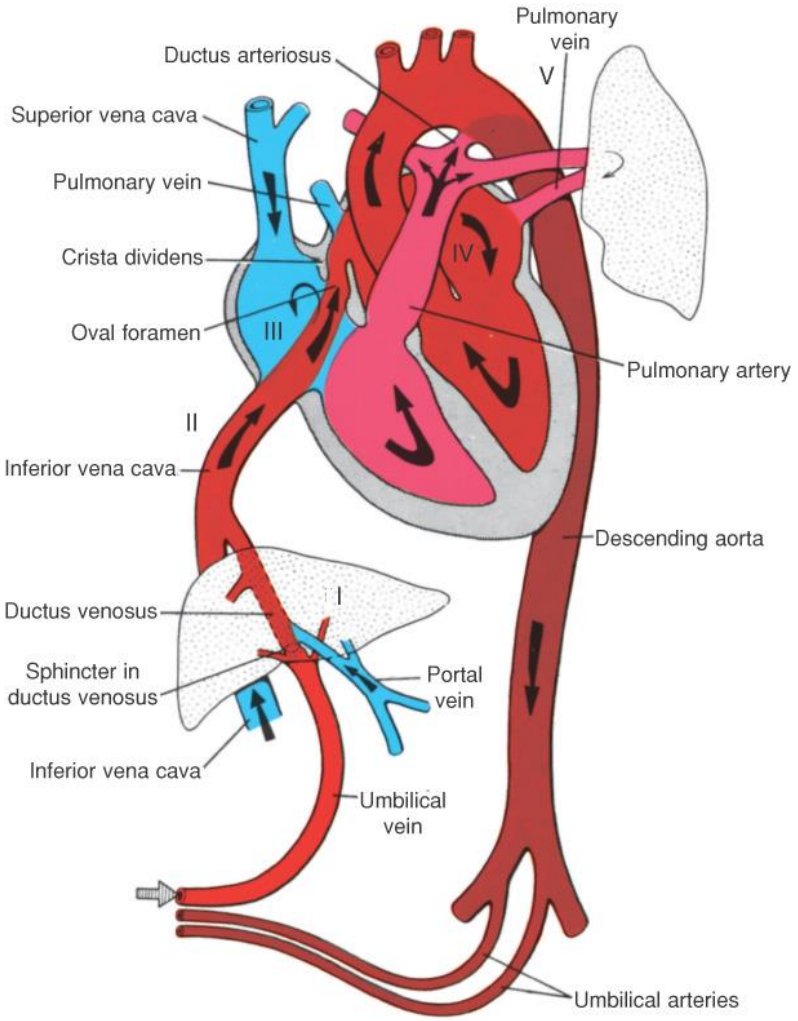
to **demonstrate**:

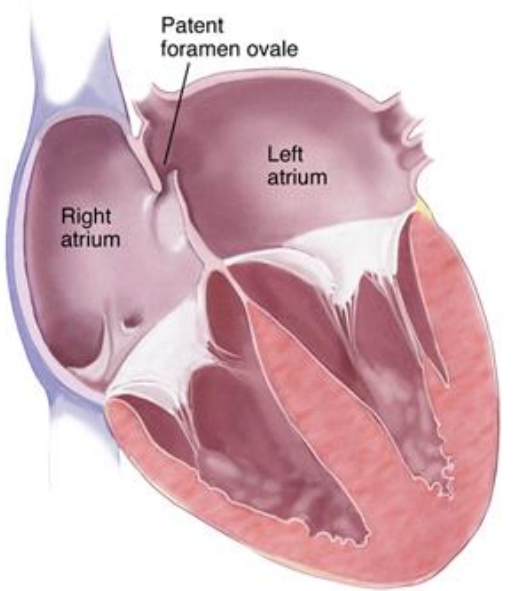
- how does adult patient with CHD look like
- what are his symptoms
- how can we investigate and treat him
- what could be different between „normal“ cardiology patient and that one with CHD

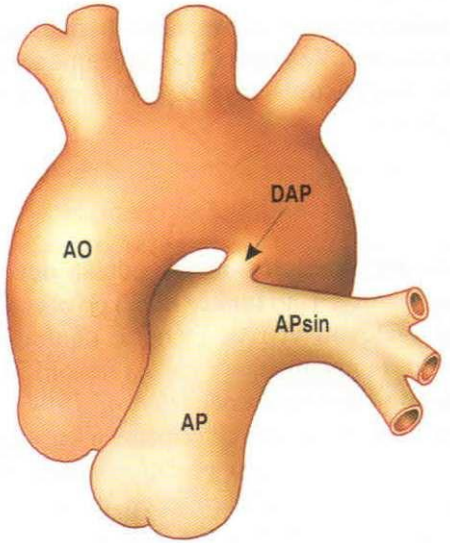
# Systemic and Pulmonary Circulation



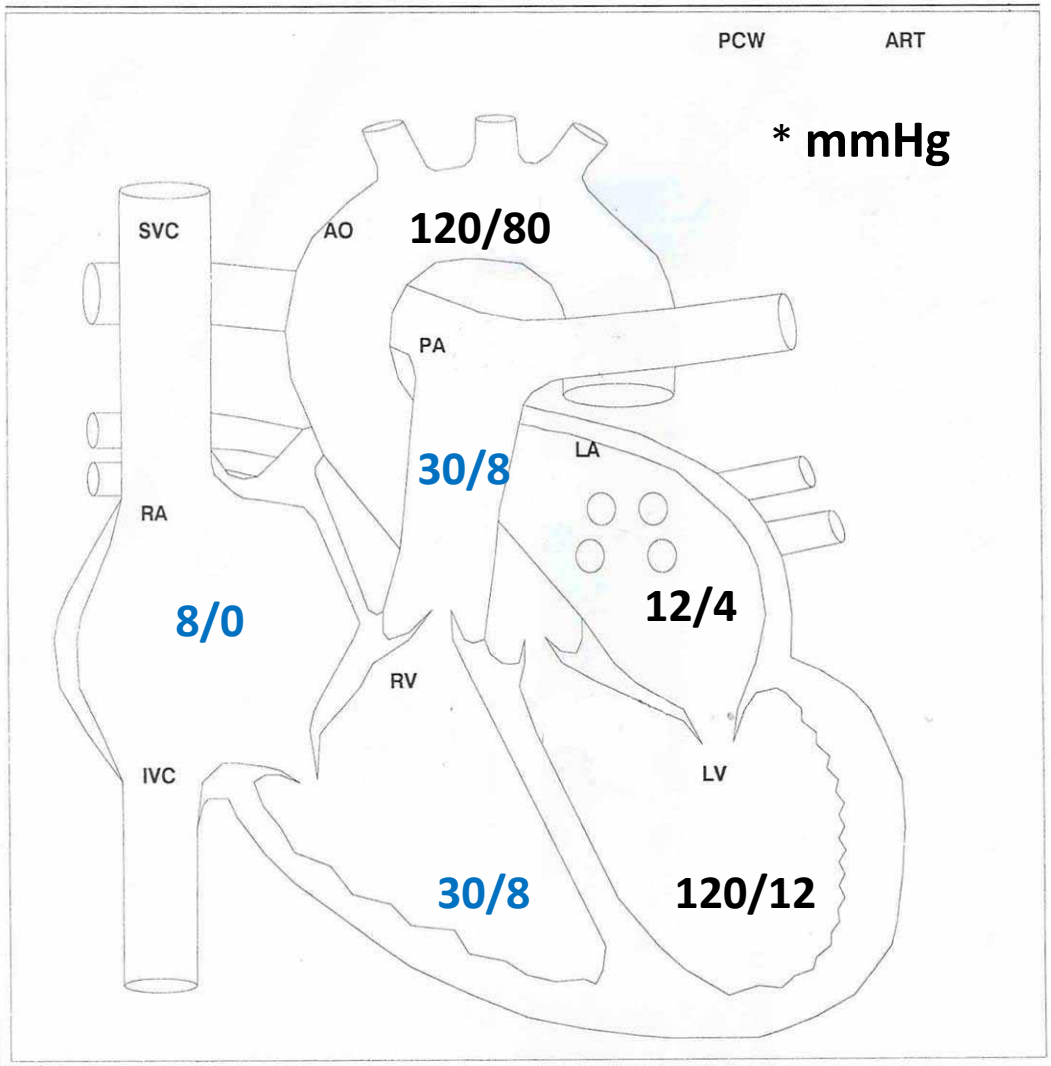
# Fetal Circulation



# Normal Blood Pressures



# Basic Terminology in CHD

- situs solitus
- situs viscerum inversus
- situs ambiguus

  = syndrome of visceral heterotaxis

  dextroisomerism (Ivermark sy)

  levoisomerism
- concordance / discordance
- restrictive / non-restrictive defect
- erythrocytosis

# Definition of Congenital Heart Disease

**Congenital Heart Disease (CHD)**

= morfological disorder of heart / great vessels that has been present since birth

# Nomenclature and Classification

- complexed and complicated (heterogenity)
- **classification according to:**

  **anatomy** (most common; **description of CHD**)

  **physiology**

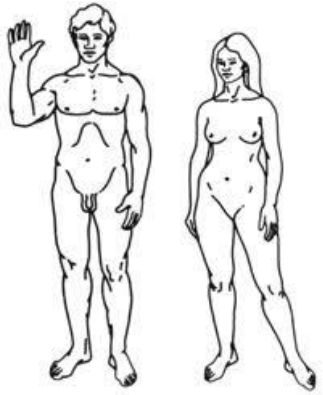
  **outcome** for the patients
- 35% of all CHD are critical disorders requiring immediate intervention

# Most frequent CHD - Review

| CHD type | % adult CHD | % children CHD |
|---|---|---|
| Atrial septal defect | 25-30 | 9 |
| Ventricular septal defect | 21 | 42 |
| Aortic coarctation | 10 | 5 |
| Tetralogy of Fallot | 10 | 3 |
| Pulmonary stenosis | 6-10 | 6 |
| Patent arterial duct | 5 | 5 |
| Transposition of great arteries | 5 | 5 |
| Atrioventricular septal defect | 4 | 4 |
| Ebstein´s anomaly of TV | 2 | 0,4 |
| Pulmonary atresia | 1 | 2 |
| Tricuspid atresia | 0,7 | 0,8 |
| *Aortic stenosis | 2-4% com.pop. | 8 |

# Most frequent CHD

- **adults**



ASD II 25-30%

VSD 21%

- **children**

VSD 42%

ASD II 9%

# Lifetime of Diagnosis of CHD

- some defects need not be presented / detected in early life

(Portion of all CHD)

- 60% in babies < 1 year old
- 30% in children
- 10% in adults

# Epidemiology of CHD

- live birth incidence approx. 6-10 cases per 1000 (1 in every 145 babies born)
- advances in diagnosis and management of pts. with CHD over the latter part of the 20th century
- 80-85% of all children with CHD survive to adulthood
- prevalence in adult population 280 per 100 000

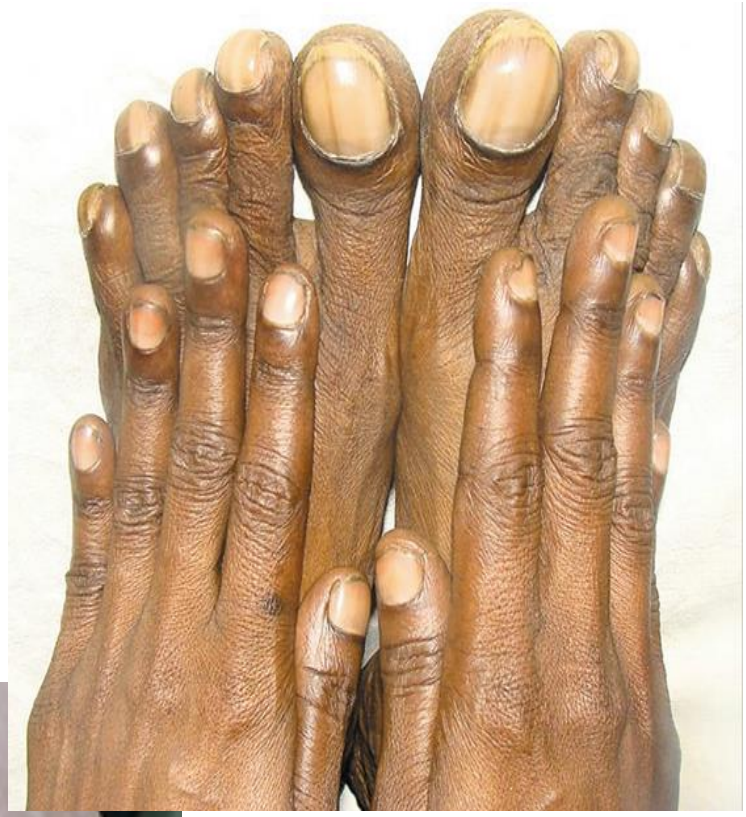
  (CR: 10 000 children and 25 000 adults with CHD)
- there are now more adults than children with CHD

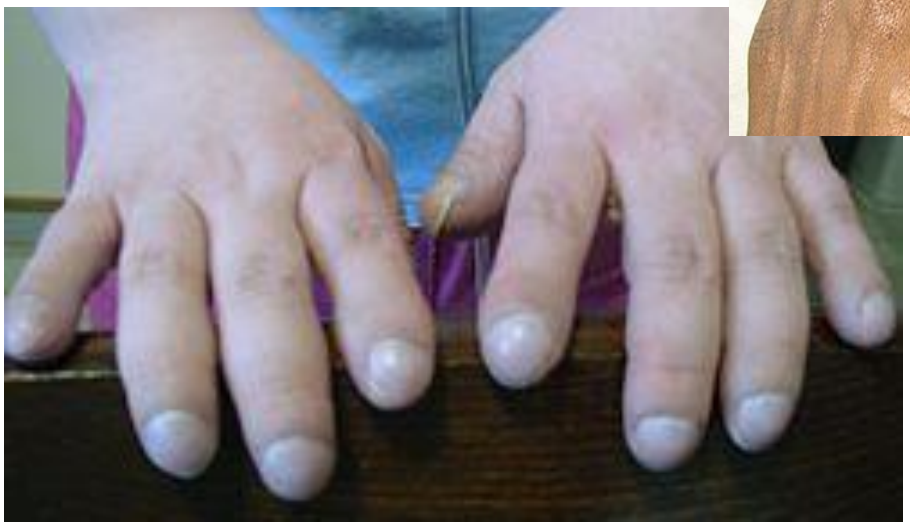
# Findings in Patients with CHD

- physical appearence (syndromes, clubbed fingers, scars)
- cyanosis
- murmurs
- hypoxemia
- hypertension
- pulmonary hypertension
- erythrocytosis (hyperviscosity, sideropenia)
- hyperuricemia, gout
- ecg changes, chest X-ray changes...

# Scars after Cardiothoracic Surgery

# Clubbed Fingers





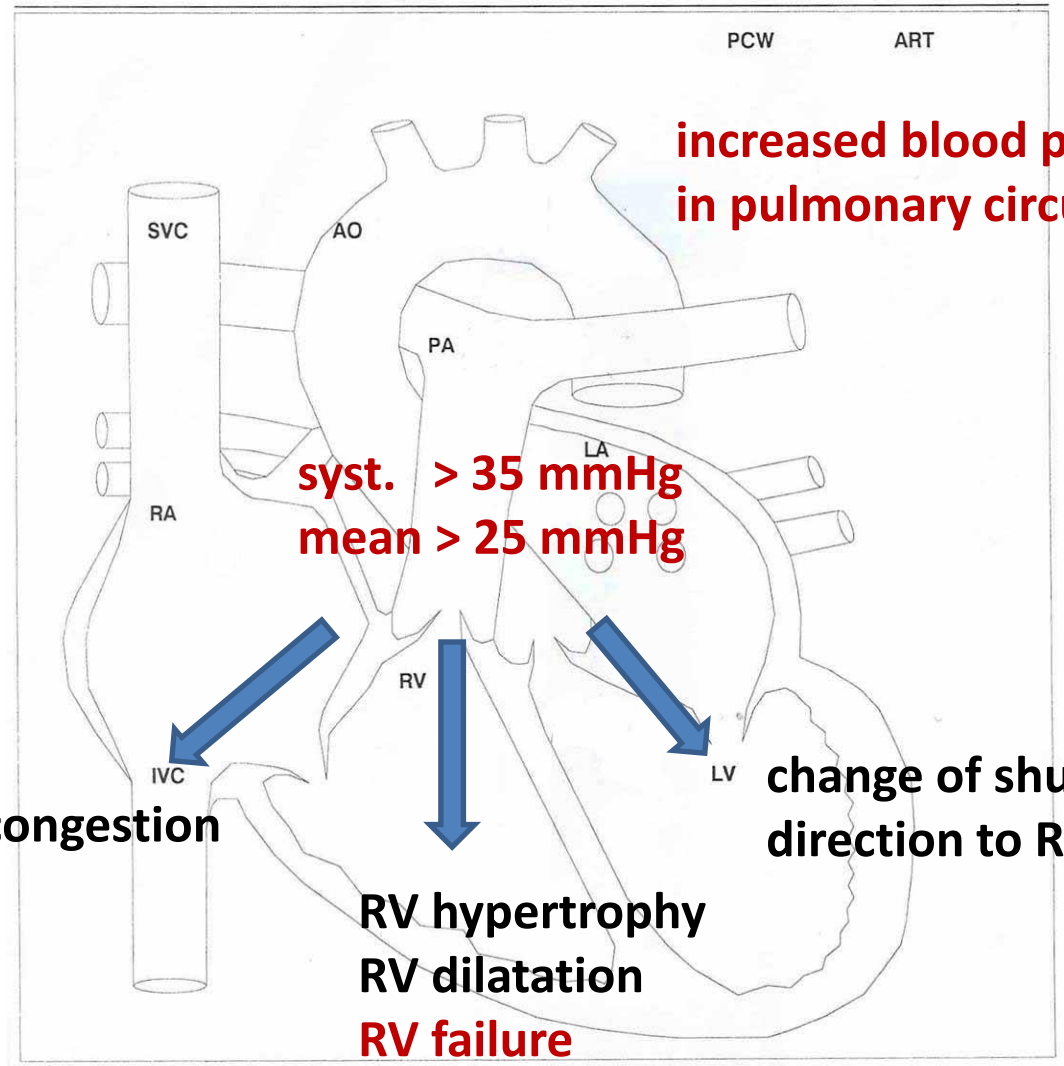
# Cyanosis

Children with Tetralogy of Fallot exhibit bluish skin during episodes of crying or feeding.

# Hypertension (Systemic Arterial)

- BP ≥ 140/90
- both arms measurement
- BP difference between arms and legs

# Pulmonary Hypertension




**increased blood pressure in pulmonary circulation**

**syst. > 35 mmHg**
**mean > 25 mmHg**

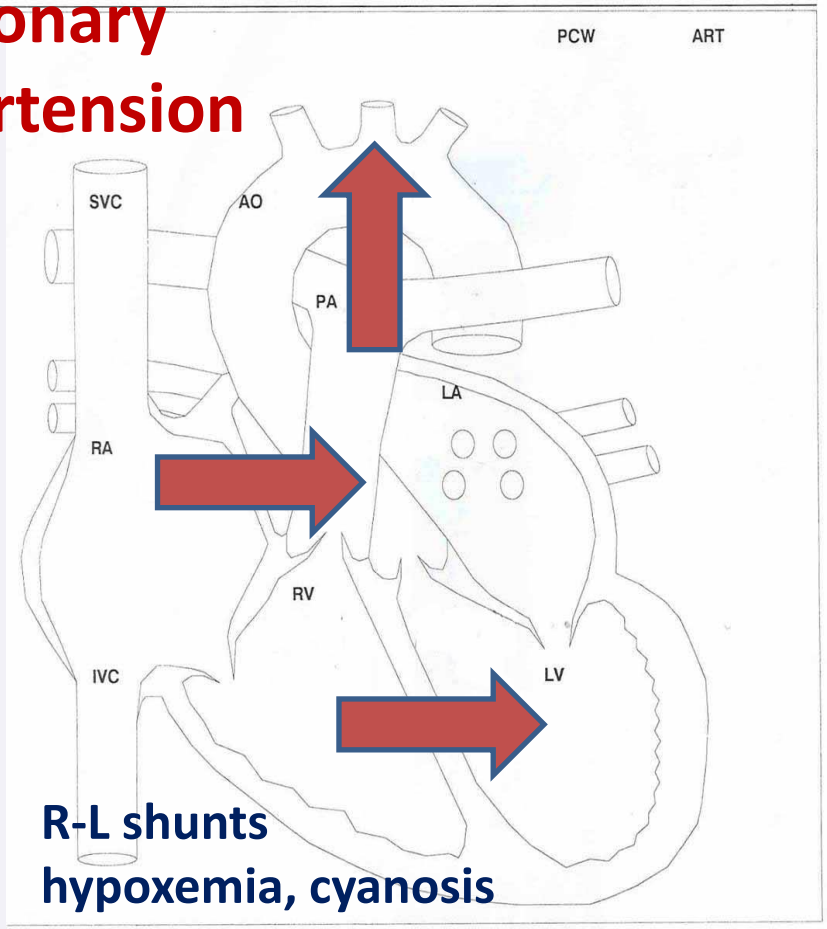
**congestion**

**change of shunt direction to R-L**

**RV hypertrophy**
**RV dilatation**
**RV failure**

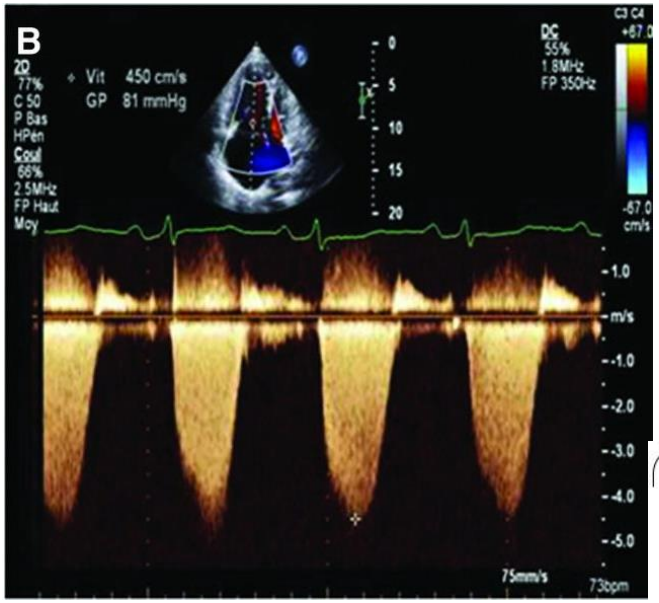
# Eisenmenger Syndrome


**Pulmonary Hypertension**



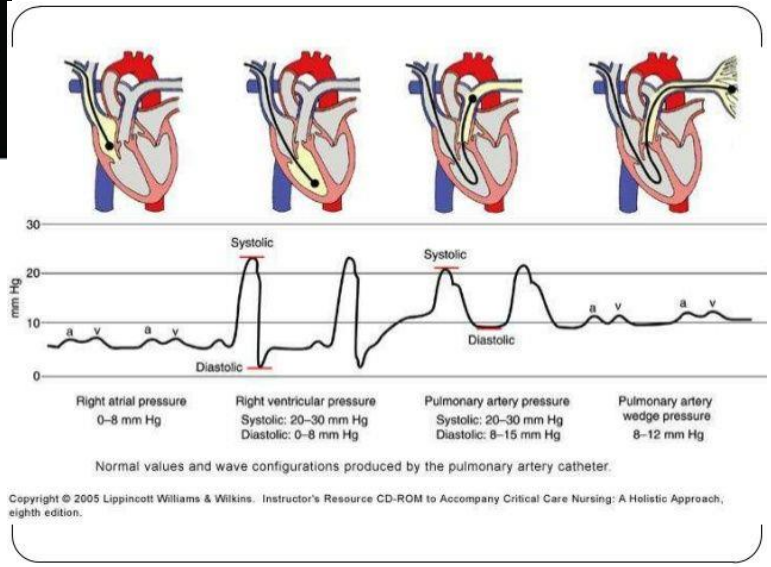

**R-L shunts**
**hypoxemia, cyanosis**

# Pulmonary Hypertension

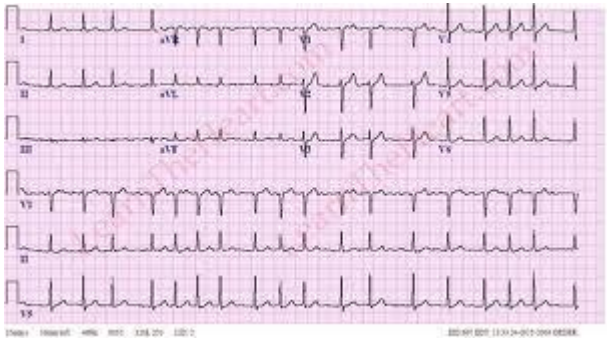


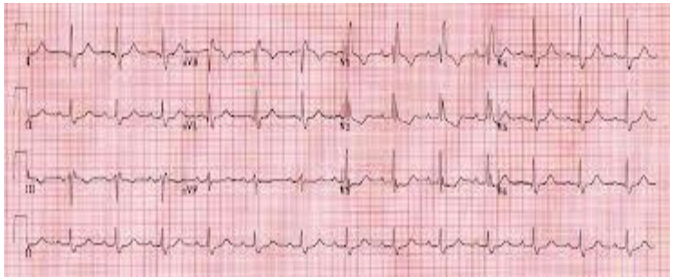

**ECHO**

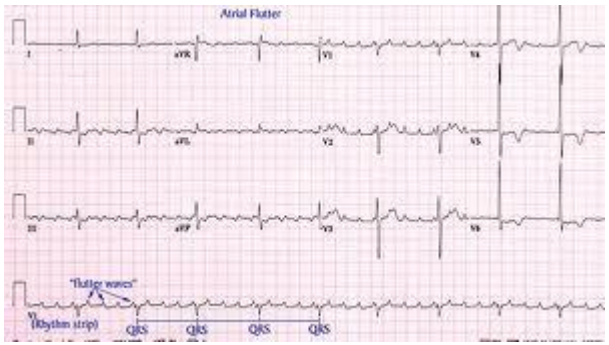
**CATHETER**

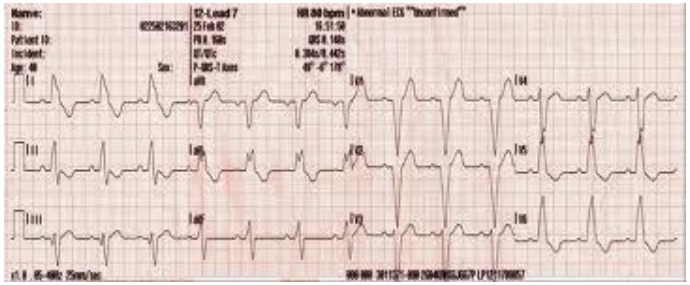


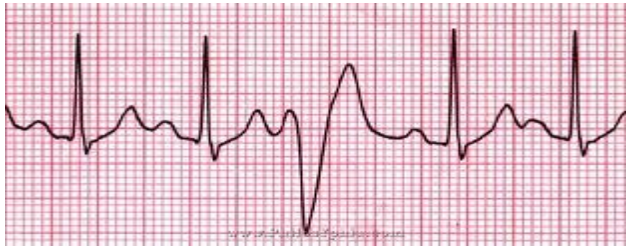
# Arrhytmias in Patients with CHD

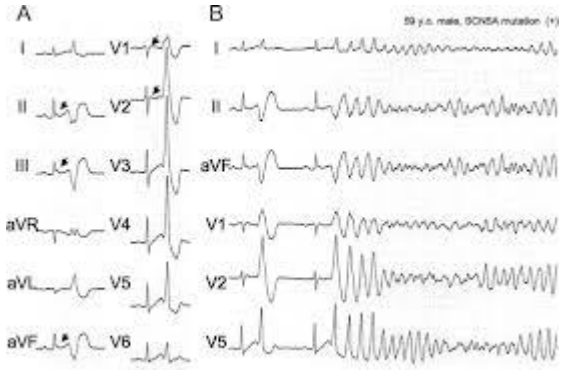










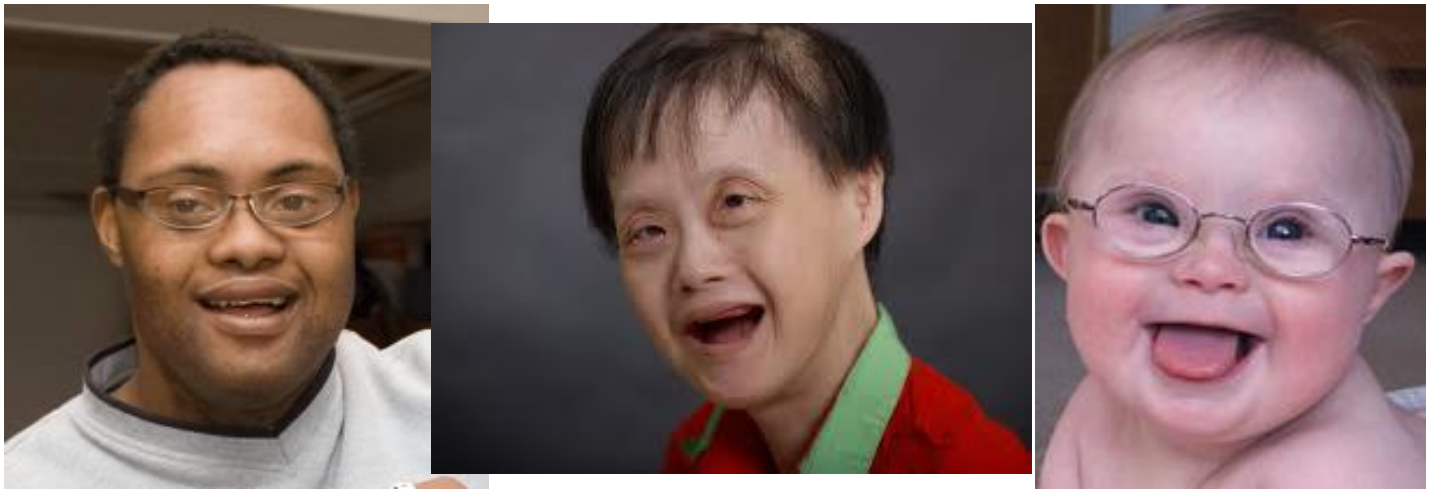
# Etiology of CHD

- **non-genetic**
  illness in the mother (rubella, diabetes, lupus)
  mother drug ingestion (anti-epileptic, alcohol, lithium)

- **genetic**
  isolated heart / GV disorder
  disorder associated with genetic syndrome

*The crucial period for fetal cardiac development occurs btw. weeks 6 and 12.

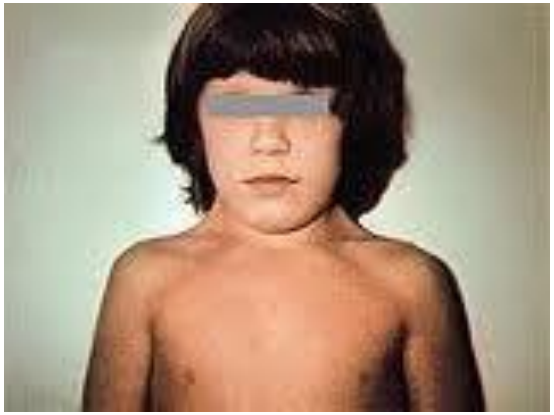
# Down syndrome (Trisomy 21)



atrioventricular septal defects

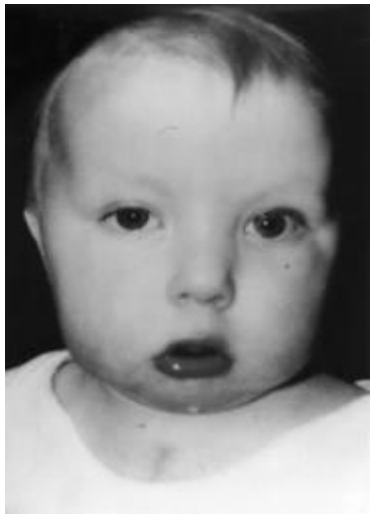
tetralogy of Fallot

# Turner syndrome



coarctation, bicuspid aortic valve

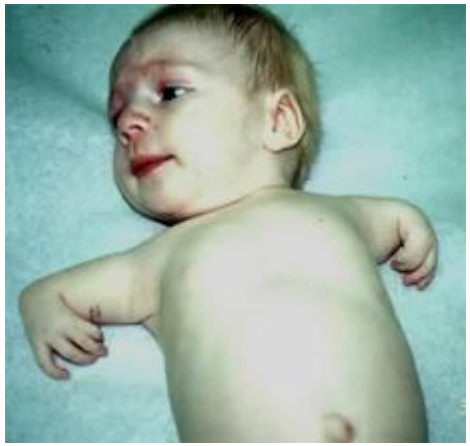
# DiGeorge syndrome (CATCH 22)



tetralogy of Fallot, right sided aortic arch,

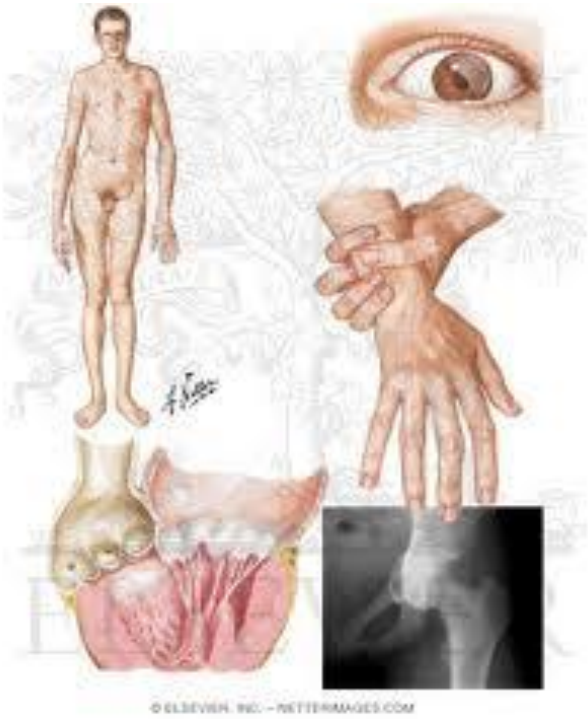
pulmonary atresia, aortic-to-pulmonary collaterals

# Holt-Oram syndrome



septation defects (ASD, VSD)

# Marfan Syndrome

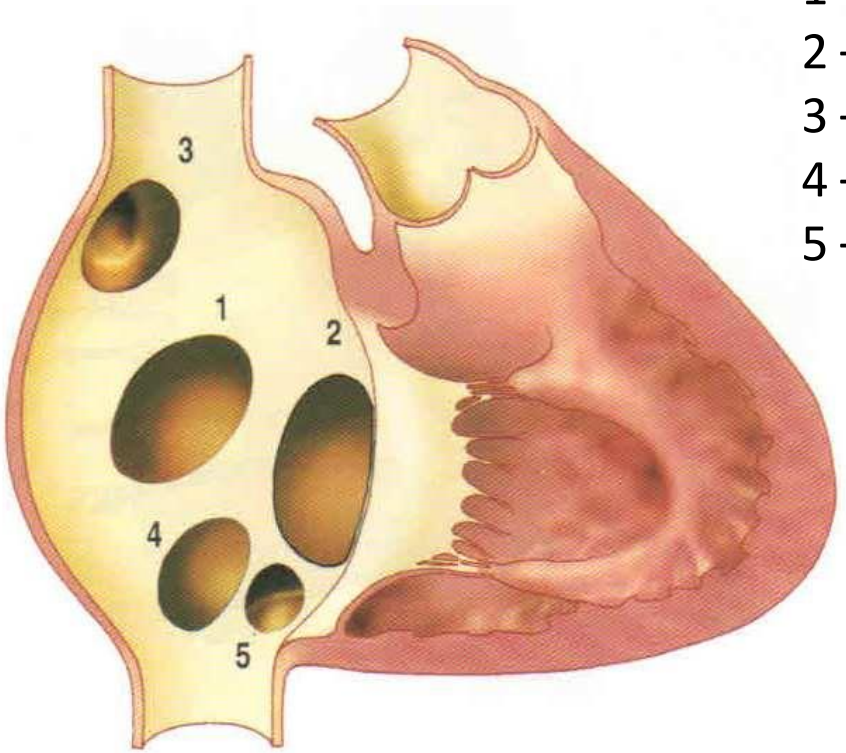


aortic dilation, aneurysm, dissection; heart valve disorders

# CHD Anatomy and Pathophysiology

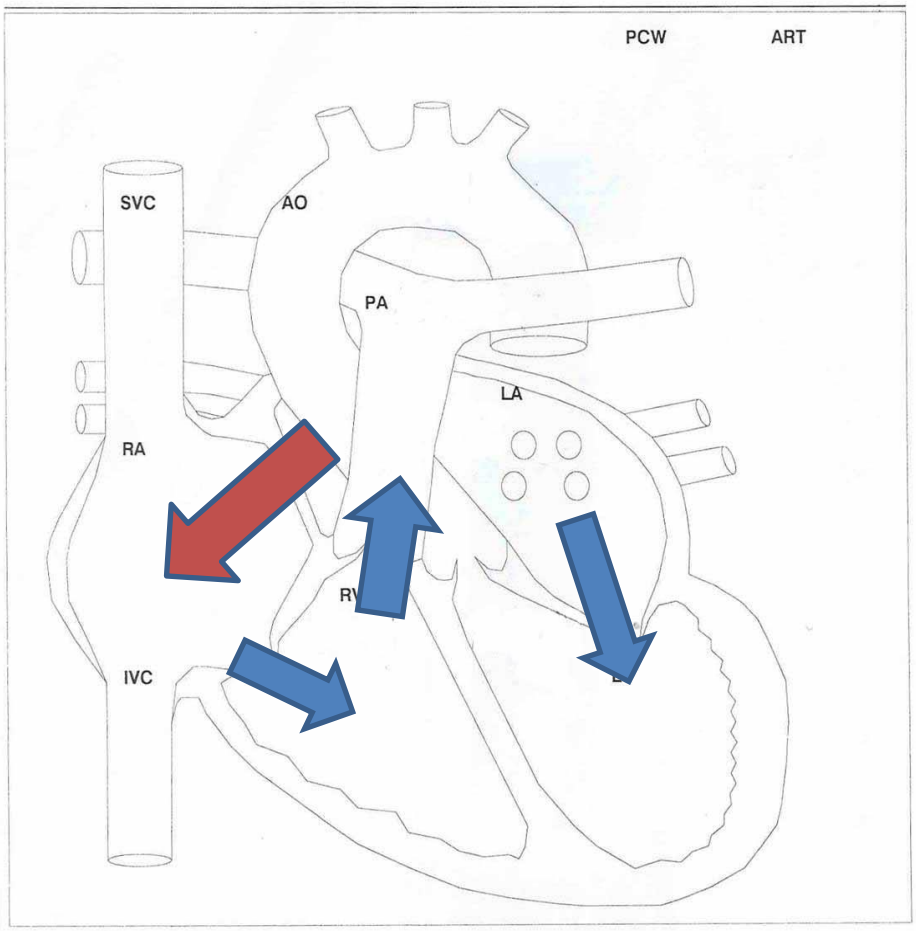
- Septation Defects (ASD, VSD)
- Patent Arterial Duct
- Aortic Coarctation
- Tetralogy of Fallot
- Transposition of Great Arteries

# Atrial Septal Defect (ASD)




1 – Secundum type
2 – Primum type
3 – Sinus venosus superior type
4 – Sinus venosus inferior type
5 – Coronary sinus type

# ASD Pathophysiology




left to right IA shunt

↓

volume overload

of right atrium

and ventricle

↓

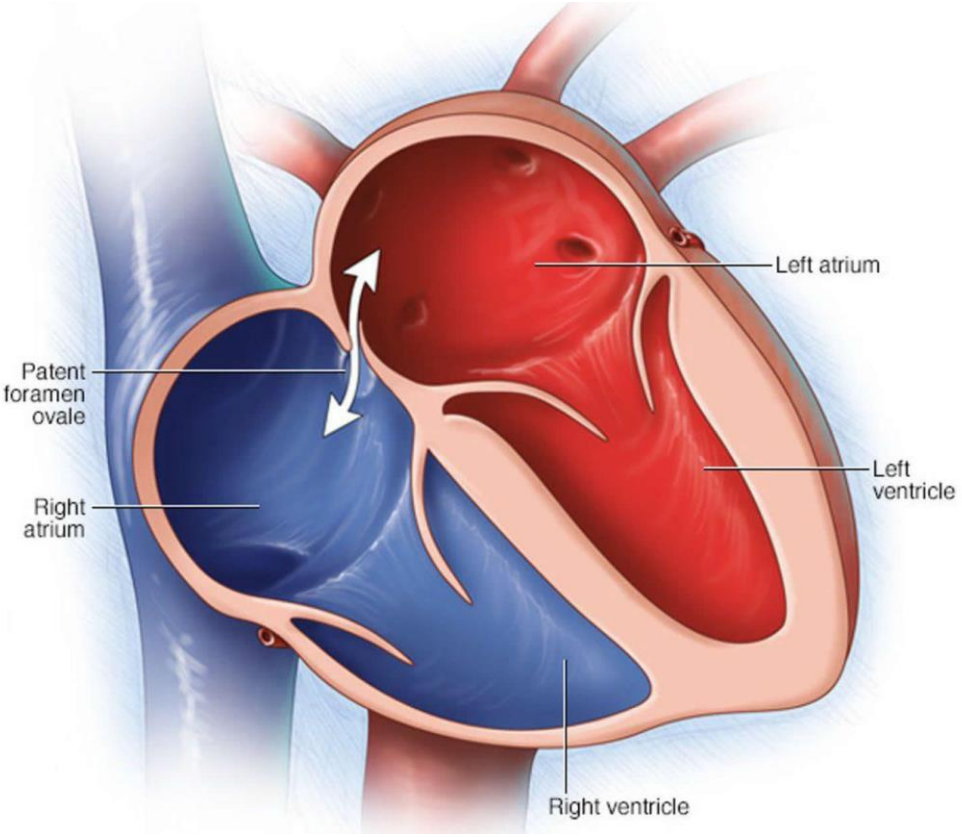
pulmonary hypercirculation

RV dilatation

↓

PH, bidirectional shunt

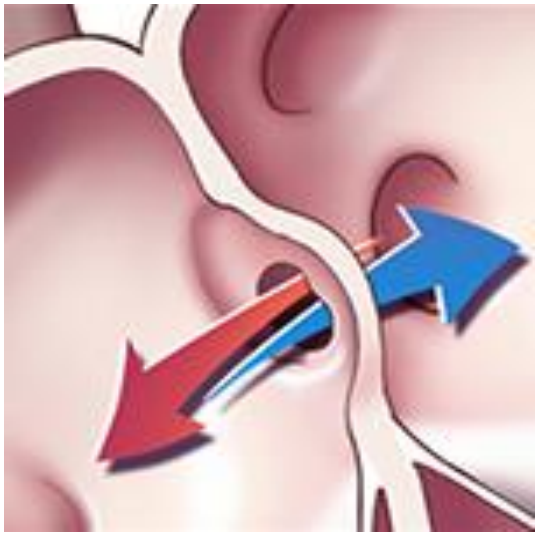
# Patent Foramen Ovale (PFO)



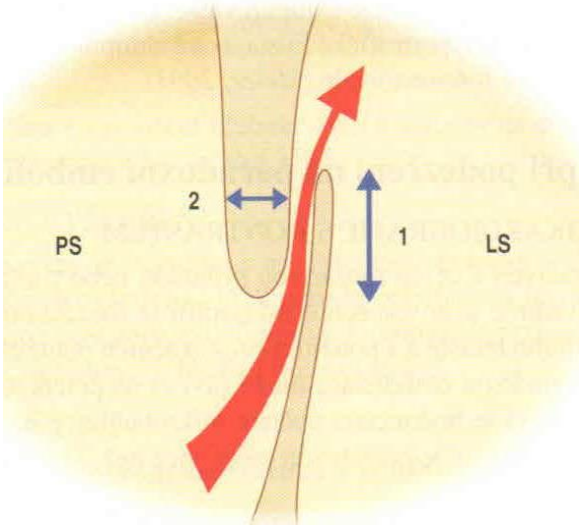

- 25-30% of general population
- not considered as CHD

# ASD x PFO

## CHD

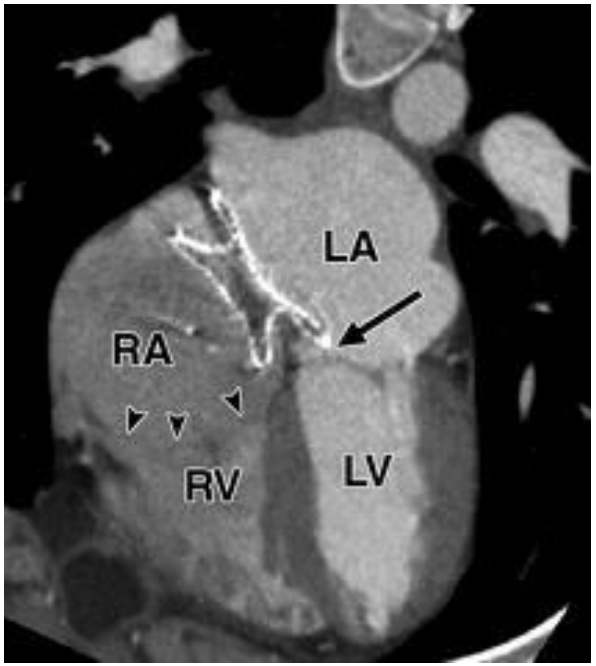


## ~~CHD~~

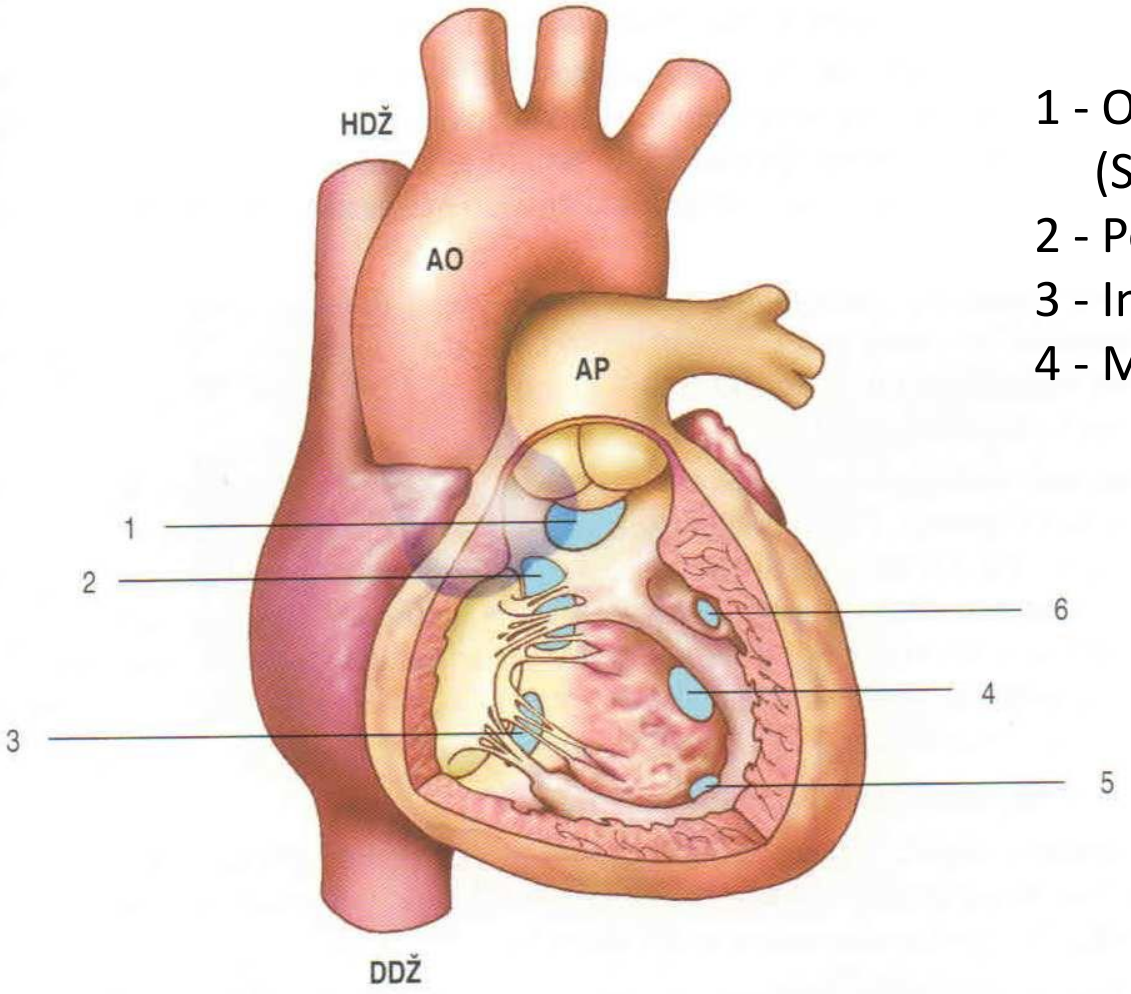



paradoxical embolism risk

# Amplatzer Ocluder - PFO/ASD II Closure

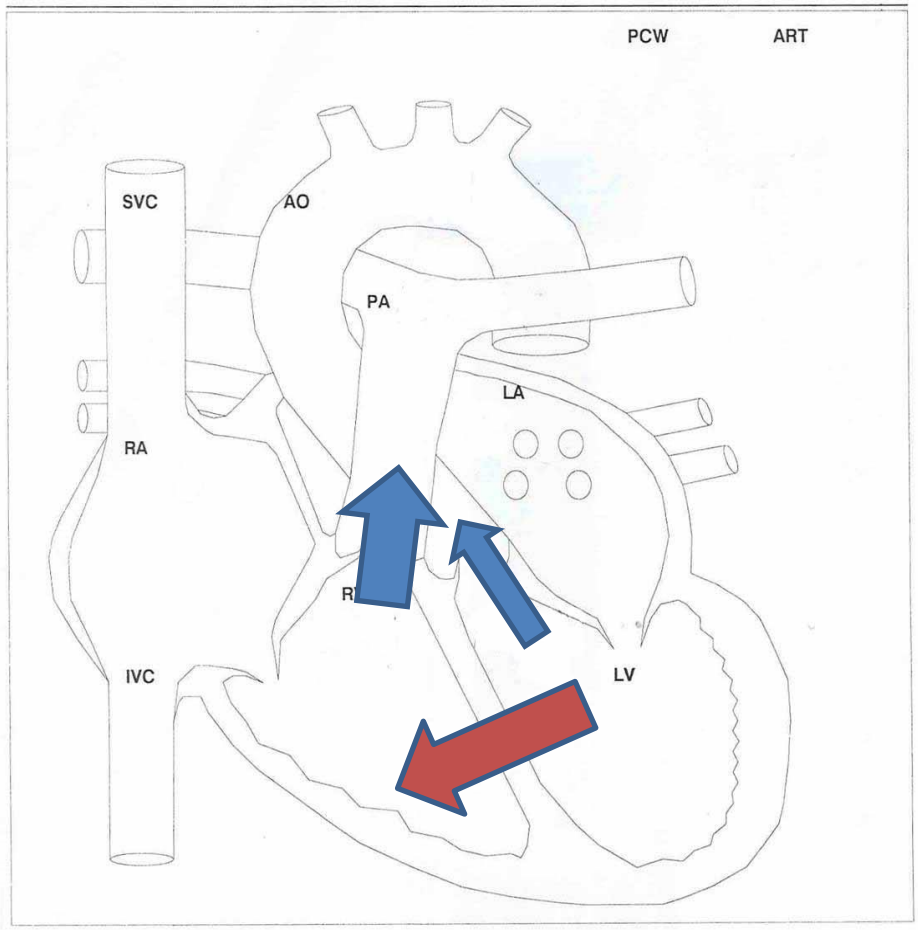


# Ventricular Septal Defect (VSD)




1 - Outflow (Subaortic)
2 - Perimembraneous
3 - Inflow
4 - Muscular

# VSD Pathophysiology




left to right IV shunt

↓

pulmonary hypercirculation

volume overload of left atrium and left ventricle

↓

PH

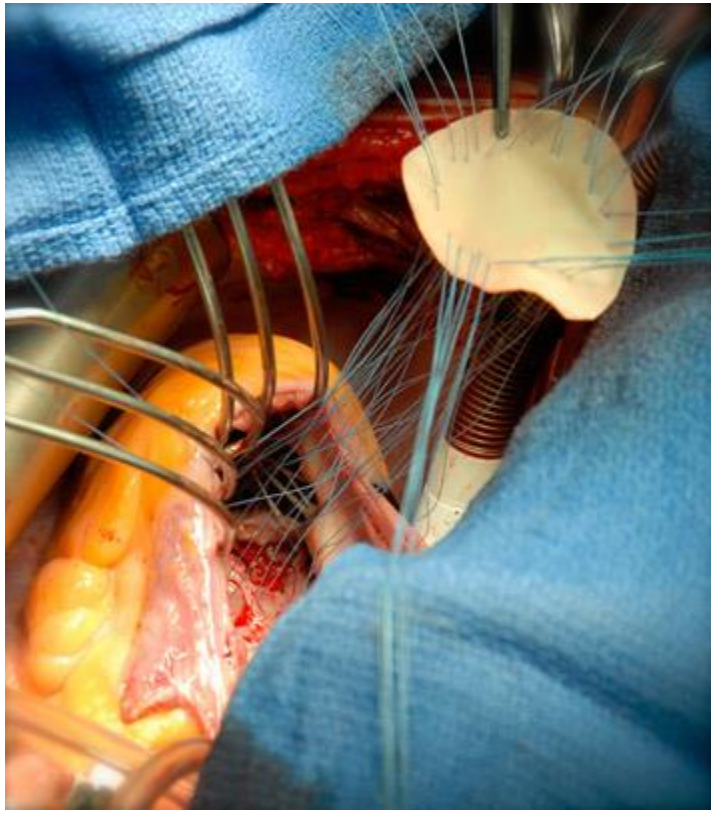
RV hypertrophy

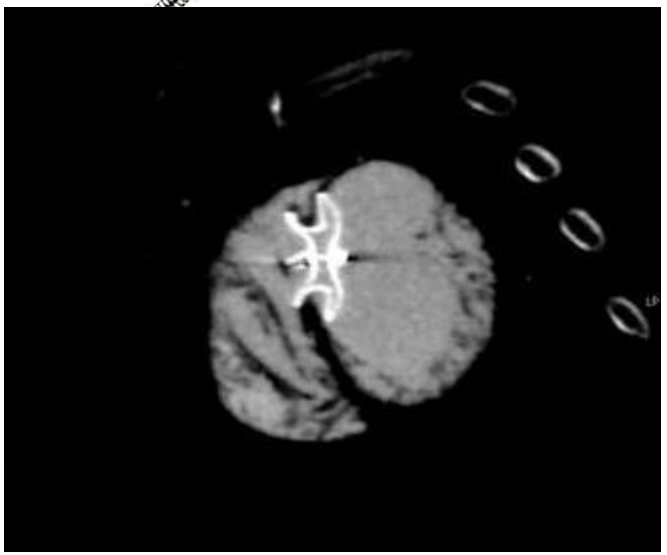
↓

right to left shunt

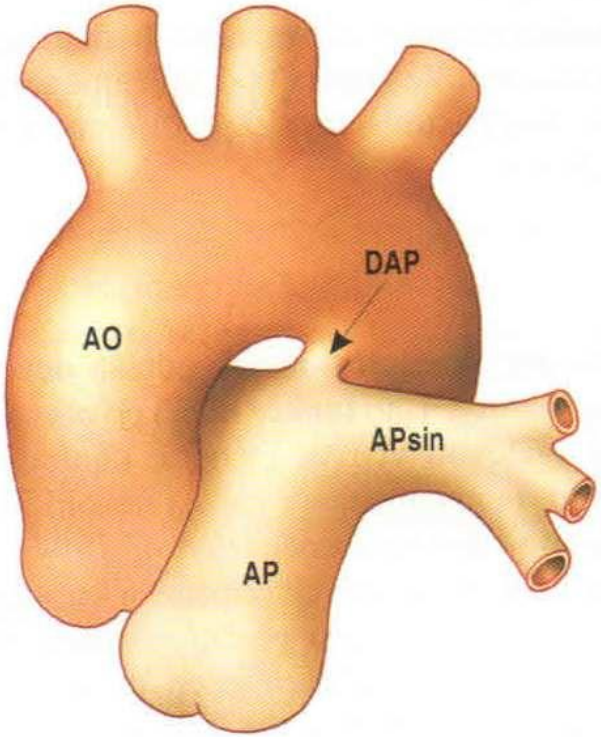
Eisenmenger syndrome

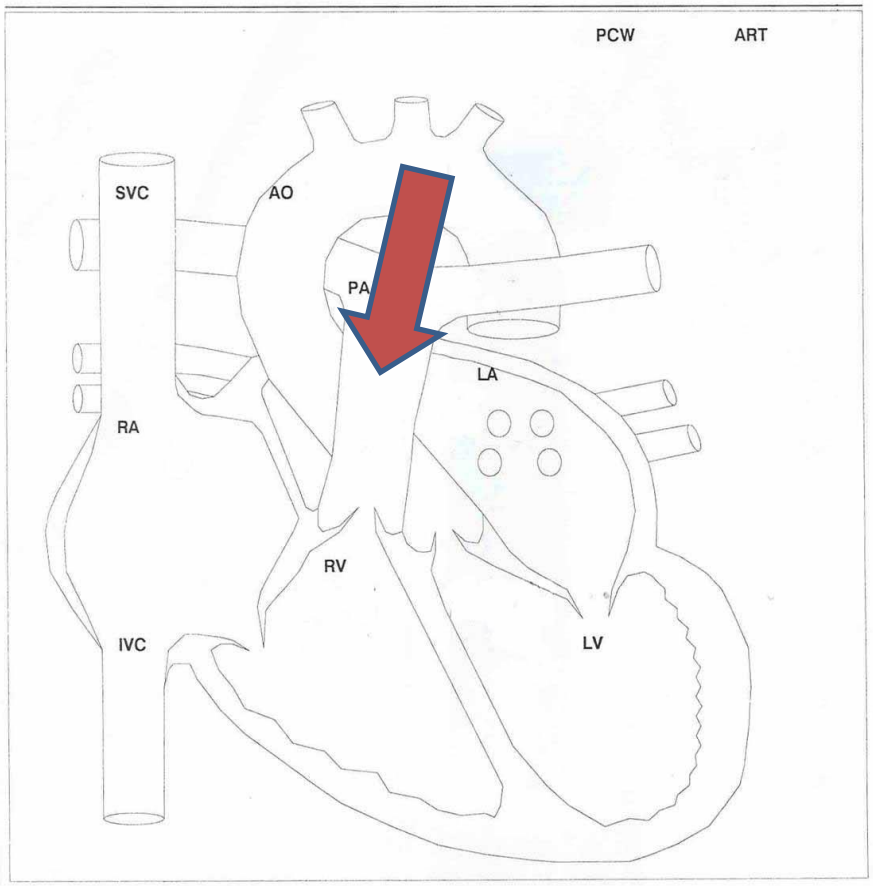
# VSD Closure





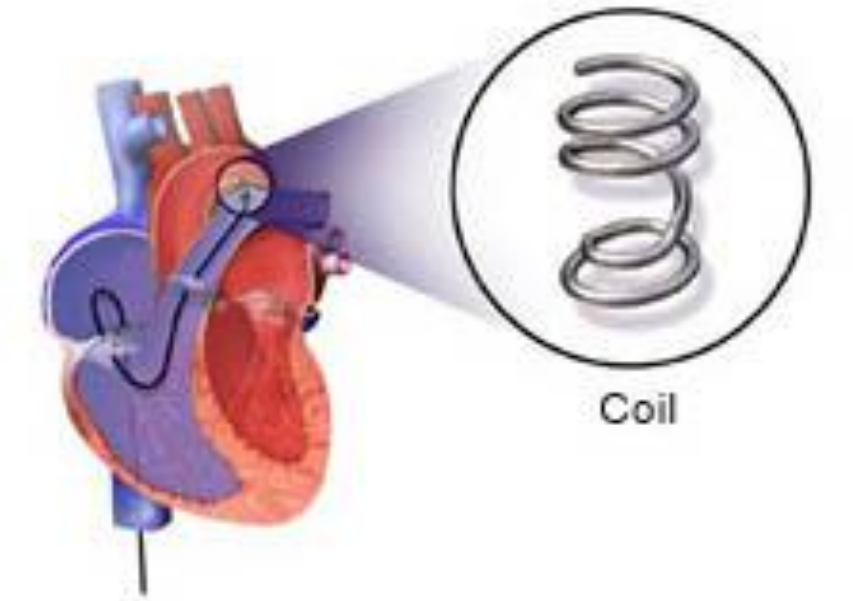
# Patent Ductus Arteriosus (PDA)



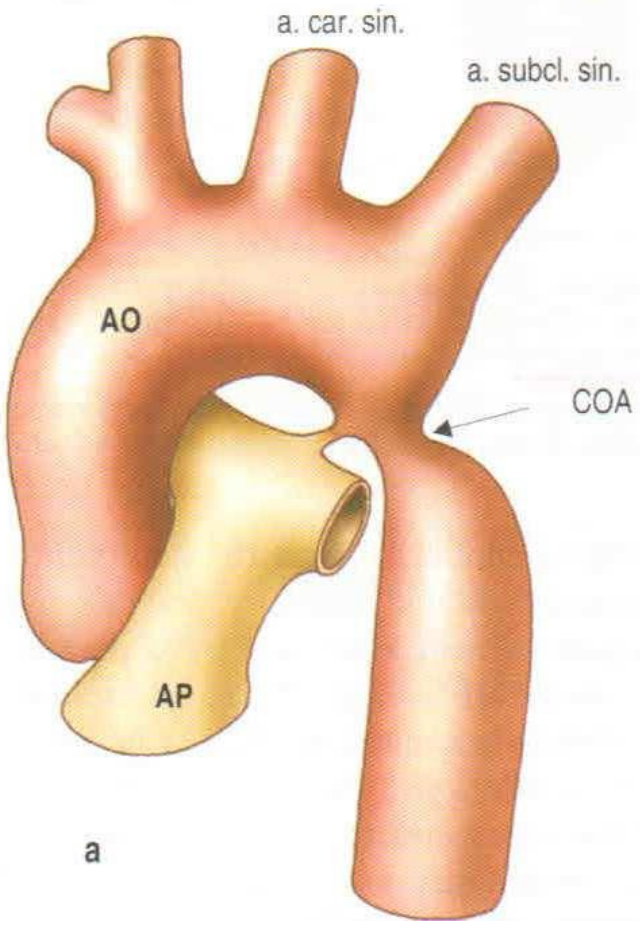

L-R shunt → PH → RV hypertrophy → R-L shunt → Eisenmenger sy

# PDA Closure




Coil Closure of PDA

# Coarctation of the Aorta




hypertension

in precoarctation area

⬇

LV hypertrophy

⬇

LV dysfunction

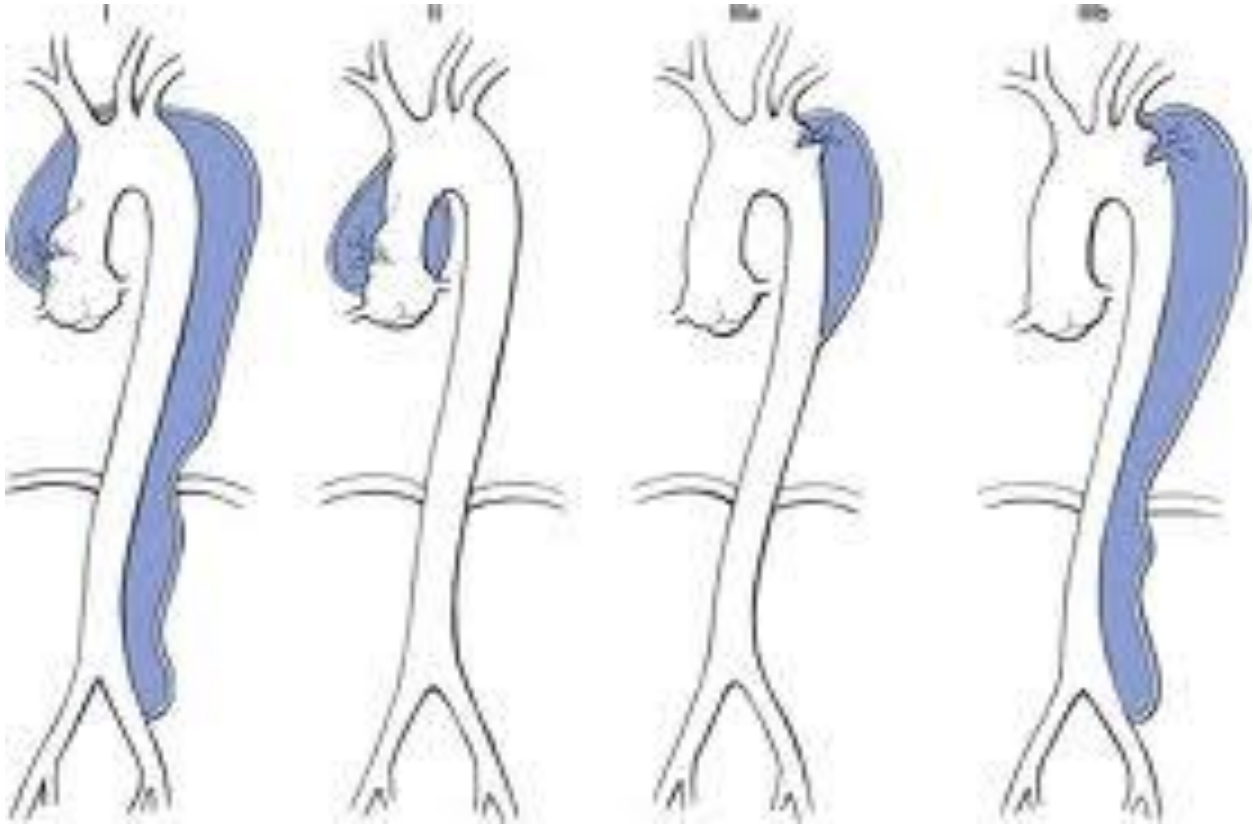
ascend. Ao dilatation

⬇

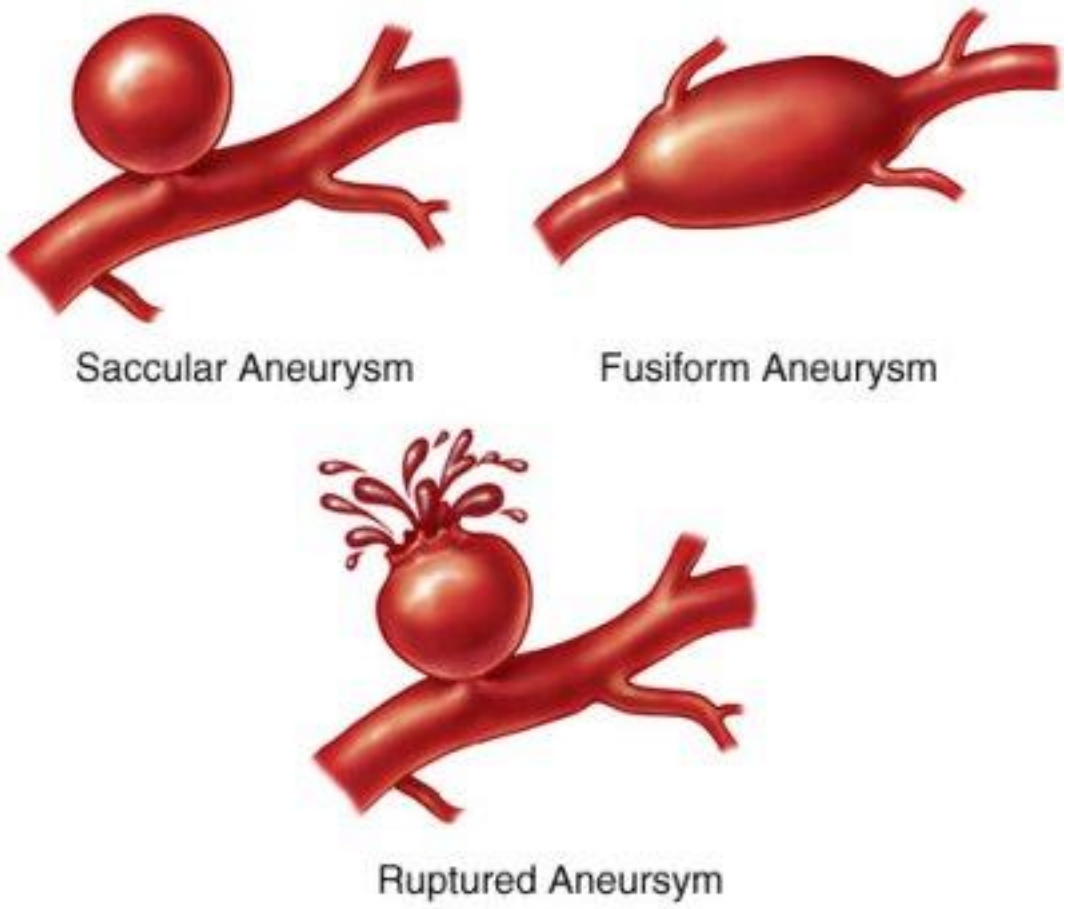
aortic dissection/rupture

*85% assoc. with bicuspid Ao

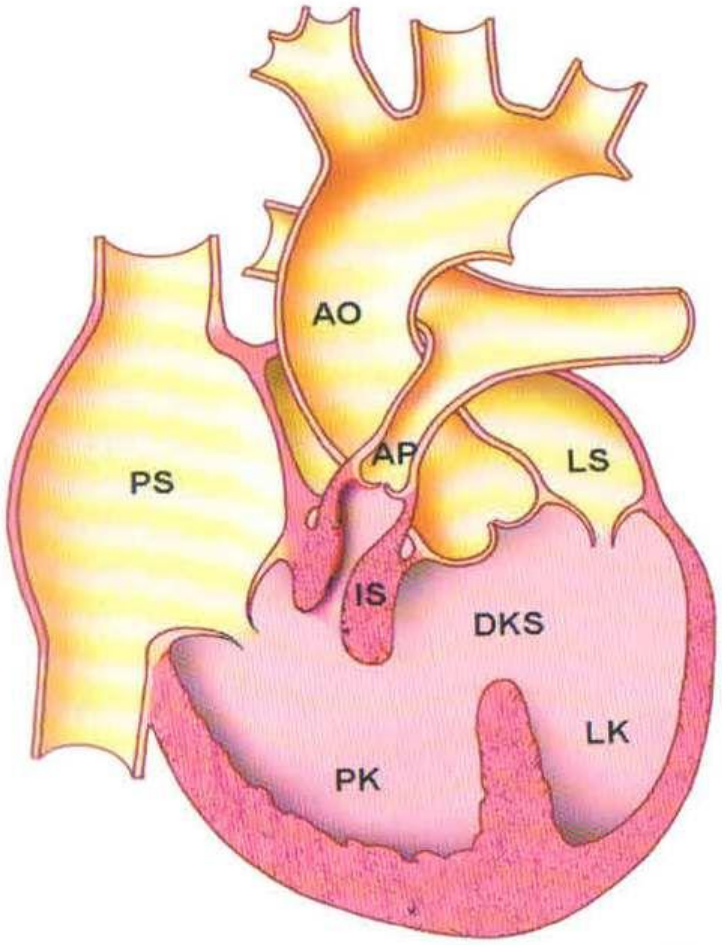
# Aortic Dissection



# Aortic Aneurysm



# Tetralogy of Fallot (TOF)

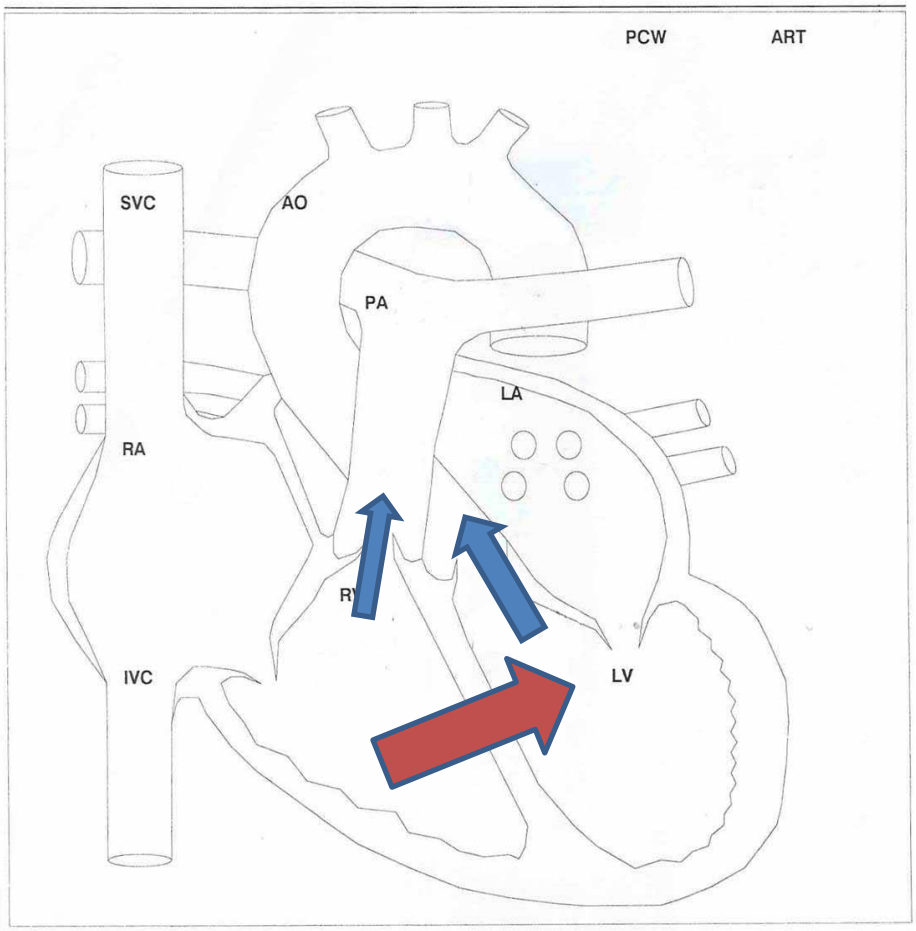



- ventricular septal defect
- overriding aorta
- pulmonary stenosis
- RV hypertrophy
- (atrial septal defect /PFO)

↓

(Pentalogy of Fallot)

# TOF Pathophysiology




Pu stenosis

RV hypertrophy

R-L shunt

↓

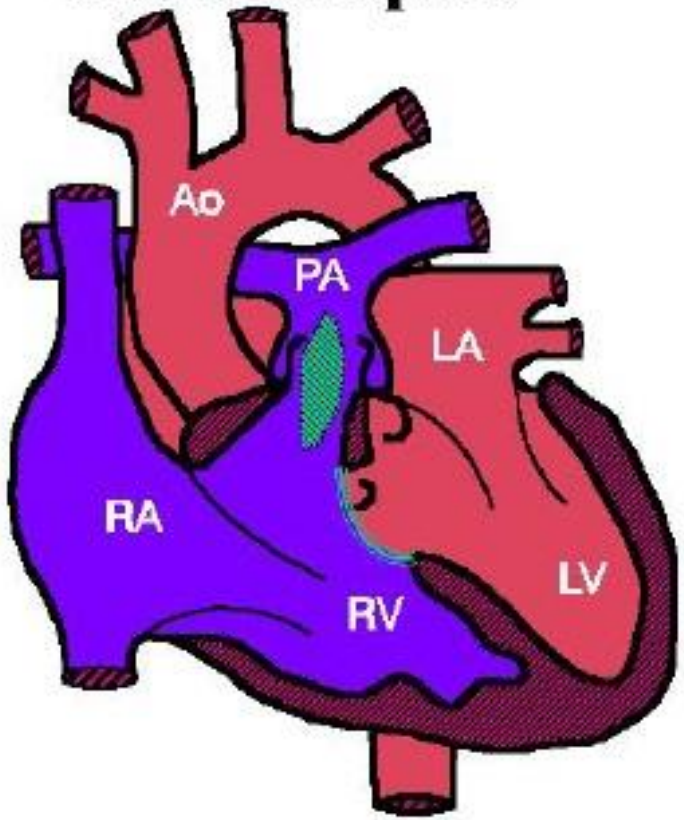
cyanosis / „pink Fallot“

↓

hypoxemia

RV failure

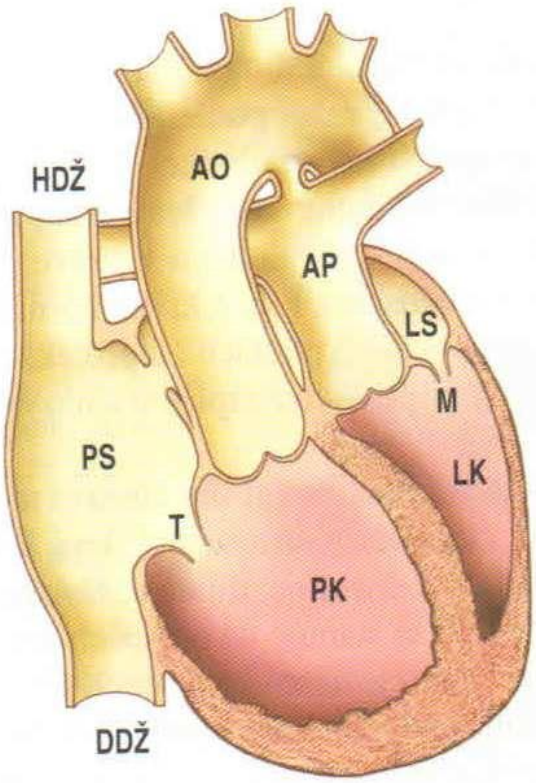
# Fallot's Tetralogy after 'Repair'




PA patch
VS patch

„normal“ circulation

# Transposition of the Great Arteries (TGA)

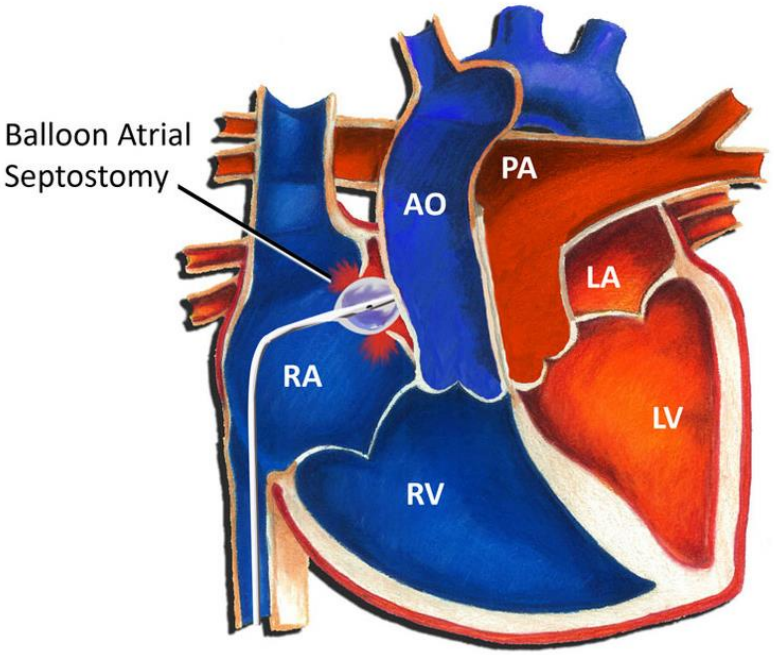



d-TGA

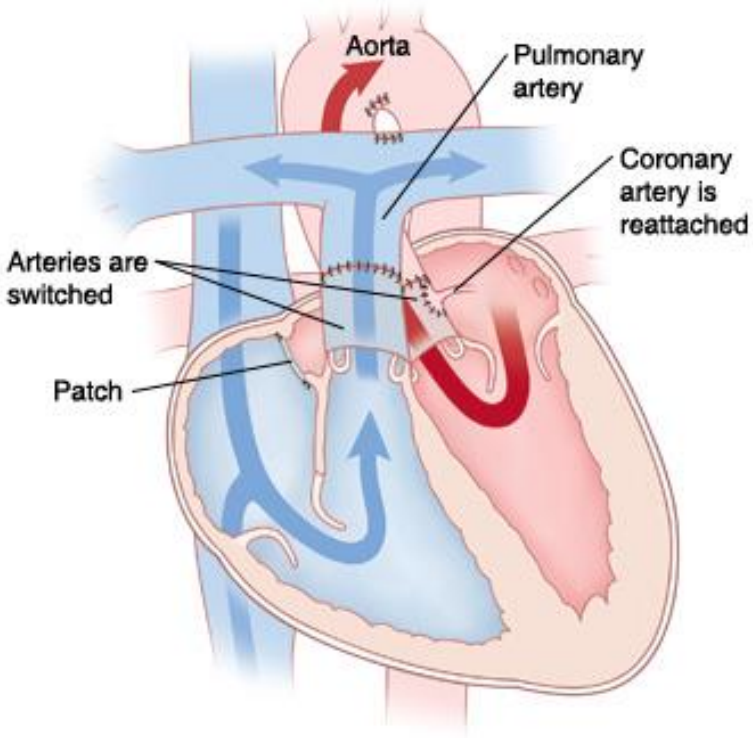
- 2 isolated circulations
- oxygenated blood in isolated pulmonary circulation
- deoxygenated blood in isolated systemic circulation
- impossible to survive without correction

# TGA Repair

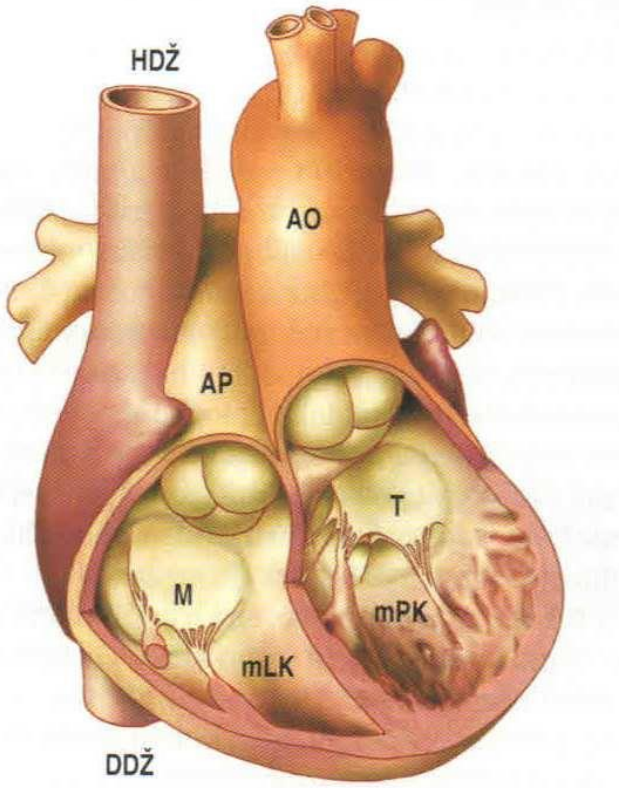
**palliative septostomy**




**arterial switch**

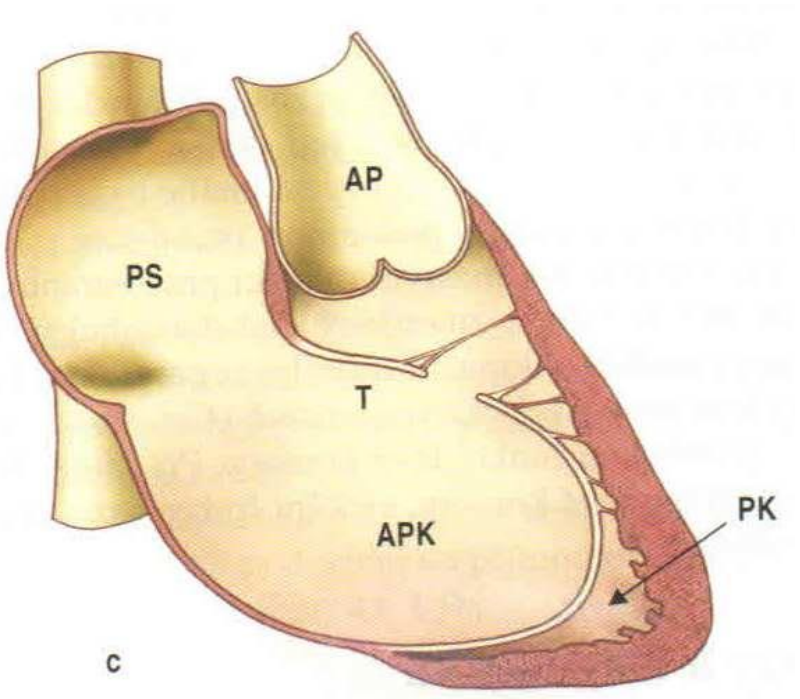


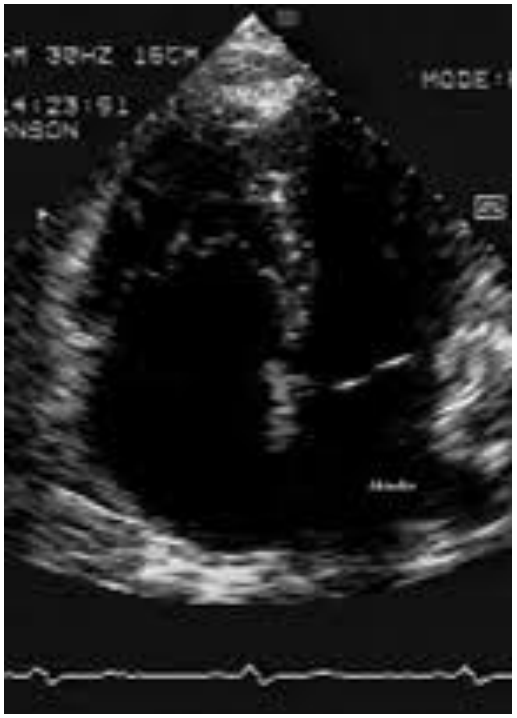
# Congenitally Corrected TGA (CCTGA)



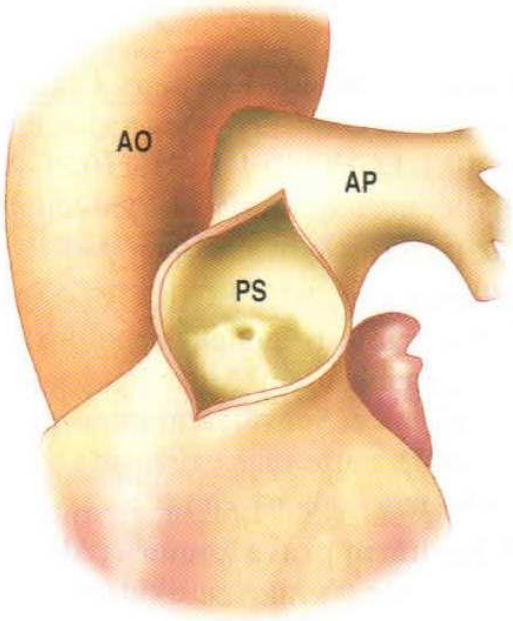

- „normal“ circulation
- RV in systemic circulation
- RV dysfunction/failure

# Ebstein´s Anomaly of the Tricuspid Valve

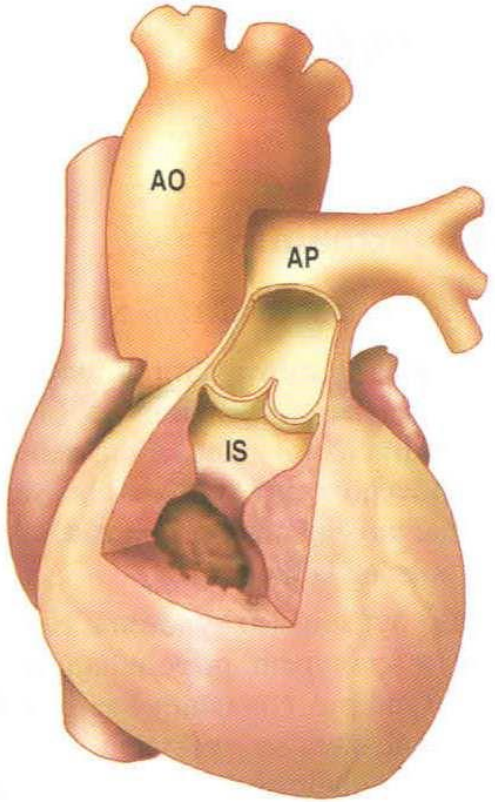


# Pulmonary Stenosis



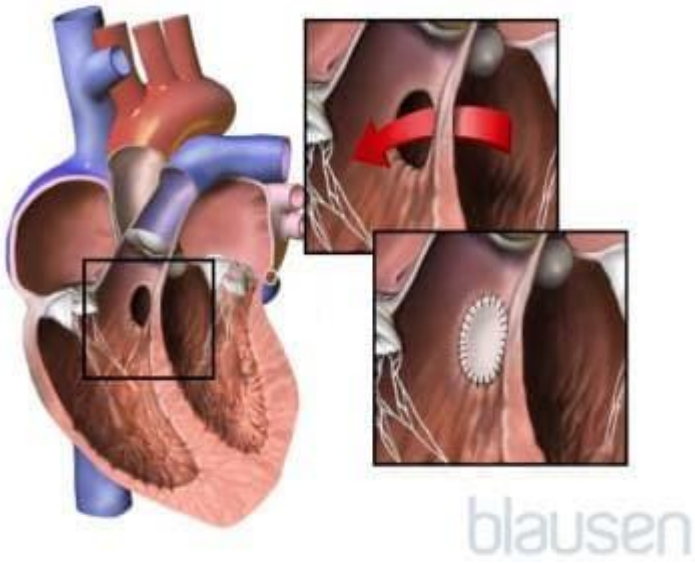

valvar

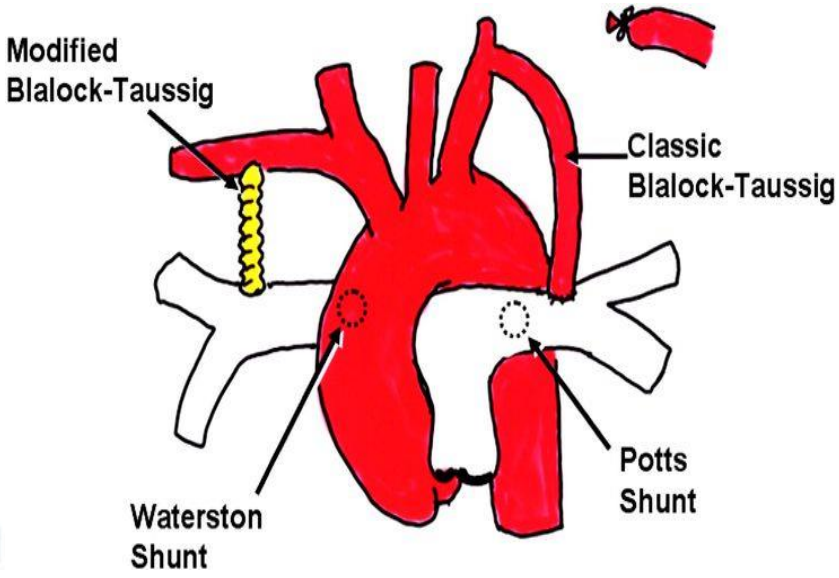



infundibular

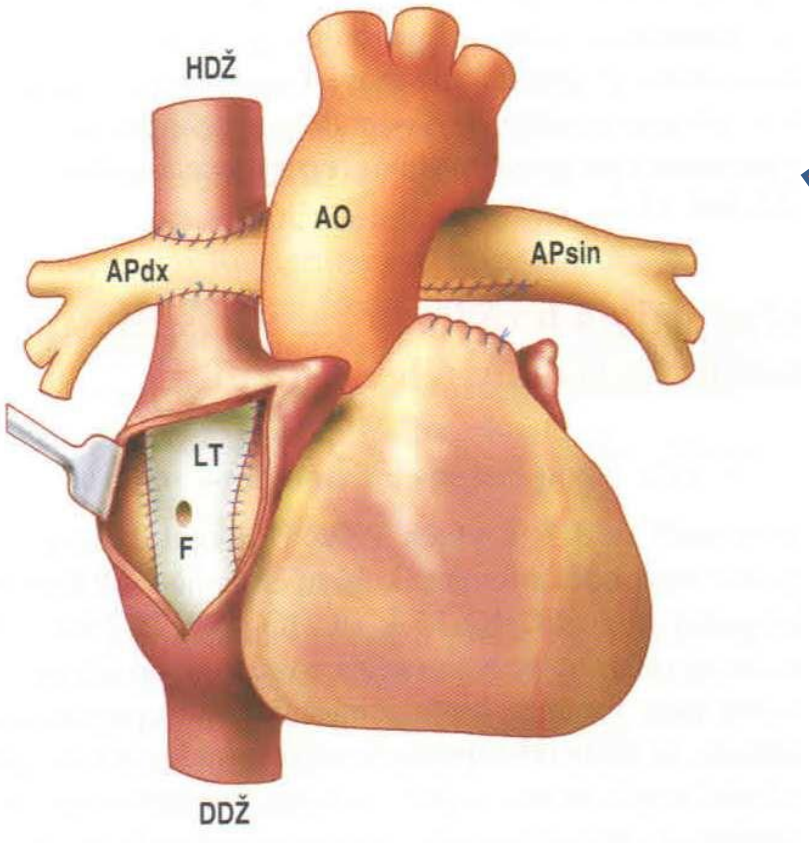
# Surgical Repair of CHD

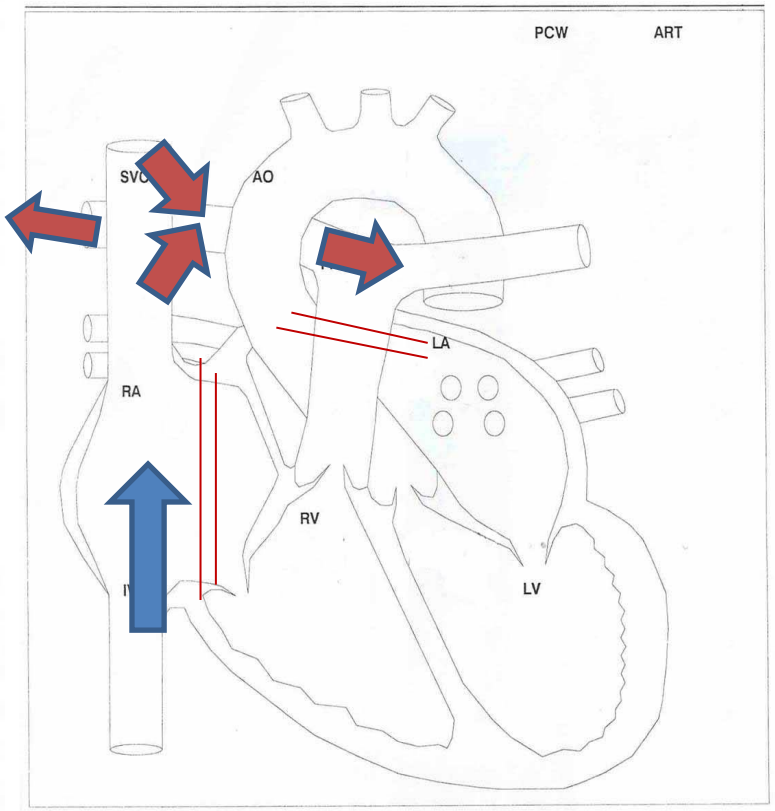
- **corrective**
  (patch...)
- **palliative**
  (connections...)



# Fontan Circulation



# CHD non-included Congenital Disorders

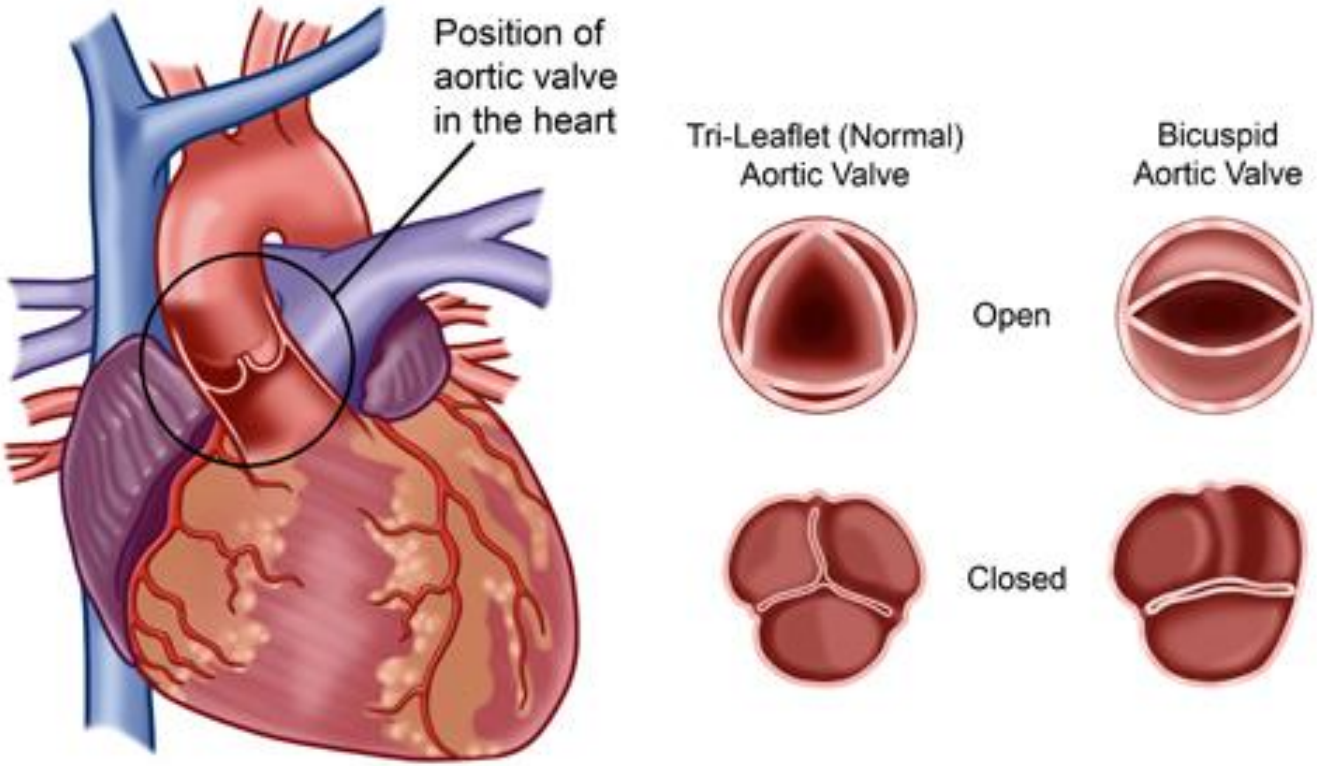
Bicuspid Aortic Valve 1-2% common population

Patent Foramen Ovale (PFO) 25-30% common population

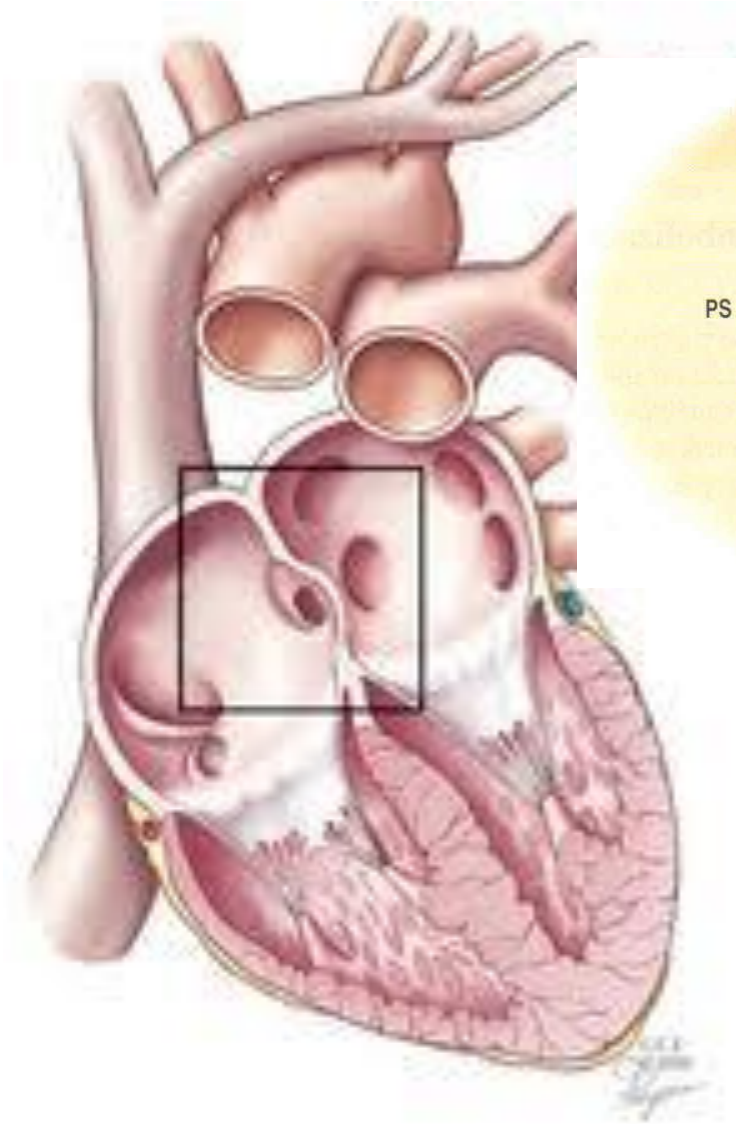
Persistent Left Upper Vena Cava 0,5%

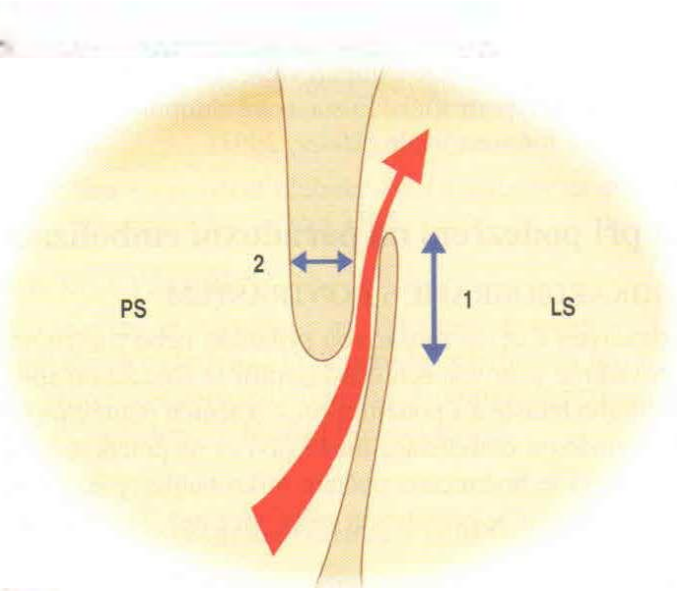
Cardiomyopathies

# Bicuspid Aortic Valve (BAO)

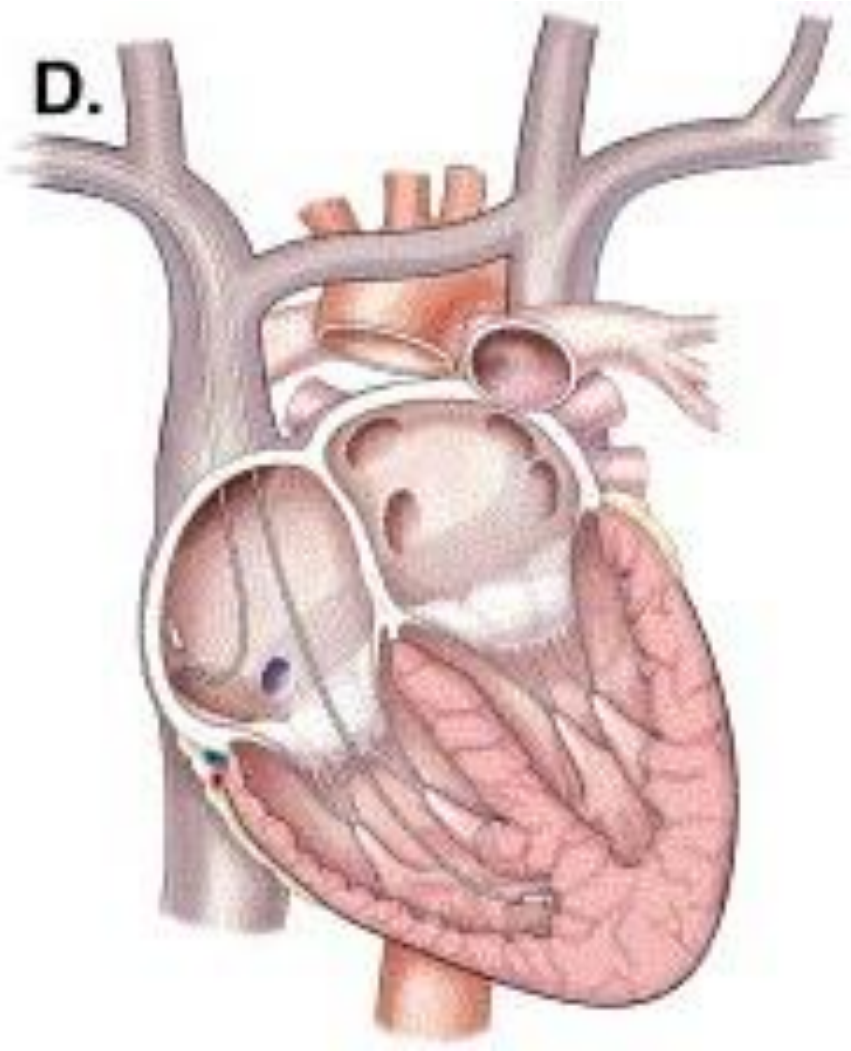


# Patent Foramen Ovale (PFO)





# Persistent Left Upper Vena Cava



# Adult Patient with CHD

- symptoms
- investigation

- difference between patient with CHD and „normal“ cardiology patient

- treatment and follow up

# Symptoms in CHD (most common)

- dyspnea
- palpitations
- syncope
- chest pain
- hemoptysis
- symptoms of hyperviscosity syndrome
- low stress tolerance
- fatigue, exhaustion

# Hyperviscosity Syndrome

chronic hypoxemia

↓

erythrocytosis

↓

stiff blood

↓

headache, vertigo, bleeding, visual disturbances, seizure, chest pain, dyspnea, difficulty walking, coma

# Investigation in Patients with CHD

- history, physical exam
- ECG, BP, blood oxygen saturation
- blood tests
- urine tests
- chest X-ray
- echocardiography
- CT scan, MRI
- stress testing (spiroergometry, 6-MWT)
- QOL questionnaire

# Problems associated with CHD in Adults

- rezidual findings
- non-detected disorders in early life
- late indication to operation
- arrhythmias
- infective endocarditis
- anticoagulation
- pregnancy and labour in women with CHD
- social and work problems
- depression

# Treatment and Follow-up

- surgical repair of disorder
- reoperation
- anticoagulation therapy/bleeding complications
- infective endocarditis prophylaxis/treatment
- pharmacology treatment of arrhytmias, PH, HF
- non-pharmacology treatment: PM, ICD, CRT
- oxygen therapy
- heart/lung transplant
- psychotherapy

# What can the patient with CHD die of?

- heart failure
- malignant arrhytmias
- aortic aneurysm rupture / dissection
- infective endocarditis
- cardioembolism (stroke)

...but we are here to help every patient with CHD living a full life!