## APPROACH TO THE DIAGNOSIS OF CYANOTIC CONGENITAL HEART DISEASE

**Cyanosis** is a blue coloring of the skin and mucous membranes caused by an increase concentration of reduced hemoglobin. This occurs at a critical level of about 5 g/ dL of reduced hemoglobin.

**Central cyanosis** is seen in the tongue and lips and is due to desaturation of central arterial blood**.** Patients who are centrally cyanosed will also be peripherally cyanosed. **Features of central cyanosis therefore include dyspnea and tachypnea, secondary polycythaemia, and bluish or purple discoloration of the oral mucous membranes, fingers and toes.**

**Peripheral cyanosis (acrocyanosis) is** seen in the hands and feet, which are cold. There is peripheral vasoconstriction and stasis of blood in the extremities, leading to increased peripheral oxygen extraction. Examples include: congestive heart failure, circulatory shock, exposure to cold temperatures and abnormalities of the peripheral circulation. **Features of peripheral cyanosis therefore include cold fingers and toes, bluish or purple discoloration of fingers and toes, and a normal pink-coloured tongue.**

**CYANOSIS: DIAGNOSTIC DIFFICULTY**

A. FALSE NEGATIVE
   1. Mild Cyanosis
   2. Skin Pigmentation
   3. Anaemia

B. FALSE POSITIVE
   1. Peripheral cyanosis
   2. Cardiac failure
   3. Lung infection

### DIFFERENTIAL DIAGNOSIS

| A Airway | B Breathing | C Circulation |
|---|---|---|
| Choanal atresia | Pneumonia | |
| Micrognathia | Congenital diaphragmatic hernia | Polycythemia |
| Pierre Robin sequence | Congenital cystic adenomatoid malformation | Anemia |
| Laryngomalacia | Pulmonary sequestration | Methemoglobinemia |
| Vocal cord paralysis | Congenital lobar emphysema | Congenital heart disease |
| Tracheal stenosis | Pulmonary hypoplasia | *Decreased pulmonary blood flow* |
| Vascular slings/rings | Phrenic nerve palsy | Tricuspid atresia |
| Cystic hygroma | Hypoventilation | Pulmonary atresia |
| Hemangioma | | Pulmonary stenosis |
| Other neck masses | | Tetralogy of Fallot |
| Foreign body aspiration | | Ebstein's anomaly |

Cyanosis is caused by

- Inadequate alveolar ventilation
    - Airway obstruction
    - Structural changes in the lungs
    - Central nervous system hypoventilation
    - Weakness of the respiratory muscles
- **Desaturated blood bypassing the lungs**
    - **Cyanotic congenital heart disease**
    - Pulmonary arterio-venous fistulas
- Increased capillary deoxygenation
    - Acrocyanosis of the newborn
    - Congestive heart failure
    - Shock
- Abnormal hemoglobin
    - Carbon monoxide poisoning
    - Methemoglobinemia

What to ask for in the Medical History:

Onset of cyanosis (congenital vs acquired cause)

Other sx: Tachypnea, dyspnea, stridor

What to look for in the Physical Examination:

Clubbing

circumoral vs peripheral cyanosis

O2 sat < 85% --clinically apparent

Tachypnea, dyspnea, stridor, wheezing, murmur

**Hyperoxic test (for neonates):** give 100% oxygen for 15-20 minutes

If pO2 > 150-200 mmHg → pulmonary cause

If pO2 < 100-150 mmHg → cardiac cause

Differential Diagnosis based on timing of presentation

Presents Almost Immediately at Birth

Transposition of the great vessels (D-transposition)

Tricuspid atresia

Presents in the Perinatal Period

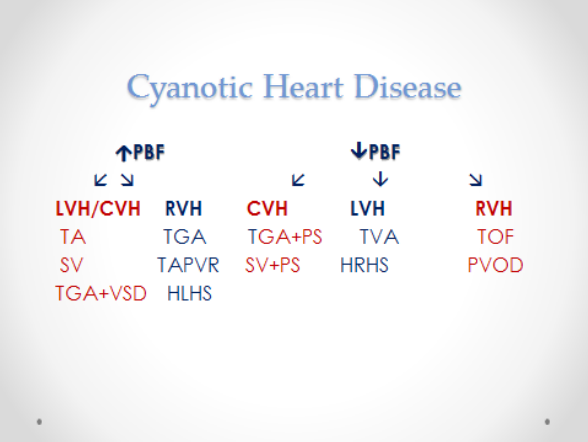
Truncus arteriosus -

Total anomalous pulmonary venous return

Tetralogy of Fallot (also known as a “TET” or “blue TET”)

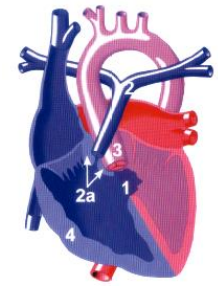
Presents after the Perinatal Period

Tetralogy of Fallot (also known as a “pink Tetralogy” or “pink TET”)



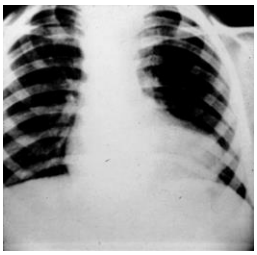

**SPECIFIC CYANOTIC CONGENITAL HEART DISEASES**

**TETRALOGY OF FALLOT**




Above:

1. Ventricular septal defect (VSD)
2. Stenotic pulmonary valve and artery
2a. Infundibular stenosis
3. Enlarged aorta overriding VSD
4. Right ventricular hypertrophy

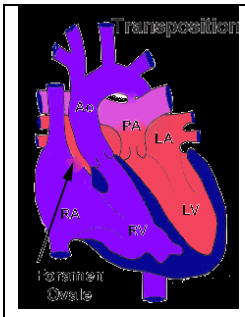


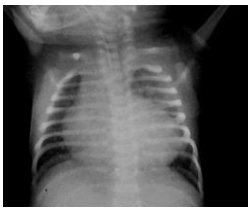
10% of CHD
Components: RVOT obstruction, VSD, overriding aorta, RVH
Manifestations: Cyanosis, hypoxic spell,Easy fatigability
Physical Examination:Systolic ejection murmur,Single $S_2$; soft $P_2$
Cyanotic oral mucosa &nailbeds, clubbing of digits
ECG - RAD; RVH
CXR- Boot shaped heart, RV cardiomegaly, Small MPA; hypovascular markings

Natural History-Cyanosis, hypoxic spell, squatting, Polycythemia, CVA, brain abscess, Infective endocarditis, RV failure

Management
- Medical
  - Prevent Fe deficiency
  - Phlebotomy
  - Propranolol
- Surgical/ Interventional
  - Palliative – shunts (BTS); PDA stenting; RVOT stenting
  - Definitive – VSD closure; infundibulectomy; transannular patching
- Management of Hypoxic spells
  - Sedate
  - Hydrate
  - Oxygenate
  - Bicarbonate
  - Beta-Blocker

**D-TRANSPOSITION OF THE GREAT ARTERIES**



5% of CHD

Pathophysiology

- Atrioventricular concordance and Ventriculo-arerial discordance
- Parallel circulation

Associated lesions

- ASD, VSD or PDA
- Shunts necessary for survival

Manifestations

- Cyanosis from birth
- CHF if without pulmonary stenosis

Natural History:

- Early and progressive cyanosis and acidosis
- Death if no shunt or intervention
- Survive if (+) VSD or PS
- Polycythemia
- CHF: Pulmonary congestion and RV failure
- Infective endocarditis

Physical examination- Cyanotic, Signs of CHF,Single loud $S_2$ soft systolic murmur

ECG- RVH; CVH if (+) VSD, PDA, or PS

CXR- Egg shaped, RV cardiomegaly, narrow pedicle, hypervascular markings if without PS, hypovascular markings if with PS

Management:

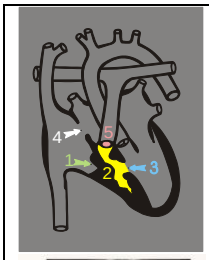
Palliative

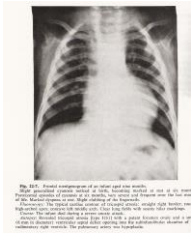
- Prostaglandin infusion
- Create interatrial communication:
  - Balloon atrial septostomy (BAS) – Rashkind procedure
  - Blade septostomy
  - Atrial septostomy – Blalock-Hanlon procedure

Definitive:

- Atrial switch – Senning or Mustard procedure
- Arterial switch – Jatene procedure

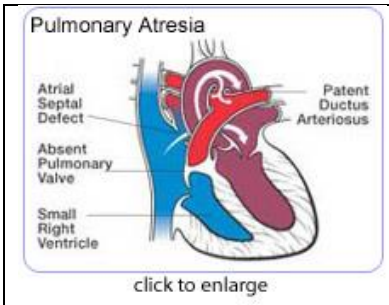
**TRICUSPID VALVE ATRESIA**



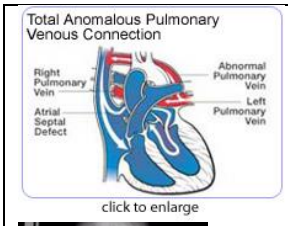
Components: Atretic Tricuspid Valve, Hypoplastic Right Ventricle, Ventricular Septal Defect, Atrial Septal Defect, Pulmonary Stenosis
Natural History: progressive cyanosis at birth
ECG-Left axis deviation (QRS axis negative in AVF) & RAE
CXR-"bookend" (flat right heart border)
Management:
Palliative
Prostaglandin infusion
Create interatrial communication:
Balloon atrial septostomy (BAS) – Rashkind procedure
Blade septostomy
Atrial septostomy – Blalock-Hanlon procedure
Definitive: Glenn Shunt and Fontan Procedure-routing of systemic venous return directly into pulmonary circulation bypassing the subpulmonary ventricle

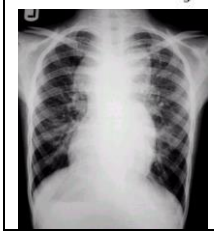
**Pulmonary Valve Atresia with Intact ventricular septum (PVA-IVS)**




Commonly symptomatic in the newborn period
Usually with TR murmur (systolic murmur at the left lower sternal border)
Management:
Palliative
Prostaglandin infusion to keep PDA open
Blalock-Taussig shunting
Definitive:
RF ablation of atretic valve
Glenn Shunt and Fontan Procedure if RV is too small

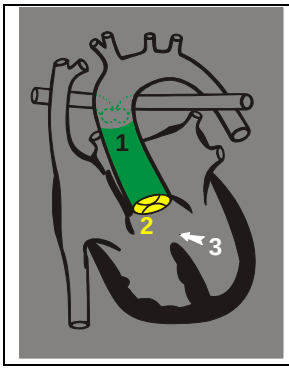
**TOTAL ANOMALOUS PULMONARY VENOUS RETURN (TAPVR)**



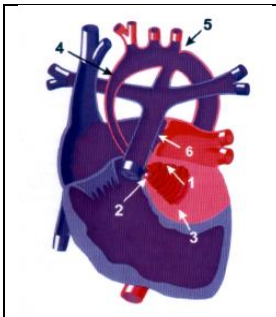



Pulmonary veins drain into the right ventricle
Types: supracardiac via a vertical vein and SVC
Infracardiac via the IVC
Intracardiac via the coronary sinus
Mixed
Physical Exam- Split S2, Systolic ejection murmur at the left upper sternal border
CXR- Snowman configuration – supracardiac type
Pulmonary edema and small heart– obstructed
Management: surgical repair to direct the pulmonary venous return into the left ventricle

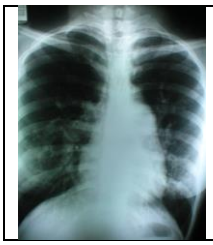
**TRUNCUS ARTERIOUSUS**

| | |
|---|---|
|  | Components:<br>Pulmonary arteries arise from aorta.<br>Truncal valve, occasionally quadracuspid, stenotic and or insufficient.<br>Overrides the ventricular septal defect.<br>Ventricular septal defect<br>Physical Exam- Loud S2, Diastolic murmur at the left upper sternal border (truncal valve regurgitation)<br>Management:<br>Surgical- VSD closure, separation of PA from aorta, creation of RV to PA conduit |

**HYPOPLASTIC LEFT HEART SYNDROME**

| | |
|---|---|
|  | Components:<br>Atretic or hypoplastic mitral valve<br>Atretic or hypoplastic aortic valve<br>Hypoplastic left ventricle<br>Hypoplastic aortic arch<br>Coarctation of aorta<br>Atrial septal defect<br>Patent Ductus Arteriosus<br>Management:<br>Palliative – Prostaglandin infusion<br>Definitive- Norwood Procedure |

**PULMONARY VASCULAR OBSTRUCTIVE DISEASE (PVOD)**

| | |
|---|---|
|  | Sequela of unoperated left-to-right shunts<br>Shunting has shifted to: Right-to-left<br>ECG- RVH<br>CXR- pruning<br>Mangement:<br>Supportive- medication to decrease PA hypertension; possibly oxygen |

**TAKE HOME MESSAGE**

- **Identify cyanosis**
- **Determine the cause of cyanosis**
- **If CCHD is suspected, use CXR & ECG to narrow down your differential**
- **In the hospital setting:**
  - **Do hyperoxia test for neonates**
  - **Use of prostaglandin for ductal dependent lesions to tide over until surgery**
  - **Management of hypoxic spells**
- **Refer as needed**