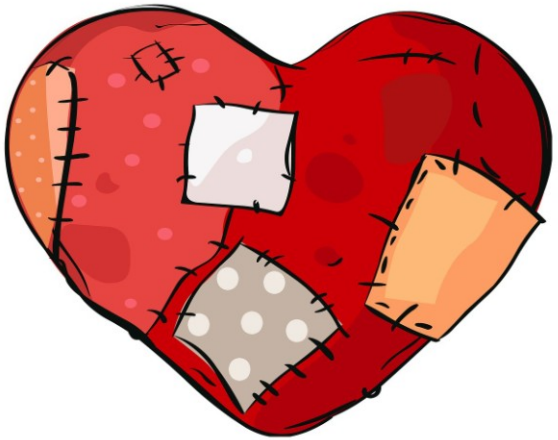
Adult Congenital Heart Disease



Lenka Kubkova
Masaryk University and University Hospital in Brno, 2020

Purpose of the Lecture

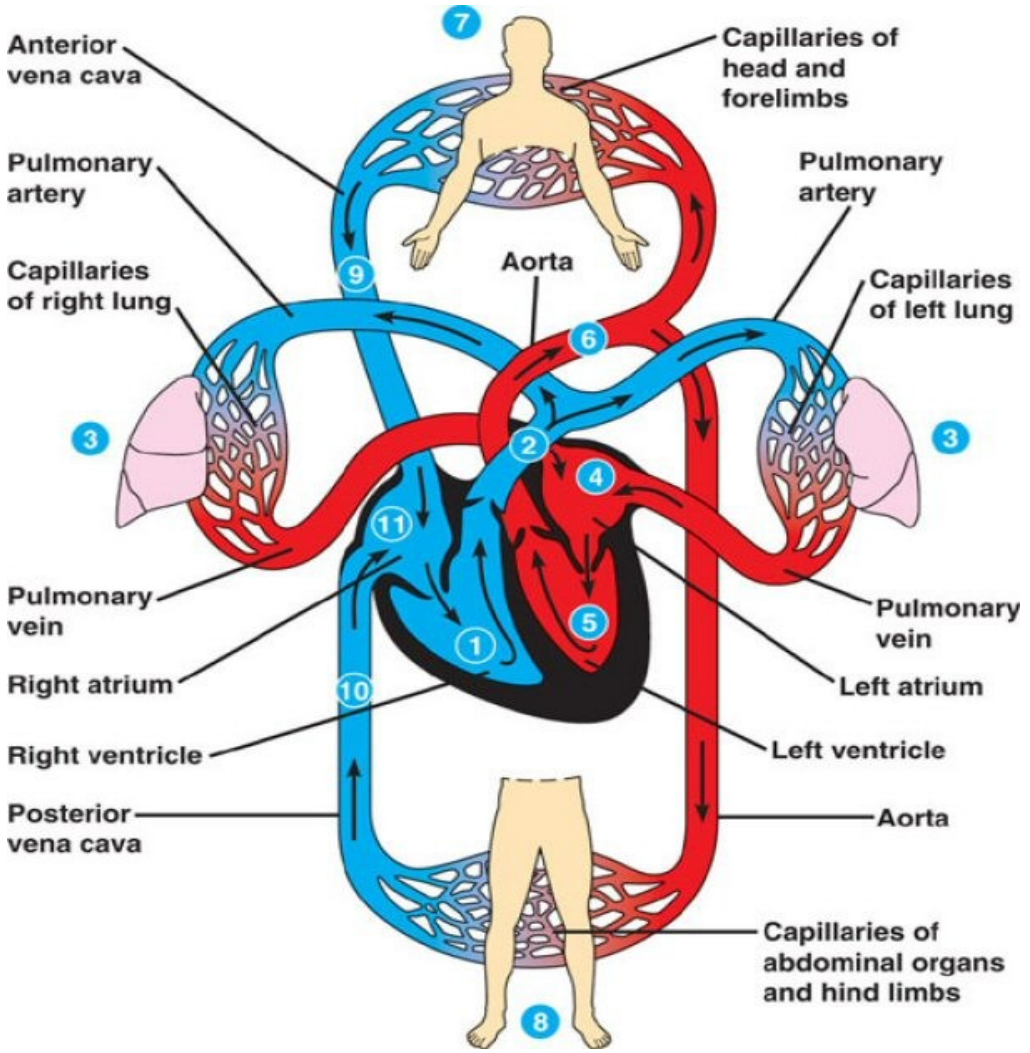
to **remember** you and **explain**:

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

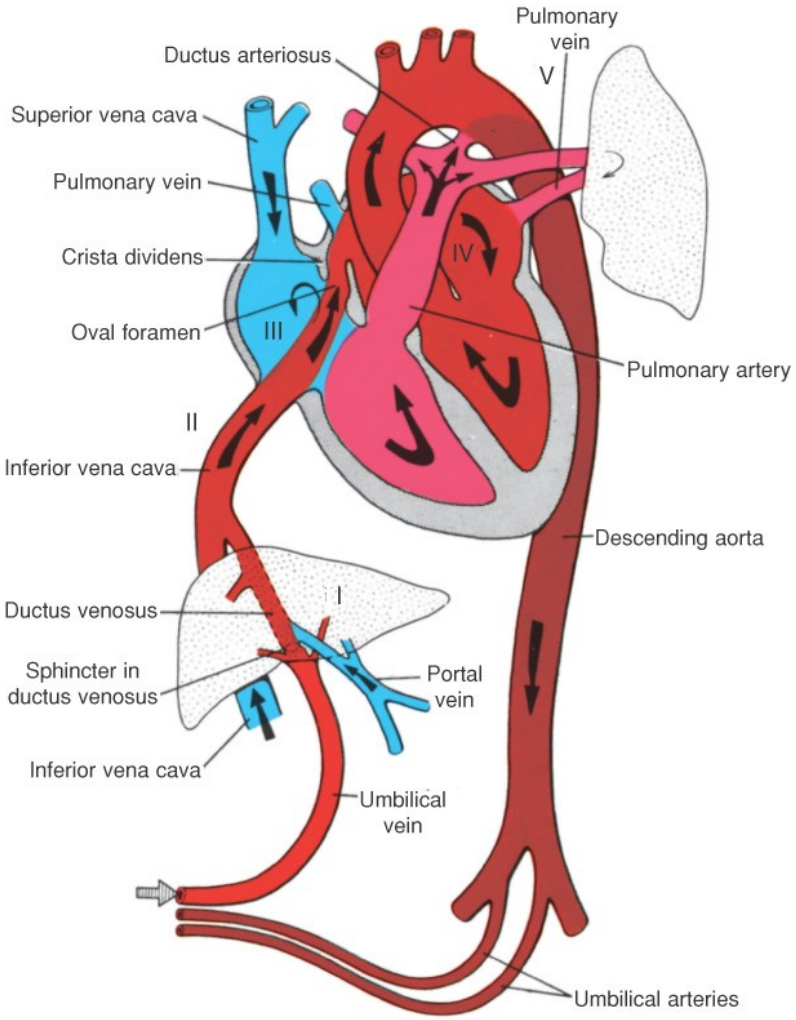
to **demonstrate**:

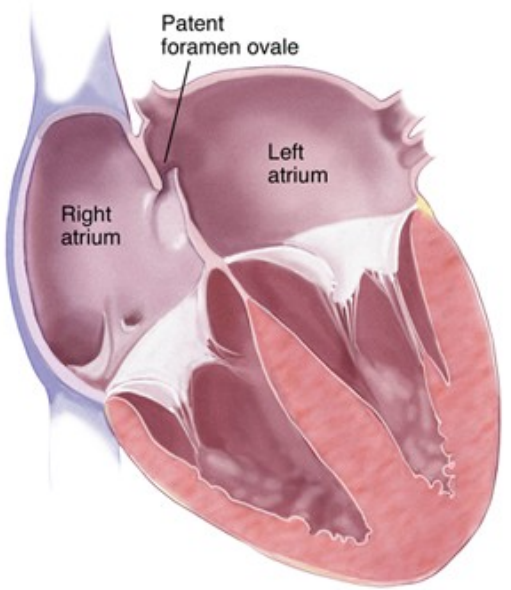
- how does adult patient with CHD look like
- what are his symptoms
- how can we investigate and treat him
- what could be different between „normal“ cardiology patient and that one with CHD

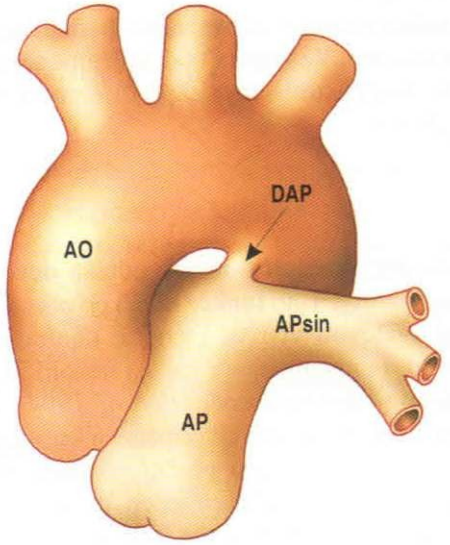
Systemic and Pulmonary Circulation



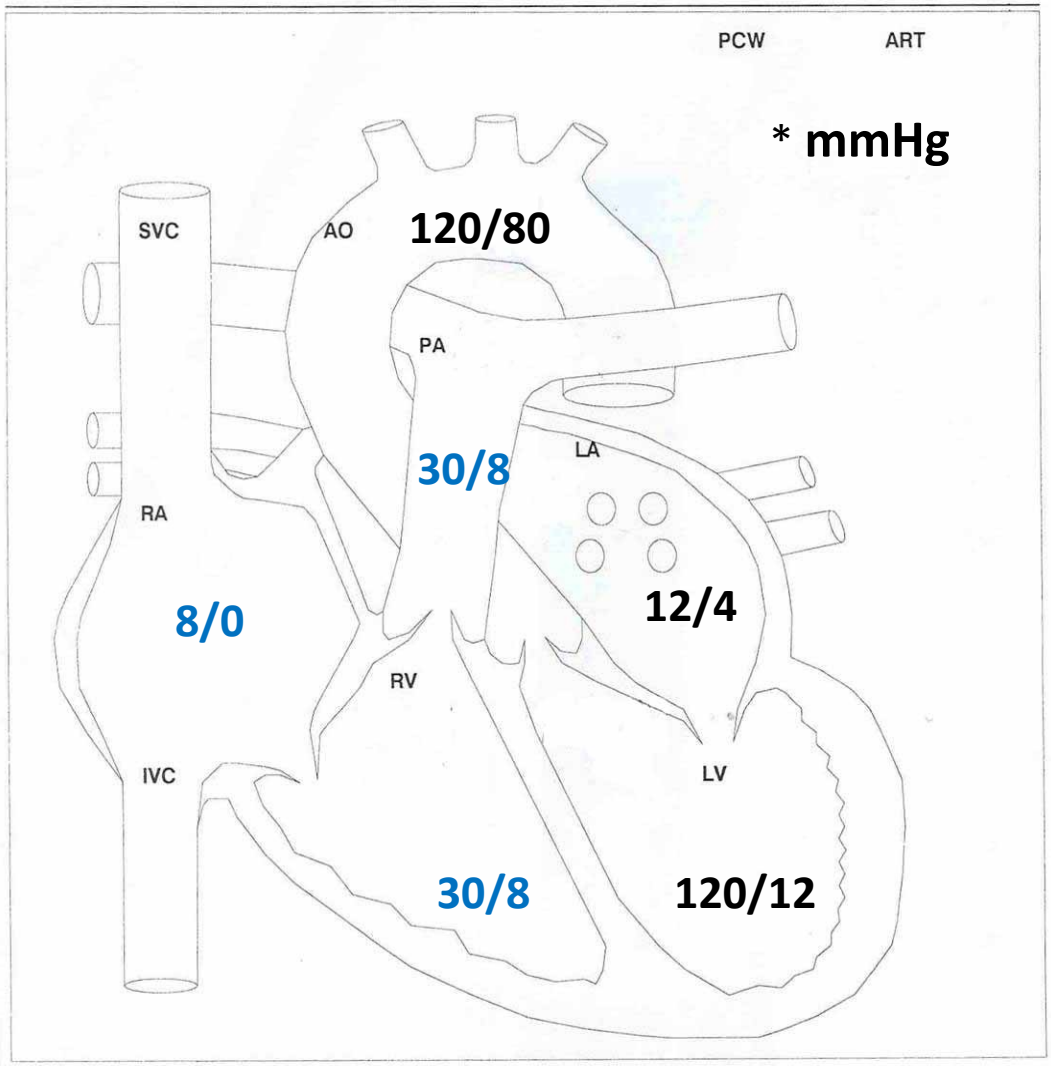
Fetal Circulation







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

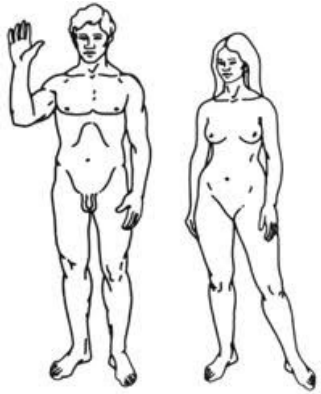
  outcome for the patients
- 35% of all CHD are critical disorders requiring immediate intervention

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

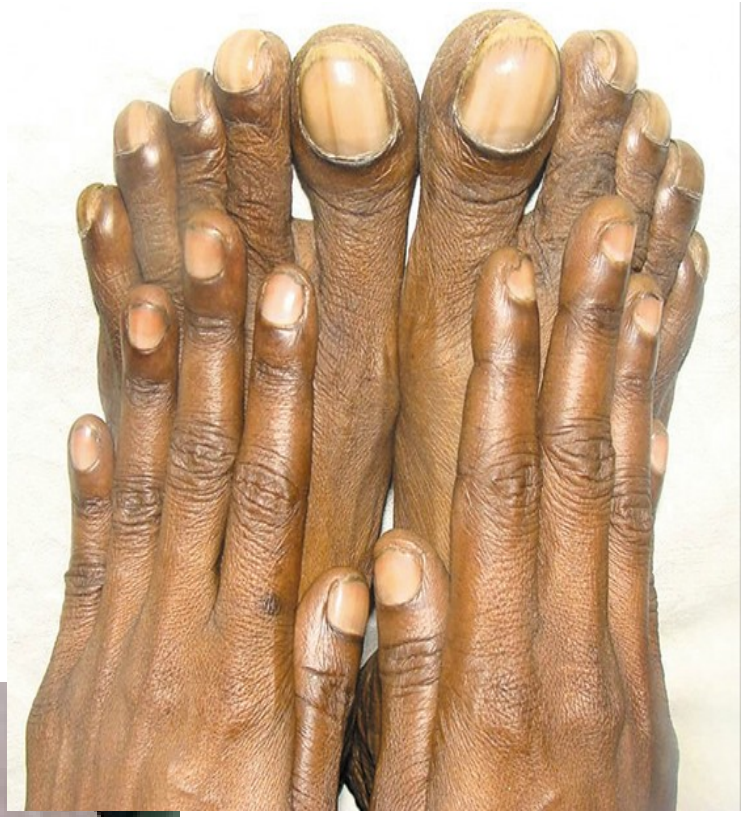
  (CR: 10 000 children and 25 000 adults with CHD)
- there are now more adults than children with CHD

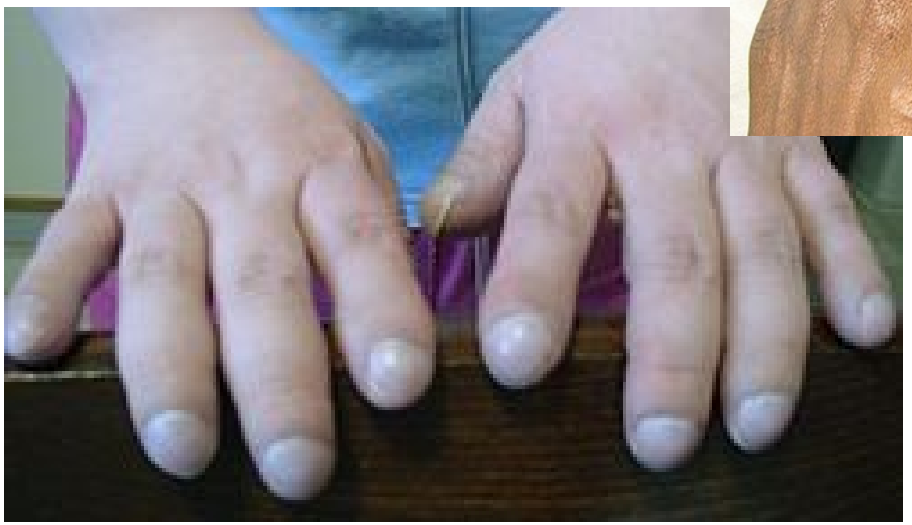
Findings in Patients with CHD

- 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





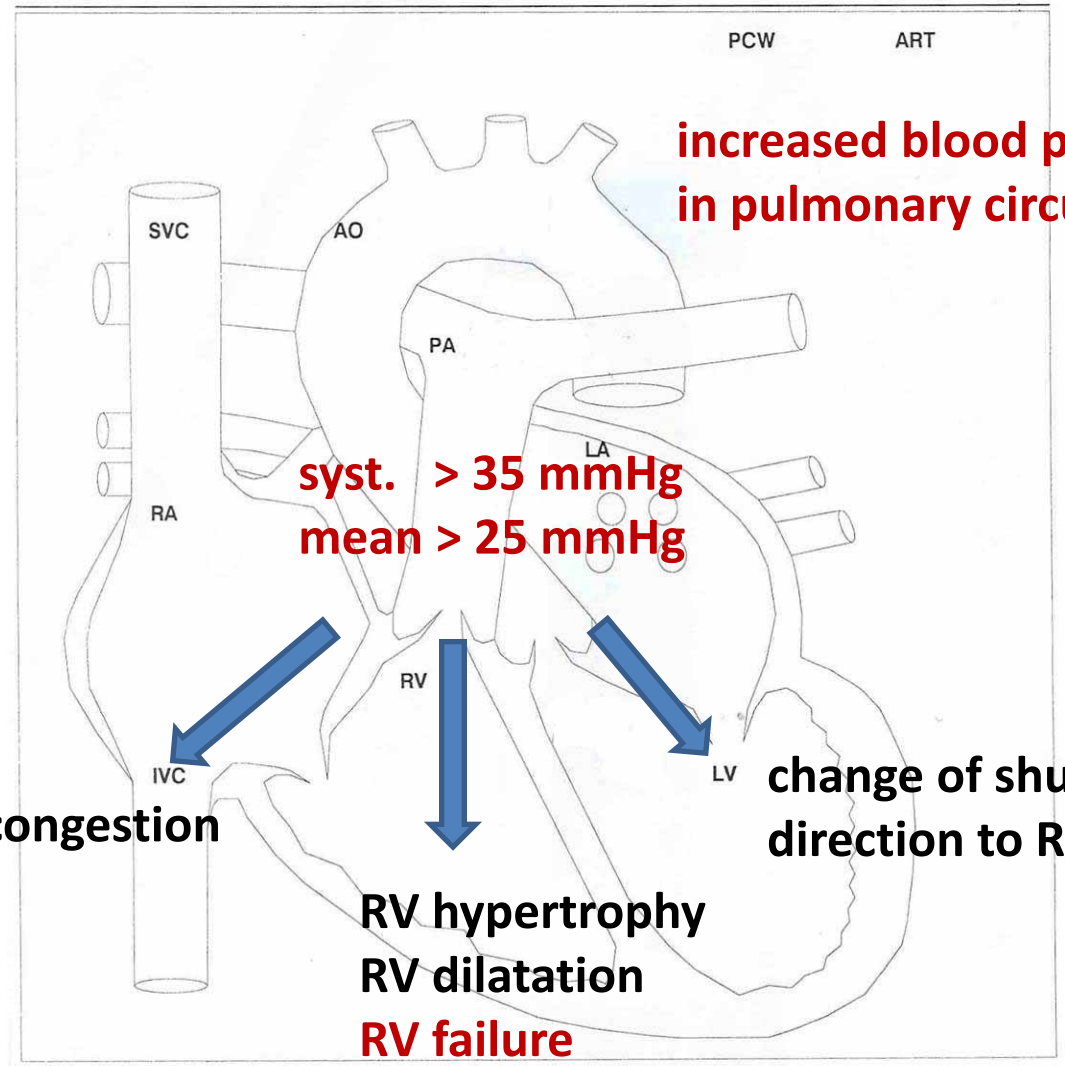
Cyanosis

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

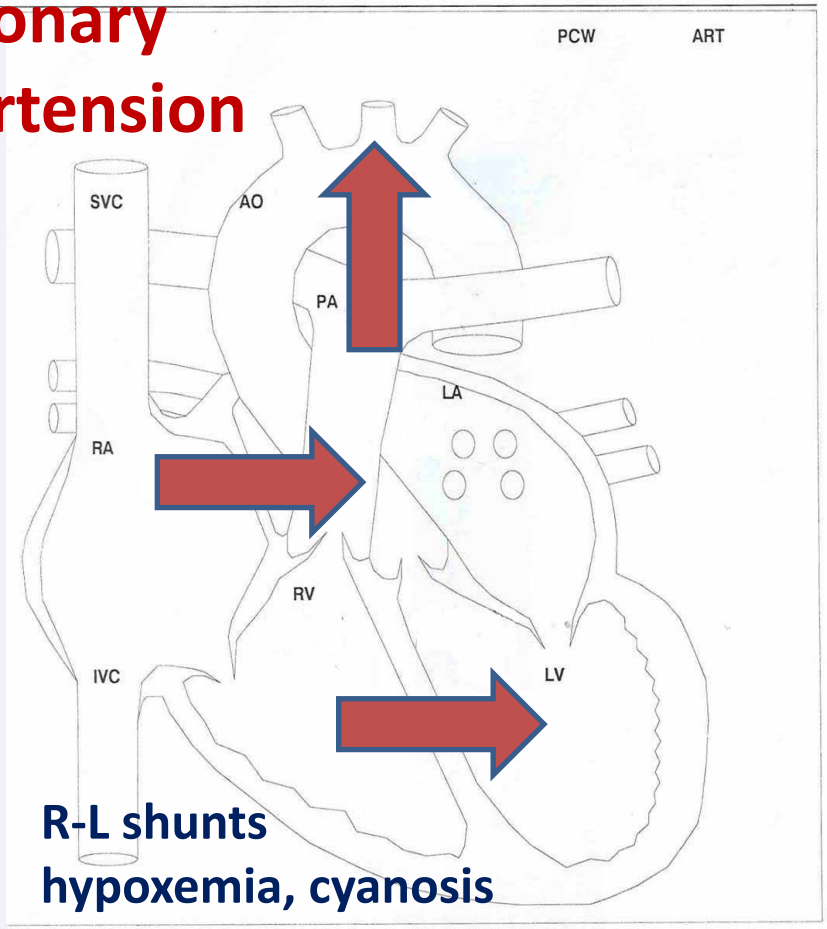
congestion

change of shunt direction to R-L

RV hypertrophy
RV dilatation
RV failure

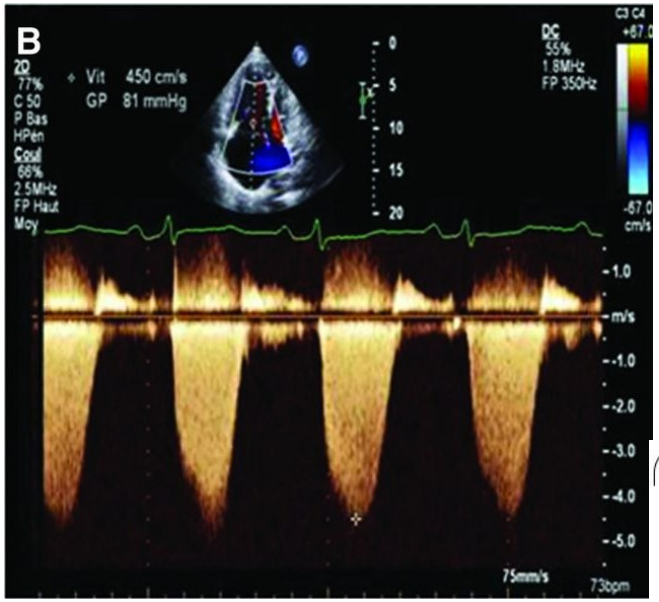
Eisenmenger Syndrome

Pulmonary Hypertension



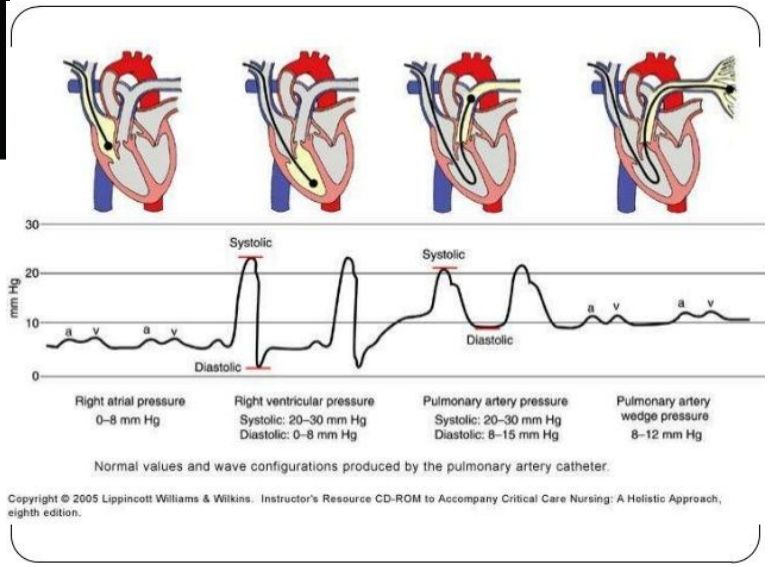
R-L shunts
hypoxemia, cyanosis

Pulmonary Hypertension

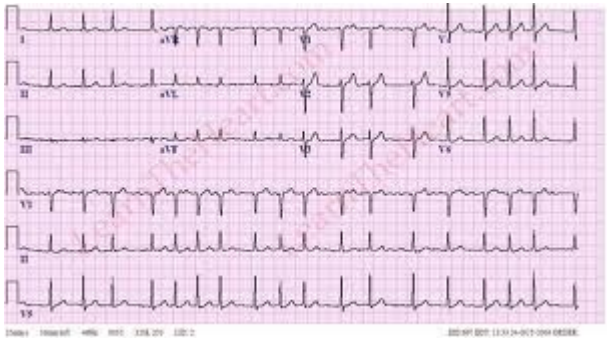


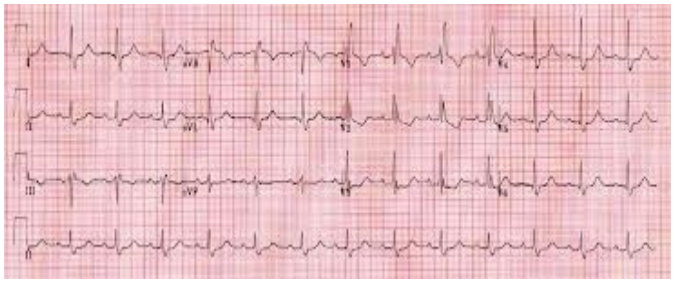
ECHO

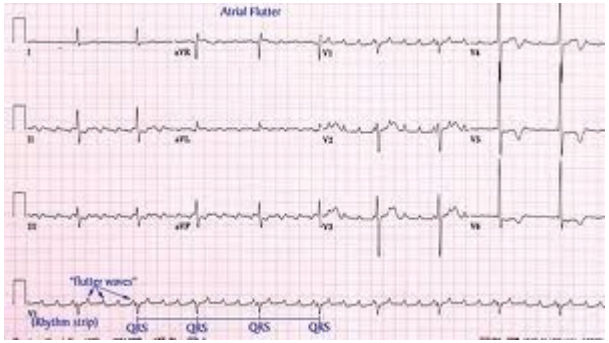
CATHETER

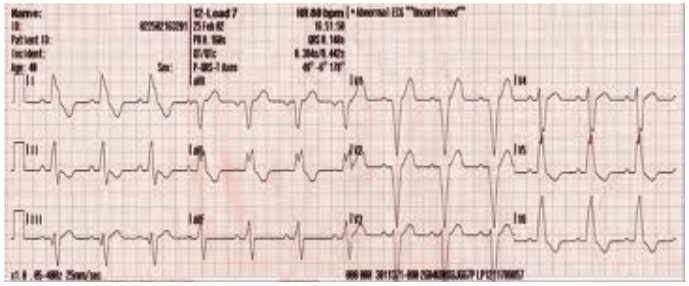


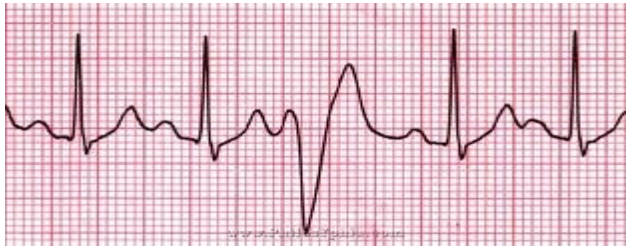
Arrhytmias in Patients with CHD

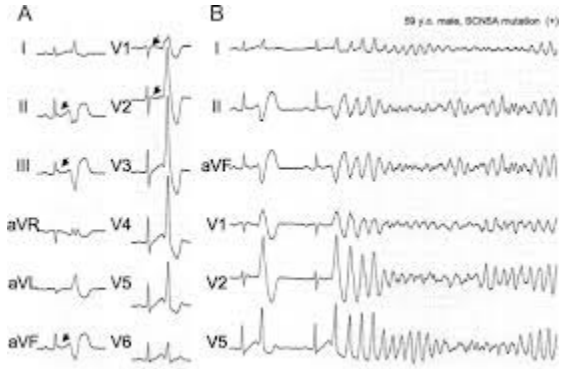












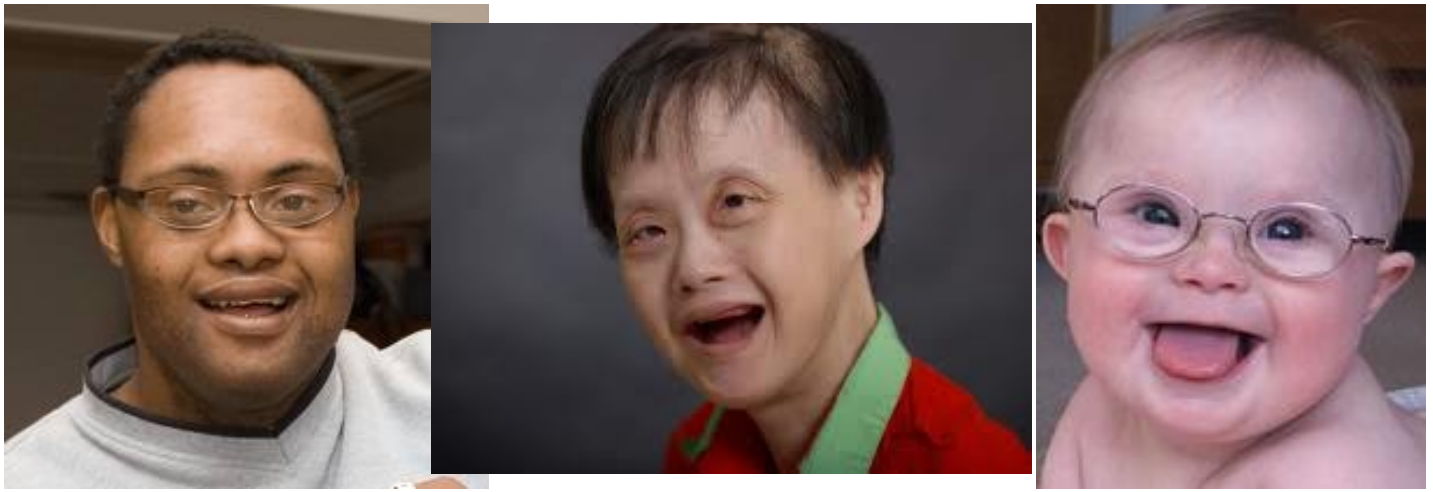
Etiology of CHD

- **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.

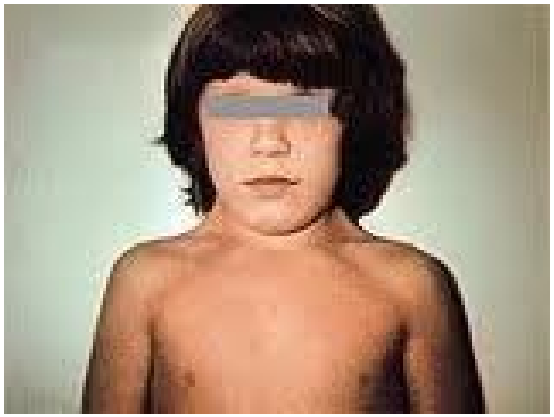
Down syndrome (Trisomy 21)



atrioventricular septal defects

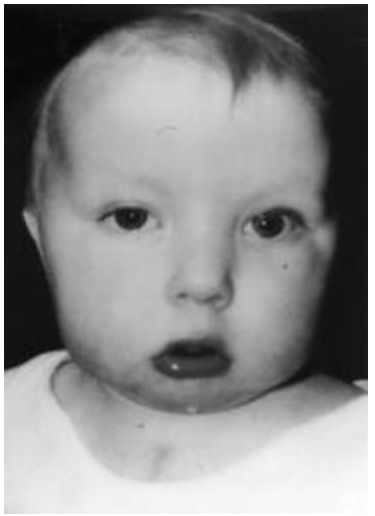
tetralogy of Fallot

Turner syndrome



coarctation, bicuspid aortic valve

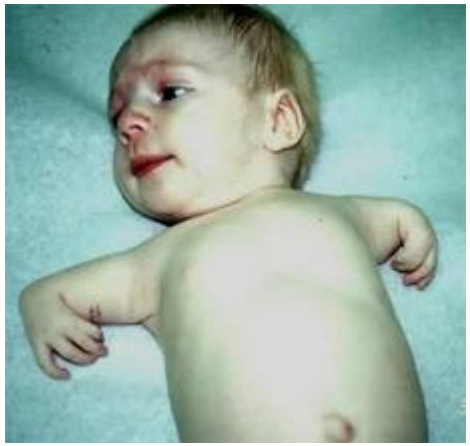
DiGeorge syndrome (CATCH 22)



tetralogy of Fallot, right sided aortic arch,

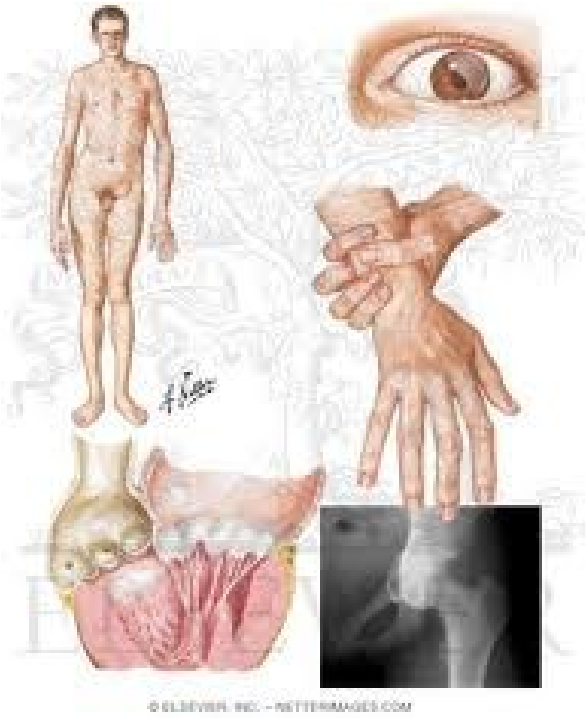
pulmonary atresia, aortic-to-pulmonary collaterals

Holt-Oram syndrome



septation defects (ASD, VSD)

Marfan Syndrome

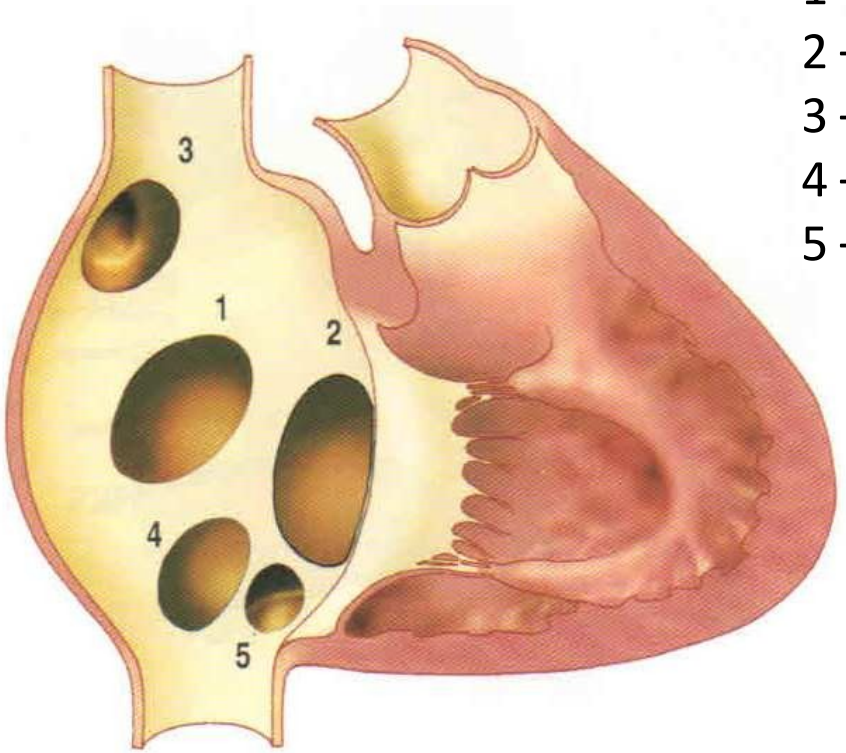


aortic dilation, aneurysm, dissection; heart valve disorders

CHD Anatomy and Pathophysiology

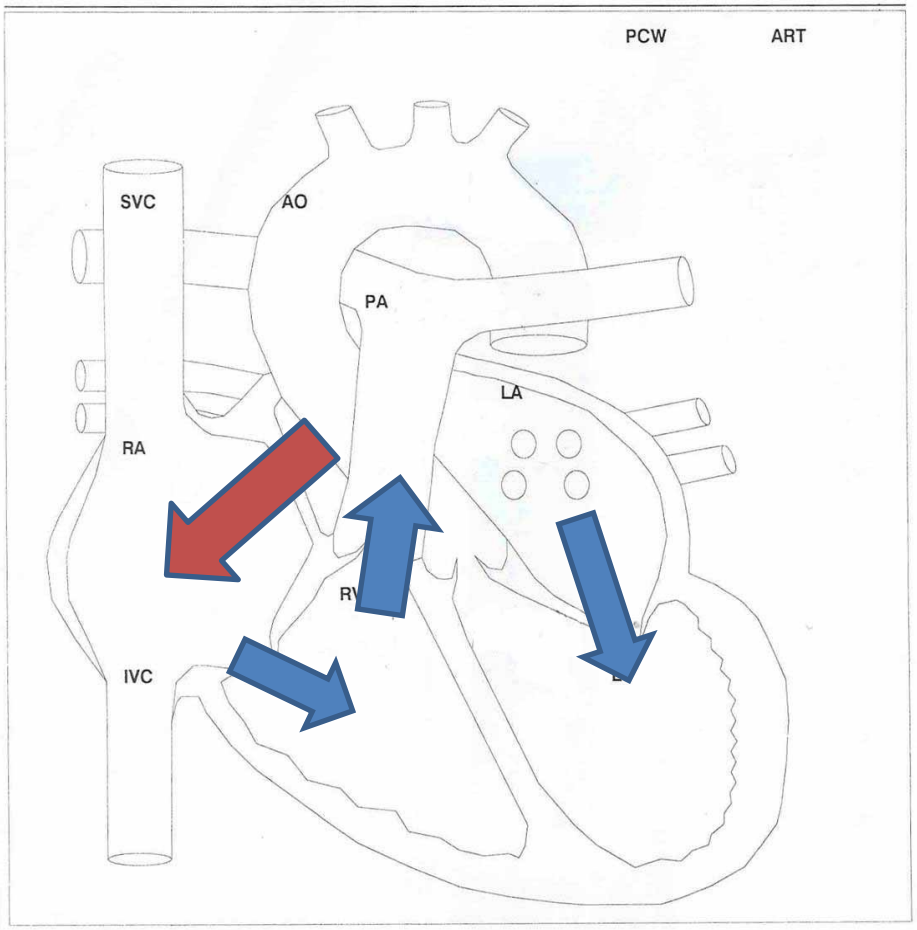
- Septation Defects (ASD, VSD)
- Patent Arterial Duct
- Aortic Coarctation
- Tetralogy of Fallot
- Transposition of Great Arteries

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

↓

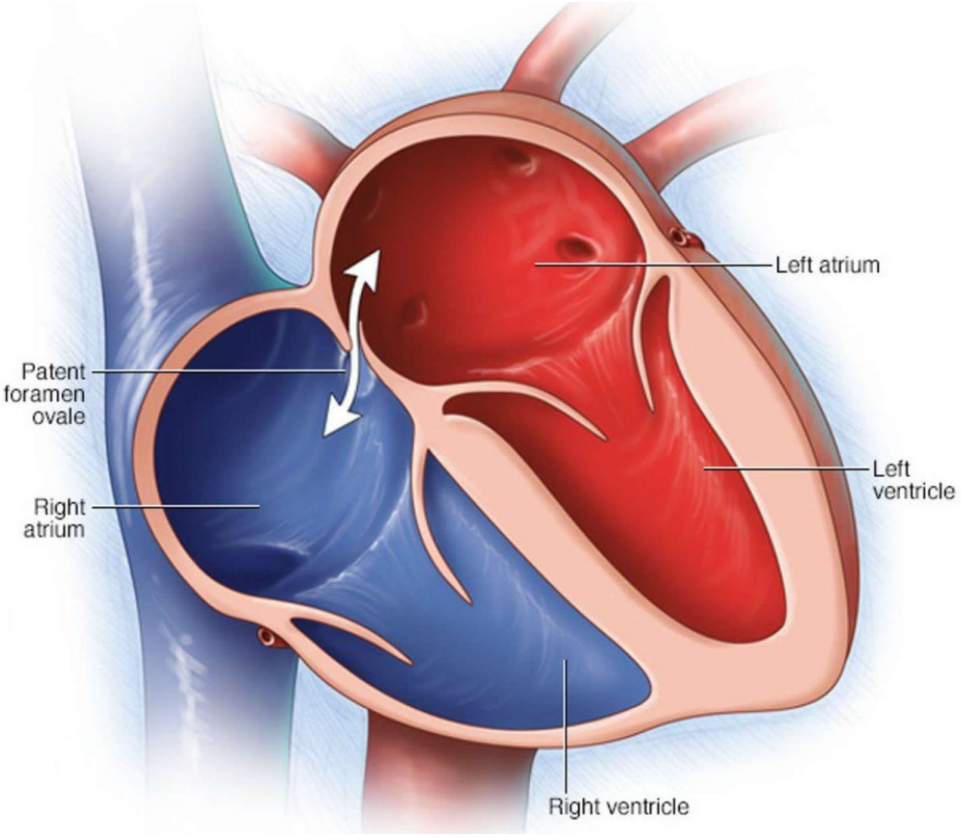
pulmonary hypercirculation

RV dilatation

↓

PH, bidirectional shunt

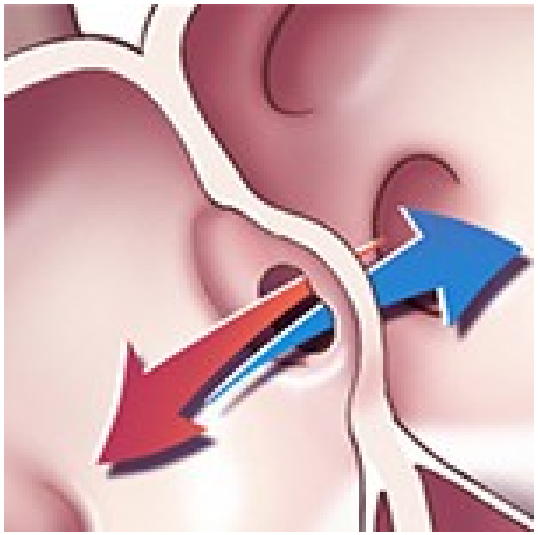
Patent Foramen Ovale (PFO)



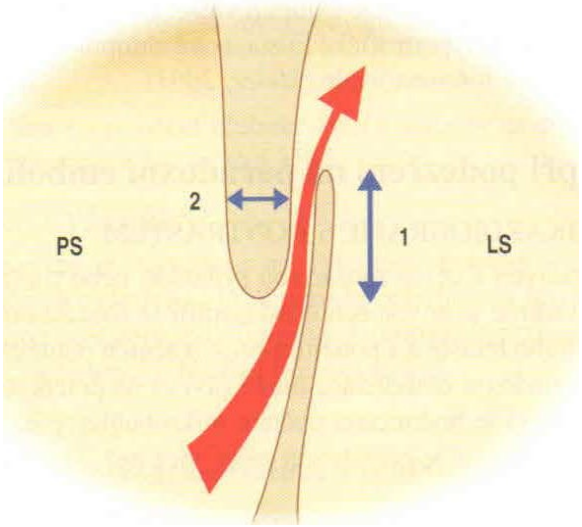
- 25-30% of general population
- not considered as CHD

ASD x PFO

CHD

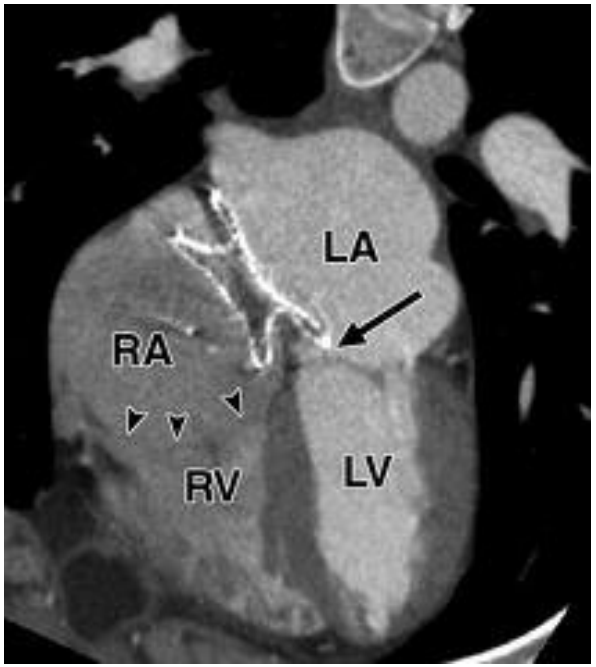


~~CHD~~

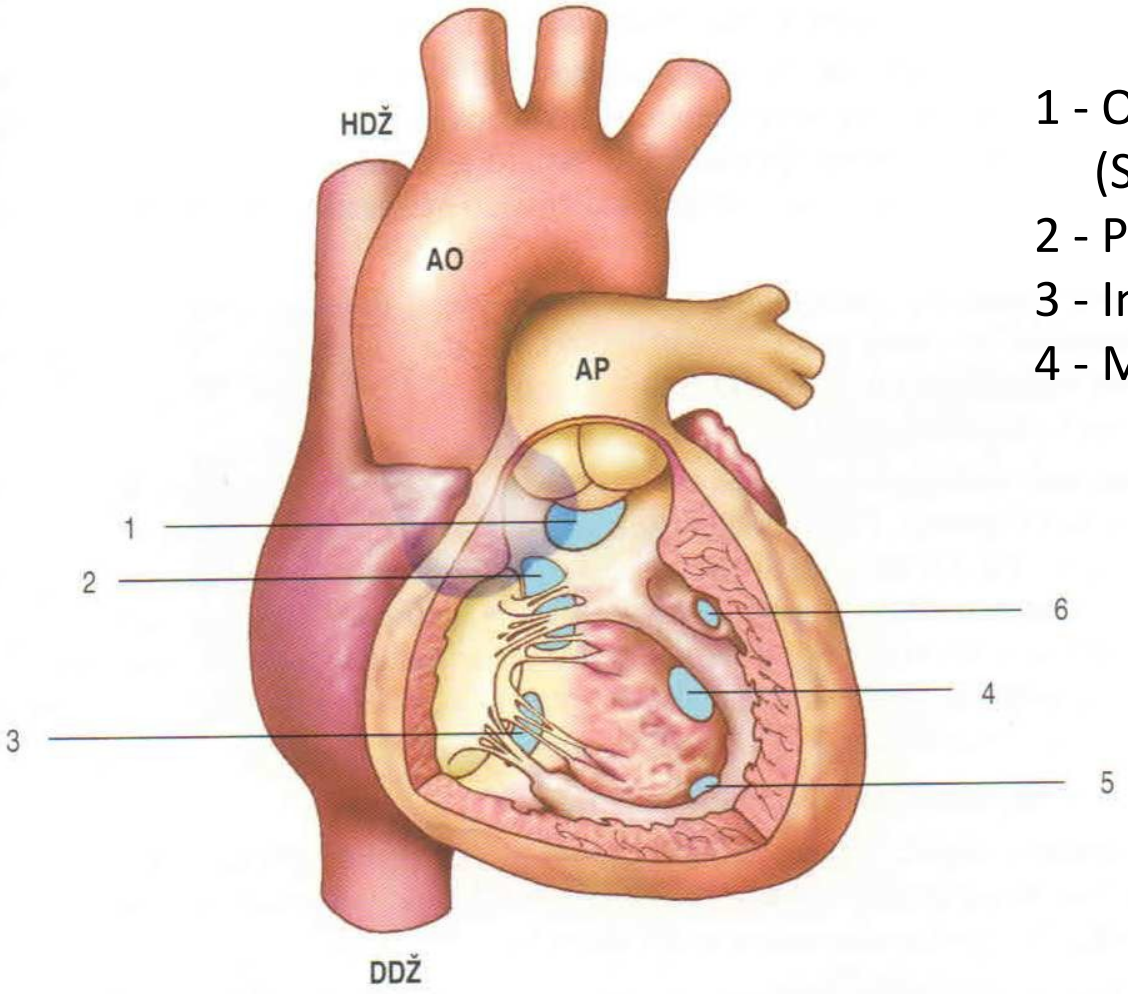


paradoxical embolism risk

Amplatzer Ocluder - PFO/ASD II Closure

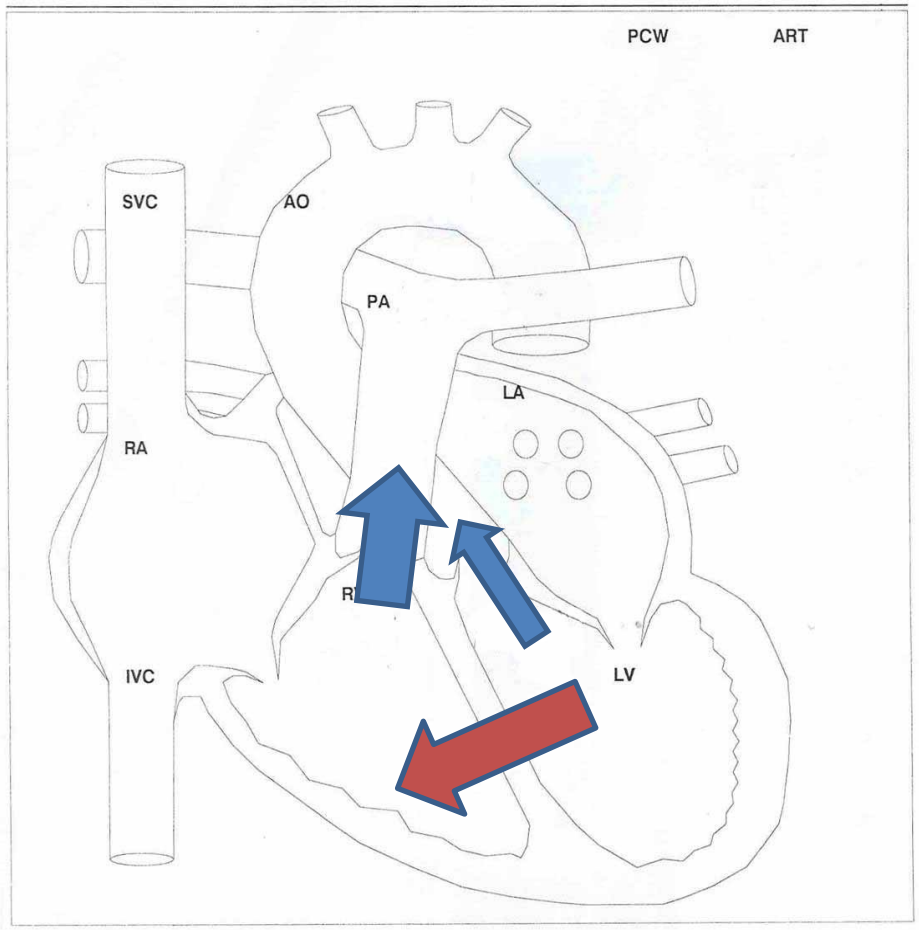


Ventricular Septal Defect (VSD)



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

VSD Pathophysiology



left to right IV shunt

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pulmonary hypercirculation

volume overload of left atrium and left ventricle

↓

PH

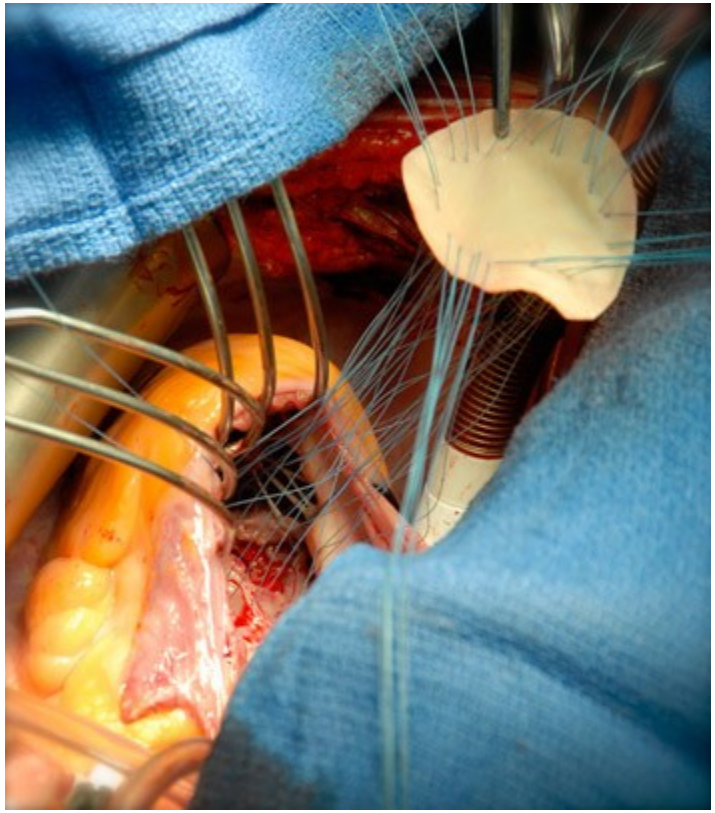
RV hypertrophy

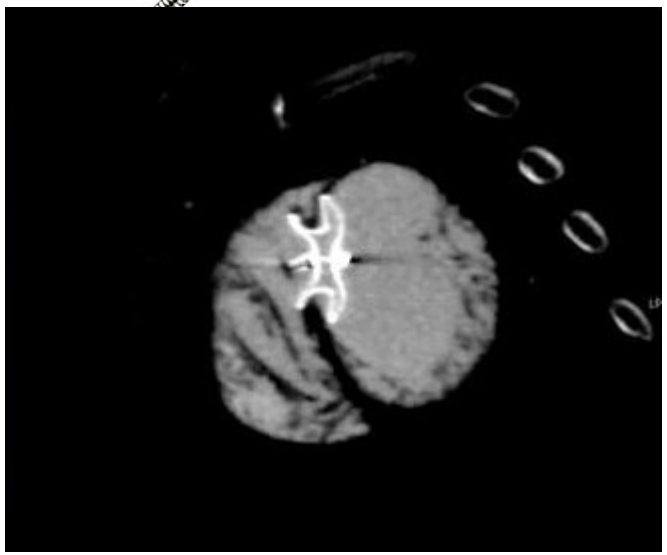
↓

right to left shunt

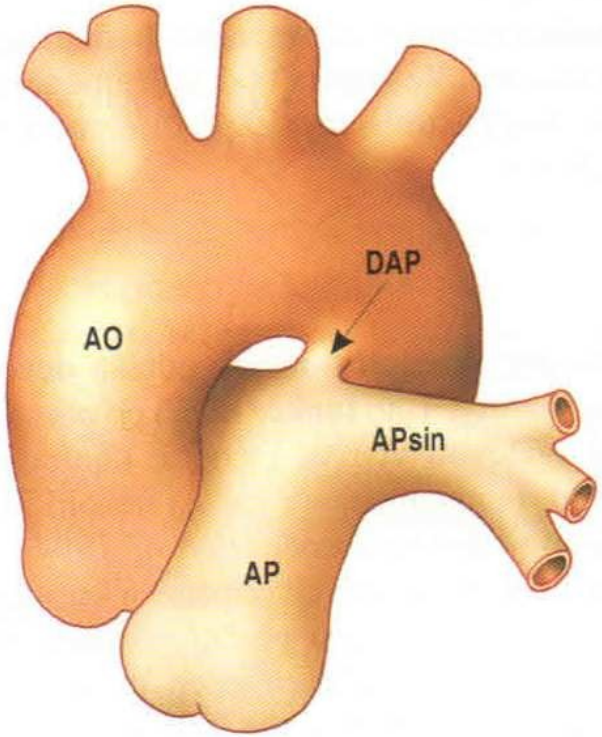
Eisenmenger syndrome

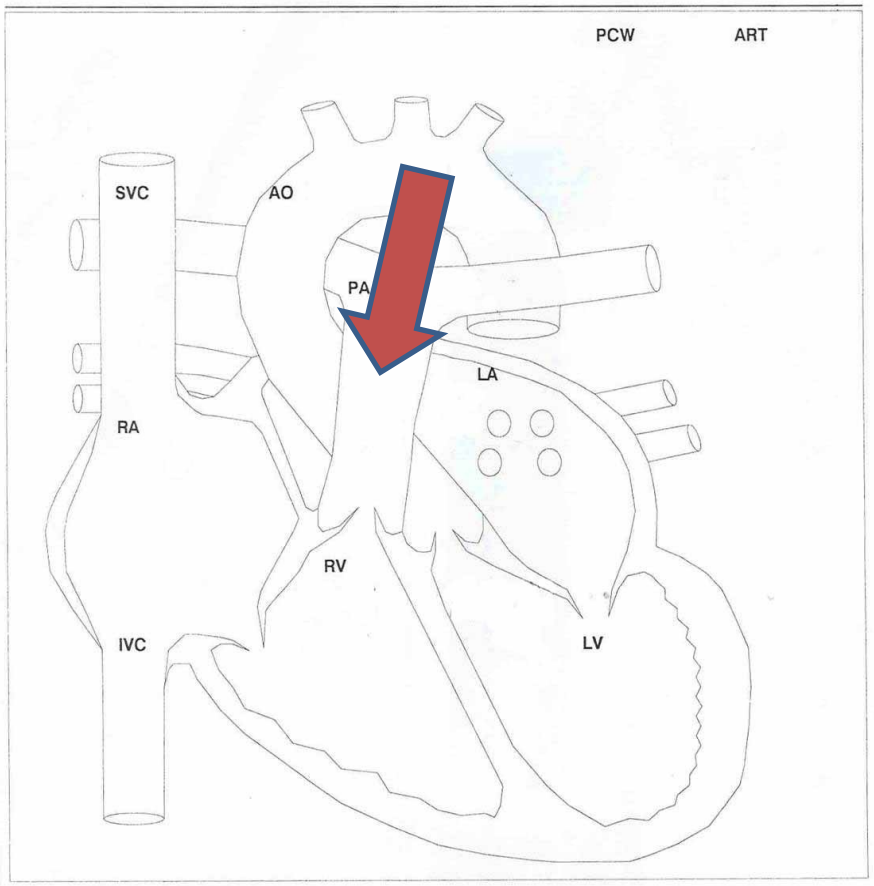
VSD Closure





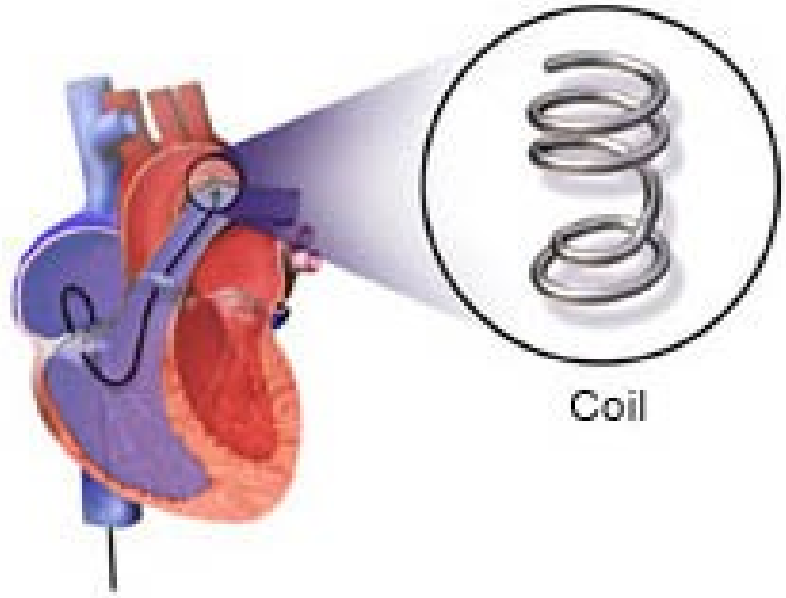
Patent Ductus Arteriosus (PDA)





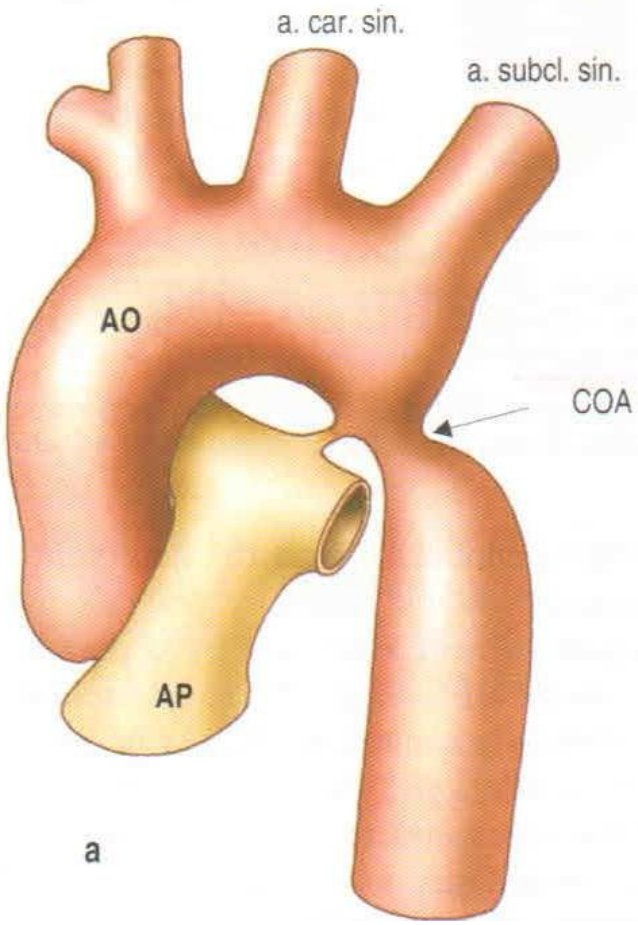
L-R shunt → PH → RV hypertrophy → R-L shunt → Eisenmenger sy

PDA Closure



Coil Closure of PDA

Coarctation of the Aorta



hypertension

in precoarctation area

⬇

LV hypertrophy

⬇

LV dysfunction

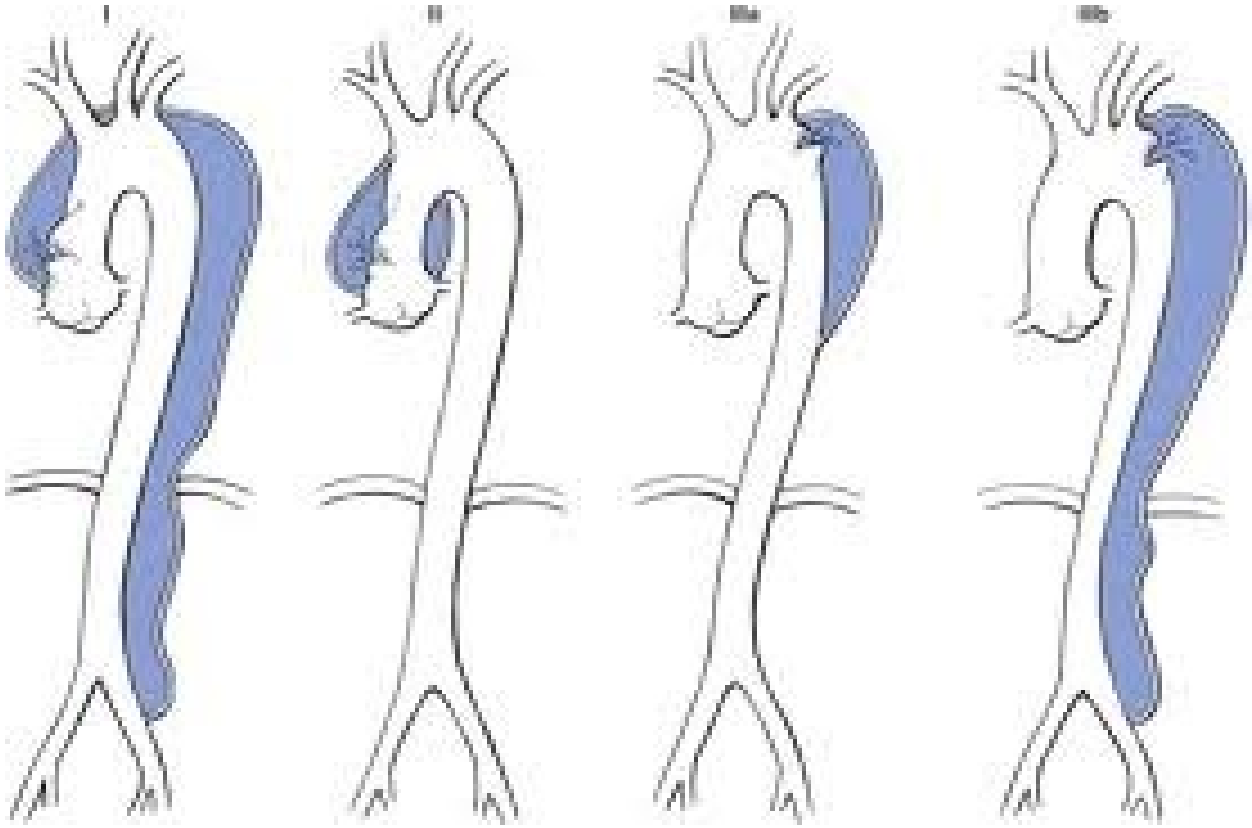
ascend. Ao dilatation

⬇

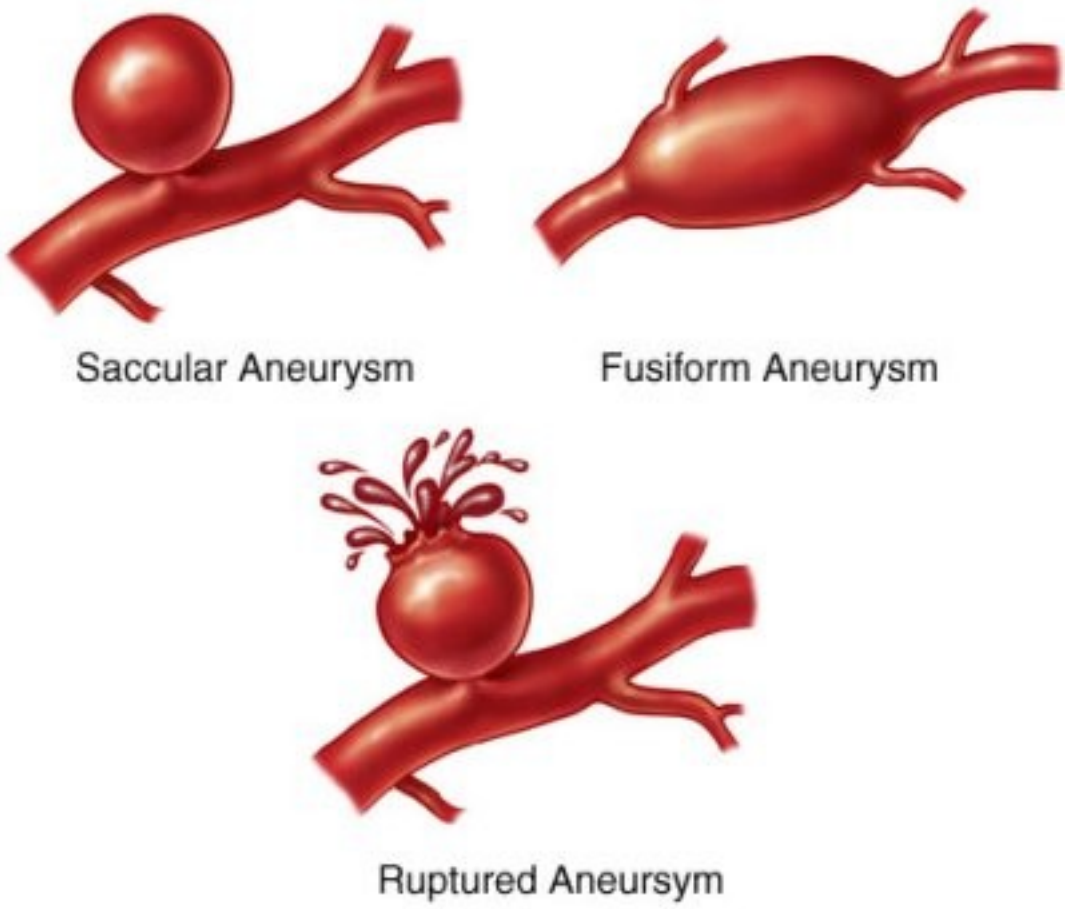
aortic dissection/rupture

*85% assoc. with bicuspid Ao

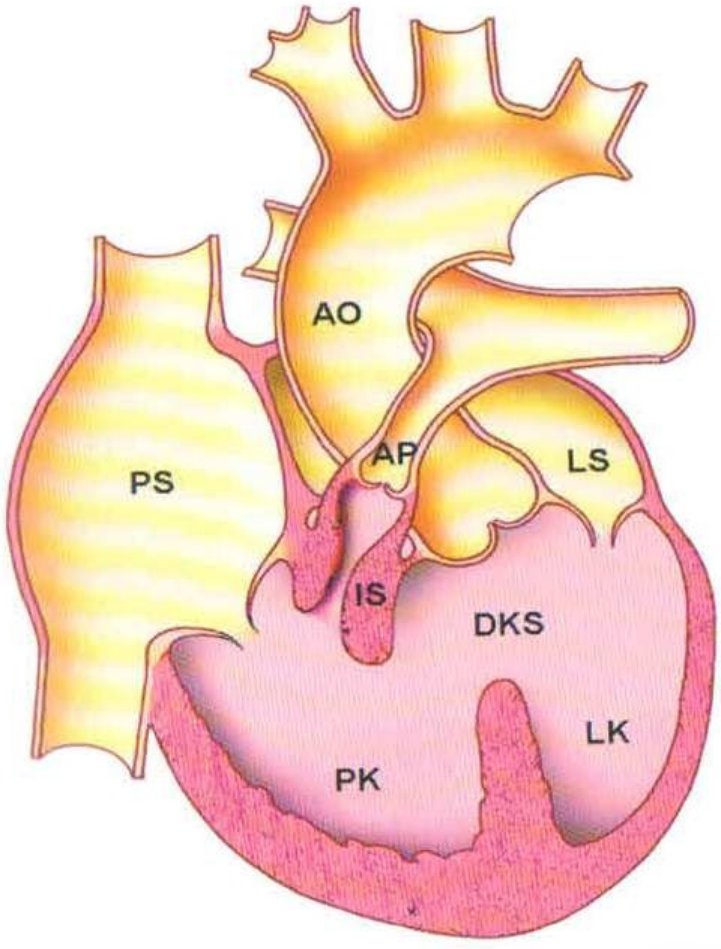
Aortic Dissection



Aortic Aneurysm



Tetralogy of Fallot (TOF)

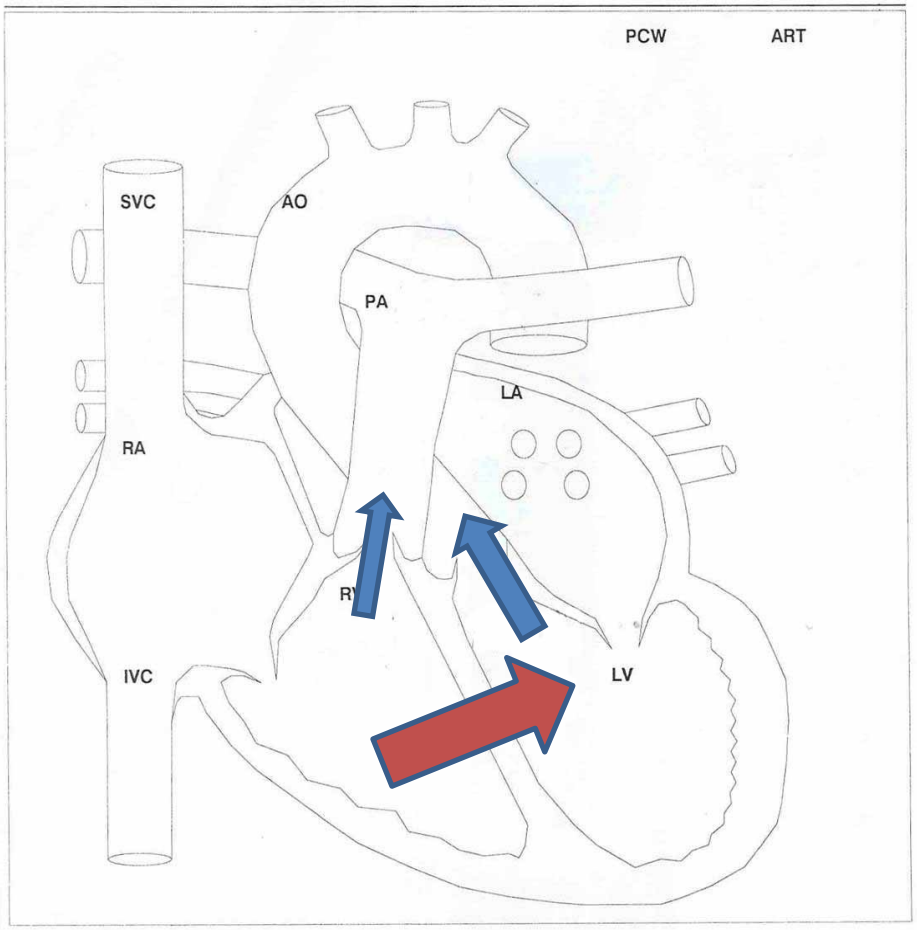


- ventricular septal defect
- overriding aorta
- pulmonary stenosis
- RV hypertrophy
- (atrial septal defect /PFO)

↓

(Pentalogy of Fallot)

TOF Pathophysiology



Pu stenosis

RV hypertrophy

R-L shunt

↓

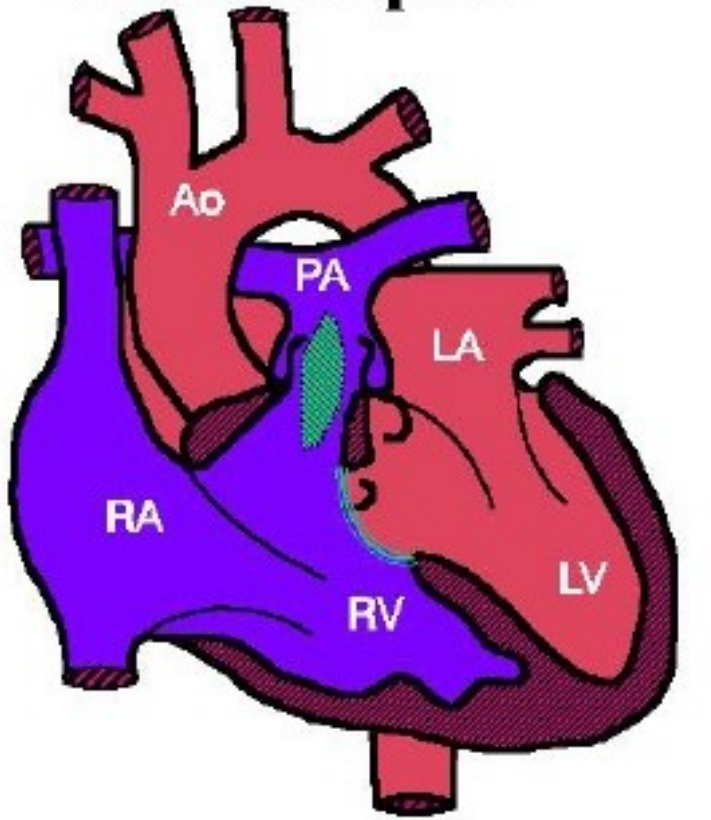
cyanosis / „pink Fallot“

↓

hypoxemia

RV failure

Fallot's Tetralogy after 'Repair'

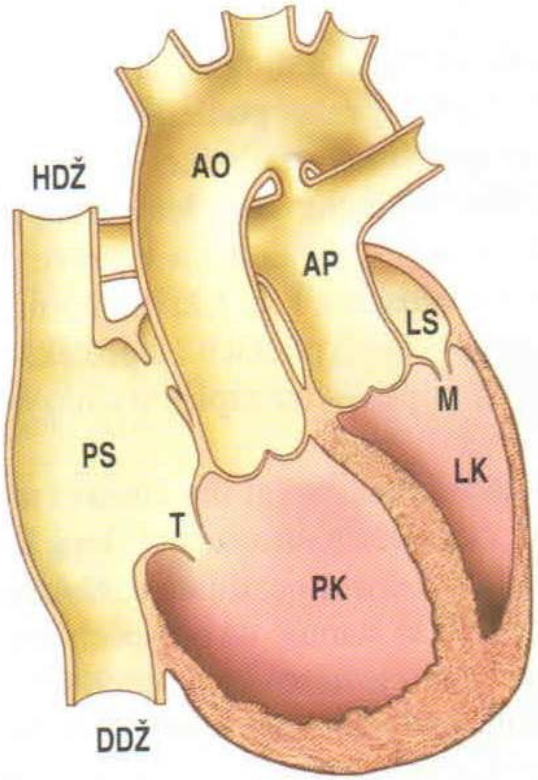


PA patch
VS patch

↓

„normal“ circulation

Transposition of the Great Arteries (TGA)

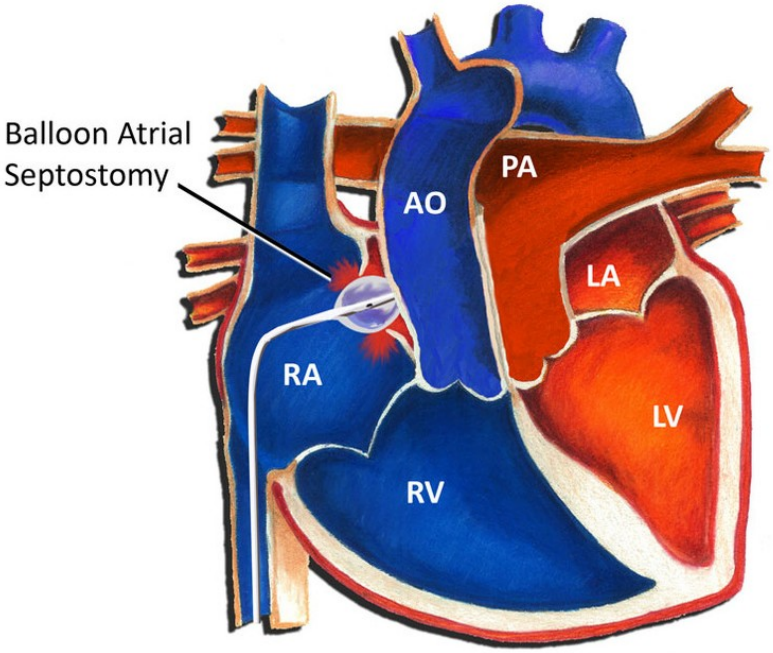


d-TGA

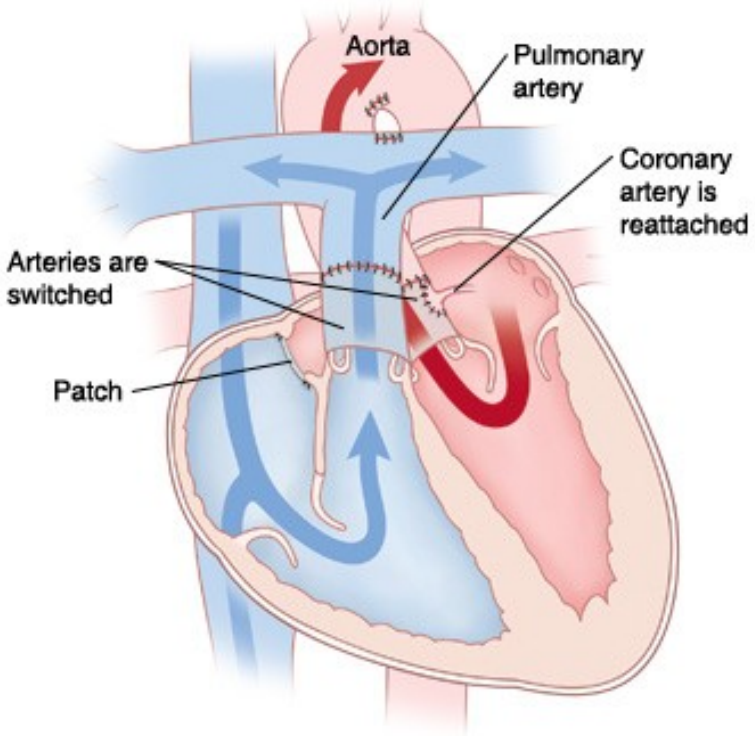
- 2 isolated circulations
- oxygenated blood in isolated pulmonary circulation
- deoxygenated blood in isolated systemic circulation
- impossible to survive without correction

TGA Repair

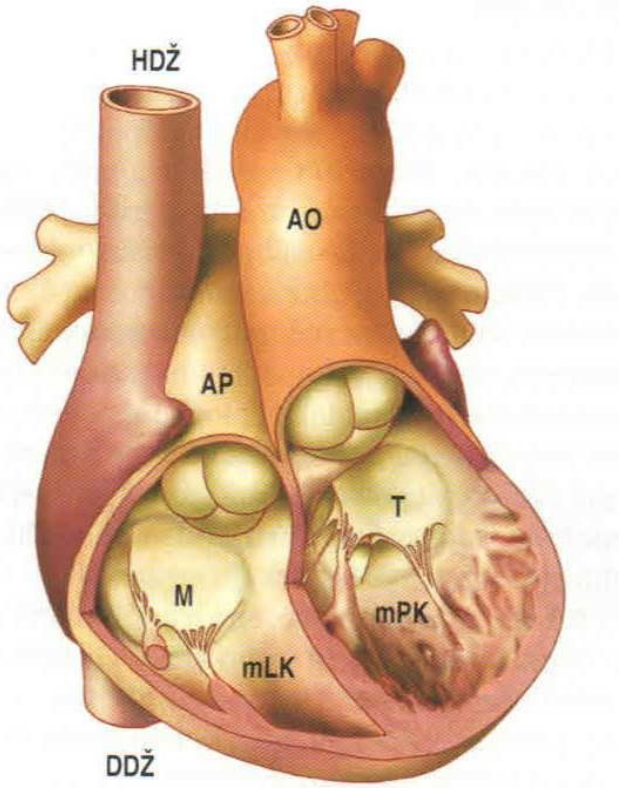
palliative septostomy



arterial switch

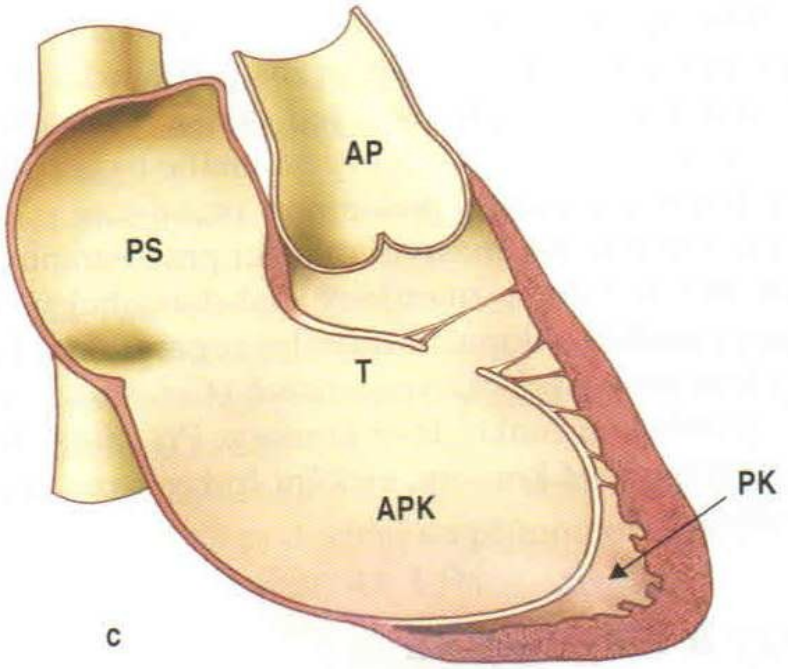


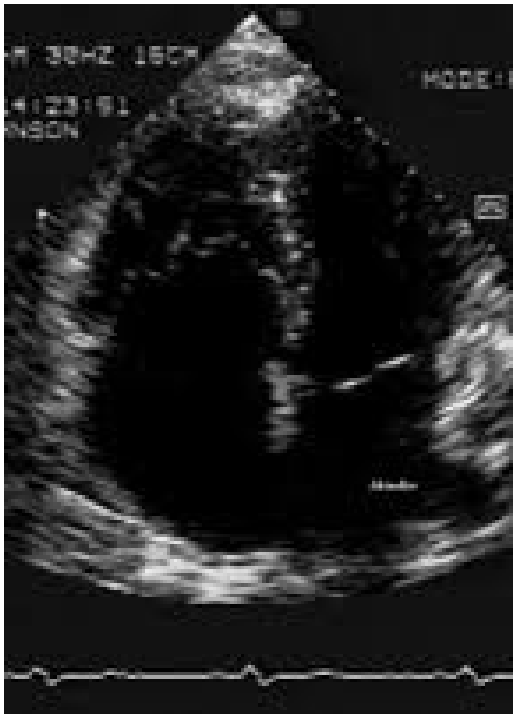
Congenitally Corrected TGA (CCTGA)



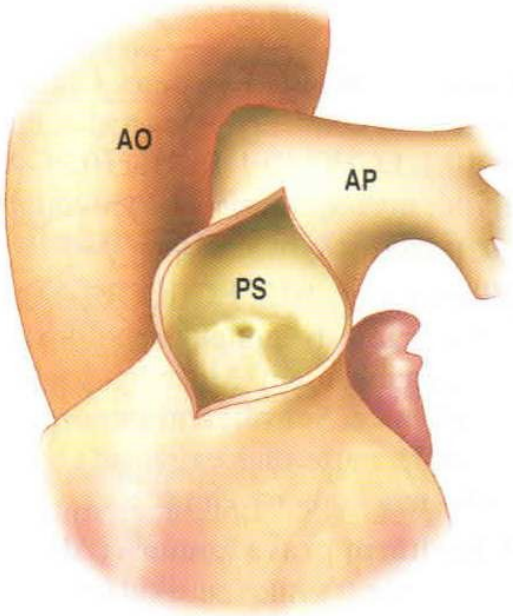
- „normal“ circulation
- RV in systemic circulation
- RV dysfunction/failure

Ebstein´s Anomaly of the Tricuspid Valve

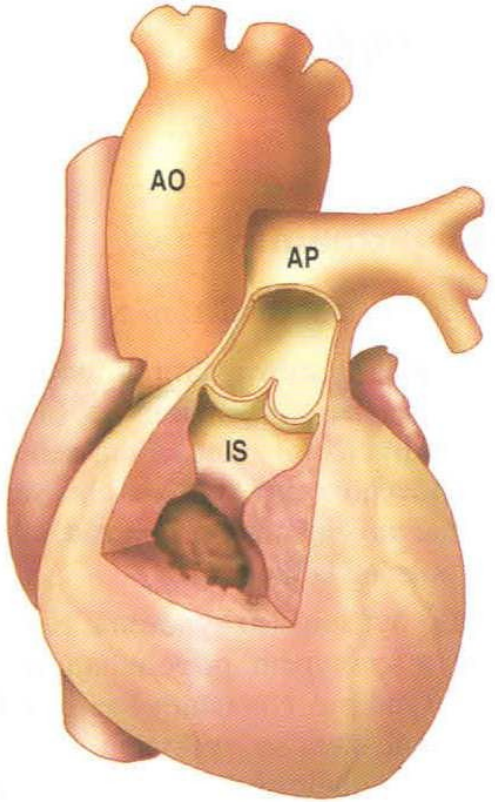




Pulmonary Stenosis



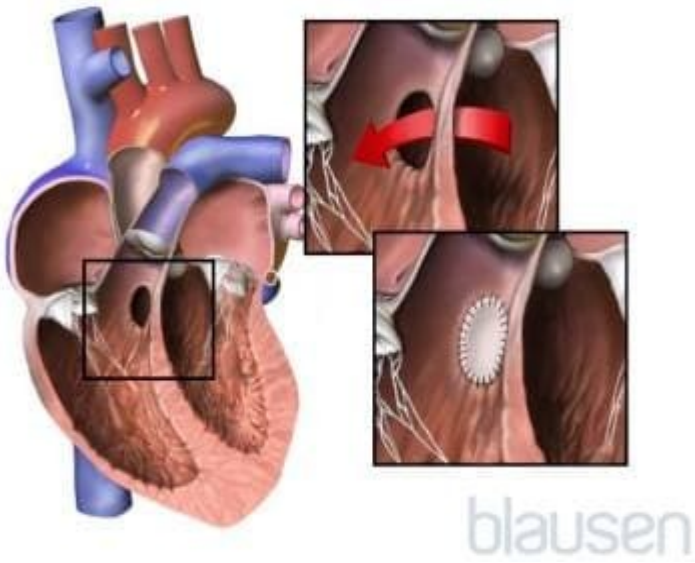
valvar

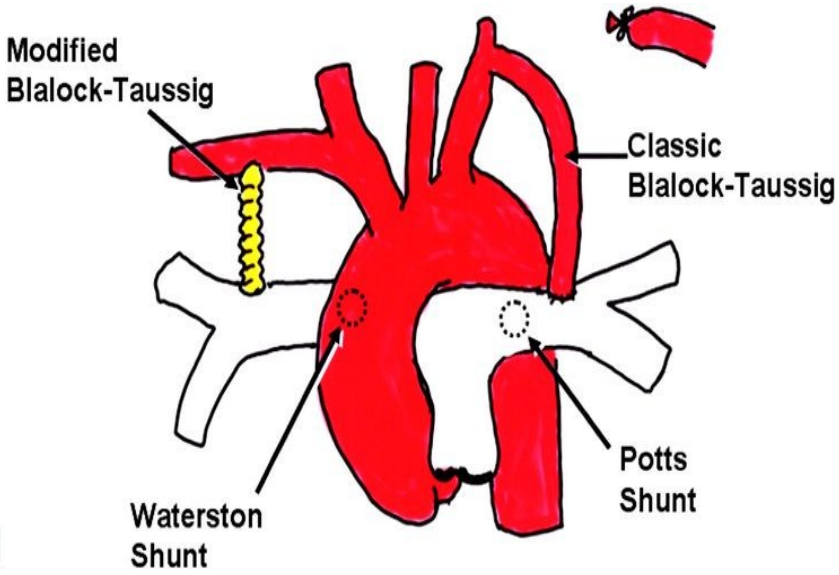


infundibular

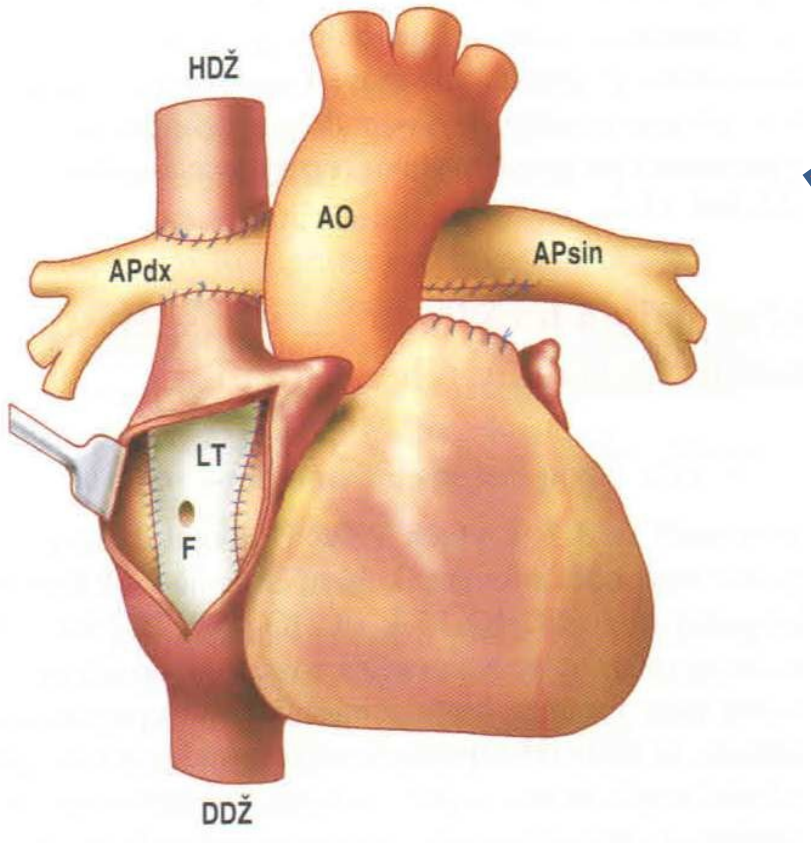
Surgical Repair of CHD

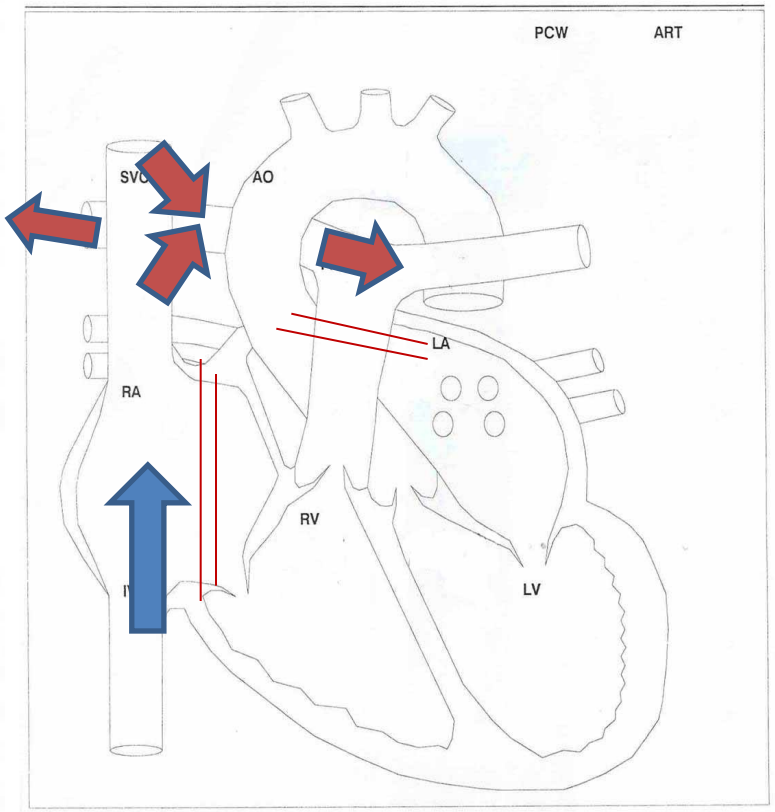
- **corrective**
  (patch...)
- **palliative**
  (connections...)





Fontan Circulation





CHD non-included Congenital Disorders

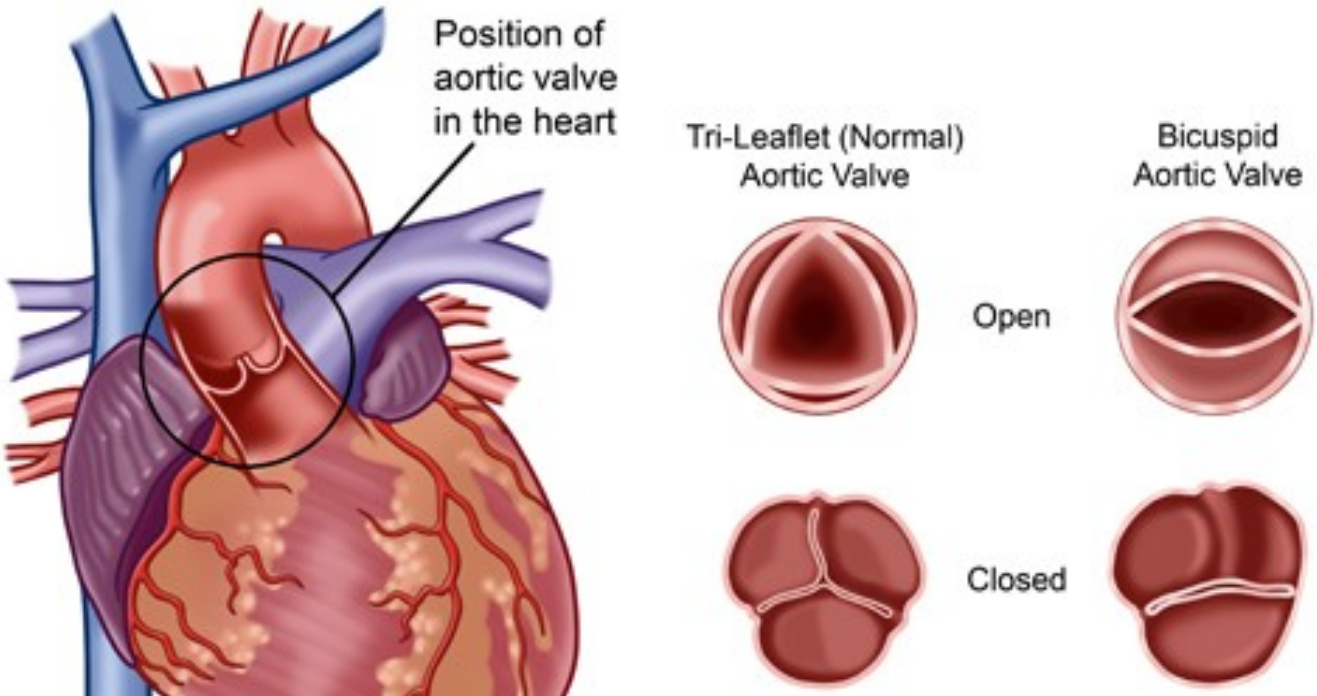
Bicuspid Aortic Valve 1-2% common population

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

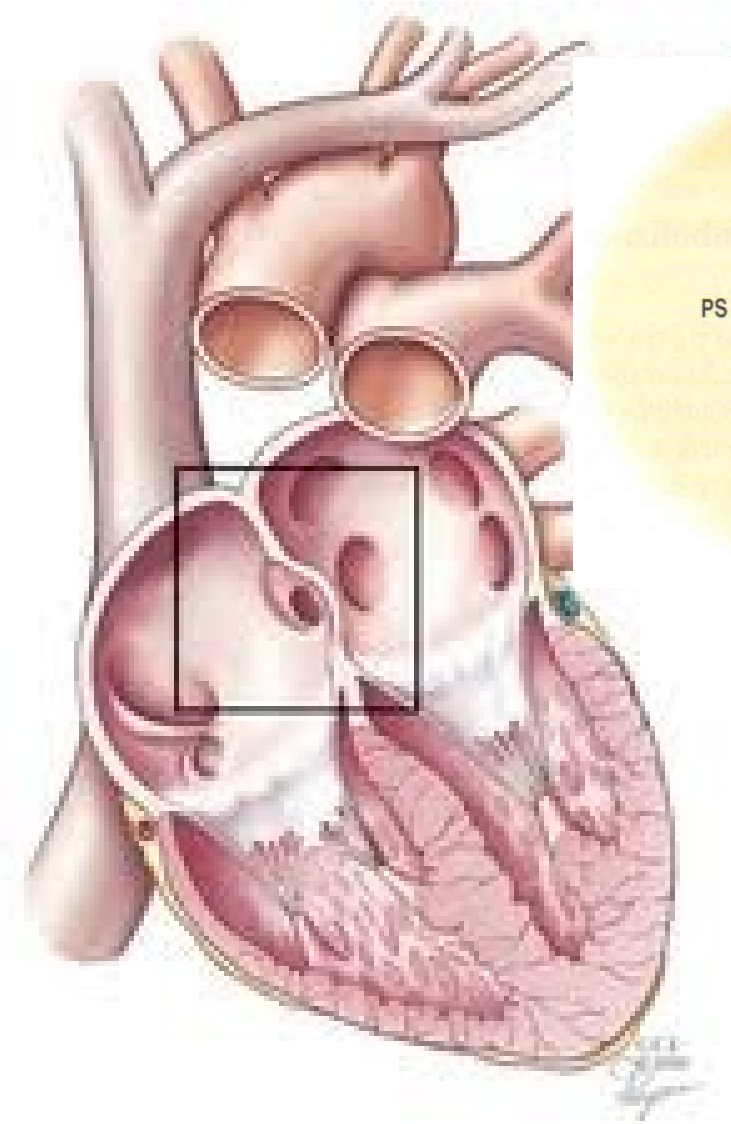
Persistent Left Upper Vena Cava 0,5%

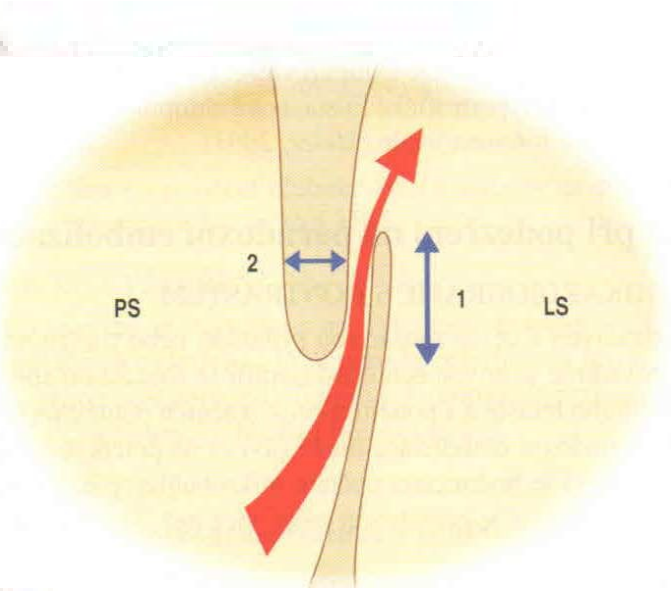
Cardiomyopathies

Bicuspid Aortic Valve (BAO)

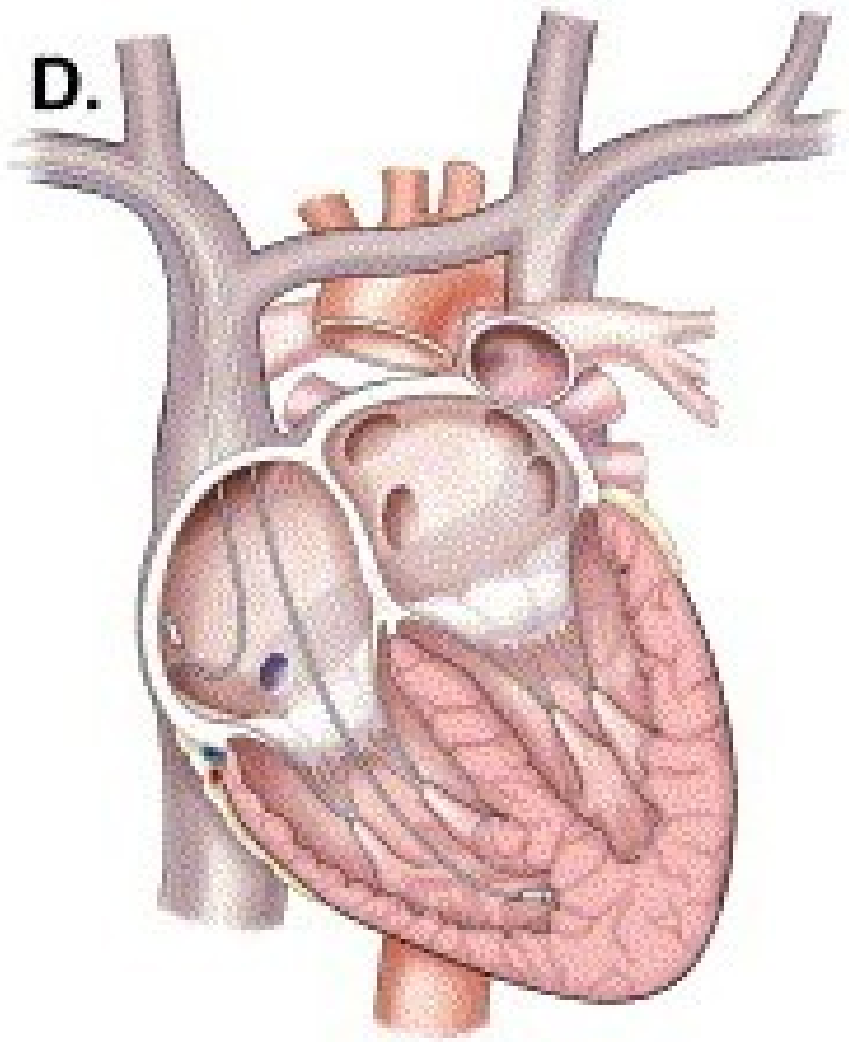


Patent Foramen Ovale (PFO)





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!

If You have any questions to discuss or need consultation regarding cardiology topics, do not hesitate to contact me by e-mail:

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(ID 39899)