Cyanotic congenital heart disease

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- Tetralogy of Fallot
- D-Transposition of great arteries (D-TGA)
- Single ventricle
- Truncus arteriosus
- Total anomalous pulmonary venous connections
- Ebstein’s anomaly
- Eisenmenger’s syndrome

Tetralogy of Fallot

= most common cyanotic congenital heart disease

- **Characterized by: pulmonary stenosis (PS), ventricular septal defect (VSD), overriding aorta, right ventricular hypertrophy**
- **Decreased pulmonary blood flow (due to PS) and right-to-left shunting (due to VSD) → cyanosis, clubbing, polycythemia**
- **Pansystolic murmur at left upper and middle sternal border**
- **Symptoms:**
- **Exertional dyspnea, cyanosis, syncope, relieved by:**

  Squatting position (decrease right-to-left shunting by increasing systemic vascular resistance and pooling of desaturated blood to lower extremities, hence decreasing desaturated blood returning to heart)

- Cyanotic spells → severe hypoxemia, hypercapnia, acidosis → unconscious, seizure, hemiparesis
- Death due to: severe hypoxemia, hypercyanotic spells, paradoxical embolism, stroke, brain abscess, infective endocarditis, sudden death
- Chest X-ray = boot-shaped heart (“couer on sabot”) due to right ventricular hypertrophy and diminished main pulmonary artery shadow.
- Treatment: without surgical intervention, 50% die before school age, another 25% die before adolescence, and <10% reach adulthood.
- Palliative surgery: Blalock-Taussig shunt (graft shunting from subclavian artery to pulmonary artery)
- Surgical repair

D-Transposition of great arteries

= great arteries arise from opposite ventricles. Thus, systemic venous return to right atrium → right ventricle → aorta. Pulmonary venous flow from pulmonary veins → left atrium → left ventricle → pulmonary artery

- Predominantly male, large birth weight. Without surgical repair, infant die of severe hypoxemia and cyanosis and never reach adulthood.
- Treatment: surgical repair

1. Atrial rerouting eg. Mustard procedure (patch used as intra-atrial baffle to reroute systemic venous return from superior and inferior vena cava to mitral valve and left ventricle, and pulmonary artery)
2. Arterial switch (Jatene operation): aorta and pulmonary artery are transected and reanastomosed to proper roots.

Single ventricle

= cardiac defect resulting in single functional ventricular chamber

- ***Classification:***

1. **Double-inlet left ventricle with small-outlet right ventricle, usually with transposed aorta**
2. **Double-inlet right ventricle with small left ventricle, usually with mitral obstruction**
3. **Indeterminate ventricle, usually right ventricle, without rudimentary chamber**
4. **Tricuspid atresia = agenesis of tricuspid valve. Blood enters right atrium mixes with pulmonary venous blood, and enters left ventricle, usually associated with ventricular septal defect (VSD), patent ductus arteriosus (PDA).**

5. **Mitral atresia and other forms of hypoplastic left heart syndrome. Pulmonary venous return reaches rigth atrium and ventricle via foramen ovale or atrial septal defect (ASD). Cardiac output provided by right ventricle through ductus arteriosus to aorta.**

- ***Symptoms:***
- **Left-to-right shunting → increase pulmonary blood flow and volume overload of the single ventricle → heart failure and death**
- **Pulmonary vascular obstructive disease → Eisenmenger's syndrome**
- **Atrial ventriular (AV) valve regurgitation**

- ***Treatment:*** **surgical repair (rare to encounter an unoperated adult patient)**

Truncus arteriosus

- Rare, always associated with VSD
- Due to failure of conus arteriosus development → pulmonary arteries emerge from ascending aorta as a common trunk
- Symptoms: tachypnea, diaphoresis, poor feeding, growth failure, cyanosis is mild or absent.
- Complete mixing of systemic and pulmonary venous blood in ascending aorta and unrestricted pulmonary blood flow → left-to-right shunting → heart failure
- Pulmonary vascular obstructive disease → Eisenmenger's syndrome
- Without surgery, majority die with heart failure in first few months of life. Rare to encounter an unoperated adult patient.
- Treatment = surgical repair and closure of VSD

Total anomalous pulmonary venous connections

- Rare
- All pulmonary veins connect anomalously to right atrium, either directly or via coronary sinus, superior or inferior vena cava
- Usually patent foramen ovale or ASD allow mixed blood in right atrium to enter left heart for systemic circulation
- Symptoms: pulmonary venous obstruction → pulmonary hypertension → cyanosis, dyspnea, heart failure
- Treatment = surgical repair (without surgery, 80% die during infancy)

- ***Cor triatriatum***

  = failure of common pulmonary vein resorption → left atrium divided by abnormal fibromuscular diaphragm into posterosuperior chamber (receiving pulmonary veins) and anteroinferior chamber (→ mitral orifice)

  → increase pulmonary venous pressure and pulmonary vascular resistance

  → pulmonary hypertension

Treatment: surgery

Ebstein's Anomaly

- **Rare**

= Downward displacement of tricuspid valve into right ventricle → proximal "atrialized" (functioning as atrial chamber) and distal functional small right ventricle →1) Tricuspid regurgitation.2) Right-to-left intra-atrial shunt (via large foramen ovale).

- **Symptoms: clinical course range from intrauterine death to asymptomatic survival to late adulthood (80-90's)**
- **Widely split second heart sound, pansystolic murmur at left sternal border (due to tricuspid regurgitation)**

- Fatigue, cyanosis, arrhythmias (= WPW syndrome or supraventricular arrhythmias due to dilated right atrium), right-to-left shunting → hypoxemia → heart failure
- Paradoxical embolism, brain abscess, infective endocarditis, sudden death

- ***Treatment:***
- If anomaly mild → asymptomatic throughout life
- If symptomatic, surgical repair (tricuspid valve reconstruction with plication of atrialized portion of right ventricle)

Eisenmenger's syndrome

= VSD + pulmonary vascular obstructive disease → right-to-left shunting → cyanosis

- Pulmonary vascular obstructive disease (→ right-to-left shunting → cyanosis, clubbing, polycythemia) secondary to the preexisting left-to-right shunts such as VSD, AV canal, PDA, truncus arteriosus, single ventricle anomalies → pulmonary hypertension → irreversible vascular changes in pulmonary vessels.

- ***Symptoms:*** fatigue, dyspnea, cyanosis, digital clubbing, heart failure, syncope, chest pain, hemoptysis (due to bleeding bronchial vessels or pulmonary infarction), arrhythmias, sudden death
- ***Treatment:***
- If stable, patient with Eisenmenger's syndrome may live for many decades but survival to late adulthood uncommon. Avoid heavy activities and high altitude.
- If symptomatic, medical therapy = monitor hemoglobin and hematocrit, pulmonary vasodilatory agents eg. Prostacyclin, calcium blocker, ACEI.
- Surgery = heart-lung or lung transplantation (high operative risk and limited long-term survival)