CONGENTAL HEART DISEASE
# Introduction

- CHDs are abnormalities of the heart or great vessels that are present at birth.
- Common type of heart disease among children.

## Pathogenesis

- The cause is unknown in almost 90% of cases.
- **Environmental factors**, such as congenital rubella infection

## Genetic factors

- Mutations of the TBX5 transcription factor cause the atrial and ventricular septal defects seen in Holt-Oram syndrome.
- Mutations in the transcription factor NKX2.5-atrial septal defects (ASDs).
Congenital heart disease can be organized 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
CHD

Acyanotic CHD
- L to R shunts: ASD, VSD, PDA
- Obstructive lesions: Pulmonic stenosis, Aortic stenosis, Coarctation of aorta, Mitral regurgitation

Cyanotic CHD
- R to L shunts: TOF, Complete TGA
Pathogenesis

- Genes located on chromosome 22 have a major role in forming the conotruncus, the branchial arches, and the human face.
- Deletions of chromosome 22q11.2 underlie 15% to 50% of outflow tract abnormalities

- **CHD can be subdivided into 3 major groups:**
  1) Malformations causing a *left-to-right shunt*
  2) Malformations causing a *right-to-left shunt* (cyanotic congenital heart diseases)
  3) Malformations causing *obstruction*
Left-to-Right Shunts

- Left-to-right shunts: most common type of congenital cardiac malformation.
- They include atrial and ventricular septal defects, and patent ductus arteriosus.
- Atrial septal defects: increased pulmonary blood vol, while ventricular septal defects and patent ductus arteriosus: increased pulmonary blood flow and pressure

- Can be asymptomatic or can cause fulminant CHF at birth.

Ventricular septal defect is the most common.
Dev’t of Atrial Septum & Defects

- AS begins as an ingrowth of the septum primum
- Minor degree of patency persists in about 25% of the general population.

1) The most common (90%) is the ostium secundum ASD.
2) Ostium primum ASDs are less common (5% of cases).
3) The sinus venosus ASDs (5% of cases) associated with frameshift mutations in the NKX2.5 transcription factor.
## Morphology

### Ostium secundum ASDs

- **a)** smooth-walled defects near the foramen ovale.
- **b)** Hemodynamic lesions are accompanied by RA & ventricular dilation.
- **c)** RV hypertrophy, and dilation of the pulmonary artery.
- **d)** This reflect the effects of a chronically increased volume load on the right side of the heart.

### Ostium primum ASDs

- ✓ occur at the lowest part of the atrial septum and can extend to the mitral and tricuspid valves
- ✓ Abnormalities of the AV valves are usually present
- ✓ VSD and severe mitral and tricuspid valve deformities, with a resultant common AV canal.
**Clinical Features**

- ASDs initially cause left-to-right shunts, as a result of the lower pressures in the pulmonary circulation and right side of the heart.
- Pulmonary vascular resistance can increase, resulting in pulmonary hypertension.
- Ostium primum defects are more likely to be associated with evidence of CHF, in part because of the high frequency of associated mitral insufficiency.
- In general these defects are well tolerated.
Patent Foramen Ovale

• foramen ovale/ostium secundum permits continued right-to-left shunting of blood during intrauterine development. • the unsealed flap can open if right-sided pressures become elevated (coughing, sneezing) can produce brief periods of right-to-left shunting
Clinical findings of ASD

Symptoms

- Generally asymptomatic until adulthood
- Pulmonary plethoric: frequent chest infections
- Systemic Circulation Insufficiency: Failure to thrive, poor weight gain, feeding difficulty, fatigue, shortness of breathe, sweating
- Cyanosis: Severe cyanosis in large lesions, softer heart murmur and accentuated P2.
X-ray findings

- Plethoric Lung fields
- RA and RV enlargement
- Prominent PA segment
- Normal or small aortic shadow
USG findings

- RA, RV enlargement
- RV overloaded
- Parallel shunt between atria in Doppler
Complication of ASD

1. Bronchopneumonia
2. Congestive heart failure
3. Infective endocarditis
## Ventricular Septal Defects

- Incomplete closure of the VS allows L-to-R shunting, the **most common congenital cardiac anomaly at birth.**

- VS is formed by fusion of an intraventricular muscular ridge that grows upward from the apex of the heart with a thinner membranous partition that grows downward from the endocardial cushion.

- The basal (membranous) region is the last part of the septum to develop and is the site of approximately **90% of VSDs.**

There is also: Muscular, Infundibular

- 30% of VSDs occur in isolation; more commonly, they are associated with other cardiac malformations.

The functional consequences of a VSD depend on the size
Morphology

1. The size and location of VSDs are variable.
2. Range from minute defects to large defects involving virtually the entire septum.
3. RV is hypertrophied and often dilated.
4. Increased diameter of the pulmonary artery.
5. Pulmonary hypertension.
L→R Shunts – General Points

**PDA & VSD**
- Presents in infancy w/ heart failure, murmur, and poor growth
- Left heart enlargement (LHE)
- Transmits flow and pressure

**ASD**
- Presents in childhood w/ murmur or exercise intolerance (AVSD or 1° ASD presents earlier)
- Right heart enlargement (RHE)
- Transmits flow only

AVSD can present as either depending on size of ASD & VSD component
Atrioventricular (AV) septal defect
- Endocardial cushion defect
- Down syndrome
Clinical Features

a) Small VSDs may be asymptomatic.

b) Larger defects, however, cause a severe left-to-right shunt.

c) Pulmonary hypertension and CHF.

d) Reversal of the shunt and cyanosis.

e) Small- or medium-sized defects that produce jet lesions in the right ventricle are also prone to superimposed infective endocarditis.
<table>
<thead>
<tr>
<th>Feature of the shunts</th>
<th>Left to right shunts</th>
<th>Right to left shunts</th>
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<tbody>
<tr>
<td></td>
<td>Generally no cyanosis</td>
<td>Cyanosis appears early</td>
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<tr>
<td></td>
<td>Increased pulmonary circulation</td>
<td>Pulmonary circulation increase or decrease</td>
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<td>Decreased systemic circulation</td>
<td>Deoxygenated blood mix with oxygenated blood in systemic circulation</td>
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<td>Pulmonary arterial hypertension (hyperkinetic obstructive)</td>
<td>Persistent cyanosis in late stage (Eissenmager’s syndrome)</td>
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**Patent Ductus Arteriosus**

- **During intrauterine life**, the ductus arteriosus permits blood flow from the pulmonary artery to the aorta, thereby bypassing the unoxygenated lungs.

- Shortly after birth, the ductus constricts; this occurs in response to increased arterial oxygenation, decreased pulmonary vascular resistance, and declining local levels of prostaglandin E$_2$.

- **Complete, structural obliteration occurs within the first few months of extrauterine life** to form the ligamentum arteriosum.

- PDAs account for about 7% of cases of congenital heart lesions.

*Used to save life in pulmonary or aortic valve obstruction or atresia*
Morphology

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

- In PDAs some of the oxygenated blood flowing out from the left ventricle is shunted back to the lungs.

- Because of the resultant volume overload, the proximal pulmonary arteries, left atrium, and ventricle can become dilated.

- Dev’t of pulmonary hypertension, atherosclerosis of the main pulmonary arteries and proliferative changes in more distal pulmonary vessels are seen, followed by right heart hypertrophy and dilation.
PDA: Clinical Signs

- Murmur
- Hyperactive precordium
- Bounding peripheral pulses
- Increase in pulse pressure
- Hypotension
- Respiratory deterioration
Clinical Features

- PDAs are high-pressure left-to-right shunts.
- Audible as harsh "machinery-like" murmurs.
- Eisenmenger syndrome with cyanosis and CHF.
- Infective endocarditis due to high pressure.
Eisenmenger’s Syndrome

- A long standing L→R shunt will eventually cause irreversible pulmonary vascular disease

- This occurs sooner in unrepaired VSDs and PDAs (vs an ASD) because of the high pressure

- Once the PVR gets very high the shunt reverses (ie- now R→L) and the patient becomes cyanotic
If the ductus arteriosus remains open after birth and fails to close it is referred to as a **patent ductus arteriosus**. The term “patent” means open. **Complications associated with patent ductus arteriosus** are **poor growth and eating, easy tiring, and a rapid heart rate**. It is also common to notice that the infant is blue in color, especially while feeding, due to a lack of oxygen.
PDA: Factors that increase incidence

- **RDS**
  - Correlated with severity of RDS.
  - After surfactant treatment increased risk of clinically symptomatic PDA

- **Prematurity:**
  1. Inversely related to gestational age
  2. Found in approx. 45% of infants <1750gm
     80% of infants <1000gm
Right-to-Left Shunts

- Cardiac malformations associated with right-to-left shunts are distinguished by cyanosis at or near the time of birth.

Important right-to-left shunts (cyanotic congenital heart disease). A, Tetralogy of Fallot. B, Transposition of the great vessels with and without VSD. (Ao, aorta; LA, left atrium; LV, left ventricle; PT, pulmonary trunk; RA, right atrium; RV, right ventricle.)
Right-to-Left Shunts

- Two important conditions associated with cyanotic congenital heart disease are **tetralogy of Fallot and transposition of the great vessels**

**Clinical findings**

- Cyanosis
- Clubbing of the fingertips (hypertrophic osteoarthropathy)
- Polycythemia

In addition to:
- Persistent truncus arteriosus
- Tricuspid atresia
- Total anomalous pulmonary venous connection

Note: Tatralogy of Fallot is the most common.
R→L Shunts

Reduced PBF

- Presents more often with cyanosis
- See oligemic lung fields
- Closure of PDA may worsen cyanosis

Why are pressures equal?

Dynamic subvalvular obstruction here causes “Tet spells”
Tetralogy of Fallot

- Most common cause of cyanotic congenital heart disease. (CCHD)
- Accounts for about 5% of all congenital cardiac malformations.

**4 features of the tetralogy**

1. VSD
2. Obstruction to the right ventricular outflow tract (subpulmonic stenosis)
3. An aorta that overrides the VSD
4. Right ventricular hypertrophy
Morphology

- The heart is large and "boot shaped" as a result of RV hypertrophy
- Proximal aorta is larger than normal, with a diminished pulmonary trunk.
- Right ventricular wall is markedly thickened.
- PDA or ASD; are actually beneficial because they permit PBF
- The pulmonary outflow tract is narrowed
Predominant manifestation of TGA is early cyanosis.

Infusions of prostaglandin E$_2$ can be used to maintain patency of the ductus arteriosus.

Maneuvers such as atrial septostomy are performed to create ASDs that enhance arterial oxygen saturation. Even with stable shunting.
Transposition of the Great Arteries:

- Aorta---anterior---right ventricle
- Pulmonary artery---posterior---left ventricle
- Parallel instead of series
- Incompatible with postnatal life unless a shunt exists for adequate mixing of blood
Persistent truncus arteriosus
• failure of separation of truncus into aorta and pulmonic trunk.
• The truncus overrides both ventricles.
• Always accompanied by a membranous VSD.
Total anomalous pulmonary venous return (TAPVR)

- Pulmonary veins----innominate vein/ coronary sinus

- Patent foramen ovale/ or ASD always present
Obstructive Lesions

- Congenital obstruction to blood flow can occur at the level of the heart valves or within a great vessel.

- Common examples of congenital obstruction:
  1. Pulmonic valve stenosis,
  2. Aortic valve stenosis or atresia,
  3. Coarctation of the aorta.
Coarctation of the Aorta

- **Coarctation** - is narrowing of the aorta at varying points anywhere from the transverse arch to the iliac bifurcation.

- 98% of coarctations are juxtaductal

- Male: Female ratio 3:1.

- Accounts for 7% of all CHD.

- It is accompanied by a bicuspid aortic valve. Congenital aortic stenosis, ASD, VSD, or mitral regurgitation may also occur. In some cases berry aneurysms in the circle of Willis coexist.
Coarctation of the Aorta

Hemodynamics

- Obstruction of left ventricular outflow $\Rightarrow$ pressure hypertrophy of the LV.
2 Types of Coarctation
1- Infantile: + PDA---- early after birth--- cyanosis in lower ½ of body
2- Adult: - PDA---- late presentation---- HTN in upper extremities
Coarctation of the Aorta

**Clinical Signs & Symptoms**

- Classic signs of coarctation are diminution or absence of femoral pulses.

- Higher BP in the upper extremities as compared to the lower extremities.

- 90% have systolic hypertension of the upper extremities.

- Pulse discrepancy between rt & lt arms.
Coarctation of the Aorta

Clinical Signs & Symptoms

- With severe coarc. LE hypoperfusion, acidosis, HF and shock.

- Differential cyanosis if ductus is still open

- II/VI systolic ejection murmur.

- Cardiomegaly, rib notching on X-ray.
Coarctation of the Aorta

**Treatment**

- With severe Coarctation maintaining the ductus with prostaglandin E is essential.

- Surgical intervention, to prevent LV dysfunction.

- Angioplasty is used by some centers.

- Re-coarctation can occur, balloon angioplasty is the procedure of choice.
# Transposition of the Great Arteries

- TGA is a discordant connection of the ventricles to their vascular outflow.
- Embryologic defect in abnormal formation of the truncal and aortopulmonary septa.

- The predominant manifestation of TGA is early cyanosis
- Tissue hypoxia
- Right ventricular hypertrophy
- Left ventricle becomes atrophic
Aortic Stenosis and Atresia:

1- Valvular --- hypoplastic left heart syndrome

2- Supravalvular

3- Subvalvular ---- sudden death with exertion
THE END