Pediatric Board Review - Congenital Heart Disease

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Our Mission

• To discuss various types of congenital heart disease that are commonly tested on the Pediatric board exam.
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)
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?
Hyercyanotic “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
Bundle Branch Block
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.
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. Oxygen saturation was 70% on room air. 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
  ▫ Significant oxygen desaturation beyond what is typically seen with sepsis should prompt investigation into cardiac etiologies.
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
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 S2, and S3 and S4.
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
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)

- Clinical Manifestations (unobstructed pulmonary veins)
  - Mild cyanosis from birth, CHF, and growth restriction.
- Murmur
  - Widely split S2, and 2-3/6 systolic ejection murmur at ULSB
  - Mid-diastolic rumble at LLSB (secondary to flow through the tricuspid valve)
Total Anomalous Pulmonary Venous Return (TAPVR)

- **EKG:**
  - rSR’ pattern in V1

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

- **Treatment:**
  - Surgery
Total Anomalous Pulmonary Venous Return (TAPVR)

- **Clinical Manifestations (obstructed pulmonary veins)**
  - Marked cyanosis and respiratory distress in the neonatal period with FTT

- **Murmur**
  - May be absent, or faint systolic ejection murmur at the ULSB

- **CXR**
  - Lung fields show pulmonary edema (may be confused with pneumonia or hyaline membrane disease)
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
  - Murmurs
    - 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
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 – Lasix
  ▫ Surgical repair at approximately 4 months of age.

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