The Blue Baby

“Cyanotic Congenital Heart Disease”

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Cyanosis

• Cyanosis is a sign not a diagnosis
• Bluish discoloration of skin, nail beds, and mucous membrane
• Noticeable when the concentration of the deoxy-hemoglobin is at least 5 g/dl
• Central vs. peripheral (acrocyanosis)
Physiologic Classification of Cyanotic Lesions

• Intracardiac mixing of systemic and pulmonary venous returns (*increased PBF*)
  • D-TGA
  • TAPVC
  • Persistent truncus arteriosus

• Obstruction to pulmonary blood flow and intracardiac shunt (*decreased PBF*)
  • TOF
  • Tricuspid Atresia
  • Pulmonary Atresia
  • Ebstein’s anomaly (when severe)
The 5 T’s

- TOF (Tetralogy of Fallot)
- TGA (D- transposition of the great arteries)
- Tr A (Persistent truncus arteriosus)
- TAPVR (Total anomalous pulmonary venous return)
- TA (Tricuspid atresia “Tingle” \{single\} ventricle)
  - Single-ventricle lesions:
    - Hypoplastic left heart syndrome (HLHS)
    - Hypoplastic right heart syndrome (PA/TA)
    - Ebstein’s anomaly of the tricuspid valve (when severe)
    - Double-inlet left ventricle (DILV)
    - Unbalanced atrioventricular septal defect (unbalanced AVC)
Tetralogy of Fallot (TOF)

- The most common cyanotic CHD (~ 10% of all CHD)

- Wide spectrum ranging from very pink to very blue to spells
TOF

• Four classic components:
  – large malalignment VSD
  – Overriding aorta
  – Pulmonic stenosis
    • subvalvar
      “infandibular”
    • valvar
    • supravalvar
  – RVH

• Other findings:
  – Right aortic arch (~ 25%)
  – LAD from RCA (~ 5%)
The four features of TOF can be explained by a single abnormality:

“The anterior deviation of the RVOT (conus, infundibulum), without which the diagnosis of TOF is in doubt”

The degree of RV outflow obstruction is quite variable, ranging from very slight obstruction (pink TET), to severe pulmonary stenosis, to complete pulmonary atresia (duct-dependent)
TOF.. Historical Background

• In 1672, Niels Stensen from Denmark gave the first anatomic description (Monology of Stensen)

• In 1888, Fallot from France described the pathologic and clinical manifestations of the defect, which he termed “La maladie bleue”

• In 1945, Alfred Blalock and Helen Taussig with Vivien Thomas from Johns Hopkins performed the first surgical aorto-pulmonary shunt (BT shunt) for palliation of cyanosis in a girl with TOF

• In 1955, Lillihei from Minnesota performed an intracardiac repair, using controlled cross-circulation, in a boy with TOF
TOF.. History
TOF.. Hemodynamics

- Two major lesions:
  - A *large nonrestrictive VSD*, allowing equalization of ventricular systolic pressures
  - *Severe pulmonic stenosis*, protecting the pulmonary vasculature from developing pulmonary hypertension

- Obstruction of the pulmonary outflow tract becomes more severe over the time, rarely progressing to pulmonary atresia
**TOF.. Characteristic Features**

- **The degree of cyanosis and symptoms are variable**
  
  Any event “temper, exercise, etc. → ↓ SVR leading to increase cyanosis by increasing R→L shunt across the VSD

- **Hypercyanotic (TET) spells** (rare due to early surgical intervention)

- **Squatting**

  ↑ SVR → ↓ R-L shunt and briefly ↑ systemic venous return → ↑ RV stroke volume and PBF

- **Easy fatigability**

- **Congestive heart failure does not occur in patients with TOF, except in those patients with acyanotic form (pink TET).**
TOF. Clinical Features

- Cyanosis/Clubbing

- Harsh SEM along MLSB and ULSB +/- a thrill *(the murmur is due to PS and not VSD)*

- Although the murmur is not diagnostic of TOF, *the loudness of the murmur is inversely related to the severity of the stenosis*

- A *continuous murmur* might be heard in patients with pulmonary atresia representing either PDA, bronchial collaterals, but may represent an operative (BT shunt)
TOF.. Clinical Features

Children with Tetralogy of Fallot exhibit bluish skin during episodes of crying or feeding.

“Tet spell”
Hypercyanotic (TET) Spells

- Peak incidence: 2-4 months of age
- No relationship between the degree of cyanosis at rest and the likelihood of having hypercyanotic spells
- Mechanism: ? multifactorial (infandibular spasm & \(SVR\))
- Features: sudden onset of the followings:
  - Hyperpnea (rapid and deep respiration)
  - Irritability and prolonged crying
  - Increase cyanosis
  - Limpness, seizure activity
  - Coma/Death
  - *Decreasing intensity of the murmur on auscultation*
**TET Spell Management**

- Have parent hold and calm child
- Knee/chest position
- Avoid iatrogenic agitation
  - Limit exam, venipuncture, etc.
    - No inotropes (Digoxin, Dopamin, or Dobutamine) and no diuretics
- Oxygen (increases SVR and decreases PVR)
- Morphine 0.1-0.2 mg/Kg SQ (decreases sympathetic tone, decreases oxygen consumption, suppresses the respiratory center and abolishes hyperpnea) or Ketamine 1-3 mg/Kg IM (sedates and increases SVR)
- IVF (improves RV performance) and correct anemia
- Phenylephrine (Neo-Synephrine; alpha 1 agonist which causes vasoconstriction then increases SVR)
  - Bolus: 0.1 mg/Kg IM, SQ, or IV
  - Followed by continuous IV infusion: 0.1-0.5 mcg/Kg/min
- Beta-blockers (Decrease oxygen consumption, may lessen infandibular spasm by decreasing myocardial contractility)
  - Esmolol load 500 mcg/Kg over 1 min (then infuse 50-950 mcg/Kg/min)
  - or Propranolol 0.05-0.25 mg/Kg IV over 5 min
- Treat acidosis with NaHCO3: 1-2 mEq/Kg/dose IV
- Intubate/paralyze/anesthetize
- Surgical shunt, emergently
**Diagnostic Tools**

- **ECG:** RAD + RVH pattern (except in acyanotic form)

- **CXR:**
  - Normal/small heart size
  - Black lung field (decreased PVMs)
  - “Boot-shaped” heart (*coeur en sabot*) {due to RVH and small MPA segment}
  - Right aortic arch (25%)
TOF .. Angiograms
TOF.. Natural History

- Infants with acyanotic TOF gradually become cyanotic
- Patients who are already cyanotic become more cyanotic as a result of worsening RVOT obstruction and polycythemia
- The time of appearance and the severity of cyanosis are directly related to the severity of pulmonary stenosis and the degree of reduction of pulmonary blood flow
- Long-standing cyanosis complications
To improve PBF and arterial saturation

- Blalock-Taussig shunt (anastomosing a subclavian artery to a branch PA)
- Modified B-T shunt (a synthetic tube “polytetrafluoroethylene PTFE” or GoreTex, usually 4 mm in diameter, that connects a subclavian artery and a branch PA)
- Other shunts: Waterston, Potts, central
TOF.. B-T Shunt
The Blalock-Taussig operation diverts blood from a branch of the aorta to the pulmonary artery. This allows blood to flow to the lungs to receive oxygen.
Other Shunts
TOF.. Timing For Complete Repair

- **Symptomatic**: 2-4 months of age (or even younger)
- **Asymptomatic and minimally cyanotic**: 4-6 months of age
- **Mildly cyanotic with previous shunt surgery**: 1-2 years post shunt surgery
- **Asymptomatic and acyanotic children (pink TET)**: 1-2 years of age
- **Asymptomatic with coronary artery anomalies**: 3-4 years of age (RV to PA conduit may be required)
TOF.. Surgical Options.. Corrective Repair

• Palliative procedures has decreased due to earlier complete repair

• Procedure: Patch closure of the VSD and widening of the RVOT by resecting the infundibular tissue and placement of a fabric patch (subannular Vs annular patch)

• Complications: Bleeding, CHF, PI, RBBB from ventriculotomy, RV/LV dysfunction, CHB, VT (rare), and sudden death
Transposition of the Great Arteries (D-TGA)

- ~5% of all CHD
- M:F = 3:1
- A communication at atrial, ventricular, or arterial level is necessary for survival
- Associated defects: VSD, PS, COA, IAA, TA, and coronary abnormalities
D-TGA

- In complete transposition, the aorta lies anteriorly to the PA

- The “D” in D-transposition refers to the position of the aorta with respect to the PA. It is rightward (dextroposition)

- Aorta from RV is always “BAD”
The aorta arises from the RV, and the pulmonary artery arises from the LV. As a result, there is a complete separation of the pulmonary and systemic circulation.
D-TGA

In half of the patients, the ventricular septum is intact; and the shunt occurs at the atrial level. In the other half, a VSD is present.

Patients with VSD and PS are often the least symptomatic because the PS prevents pulmonary over circulation and enhances flow of fully saturated blood through the VSD into the aorta; these patients resemble those with TOF.
Clinical features

- Infants are often large for gestational age (LGA)
- Moderate to severe cyanosis from birth
- Happy tachypnea (tachypnea without retractions unless CHF develops)
- Physical findings vary with associated defect(s)
- S2 is single and loud (representing closure of the anteriorly placed aortic valve)
- Usually no murmur is heard if no other lesions exist (VSD/PS)
• **ABG:** severe hypoxemia +/- acidosis

• **ECG:** RAD, RVH/CVH, RAE

• **CXR:**
  - Cardiomegaly
  - Increased PVMs
  - *“Egg on a string”* appearance due to narrow mediastinum because the great vessels lie one in front of the other
D-TGA .. Angiograms

PA
LV
RV
AO
PDA
Untreated patients die in a few weeks

Patients with no atrial or ventricular septal defect are severely cyanotic, and may die in a matter of hours
D-TGA Treatment.. Medical

1- Ensure mixing and improve arterial oxygen saturation:
   a. Prostaglandin E1 (PGE1) at 0.025-0.1 mcg/Kg/min IV infusion to maintain ductus arteriosus patency.
   {S/E: apnea, hypotension, jitteriness/seizures fever, rash, diarrhea, plts inhibition}
   b. Rashkind balloon atrial septostomy (BAS)

2- Treat CHF

Note: Oxygen can be bad ???
(PDA closure and pulmonary vessels dilation leading to CHF)
Balloon Atrial Septostomy (Rashkind Procedure)

Balloon-tipped catheter is inserted through the atrial septal defect (ASD)

Once the balloon is inflated, the catheter is pulled back through to widen the ASD

An opening in the septum allows oxygen rich and oxygen poor blood to mix to improve circulation.
D-TGA Treatment.. Surgical

- **At atrial level** (*Senning/Mustard* procedures)
- **At the ventricular level** (*Rastelli* procedure) for D-TGA + VSD + PS
- **At the arterial level** {Arterial switch procedure “*Jatene/LeCompte operation*”} *(performed within the first 2 weeks of life before the PVR falls; the LV will then become “deconditioned” to a systemic pressure load)*
- **Damus-Kaye-Stansel** operation for D-TGA + VSD + subaortic stenosis
Arterial Switch Operation

- Aorta
- Pulmonary artery
- Patch in place of old coronary artery origin.
- Aorta is "switched" with the pulmonary artery.

Coronary arteries
- Both transposed arteries are divided at the red dotted line.
- Coronary arteries are detached from aortic valve (on right side of heart) and connected to pulmonic valve (on left side of heart).

ARTERIAL SWITCH OPERATION

- AO
- LPA
- RPA
- LCA
- RCA
- RA
- RV
- Distal AO
- Prox. PA
- Distal PA
- Prox. AO
Long Term Complications

- Atrial switch
  - Arrhythmias
  - Sudden death
  - Obstruction of venous return
  - Progressive RV dysfunction
- Arterial switch
  - Coronary artery stenosis
  - Pulmonary artery stenosis
Truncus Arteriosus

A single arterial blood vessel leaves the heart and feeds both the pulmonary and systemic circulations.
Truncus Arteriosus (Tr A)

- **Etiology**: failure of the embryonic truncus arteriosus to septate during development

- **Always associated with a large VSD**

- The valve at the origin of this vessel is often abnormal (tricuspid or more), and may be narrowed or insufficient

- **Hemodynamics similar to those of VSD and PDA**
- Pulmonary over circulation effect

- ~75% associated with *DiGeorge syndrome* (chromosome 22 deletion) (hypocalcemia and lymphopenia)

- Often presents with mild cyanosis and loud murmur in the nursery
Pulmonary Vascular Resistance Changes
Truncus Arteriosus.. Clinical Features

- Tr A vs. TOF (aortic valve involvement in case of Tr A)
- Cyanosis (because the elevated PVR limits the PBF)
- CHF (when PVR drops)
- Coronary steal (from PA’s)
- The pulses are bounding
- Single S2
- Systolic ejection click at the apex (from the stenotic valve)
- Different murmurs can be heard
  - Loud systolic murmur from a VSD +/- MDR at the apex
  - +/- SEM if the the truncal valve is stenotic
  - +/- high-pitched early diastolic decrescendo murmur if the truncal valve is insufficient
Diagnostic Tools

- **EKG:** normal axis, BVH
- **CXR:**
  - Cardiomegaly
  - Increased PVMs
  - Prominent ascending aorta
  - Right aortic arch (25%)
Truncus Arteriosus.. Natural History

- Similar to VSD but is more severe with rapid development of pulmonary vascular disease
- Mortality of 85-100% before the first year of life
- Trucal insufficiency is usually progressive
- High risk of sudden death before surgery due to coronary steal
Truncus Arteriosus.. Treatment

• PGE1 infusion is harmless but unhelpful
• Treat CHF
• Corrective surgery at the time of diagnosis
• Procedure: patch closure of the VSD and RV-PA conduit placement
Total Anomalous Pulmonary Venous Return/Connection (TAPVR)

- ~ 1% of all CHD
- M : F = 4 : 1
- No direct connection between the pulmonary veins and the left atrium
TAPVR.. Types

- Supracardiac (50%)

- Cardiac (20%)

- Infracardiac (subdiaphragmatic) (20%) .. obstruction is common

- Mixed type (10%)
TAPVR. Echo Findings
During embryogenesis, the developing pulmonary veins share connections with various systemic veins. In normal situations, these connections regress and the pulmonary veins connect to the left atrium. In TAPVR (Total Anomalous Pulmonary Venous Return), the veins do not connect to the LA, and one or more primordial connections remain.

An atrial communication (ASD/PFO) is necessary for survival.
Unobstructed TAPVR

- Variable presentations
  - Asymptomatic (discharge home)
  - Most patients develop CHF in infancy
  - Failure to thrive
  - Recurrent respiratory infections
  - Nearly all patients come to medical attention before 6 months of age
TAPVR.. Supracardiac

Vertical vein
Diagnostic tools

- **P/E:**
  - Variable cyanosis
  - Signs of CHF
  - May mimic ASD physical findings
  - or pulmonary hypertension (Single and loud S2)

- **EKG:** RAD, RAE, RVH

- **CXR:**
  - Cardiomegaly
  - Increased PVMs
  - "Snow-man" appearance (due to dilated vertical vein, innominate vein, and RSVC)
Obstructed TAPVR.. Why?

- The venous channel is long
- The channel traverses the diaphragm through the esophageal hiatus and is compressed by either esophageal or diaphragmatic movement
- The channel narrows at its junction with the portal venous system
- The pulmonary venous blood must traverse the hepatic capillary system before returning to the RA by way of hepatic veins
- The ductus venosus closes shortly after birth
TAPVR.. Infracardiac
Clinical Features

- The obstruction elevates pulmonary venous pressure leading to severe pulmonary edema

- The clinical features are related to the consequences of pulmonary venous obstruction and to the limited PBF

- Severe cyanosis and respiratory distress
• **P/E:**
  - Cyanosis with respiratory distress
  - S2 single and loud
  - No murmur

• **EKG:** non-specific

• **CXR:**
  - Normal heart size
  - Diffuse reticular pattern of pulmonary edema
    - “white-out” lungs
    - {confused with RDS}
TAPVR.. Treatment

- Emergency surgery as soon as the diagnosis is made. The only alternative to emergency surgery is ECMO.
- Surgical repair usually consists of reimplantation of the pulmonary veins to the left atrium.
- In some infants, pulmonary hypertension persists in the postoperative period for a few days and requires management with mechanical hyperventilation, NO and/or ECMO.
Tricuspid Atresia

- The tricuspid valve and the inflow portion of the RV are not developed leading to lack of direct communication between the RA and the RV.
- Associated defects (ASD, VSD, or PDA) are necessary for survival.
Tricuspid Atresia

It is classified according to the presence or absence of PS and TGA.
TA.. Clinical Features

- Cyanosis
- CHF with slight cyanosis (due to increased PBF)
- Single S1
- +/- heart murmur
• **EKG:** *(LAD and LVH)*
  – Left axis deviation
  – RAE
  – LVH (paucity of the RV forces)

• **CXR:**
  – Decreased PVMs (except if TGA and/or large VSD exist)
  – Cardiomegaly
    {prominent right heart border (RAE) and the prominent left heart border (LVE)}
Surgical Options

• For all single ventricle lesions, **Palliation**, but not physiologic corrective, operations are available

• Palliation is achieved with a series of three stages:
  – A modified B-T Shunt
  – **Bidirectional Glenn** (anastomosis of the end of the SVC to the roof of the RPA)
  – **Fontan** Operation (TCPC): a complete (total) cavopulmonary connection

• Heart transplantation
Fontan Procedure

First stage: Bi-Directional Glenn
A graft is used to route blood flow from the superior vena cava to the pulmonary artery instead of to the right atrium.

Second stage: Fontan
Both a graft and an internal baffle (wall) are used to route blood flow from the inferior vena cava to the pulmonary artery instead of to the right atrium.
* Modified from Mullins CE and Mayer DC, Congenital Heart Disease – A Diagrammatic Atlas
Ebstein’s Anomaly (of the Tricuspid Valve)
Left: The base of the pulmonary artery is connected with the aorta, and a shunt is placed between a branch of the aorta and the other part of the pulmonary artery.

Right: A new pathway to bypass the left side of the heart is created. Blood moves through the pulmonary artery to the aorta and out to the body. Some blood moves through the Blalock-Taussig shunt to the pulmonary artery, connected to the lungs.
Your Role As Pediatrician!

High index of suspicion
Initiate work-up
Initiate therapy
Call your cardiologist
High Index of Suspicion

- Suspicious family and social history
- How does the baby look?
  - Color
  - Respiratory status
  - Dysmorphic features
  - O2 Saturations
  - Feeding difficulties
- On exam:
  - Heart murmur (in the delivery room never good)
  - Irregular rhythm
  - Diminished femoral pulses
Your Role As Pediatrician!

• **Initiate work-up:**
  – Four extremities BPs
  – Oximetry (pre/postductal)
  – Hyperoxia test
  – CXR +/- EKG
  – ABGs

• **Initiate therapy:**
  - Don’t hesitate to start Prostaglandin \((0.0125-0.1 \text{ mcg/kg/min})\)
  - Avoid O2 unless necessary \((\text{Keep O2 sats > 75\% unless associated acidosis})\)

• Always, when in doubt, call your cardiologist!
Cyanosis Work-Up

- Hyperoxia test
  - $pO_2 > 150 \text{ mm Hg}$  most likely respiratory
  - $pO_2 < 40 \text{ mm Hg}$  most likely cardiac
  - $pO_2 40-150 \text{ mm Hg}$  either possible

- Always obtain ABG/Oximetry from two areas:
  - Preductal (right arm, ear, radial artery)
  - Postductal (umbilical artery or feet)
Cyanosis Work-Up

**Differential Cyanosis**
- Preductal 10% higher than postductal (pink upper and blue lower part of the body)
  - PPHN
  - Left heart obstructive lesions (severe AS, IAA, COA) with R → L ductal shunt

**Reverse Differential Cyanosis**
- Postductal 10% higher than preductal (blue upper and pink lower part of the body)
  - D-TGA+PDA+PPHN
  - D-TGA+PDA+COA/IAA
Take Home Message

- Cyanotic Heart lesions are not uncommon
- Early detection and work-up is necessary
- Management options should be individualized
- Respect and support family decision
- Complete cure is not always available
- Long term complications may occur
Thank You