Review of Congenital Heart Disease

Steven H. Todman, M.D.
Pediatric Cardiologist
Louisiana State University
Our Mission

• To discuss various types of congenital heart disease
The Presentation of Congenital Heart Disease is Age Dependent

- Important time periods
  - Neonatal period: Birth to 1 month
  - Infancy: 2 months to one year
  - Children and adolescent
Transitional Circulation

- In the first week of life, the PFO and PDA closes.
- Subsequently, pulmonary vascular resistance drops to normal levels by 2 months.
Cyanosis

• Hyperoxia test is the gold standard to evaluate cyanosis.
  ▫ Obtain ABG to measure PO2
  ▫ Place in 100% oxygen for >10 minutes
  ▫ Measure the PO2
    • If PO2 <50 or unchanged, cardiac etiology is likely
    • If PO2 is 50-150, equivocal for cardiac or pulmonary etiology
    • PO2>150, cardiac etiology is unlikely.
Neonatal Cardiogenic Shock

- Decrease in systemic blood flow with PDA closure.
- Symptoms
  - Poor feeding
  - Lethargy or irritability
  - Oliguria
  - Cool extremities
  - Poor perfusion and peripheral pulses (x4)
Pulmonary Stenosis

• Pathology:
  ▫ Pulmonary stenosis may be valvular, subvalvular, or supravalvular.
• Clinical Manifestations:
  ▫ Mild-moderate: asymptomatic
  ▫ Murmur: systolic ejection murmur at ULSB, radiates to back. +/- click, +/- thrill
  ▫ ECG is normal
  ▫ CXR is normal
Pulmonary Stenosis

- Clinical Manifestations:
  - Severe/ductal dependent:
    - Cyanosis
  - Moderate to severe:
    - ECG demonstrates RAD and RVH
    - CXR is normal, or can show diminished vascular markings.
Pulmonary Stenosis

• **Treatment:**
  - **Mild-moderate:**
    - Observation
  - **Severe**
    - Balloon valvuloplasty
  - **Ductal dependent**
    - Prostaglandins
Pulmonary Stenosis

• Natural History
  ▫ Mild pulmonary stenosis
    • Non-progressive
  ▫ Moderate to severe pulmonary stenosis
    • Progressive

• Associations:
  ▫ Noonan’s syndrome
Aortic Stenosis

• Pathology:
  ▫ May be valvar, subvalvar, or supravalvar

• Clinical Manifestations
  ▫ Murmur: Systolic murmur at URSB, or ULSB with radiation to the neck.
    • +/- click
    • +/- thrill
Aortic Stenosis

• **Clinical Manifestations**
  - Moderate to severe aortic stenosis can be associated with chest pain, syncope, or sudden death.
  - Neonatal presentation of severe aortic stenosis may be heart failure.
Aortic Stenosis

- **EKG:**
  - Mild: normal
  - Moderate to severe: LVH +/- strain
- **CXR:**
  - Usually normal
- **Natural History:**
  - Progressive
Aortic Stenosis

- **Treatment:**
  - Mild to moderate: observation
  - Severe – Neonatal: PGE and balloon valvuloplasty
  - Severe – Child/Adolescent: balloon valvuloplasty

- **Associations:**
  - Bicuspid aortic valve
  - Coarctation of the aorta
  - Williams’ syndrome (supravalvar AS, PS)
“If we pull this off, we’ll eat like kings.”
Coarctation of the Aorta

• Pathology:
  ▫ Almost always juxtaductal

• Clinical Manifestations:
  ▫ First week of life: Poor feeding, respiratory distress, shock, acidemia, weak lower extremity pulses
  ▫ May have no murmur, or non-specific systolic ejection murmur.
Coarctation of the Aorta

- **Treatment:**
  - Medical – PGE1 infusion first weeks of life.
  - Surgical – Repair (end-to-end anastomosis)
- **Natural History:**
  - Re-coarctation
- **Associations:**
  - Bicuspid aortic valve
  - Turner’s syndrome
Interrupted Aortic Arch

• Pathology:
  ▫ Severe form of coarctation where a portion of the aortic arch is atretic, or absent.
Interrupted Aortic Arch

- **Pathology:**
  - Severe form of coarctation where a portion of the aortic arch is atretic, or absent.
Interrupted Aortic Arch

- **Treatment**
  - PGE1
  - Surgical repair
- **Associations**
  - Type B interrupted aortic arch and DiGeorge syndrome
**d-Transposition of the Great Arteries**

- **Pathology:**
  - Parallel circulation
  - Mixing is required (ASD, PDA)
- **Clinical Manifestations**
  - Cyanosis in a large newborn
  - Single S2
  - Usually no murmur
d-Transposition of the Great Vessels

- **EKG:**
  - Normal
- **CXR:**
  - Egg on a string
- **Treatment:**
  - Prostaglandins
  - +/- Balloon atrial septostomy
  - Surgery
d-Transposition of the Great Arteries

• **Associations:**
  ▫ Most common cyanotic lesion to present in the newborn period
  ▫ Big fat blue baby
Clinical Case

- 6 month old male presents to your clinic for a well child checkup. Pulse oxymetry measures 88% in the right upper extremity. Pulses are equal in the upper and lower extremities, and the lungs are clear to auscultation.

- Cardiac auscultation reveals a normal S1 and S2, and a loud, harsh III/VI systolic ejection murmur at the upper left sternal border.
Clinical Case

- Differential diagnosis?
Vignette #3
Vignette #3
Clinical Case

- You leave the room to check on another patient and return to find that the patient is crying unconsolably, and is visibly cyanotic. No murmur is heard.
  - What is going on, and what is the next step?
Hypercyanotic “TET” Spell
Hypercyanotic “TET” Spell

- (1) Comfort the child
- (2) Oxygen (preferably BBO2)
- (3) Knee to chest position
  - Raises systemic vascular resistance.
- (4) Morphine sulfate SQ (0.1mg/kg)
  - Slows respiration, and may also relax the infundibulum
- (5) Phenylephrine 2 to 5 mg/kg/min
  - Increases SVR
Tetralogy of Fallot

• Pathology:
  ▫ (1) RVOT obstruction
  ▫ (2) RVH
  ▫ (3) VSD
  ▫ (4) Overriding aorta

• Clinical Manifestations
  ▫ Degree of RVOT obstruction determines oxygen saturation
  ▫ Murmur: Systolic ejection murmur at the mid to upper left sternal border
Tetralogy of Fallot

- **EKG:**
  - RVH and RAD
- **CXR:**
  - Boot shaped heart (upturned cardiac apex), decreased lung vasculature.
- **Natural History:**
  - Hypercyanotic “Tet” Spells
- **Associations:**
  - Most common cyanotic lesion in general
  - DiGeorge syndrome
Post-op
Post-op
Clinical Case

• A 10 day-old boy presents to the emergency room with increased irritability, poor feeding and ashen discoloration of the skin for the past 2-3 days. He was born full term via normal vaginal delivery with no perinatal complications.

• He was well, and asymptomatic for the first week of life. There are no known sick contacts.
“And now Edgar’s gone. … Something’s going on around here.”
Clinical Case

- Physical Exam - child in moderate to severe respiratory distress with cyanosis and gray skin tone. Capillary refill is more than 3 seconds, with weak pulses in all extremities. Blood pressure was not obtainable. Mild hepatomegaly was noted, and the cardiac apex appears displaced to the right. Auscultation revealed a single second heart sound with no significant murmurs.
Clinical Case

• Your assessment?
Clinical Case

• Presentation is classic for cardiogenic shock
  ▫ Abnormal apical impulse
  ▫ Single second heart sound
Diagnosis and management

- Hypoplastic left heart
- Prostaglandins
- Correction of metabolic acidosis
- Avoid excessive oxygen
- Maintain normal electrolytes
  - Calcium
Hypoplastic Left heart syndrome

- Pathology:
  - Hypoplasia of LV, and atresia or critical stenosis of the aortic and/or mitral valves, and hypoplasia of the ascending aorta and aortic arch.
Hypoplastic Left Heart

• Clinical Manifestations
  ▫ **Cardiogenic shock**
    • Tachycardia, dyspnea, weak peripheral pulses
    • Generally greyish-blue skin color with poor perfusion
  ▫ **Murmur**
    • May have no murmur
    • S2 is single
    • PMI may be displaced to the right
Hypoplastic Left Heart

• EKG:
  ▫ RVH

• CXR:
  ▫ Usually normal

• Natural History:
  ▫ Critically ill (shock) during first week of life with PDA closure

• Treatment:
  ▫ Prostaglandins
  ▫ Surgery
    • Norwood, Bidirectional Glenn/Hemi-fontan, Fontan
“Say ... what's a mountain goat doing way up here in a cloud bank?”
Tricuspid Atresia

- **Pathology:**
  - Absent tricuspid valve, with hypoplastic right ventricle.
  - ASD with right to left shunting is necessary.
Tricuspid Atresia

• Clinical Manifestations
  ▫ Presentation varies, however generally presents with cyanosis
Tricuspid Atresia

• EKG:
  ▫ Superior QRS axis (0 to -90 degrees), LVH

• Treatment:
  ▫ Prostaglandins if severe cyanosis
  ▫ Surgery
    • Ultimately requires Fontan

• Associations:
  ▫ Cyanosis with superior QRS/LVH = TA
Ebstein’s Anomaly

Pathology:
- Apical displacement of the tricuspid valve, so that a portion of the RV is incorporated into the RA (atrialized).
- A PFO/ASD is present in all patients.
Ebstein’s Anomaly

- Clinical Manifestations
  - Cyanosis often present in the first few days of life.
  - Murmur
    - Triple or quadruple rhythm with widely split $S_2$, and $S_3$ and $S_4$. 
Ebstein’s Anomaly

- **EKG:**
  - RBBB, RAE, WPW pattern, first degree heart block
- **CXR:**
  - Wall to wall heart
- **Treatment:**
  - Eventually requires surgery
- **Associations:**
  - WPW
Truncus Arteriosus

- Pathology:
  - A single arterial trunk with a truncal valve exits the heart and gives rise to the pulmonary, systemic, and coronary circulations.
  - A large VSD is present below the truncal valve.
Truncus Arteriosus

• Clinical Manifestations
  ▫ Cyanosis can be seen after birth.
  ▫ CHF develops weeks after birth after PVR decreases.
  ▫ Bounding peripheral pulses

• Murmur
  • Single S2
  • May have an early diastolic murmur from truncal valve insufficiency
Truncus Arteriosus

- **EKG:**
  - Biventricular hypertrophy

- **CXR:**
  - Cardiomegaly, with increased vascularity

- **Treatment:**
  - Surgery

- **Associations:**
  - DiGeorge syndrome
After many years of marital bliss, tension enters the Kent household.
Total Anomalous Pulmonary Venous Return (TAPVR)

- **Pathology (Supracardiac):**
  - Most common type
  - Common pulmonary venous sinus drains into the right SVC through the left vertical vein and the left innominate vein.
Total Anomalous Pulmonary Venous Return (TAPVR)

- **EKG:**
  - rSR’ pattern in V1

- **CXR:**
  - Cardiomegaly, with increased vascularity.
  - Snowman sign generally after 4 months.

- **Treatment:**
  - Surgery
Left to Right Shunt Lesions

- ASD
- VSD
- PDA
- Endocardial Cushion Defect (AV canal)
Atrial Septal Defect

- Pathology:
  - Most common
    - Secundum
  - Sinus venosus defects are associated with PAPVR.
Atrial Septal Defect

• Clinical Manifestations
  ▫ Pediatric patients are typically asymptomatic
    • Generally, no CHF
  ▫ Murmur
    • Widely split and fixed S2 and a systolic ejection murmur at the ULSB
    • Mid-diastolic rumble from relative tricuspid stenosis at the LLSB
Atrial Septal Defect

- **EKG:**
  - rSR’ pattern in V1

- **CXR:**
  - Cardiomegaly with right heart enlargement
  - Prominent pulmonary artery and increased lung markings
Atrial Septal Defect

• Natural History:
  ▫ Small defects tend to close spontaneously prior to 4 years of life.
  ▫ Larger defects rarely close spontaneously

• Treatment:
  ▫ Interventional closure in cath lab ~4 years of age
  ▫ Surgical closure if not amenable to device closure

• Association:
  ▫ Holt-Oram
And no one ever heard from the Anderson brothers again.
Ventricular Septal Defect

- Pathology (small):
  - Holosystolic murmur at the LLSB
Ventricular Septal Defect

- Clinical Manifestations (small VSD)
  - Pediatric patients are typically asymptomatic with normal growth and development
- EKG:
  - normal
- CXR:
  - normal
Ventricular Septal Defect

• Pathology (moderate to large VSD):
  ▫ **Murmur**
    • Holosystolic murmur at LLSB
    • +/- Apical diastolic murmur
Ventricular Septal Defect

- Clinical Manifestations (moderate to large VSD)
  - Poor weight gain, decreased exercise tolerance, frequent lower respiratory infections, and CHF
- EKG:
  - LVH, or biventricular hypertrophy
- CXR:
  - Cardiomegaly with increased pulmonary vascularity
Ventricular Septal Defect

• Treatment
  ▫ Anticongestive medications
    • Diuretics first line
  ▫ Surgical repair 4-6 months of age.
Patent Ductus Arteriosus

- Pathology (small):
  - Continuous murmur at the LUSB.
Patent Ductus Arteriosus

- Clinical Manifestations (small PDA)
  - Pediatric patients are typically asymptomatic with normal growth and development
- EKG:
  - normal
- CXR:
  - normal
Patent Ductus Arteriosus

- Pathology (moderate to large PDA):
  - Murmur
    - Continuous murmur at LUSB
    - Bounding peripheral pulses with wide pulse pressure
Patent Ductus Arteriosus

• Clinical Manifestations (moderate to large PDA)
  ▫ Poor weight gain, decreased exercise tolerance, frequent lower respiratory infections, and CHF

• EKG:
  ▫ LVH, or biventricular hypertrophy

• CXR:
  ▫ Cardiomegaly with increased pulmonary vascularity
Patent Ductus Arteriosus

- **Treatment**
  - Indomethacin if in the immediate newborn period, particularly with pre-term infants
  - Device closure in the cardiac catheterization laboratory
  - Surgical ligation
Endocardial Cushion Defect

- Pathology: Complete AV canal most common form
  - Ostium Primum ASD, VSD in the inlet ventricular septum, and cleft mitral valve
  - Results in interatrial and interventricular shunts, and AV valve regurgitation
Endocardial Cushion Defect

• Clinical Manifestations
  ▫ Patients typically have signs of CHF.
  ▫ Murmur
    • Systolic ejection murmur at upper left sternal border (relative pulmonary stenosis)
    • Apical holosystolic murmur (mitral regurgitation)
    • May also have a gallop rhythm and hepatomegaly if CHF is present.
Endocardial Cushion Defect

- **EKG:**
  - Superior QRS axis
  - First degree heart block
  - RVH

- **CXR:**
  - Cardiomegaly with increased lung markings
Endocardial Cushion Defect

• Natural History:
  ▫ Heart failure 1 to 2 months after birth.
  ▫ Recurrent lower respiratory infections are common.

• Treatment:
  ▫ Anticongestive medications – Furosemide
  ▫ Surgical repair at approximately 4 months of age.

• Association:
  ▫ Down Syndrome
“Mr. Osborne, may I be excused? My brain is full.”