Congenital Heart Disease

- Abnormalities of the heart/ great vessels since birth
- Incidence higher in premature infants
- Faulty embryogenesis during 3-8 weeks of IU life
- Cause – unknown
  - genetic or environmental
  - rubella infection, drugs, heavy drinking during pregnancy
Classification

1. Malpositions of Heart
   Dextrocardia
   may be accompanied by situs inversus
2. Shunts (Cyanotic CHD)
3. Obstructions (Obstructive CHD)
Shunts

A. Left to Right shunts (Acyanotic or Late Cyanotic group) 
   cyanosis months or years after birth
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 (TOF)- 6-15%
2. Transposition of great arteries -4-10%
3. Persistent truncus arteriosus – 2%
4. Tricuspid atresia and stenosis 1%
Obstructions

1. Coarctation of Aorta  5-7%
2. Aortic stenosis and atresia  4-6%
3. Pulmonary stenosis and atresia  5- 7%
Shunts

Abnormal communication b/w 2 chambers or blood vs; blood flows according to pressure gradient

R→L shunt:

• ↓pulm blood flow → poor oxygenation of blood → enters Lt heart → systemic circulation → dusky blueness of mucus membranes and skin (Cyanosis)
• Functional anemia → increased synthesis of Hb + RBC mass (polycythemia)
• Emboli from peripheral veins donot undergo filtration action of lungs → enter Lt heart → embolize to systemic circulation (paradoxical emboli) → cause brain infarction & abscess
• Clubbing (hypertrophic osteoarthropathy) of tips of fingers and toes
Shunt

L→R shunt:
- ↑pulm blood flow → ↑pulm pressure → RVH → potential cardiac failure
- ↑pulm blood flow → medial hypertrophy + intimal proliferation to prevent pulmonary edema.
- Prolonged ↑pulm pressure → (> even systemic pressure) → reversing the flow from R→L: unoxygenated blood in systemic © → late cyanosis or Eisenmenger syndrome
- Once significant pulmonary HT develops, surgical Rx of cardiac defects not possible
Ventricular Septal Defect (VSD)

- Hole between the two ventricles, incomplete closure of ventricular septum
- Left to right shunt – majority
- Dilated right heart – too much blood to lungs – increase in pulmonary pressure
- Morphology:
  - 90% in membranous septum
  - 10% lie below pulm valve or within muscular septum
  - Mostly single. Multiple VSDs in muscular septum: Swiss cheese septum
VSD

CI features: depend on size of lesion

- Small lesions recognized later or may spontaneously close
- Large VSDs recognized early in life, cause Lt- Rt shunts → volume hypertrophy of RV → pulmonary HT since birth. Ultimately, shunt reversal, cyanosis, death
- Not corrected till 1 yr to wait for spontaneous closure
Atrial Septal Defect

- Abnormal fixed opening in atrial septum caused by incomplete tissue formation
- Not to confuse with patent foramen ovale present in 30% of normal individuals
- Unnoticed in infancy and childhood
- Usually presents late in life (30), late cyanotic heart dis.
- L→R shunt at atrial level (pulm vascular resistance is less than systemic and compliance of Rt ventricle is greater than Lt)
- Pulm blood flow increased to 2-4 times, hypertrophy of RA and RV
- Pulmonary HT, RHF unusual
Morphology: 3 types according to location

- **Secundum ASD (90%)**: deficient or fenestrated oval fossa near centre of septum
- **Primum ASD**: occur adjacent to AV valves
- **Sinus venosus**: near entrance of SVC
- **AVSD – Atrio ventricular septal defect**
Patent Ductus Arteriosus

• Ductus arteriosus is normal connection b/w aorta and bifurcation of pulmonary A
• Normally closes at 1\textsuperscript{st} or 2\textsuperscript{nd} day of life, > 3months persistence is abnormal
• Cause: possibly due to ↑ levels of PGE2 after birth
- seen in children with respiratory distress syndrome
- pharmacologic closure with indomethacin (PGE2 inhibitor)
• Most often does not produce functional difficulties at birth
• A narrow ductus: no effect on growth and development during childhood
• Harsh machinery like murmur
Right to Left shunts (Cyanotic CHD)

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

Cyanosis in early postnatal life
Tetralogy of Fallot

• Combination of shunts with obstruction with functional shunting of blood
• Most common cyanotic heart disease
• 4 features:
  1. VSD
  2. Displacement of aorta to right side so that it overrides the septal defect
  3. Sub-pulmonary stenosis (obstruction)
  4. Right ventricular hypertrophy
• Clinical manifestations dependant on extent of pulmonary stenosis & VSD
• 2 types: Cyanotic and Acyanotic (pink tetralogy)
• Cyanotic:
  - Pulm stenosis is greater $\rightarrow \uparrow$ resistance to flow of blood in RV $\rightarrow$ it flows to LV $\rightarrow$ Cyanosis
  - Effects: - pressure hypertrophy of RA and RV
    - small tricuspid valve
    - small Lt atrium & ventricle
    - enlarged aortic orifice
• Acyanotic tetralogy:
  - VSD larger, pulmonary stenosis mild: L $\rightarrow$ R shunt, behaves like isolated VSD
Boot shaped heart
1. Ventricular Septal Defect
2. Pulmonary Stenosis
3. Hypertrophy of Rt. Ventricle
4. Overriding Aorta

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Transposition of Great arteries

- **Regular transposition:**
  - Aorta arises from RV and Pulmonary A from LV - cyanosis from birth

- **Corrected Transposition:**
  - Aorta arises from RV, Pulmonary A from LV + Pulm veins drain into RA, Sup and Inf vena cava into LA
  - Physiologically corrected circulation
Persistent Truncus Arteriosus

- Arch that separates aorta from pulmonary A fails to develop. A single large vessel receives blood from both the ventricles
- Often associated VSD
- Early systemic cyanosis
- Poor prognosis

Tricuspid Atresia and stenosis
- Often associated with pulmonary stenosis or atresia
- Atresia- absence of tricuspid orifice, there is dimple in floor of rt atrium
- Stenosis- tricuspid ring is small
Obstructions (Obstructive CHD)

Coarctation of Aorta:
- Localised narrowing in any part of the aorta
- More common in males, females with Turner syndrome
- **Postductal or adult type**
  - Obstruction is just distal to ductus arteriosus which is closed
  - Characterized by HT in upper extremities, weak pulses and low BP in the lower extremities, effects of arterial insufficiency such as coldness and claudication
  - With time, development of collaterals b/w pre-stenotic and post-stenotic segment with enlargement of intercostal arteries → rib erosion
• **Preductal or Infantile type:**
  - narrowing proximal to ductus arteriosus which remains patent
  - lower half of body cyanosed while upper part of body receives blood from aorta
Aortic stenosis and atresia

- Most common anomaly of aorta is congenital bicuspid valve. Not much functional significance except predisposes it to calcification
- Congenital aortic atresia rare & incompatible with life
- Aortic stenosis- congenital or acquired(RHD)
- 3 types of congenital AS:
  1. **Valvular**: cusps thickened and malformed
  2. **Subvalvular**: thick fibrous ring under the aortic valve
  3. **Supravalvular**: uncommon
- May be assoc with hypoplastic heart synd: fatal in neonates
Pulmonary Stenosis and Atresia

• Stenosis
  - commonest form of obstructive CHD
  - occurs as component of TOF or isolated defect
  - fusion of cusps of pulmonary valve forming diaphragm like obstruction

• Atresia
  - no communication b/w rt ventricle & lungs
  - blood goes to left heart through interatrial septal defect and enters lungs via PDA