Congenital Heart Disease

1. Acyanotic lesions - VSD, ASD, PDA

- Left to right shunting, mixing of oxygenated blood with deoxygenated blood
- Increased pulmonary blood flow → risk of pulmonary hypertension and untreated acyanotic heart disease can lead to Eisenmenger syndrome
- Lesions that are above the level of the nipple usually give rise to ejection systolic murmurs while lesions below the level of the nipple typically cause pan systolic murmurs

Ventricular septal defect	
Symptoms	Depends on size of defect ● Small - may be asymptomatic, normal growth ● Moderate - poor feeding, FTT, SOB ● Large - poor feeding, FTT (falls below centiles), SOB, sweaty and pale with feeds
Epidemiology	Most common congenital heart lesion (15-20%) Associated with Down's syndrome (AVSD)
Time of presentation	Antenatal diagnosis at 16-18 weeks Presentation at 6-8 weeks Congestive heart failure typically presents after 4-6 weeks Persistent pulmonary hypertension of the newborn (PPHN) may become established by 6-12 months
Clinical findings	Palpate ● Check for presence of thrill ● Might be useful to palpate liver (enlarged in heart failure) **Auscultate** ● Loudness of murmur is inversely proportionate to the size of defect ● Pan-systolic murmur heard loudest at LLSB ● Typically grade 3-4 ● Loud P2 suggests presence of pulmonary hypertension
Investigations	● Pulse oximetry to determine level of oxygen saturation ● ECHO - visualise defect directly ● CXR - cardiomegaly and pulmonary odema (increased pulmonary vascular markings) if severe VSD (presence of heart failure), enlarged pulmonary artery ● ECG - ○ In patients with moderate or large VSD, the ECG may demonstrate LV hypertrophy (LVH) manifested as increased voltage in V5 and V6, or leads II, III, and aVF ○ In patients with elevated RV pressure, the ECG demonstrates RV hypertrophy (RVH), often manifested by tall R waves in

	leads V4R and V1, or upright T waves in these leads beyond the first 24 hours of life, in addition to LVH.
Management	● Small lesion < 5mm usually close spontaneously, no repair required (30-40%) ● Moderate lesion ○ Diuretic therapy (Furosemide and Spironolactone) ○ Feeding with high caloric feeds (Infantrini) ● Large lesion ○ Manage as per moderate lesion ○ Optimise weight gain for surgery ○ Schedule for surgery before 12 months to prevent PPHN

Atrial septal defect	
Symptoms	● Typically asymptomatic ● Some children will have recurrent chest infections
Epidemiology	Second most common acyanotic heart lesion (5-10%)
Time of presentation	Mean age of diagnosis is 4.5 years from incidental finding of murmur Symptomatic presentation is usually before age of 40 years with arrhythmias, dyspnoea
Clinical features	● May also have no auscultatory finding in infants (asymptomatic) **Auscultate** ● **Ejection systolic murmur** heard loudest at ULSB ● **Widely fixed splitting of second heart sound** (L→ R shunting increases RV filling, thus RV ejection time is increased and pulmonary valve closure is delayed for a significant amount of time after aortic valve closure)
Investigations	● Pulse oximetry ● ECHO - visualise defect directly, shows dilated RV and increased RV filling and ejection time ● CXR - usually no findings ● ECG - incomplete RBBB
Management	● Most children asymptomatic and rarely require CHF therapy ● Spontaneous closure in lesions smaller than 7-8mm ● Large defects require repair - percutaneous (catheter closure) or surgery using median sternotomy incision

Patent ductus arteriosus	
Symptoms	Dependent on size of lesion ● Small - asymptomatic ● Moderate - congestive heart failure with poor feeding, FTT ● Large - poor feeding, severe FTT, recurrent LRTIs ○ For preterm infants they experience failure to wean from ventilation
Epidemiology	5-10% of all congenital heart defects Very common in preterm infants
Time of presentation	Symptoms usually present 3-5 days after birth when the duct begins to close
Clinical features	Palpate ● Might be useful to palpate liver (enlarged in heart failure) ● Bounding pulses and wide pulse pressure Auscultate ● Continuous machinery murmur typically in ULSB, best heard below left clavicle Check for presence of thrill (ULSB)
Investigations	● 2D ECHO and Doppler ● CXR and ECG are less useful in diagnosing PDA
Management	● If preterm - good probability of spontaneous closure ● If term - less likely to close spontaneously ● Medical - indomethacin / ibuprofen (not effective in term infants) ● Surgical - catheter closure or PDA ligation (left lateral thoracotomy incision) when weight is at least 5kg

2. Outflow tract obstruction - coarctation of the aorta

Coarctation of aorta	
Pathophysiology	Obstruction to the LV outflow tract leads to increase in LV afterload which can cause LV hypertrophy ● Neonates with severe coarctation can develop heart failure
Epidemiology	Approximately 5% of all congenital heart defects Associated with Turner's syndrome (5-15% of girls with coarctation)
Time of presentation	Symptoms present 3-5 days after birth when the duct begins to close as the PDA and foramen ovale allows blood to bypass the outflow obstruction
Clinical features	Palpate ● Systolic blood pressure is high when measured with BP cuff ● Absent femoral pulses (do 4-limb BP measurement)

	● Cold extremities (especially feet) ● Hepatomegaly in heart failure due to severe coarctation Auscultate ● Murmur heard at back between scalpulae
Investigations	● 2D ECHO and Doppler - direct visualisation of defect ● CXR and ECG are less useful in diagnosis
Management	Medical therapy: ● Continuous intravenous infusion of prostaglandin E1 to keep the ductus arteriosus open ● Dopamine or Dobutamine to improve contractility in those with heart failure ● Supportive care to correct metabolic acidosis, hypoglycemia, respiratory failure, and anemia that may contribute to or be a consequence of heart failure. Surgical repair: ● Balloon angioplasty ● Resection with end-to-end angioplasty ● Bypass graft ● Subclavian flap

3. Cyanotic lesions - 6Ts

Occurs due to the mixing of deoxygenated blood with oxygenated blood (right → left shunt)

<u>Differential diagnoses of cyanotic lesions using the 6 'T's are:</u>

a. **T**OF
b. **T**GA
c. **T**runcus arteriosus
d. **T**otal anomalous pulmonary venous connection
e. **T**ricuspid valve abnormalities
f. **T**on of others - hypoplastic left heart, double outlet right ventricle, pulmonary atresia

Tetralogy of Fallot 1. Ventricular septal defect 2. Overriding aorta 3. Pulmonary stenosis 4. Right ventricular hypertrophy	
Symptoms	● Cyanosis ● Poor feeding, sweating during feeds
Epidemiology	7-10% of congenital heart disease
Time of presentation	During neonatal period when the patent ductus arteriosus begins to close (Day 3-5)
Main clinical findings	● Cyanotic “tet” spells due to increased RV to LV shunt due to pulmonary stenosis causing RV outflow tract obstruction ● Murmur may be present due to right ventricular outflow tract obstruction (RVOTO) caused by pulmonary stenosis and not VSD ○ The murmur is crescendo-decrescendo with a harsh ejection systolic quality, heard loudest over the ULSE with posterior radiation
Investigations	● 2D ECHO and Doppler - to assess location and number of VSDs and severity of RVOTO ● ECG - shows right atrial enlargement and right ventricular hypertrophy (right axis deviation, prominent R waves anteriorly and S waves posteriorly) ● CXR - classic “boot shaped heart”, with a right aortic arch seen in 25% of patients ● Cardiac catheterisation can help further delineate cardiac lesion, particularly helpful for assessing levels of right ventricular outflow obstruction.
Management	● Neonates with severe cyanosis - prostaglandin infusion to maintain patency of ductus and pulmonary flow until time of surgical repair ● Medical ○ Tet spells ■ Knee to chest position to increase systemic vascular resistance and promote blood flow into the pulmonary circulation ■ Oxygen ■ Morphine ■ B-blockers ○ Heart failure - digoxin and loop diuretic (Furosemide) ○ Prophylaxis for endocarditis - antibiotics ● Surgical repair - BT shunt

Transposition of Great Arteries	
Pathophysiology	Aorta arises from the RV and pulmonary artery from the LV, resulting in deoxygenated blood from the RV being circulated around the body
Symptoms	● Cyanosis ● Poor feeding, sweating during feeds
Epidemiology	TGA accounts for about 3% of all congenital heart disease, and 20% of all cyanotic heart disease Without treatment, 90% will die within the first year of life
Time of presentation	During neonatal period when the patent ductus arteriosus begins to close (Day 3-5)
Main clinical findings	● Cyanosis ● Tachypnea ● Murmur
Investigations	● Fetal ultrasound ● ECHO ● CXR - classic “egg on a string” appearance ● ECG and cardiac catheterisation is typically not used in the diagnosis of TGA
Management	● Balloon atrial septostomy to increase mixing of the two circulatory systems ● Arterial switch procedure

4. Innocent murmurs

a. Types - Still’s murmur, venous hum, turbulent flow in the pulmonary artery bifurcation
b. 25% of full term neonates have a murmur
c. Features of an innocent murmur (10 ‘S’)

10 ‘S’ of innocent murmur: soft, systolic, short, S1 and S2 normal, symptomless, special test (x-ray and ECG) normal, standing / sitting vary with position, sternal depression

Still’s murmur	A soft vibratory murmur heard in the LLSB most frequently in childhood when there is normal blood flow and no cardiac lesion
Venous hum	Continuous murmur heard loudest over the clavicles due to venous return from the head and neck, and this varies with position
Turbulent flow in pulmonary artery bifurcation	A soft ejection systolic murmur caused by turbulent flow in the pulmonary artery (PA) bifurcation as the PA bifurcation and branches are small