Congenital Heart Disease

Incidence:

- May continue to have arrhythmias or ventricular dysfunction in 85% of cases.

- Trisomy 21 (Down syndrome) is most common.
- Deletion of 22q11.2 (50% of patients with DiGeorge syndrome).
- Transcription factors work together, so mutation in one of many.

- Day 50, septation produces 4-chambered heart.
- Day 28, heart chambers begin to form, neural crest cells migrate into the outflow tract where they participate in septation.
- Day 20, develops into beating tube that loops to right.
- Day 28, heart chambers begin to form, neural crest cells migrate into the outflow track where they participate in septation.
- Transcription factors work together, so mutation in one of many.

Pathology 12: Incomplete - Congenital Heart Disease-Obstructive Congenital Anomalies

- Total Anomalous Pulmonary Venous Connection (TAPVC): pulmonary veins fail to join the left atrium; patent foramen ovale or ASD always present -> allows pulmonary venous blood to enter systemic circulation (paradoxical embolism) possibly leading to death.
- Persistent Truncus Arteriosus: produces ventriculoarterial discordance (aorta arises from right ventricle); if severe, greater resistance to right ventricular outflow than left; right-to-left shunting develops, produces cyanosis and death; closure of isolated PDA should occur as early as feasible; if infant has congenital heart disease (6-10%) have VSD.
- Ventricular Septal Defect (VSD): incomplete closure of ventricular septum; most associated with other congenital cardiac anomalies in 20-40%.
- Right-to-left shunting (ASD or patent foramen ovale) and VSD; cyanosis present from birth; high mortality in first weeks/months of life.
- Tetralogy of Fallot: 4 features: VSD, obstruction of right ventricular outflow -> right-to-left shunting develops, results in shunt reversal, cyanosis, and death; closure of isolated PDA should occur as early as feasible; if infant has congenital heart disease.
- Transposition of the Great Arteries (TGA): produces ventriculoarterial discordance (aorta arises from right ventricle); left ventricle becomes atrophic because it only supports low-pressure pulmonary circulation -> incompatible with postnatal life unless a shunt exists; outlook depends on clinical consequences depending on severity.

Obstructive Congenital Heart Disease:

- Causes include obstruction to flow from left to right and vice versa, anatomic, or valvular defects; produces cyanosis, often in infancy; may not effect growth and development; usually asymptomatic until ~age 30; surgical repair possible.
- Patent (PERSISTENT) Ductus Arteriosus (PDA): occur as isolated anomaly, commonly associated with other anomalies; large V/Ss -> increase in pulmonary blood flow; increase in size and obesity.
- Right-to-left shunting (ASD or patent foramen ovale) and VSD; cyanosis present from birth; high mortality in first weeks/months of life.
- Tetralogy of Fallot: 4 features: VSD, obstruction of right ventricular outflow -> right-to-left shunting develops, results in shunt reversal, cyanosis, and death; closure of isolated PDA should occur as early as feasible; if infant has congenital heart disease.
- Transposition of the Great Arteries (TGA): produces ventriculoarterial discordance (aorta arises from right ventricle); left ventricle becomes atrophic because it only supports low-pressure pulmonary circulation -> incompatible with postnatal life unless a shunt exists; outlook depends on clinical consequences depending on severity.

Congenital heart defects are a general denominator that cause loss of function of 1 of 1 factor.