Congenital Heart Disease – What’s the Diagnosis?

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Practice Change

Upon completion of this session, you should be able to:

- Utilize clinical exam and hyperoxia testing to develop a differential for congenital heart disease without an echocardiogram.

- Utilize tools to quickly review the potentially life-threatening forms of congenital heart disease.
Transitions Can Be Stressful: From Womb to Delivery Room

- Objectives
  - Differentiate structural cardiac diseases in the neonate based on physical exam
  - Describe the conditions in which prostaglandin therapy is best used
  - Identify cardiogenic shock
  - Summarize the utility of various diagnostic studies in the evaluation of congenital heart disease and congestive heart failure
Baby Blue – The History

- Full term, LGA, NSVD
- Noted in nursery to be grunting and cyanotic
- Sat 80%
- HR 150s, “normal” BP
- Systolic murmur at left sternal border
- Lungs clear, slight retractions
- Good pulses and perfusion

What is the differential??
Differential Diagnosis

- Sepsis
- Hypoglycemia
- Lung Pathology
  - Alveolar hypoventilation
  - Ventilation-perfusion mismatch
  - Impairment of diffusion
- Congenital Heart Disease
Diagnostic Helpers

- **History**
- **Exam**
  - Signs of respiratory distress?
  - Evidence of prematurity?
  - Central versus peripheral cyanosis?
  - Is there a murmur?
- Cardiac screen with pulse oximetry
- Hyperoxia Test
- 4 extremity BPs
- Chest x-ray
Cyanosis**

- **Peripheral**
  - Blueness of nail beds, hands and feet
    - related to circulation but not hypoxia
  - Common finding in normal infant

- **Central**
  - Tongue, tip of nose
  - Difficult to detect unless Sat <85%
    - Why?
Recognition of Cyanosis

- Detectable with 3-5gm/dl of reduced hemoglobin
- Factors that shift oxygen-hemoglobin to left delay appearance of cyanosis
  - Presence of Fetal Hemoglobin
  - Hypothermia
- Skin pigmentation can also impact ability to detect cyanosis
Diagnostic Helpers

- Exam
- Cardiac screen with pulse oximetry
- Hyperoxia Test
  - How is this test performed?
- Chest x-ray
- 4 extremity BPs
- Then Echo
Neonatal Pulse Ox Screening**

- Usual cut-off level of 94-96%
  - Repeat screening can limit false positives from transient ductal shunting
- More reliable after 2 hours of age
- 1st day identifies other problems in addition to heart disease, later screening is more specific
- Greater sensitivity with post-ductal probe

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Hyperoxia Test

- Room air **arterial** blood gas
- Place patient in 100% oxyhood for 20 minutes. Again measure arterial PAO$_2$.
  - PA$_{O2}$ greater than 250 = lung disease
  - PA$_{O2}$ less than 150 = cardiac disease
- Pulse oximetry is not sensitive enough to detect change
Cyanotic Lesions

- The terrible T’s in frequency of appearance
  - Tetralogy of Fallot **
  - Transposition**
  - Total anomalous pulmonary venous return
  - Tricuspid Atresia
  - Truncus

- Use your hand to remember them all!
  - Truncus, Transposition, Tri Atresia, Tetralogy, TAPVR
Tetralogy of Fallot
Truncus Arteriosus

AO = Aorta
PA = Pulmonary Artery
LA = Left Atrium
RA = Right Atrium
LV = Left Ventricle
RV = Right Ventricle

Continued Aorta and Pulmonary Artery
Opening Between Ventricles

Oxygen-rich Blood
Oxygen-poor Blood
Mixed Blood
In the presence of cyanosis, is there a murmur?**

- TOF – pulmonary stenosis murmur
- *Critical* pulmonary stenosis
  - In these cases, augment pulmonary blood flow with prostaglandin
  - Supplemental oxygen
- Truncus arteriosus with abnormal truncal valve
  - prostaglandin won’t help here
Cyanosis with no murmur

- Transposition**
- Tricuspid Atresia
- Pulmonary atresia
- TAPVR
Cyanotic infant:
Ductal Dependent PBF

- Desaturation is due to limited pulmonary blood flow
  - Tetralogy of Fallot
  - Pulmonary atresia or critical pulmonary stenosis
  - Tricuspid atresia

- Due to high RV pressure, atrial level shunt from right to left occurs (i.e. blue blood mixes with red)

- May benefit from prostaglandin to maintain PDA flow from aorta to pulmonary artery**
Prognosis for Children with TOF**

- 36-year actuarial survival rate of 96% and normal life expectancy (J Am Coll Cardiol. 1997;30(5):1374-1383)
  - Repaired prior to onset of polycythemia
  - No transannular patch
- [http://www.sts.org/patient-information/what-pediatric-heart-disease/](http://www.sts.org/patient-information/what-pediatric-heart-disease/)
Baby Shocky – The History

- This 11 day old female who was found to have a heart murmur on routine newborn care on DOL #4. By report, the patient otherwise looked well. Referral to an outside cardiologist was made, and occurred on DOL 11. At this office visit, she was found to be severely ‘cyanotic’, grunting and cool to the touch.

- What’s the differential diagnosis?
Physical Exam

- HR 168, RR 80, Systolic BP 70, RA SaO2 90%
- GEN: cyanotic infant in acute distress
- HEENT: Anterior fontanelle soft
- CVS: RRR, + systolic murmur (not otherwise described), no gallop,
- Perfusion: poor with ~ 5 sec cap refill
- ABD: soft, non tender, palpable liver
- EXT: cold and mottled
- NEURO: awake, alert and responding to stimuli

SHOCK!!!!
Physical Findings of Shock**

- Tachycardia, tachypnea
- Respiratory distress, grunting, retracting
- Pale or gray with poor perfusion, mottling
- Gallop rhythm
- Hepatomegaly
- Historical findings: Loss of appetite, vomiting, lethargy
**SHOCK!**

- Sepsis
- Congenital Heart Disease – Left Heart Obstruction
  - Coarctation
  - HLHS
  - Critical aortic stenosis
  - Interruption of the aortic arch
- Cardiomyopathy
  - Myocarditis
  - Anomalous left coronary artery
  - Sustained tachyarrhythmia
DIMINISHED SYSTEMIC PERFUSION

- Sepsis-like picture, poor perfusion
- Not notably blue,
  - Do Pulse Ox screen
- Consider cardiac lesions:
  - Coarctation
  - Interrupted aortic arch
  - HLHS
  - Critical AS
- Treatment: Prostaglandin
  - Avoid hyperventilation ($C_{O2}$ rise = increase PVR)

www.stanfordchildrens.org
How to use PGE: Re-inventing the fetus!**

- It is not necessary or important to make the exact anatomic diagnosis prior to initiation of PGE therapy.
- It is necessary to make the diagnosis of highly probable CHD with either:
  - ductal dependent systemic (shock) or
  - ductal dependent pulmonary (cyanosis)
  blood flow prior to initiation of PGE therapy.
Congenital Heart Disease in Patients with Abnormal Karyotype

- Turner syndrome**
  - Bicuspid aortic valve
  - Coarctation

- Trisomy 21
  - 50% have cardiac disease,
    - 40% of these are complete AV Canal
    - VSD, PDA, and Tetralogy of Fallot
  - 75% of patients with AV Canal have Trisomy 21
Congestive Heart Failure

- Inadequate delivery of oxygen to tissues
- Can result from
  - structural abnormality (left to right shunt) or
  - decreased cardiac function
Clinical diagnosis:
- Tachycardia, Tachypnea
- Pulmonary congestion
  - Wheezing
- Hepatomegaly & cardiomegaly
- Dynamic precordium with gallop rhythm
- Poor feeding and/or weight loss
CHF in the Child/Adolescent

- Nausea/vomiting
- Edema
- Hepatomegaly
- JVD
- Gallop rhythm
- +/- Cardiomegaly on CXR
Congestive Heart Failure**

- Generally occurs AFTER first two weeks of life
  - Early (weeks 1-2) Left sided obstructive lesions
    - Aortic Stenosis, Coarctation, Interruption of the Aortic Arch, Hypoplastic Left Heart Syndrome
  - Late (weeks 2-8) Left to right shunts or pump failure
    - L to R shunt
      - Atrioventricular Canal, VSD, unobstructed TAPVR, PDA
    - Pump failure
      - Anomalous Left Coronary Artery from the Pulmonary Artery (ALCAPA), cardiomyopathy, myocarditis
**Congestive Heart Failure**

**Based on these potential diagnoses, evaluation of infants and children presenting with congestive heart failure should include:**

- Chest radiograph
- ECG
- Echocardiogram
- And in addition, consideration for
  - Troponin
  - Lactate
  - BNP (B-type natriuretic peptide)
  - Viral panel
  - Genetic testing
Narrow Complex Tachycardias

Come to the case session to learn more!!!
Anomalous Left Coronary Artery from the Pulmonary Artery

Normal Coronary Arteries

ALCAPA

Aorta

Left coronary

PA

Right coronary

Circumflex

LAD
Anomalous Left Coronary Artery from the Pulmonary Artery (ALCAPA)

- Aorta
- Pulmonary artery
- Anomalous left coronary artery from the pulmonary artery (ALCAPA)
- Tissue damage
- Right coronary artery

Oxygen-rich blood
Oxygen-poor blood

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Conclusions

- A “Cardiac Screen” should be performed on all neonates with suspected CHD.
  - Vitals including 4 extremity blood pressures
  - Saturations by pulse oximetry, ideally repeated at greater than 24 hours age
  - Historical context and exam leads to further testing

- Infants appearing to have ductal dependent pulmonary or systemic blood flow should be started on PGE1.
Practice Change

Upon completion of this session, I encourage you to incorporate these changes into your practice: Utilize tools to quickly review the potentially life-threatening forms of congenital heart disease.

- The hand rule for cyanotic congenital heart disease
- Cardiac screen with pulse oximetry
- Physical exam findings consistent with CHD
Bibliography


2014 Appropriate Use Criteria for Initial Transthoracic Echocardiography in Outpatient Pediatric Cardiology. *Pediatrics* 2014;134:e1774

Cardiac Images: http://www.cdc.gov/ncbddd/heartdefects/index.html