CYANOTIC CONGENITAL HEART DISEASES
INTRODUCTION

• Cyanosis is a bluish or purplish tinge to the skin and mucous membranes

• Approximately 5 g/dL of unoxygenated hemoglobin in the capillaries generates the dark blue color appreciated clinically as cyanosis

• Cyanosis is recognized at a higher level of oxygen saturation in patients with polycythemia and at a lower level of oxygen saturation in patients with anemia
CYANOSIS - TYPES

– Central – cyanotic CHD
– Peripheral – hypothermia, CCF
– Mixed Cyanosis – CHD in Shock
– Differential cyanosis – PDA with reversal
– Reverse differential cyanosis – TGA with PDA with reversal
– Intermittent Cyanosis – Ebsteins anomaly
– Circumoral cyanosis
– Cyclical cyanosis – Bilateral choanal atresia
HOW TO DIFFERENTIATE?

**True Cyanosis**
- Associated with clubbing
- ABG - confirms

**Cyanosis like conditions**
- Not associated with clubbing
- Lab estimation of Meth Hb and Sulph Hb Confirms
DIFFERENTIAL DIAGNOSIS FOR CYANOSIS

– Methemoglobin
– Sulfhemoglobin
– Pseudocyanosis: is a bluish tinge to the skin and/or mucous membranes that is not associated with either hypoxemia or peripheral vasoconstriction. Most causes are related to metals (e.g., silver nitrate, silver iodide, silver, lead) or drugs (e.g., phenothiazines, amiodarone, chloroquine hydrochloride).
CYANOTIC CHD: CLINICAL DIAGNOSTIC APPROACH

Cyanotic CHD

- Pulmonary blood flow
- Pulmonary blood flow
- Normal Pulmonary flow
Cyanotic Congenital Heart Disease

Cyanosis, Clubbing, Polycythemia

Increased Pulmonary Blood Flow

- Transposition of Great arteries (3-5%)
- Truncus Arteriosus (1-2%)
- Single Ventricle (1-2%)
- TAPVC (1-2%)
- HLHS (1-3%)

Decreased Pulmonary Blood Flow

- Tetralogy of Fallot (5-7%)
- Tricuspid Atersia
- Ebstein’s Anomaly
- Pulmonary Atresia
TETRALOGY OF FALLOT
INTRODUCTION

• In 1888, Fallot described the anatomy of TOF
• Incidence 10 % of all forms of congenital heart disease
• The most common cardiac malformation responsible for cyanosis after 1 year of age.
PATHOLOGY

• The four components of TOF are
  – Ventricular septal defect
  – Obstruction to right ventricular outflow
  – Overriding of the aorta
  – Right ventricular hypertrophy
• Only two abnormalities are required  
  – A VSD large enough to equalize pressures in both ventricles  
  – A right ventricular outflow tract obstruction  
• RVH is secondary to right ventricular outflow tract obstruction (RVOT) and VSD  
• Over riding of aorta varies  
• VSD is perimembranous defect with extension into the subpulmonary region  
• VSD is non restrictive and large
HEMODYNAMICS
HISTORY

• Appearance of cyanosis after neonatal period

• Hypoxemic Spells

• Low birth weight or development delay or easy fatigability
GENERAL EXAMINATION

- Cyanosis
- Clubbing
- Polycythemia
- Tachypnea
SYSTEMIC EXAMINATION

• RV tap in left sternal border
• Systolic thrill in upper and mid left sternal borders
• Ejection click which originates from aorta
• S2 is single due to absent pulmonary component
• A loud ejection type systolic murmur heard at the mid and upper left sternal border
• This murmur originates from the **Pulmonary stenosis** and may be confused with the holosystolic murmur of **VSD**
• Intensity of the murmur depends on the severity of pulmonary stenosis or RVOT obstruction
• More severe the obstruction, shorter and softer murmur will be heard
• In Pulmonary atresia, murmur is either absent or very soft
• Auscultation of back is important to find the presence of MAPCAs (Major Aorto Pulmonary Collateral Arteries)
INVESTIGATIONS

• Hematology
  – Polycythemia secondary to cyanosis (hematocrit >65%)
  – Anemia – due to relative iron deficiency
• Electrocardiography
• X-ray
• Echocardiography
• Angiogram
• Normal size heart
• Pulmonary vascular markings are decreased
• Concave main pulmonary artery segment with an upturned apex – *BOOT shaped heart* or *coeur en sabot*
• Right atrial enlargement (25%)
• Right aortic arch (25%)
Right axis deviation, Right ventricular hypertrophy
ECHOCARDIOGRAPHY
DIFFERENTIAL DIAGNOSIS OF FALLOT’S PHYSIOLOGY

- Fallot’s Tetralogy
- Transposition of great arteries
- Tricuspid atresia
- Single ventricle
- Double outlet right ventricle
- Corrected transposition of great arteries
- Atrioventricular canal defect
- Malposition's
COMPLICATIONS OF CYANOSIS / CYANOTIC CHD

- Clubbing
- Cyanotic Spell
- Depressed IQ
- Infective endocarditis
- Polycythemia
- Embolic phenomenon
HYPOXEMIC SPELL

• Hypercyanotic or Tet or cyanotic or hypoxic spell
• Mechanism - Secondary to infundibular spasm and/or decreased SVR with increased right-to-left shunting at the VSD, resulting in diminished pulmonary blood flow
• Peak incidence 2 - 4 months
• Usually occurs in morning after crying, feeding or defecation
• Severe spell may lead to limpness, convulsion, cerebrovascular accident or even death
HEMODYNAMICS OF SPELL

• Increased activity
• Increased respiration
• Increased venous return
• Fixed pulmonary blood flow
• Increased (RV) to (LV) shunt
• Increased cyanosis
HYPOXEMIC SPELL - SYMPTOMS

• Sudden onset of cyanosis or deepening of cyanosis

• Sudden onset of dyspnea

• Alterations in consciousness, encompassing a spectrum from irritability to syncope

• Decrease in intensity or even disappearance of systolic murmur
HYPOXEMIC SPELL – TREATMENT

• Knee chest position or squatting – decreases systemic venous return and increases systemic vascular resistance at femoral arteries
• Morphine sulphate – 0.2mg/kg subcutaneously or intramuscularly, suppresses the respiratory centre and abolishes hyperpnoea
• Oxygen has little effect of arterial oxygen saturation
• Acidosis should be treated with sodium bicarbonate 1mEq/kg administered intravenously
HEMODYNAMICS OF SQUATTING

• Decreased venous return
• Increased systemic vascular resistance
• Increased pulmonary blood flow
• Decreased cyanosis

• Squatting Equivalent – Knee Chest position, child sitting with flexed limbs, mother carrying the child with folded limbs.
HYPOXEMIC SPELL – FOLLOW UP

• Following treatment, patient becomes less cyanotic, and heart murmur become louder
• Indicates increased amount of blood flowing through stenotic right ventricular outflow tract
• If Hypoxemic spell not fully respond
  – Vasoconstrictor: Phenylephrine 0.02 mg/kg IV
  – Propranolol 0.01 to 0.25 mg/kg slow IV push, reduces the heart rate and may reverse the spell
  – Ketamine 1 – 3 mg/kg over 60 secs, increases systemic vascular resistance and sedates the patient
NEUROLOGICAL COMPLICATIONS OF CHD: TIP OF THE PROVERBIAL ICEBERG!

Stroke
Brain abscess
Seizures

Adverse neuro-developmental outcome:
• Lower IQ
• Poor motor skills
• Poor language skills
• Cognitive impairment
NEUROLOGICAL COMPLICATIONS
MANAGEMENT PRINCIPLES

MEDICAL  ↔  SURGICAL
TREATMENT OF TOF – MEDICAL

- Prevention of Hypoxemic spell
  - Oral Propranolol therapy 0.5 to 1.5 mg/kg every 6 hours – to prevent Hypoxemic spell
- Relative iron deficiency anemia should be detected and treated since anemic children are more susceptible to cerebrovascular complications
- Maintenance of good dental hygiene and infective endocarditis prophylaxis
- Hematocrit has to maintained <65%, Phlebotomy may be needed to manage polycythemia
INDICATIONS FOR SHUNT PROCEDURES

• Neonates with TOF and pulmonary atresia
• Infants with hypoplastic pulmonary annulus, which requires a transannular patch for complete repair
• Children with hypoplastic pulmonary arteries
• Severely cyanotic infants younger than 3 months of age
• Infants younger than 3 to 4 months old who have medically unmanageable hypoxic spells
SHUNT PROCEDURES

Systemic – Pulmonary Shunt

• Blalock-Taussig:
  – anastomosed between the subclavian artery and ipsilateral PA, preformed in infants older than 3 months

• Gore-Tex Interposition shunt:
  – Placed between the subclavian and ipsilateral PA, done even in small infants younger than 3 months

• Waterston:
  – anastomosed between ascending aorta right PA, no longer performed

• Potts:
  – anastomosed between descending aorta and left PA, no longer performed
TRICUSPID ATRESIA

- Marked Cyanosis present from birth
- ECG with left axis deviation, right atrial enlargement and LVH
EBSTEIN’S ANOMALY
EBSTEIN’S ANOMALY – CONT.

- Displacement of abnormal tricuspid valve into right ventricle
- Anterior cusp retains some attachment to the valve ring
- Other leaflets are adherent to the valve of the right ventricle
- Intermittent Cyanosis
- Multiple Clicks
- Right atrium is huge - Arterialization of Right Ventricle
- Tricuspid valve is regurgitant
EBSTEIN’S ANOMALY – CONT.
PULMONARY ATRESIA

- Cyanosis at birth
- X-ray Chest show a concave pulmonary artery segment and apex tilted upward
TRANSPOSITION OF GREAT ARTERIES
TGA: TRANSPOSITION PHYSIOLOGY

- Oxygenated blood circulates within the pulmonary circulation and de-oxygenated blood in systemic circulation.
- Hypoxia is the result of impaired mixing.
- Better admixture – better oxygen saturation
- Early presentation
TRUNCUS ARTERIOSUS

- Early CHF
- Mild or No Cyanosis
- Systolic ejection click
HYPOPLASTIC LEFT HEART SYNDROME

Normal
- to body only
- to lungs only

Hypoplastic left heart syndrome
- Patent (open) ductus arteriosus
- Small (hypoplastic) aorta
- Atrial septal defect
- Absent (atretic) mitral valve
PULMONARY AV FISTULA

- Fistulous vascular communications in the lungs may be large and localized or multiple, scattered and small
- The most common form of this unusual condition is “Osler – Weber – Rendu Syndrome”
- Clinical features depend on the magnitude of shunt
- Mild cyanosis will be present
- Routine echo will be normal but “Contrast” echo will be diagnostic
CYANOTIC CHD: APPROACH

1. CLINICAL SUSPICION
2. ASSIGN PHYSIOLOGY
3. ASSESS SEVERITY
4. PRECISE DIAGNOSIS
BEDSIDE TOOLS

• Clinical evaluation
• Chest x-ray
• ECG
• Measurement of oxygen saturation
• The hyperoxia test
STEP 1: DETECTION OF CYANOSIS

Clinical recognition of cyanosis has its pitfalls

- Lighting
- Anemia
- Pigmentation
- Peripheral cyanosis
- Mild cyanosis
IF IN DOUBT .......

• Misleading if used incorrectly
• Watch over a period of 1-2 min
• Stable waveforms
• Heart rate display correlating with actual HR
• Protect probe from light
THE HYPEROXIA TEST

- 100% O₂ via hood
- ~10 min..
- Take ABG – pO₂

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STEP 2: ASSIGN PHYSIOLOGY

CLINICAL ASSESSMENT OF PULMONARY BLOOD FLOW

REDUCED

- NO FTT
- 1. MORE CYANOSIS
- 2. CYANOTIC SPELLS
- 3. QUIET PRECORDIUM
- 4. NO HEPATOMEGALY

INCREASED

- 1. CHF +
- 2. FTT +
- 3. MILD CYANOSIS
- 4. NO CYANOTIC SPELLS
- 5. HYPERACTIVE PRECORDIUM
- 6. HEPATOMEGALY
CARDIAC EXAMINATION: CLUES BASED ON S2 SPLIT

Pure PS may have a wide split S2 with softly audible P2

- **single**
  - TOF physiology
  - TGA
  - Most admixture lesions

- **fixed**
  - TAPVC

- **normal**
  - Excludes Cardiac cause

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**TYPE AND LOCATION OF MURMUR**

- Ejection SM in pulmonary area – Most cases.
- PSM in LLSB – Tricuspid Atresia (VSD)
- Continuous murmurs: Pulmonary atresia.
- To and Fro Murmur: TOF-APV.
CXR IN CLASSIFYING PHYSIOLOGY
STEP 3: ASSESSMENT OF SEVERITY

• Early onset of cyanosis (especially in neonatal period)

• Cyanotic Spells

• Cyanosis with CHF

• Severe Cyanosis with no/very soft murmurs
STEP 4: CONFIRMING DIAGNOSIS

- Echocardiography allows complete diagnosis in majority of cases.
- Cardiac Catheterization required in very selected situations.
- Advances in CT/MRI obviate need for cath further
REFERRAL TO A SPECIALIST

• Refer as soon as you make a diagnosis of cyanotic heart disease.

• Neonates are likely to need immediate intervention

• Older children and those with stable CHD for diagnostic confirmation and planning further management
TIMING OF INTERVENTION: NEWER TRENDS

• Early correction of congenital heart disease is desirable because it avoids a number of adverse cardiac, neurodevelopment and other consequences.

• Early correction of a variety of congenital heart lesions is feasible and realistic with excellent results in most of the developed nations and selected Indian centers.
TIMING GUIDELINES: TAPVC

• As soon as diagnosis is made
• Obstructed TAPVC is a surgical emergency
• Any delay may be catastrophic
TGA: TIMING OF SURGERY

- Neonatal diagnosis: Arterial Switch Operation at 10-21 days age
- Diagnosis after 1 month age: Atrial Switch (Senning) operation at 3-4 months
- TGA with VSD: Arterial Switch with VSD closure between 1-3 months.
TRUNCUS ARTERIOSUS

- Elective Repair by 1 – 3 months
- > 3 months high risk for pulmonary vascular disease.
OTHER SITUATIONS WHERE WAITING MAY BE JUSTIFIED:

• Complex 2 ventricle states: wait till cyanosis is apparent/older age (4-5 years)
  - DORV VSD PS
  - TGA VSD PS
  - cTGA VSD PS

• Balanced Single Ventricle states (SaO2 85-90%)
  Intervene if symptoms/cyanosis +
Early detection and referral for timely intervention with a view to minimize mortality and morbidity from prolonged hypoxemia.
Thank You