Congenital Heart Disease
An Approach for Simple and Complex Anomalies

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Disclosures

• None
ASCeXAM
• Contains questions on general congenital heart disease, not “adult” CHD
• Study guide contain all of the information in this talk plus addition topics that will be helpful for the exam
• There have been a few questions on fetal echo which have appeared on the ASCeXAM which are covered in the handout
• Insider information provided in study guide – topics that have appeared on prior exams (last page of study guide)

Which heart defect is more common?
1. Tetralogy of Fallot
2. Transposition of the Great Arteries
3. Aortic Stenosis
4. Coarctation of the Aorta
5. Atrioventricular Canal
What is the most common defect seen with Down syndrome (trisomy 21) ?

1. Patent Ductus Arteriosus
2. Tetralogy of Fallot
3. Aortic Stenosis
4. Ventricular septal defect
5. Coarctation of the Aorta

Congenital Heart Disease
Spectrum of Congenital Heart Disease - Incidence

- 0.5–0.8% of live births *
- >30,000 individuals/year in U.S.
- 50% simple shunts (ASD, VSD, PDA)
- 20% simple obstruction
- 30% complex

* Excludes MVP (4-6%) & Bicuspid AV (1-2%)
### Congenital Heart Disease

**Spectrum of Congenital Heart Disease - Frequency**

<table>
<thead>
<tr>
<th>Cardiac Malformation</th>
<th>% of CHD</th>
<th>M:F Ratio</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ventr. Septal Defect</td>
<td>18-28</td>
<td>1:1</td>
</tr>
<tr>
<td>Patent Ductus Arter.</td>
<td>10-18</td>
<td>1:2-3</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>10-13</td>
<td>1:1</td>
</tr>
<tr>
<td>Atrial Septal Defect</td>
<td>7-8</td>
<td>1:2-4</td>
</tr>
<tr>
<td>Pulmonary Stenosis</td>
<td>7-8</td>
<td>1:1</td>
</tr>
<tr>
<td>Transp. of Grt. Art.</td>
<td>4-8</td>
<td>2-4:1</td>
</tr>
<tr>
<td>Coarctation of Aorta</td>
<td>5-7</td>
<td>2-5:1</td>
</tr>
<tr>
<td>AV Septal Defect</td>
<td>2-7</td>
<td>1:1</td>
</tr>
<tr>
<td>Aortic Stenosis</td>
<td>2-5</td>
<td>4:1</td>
</tr>
<tr>
<td>Truncus Arteriosus</td>
<td>1-2</td>
<td>1:1</td>
</tr>
<tr>
<td>Tricuspid Atresia</td>
<td>1-2</td>
<td>1:1</td>
</tr>
<tr>
<td>Tot. Anom Pulm Veins</td>
<td>1-2</td>
<td>1:1</td>
</tr>
</tbody>
</table>

### Congenital Heart Disease

**Common Syndromes/Chromosomal Anomalies**

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>Associations</th>
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</thead>
<tbody>
<tr>
<td>Trisomy 21</td>
<td>VSD, AV Canal</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>VSD, PDA</td>
</tr>
<tr>
<td>Trisomy 13</td>
<td>VSD, PDA, Dextrocardia</td>
</tr>
<tr>
<td>Turner</td>
<td>Coarctation, AS</td>
</tr>
<tr>
<td>Noonan</td>
<td>PS, HCM</td>
</tr>
<tr>
<td>Williams</td>
<td>Supravalvar AS, Peripheral PS</td>
</tr>
<tr>
<td>Holt-Oram</td>
<td>ASD</td>
</tr>
<tr>
<td>Marfan</td>
<td>Aortic root dilation, MVP</td>
</tr>
<tr>
<td>DiGeorge</td>
<td>VSD, arch anomalies, TOF</td>
</tr>
</tbody>
</table>
Congenital Heart Disease

- Currently over 1 million patients over 18 with CHD are alive in this country
- This increases at 4-5% per year
- 20,000 cardiac surgical procedures for CHD/year
- >90% of children with CHD survive to adulthood
- The majority of adult CHD patients will be post-op
  - The ASCeXAM does not cover much post-op disease, but has asked questions about operations

Congenital Heart Disease
A Brief History of Operations

<table>
<thead>
<tr>
<th>Year</th>
<th>Physician</th>
<th>Procedure</th>
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</thead>
<tbody>
<tr>
<td>1938</td>
<td>Gross</td>
<td>Ligation of PDA</td>
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<tr>
<td>1944</td>
<td>Blalock, Taussig</td>
<td>Syst.-pulm. shunt</td>
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<tr>
<td>1945</td>
<td>Gross, Crafoot</td>
<td>Repair of coarctation</td>
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<tr>
<td>1946</td>
<td>Potts</td>
<td>Aorta to LPA shunt/direct anastomosis</td>
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<tr>
<td>1952</td>
<td>Muller</td>
<td>Pulm. artery band</td>
</tr>
<tr>
<td>1953</td>
<td>Gibbon</td>
<td>Repair of ASD</td>
</tr>
<tr>
<td>1954</td>
<td>Lillehei</td>
<td>Repair of VSD</td>
</tr>
<tr>
<td>1954</td>
<td>Glenn</td>
<td>SVC-PA shunt</td>
</tr>
<tr>
<td>1956-7</td>
<td>Mustard/Senning</td>
<td>Atrial correction of TGA</td>
</tr>
<tr>
<td>1955</td>
<td>Lillehei, Kirlin</td>
<td>Repair of tetralogy of Fallot</td>
</tr>
<tr>
<td>1960</td>
<td>Waterston</td>
<td>Aorta to RPA shunt/direct anastomosis</td>
</tr>
<tr>
<td>1964</td>
<td>Rastelli</td>
<td>Conduit replacement of PA</td>
</tr>
<tr>
<td>1967</td>
<td>Rashkind</td>
<td>Balloon atrial septostomy</td>
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<tr>
<td>1971</td>
<td>Fontan, Kreutzer</td>
<td>Repair of tricuspid atresia</td>
</tr>
<tr>
<td>1976</td>
<td>Jatene</td>
<td>Arterial switch for TGA</td>
</tr>
<tr>
<td>1978</td>
<td></td>
<td>Cold blood cardioplegia</td>
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</tbody>
</table>
Case 1 - 3 month old with a murmur, tachypnea and failure to thrive
The defect shown in this example is:

1. Secundum VSD
2. Sinus Venosus VSD
3. Perimembranous VSD
4. Inlet VSD
5. Supracristal VSD

An isolated VSD will generally produce enlargement of which chamber(s):

1. Left atrium, Left ventricle
2. Right ventricle
3. Right ventricle, pulmonary artery
4. Aorta
5. Right ventricle, right atrium
What is the right ventricular pressure?

1. Normal
2. Supra systemic
3. Systemic
4. Can’t tell from information given
5. Want to go home now

Ventricular Septal Defect
Clinical

- Most common defect, 25% of CHD
- Shunt flow usually left to right in infants and children
- Symptoms depend on the size of the defect
  - Large - >50% of aortic annulus size
  - Medium - 25-50% of annulus size
  - Small - <25% of annulus size
- Large VSDs result in pulmonary edema → tachypnea, poor feeding, failure to thrive in infants
- In un-operated patients with large defects pulmonary vascular disease develops → shunt reversal and cyanosis (Eisenmenger’s complex)
Ventricular Septal Defect
Anatomy

1 PERIMEMBRANOUS
2 SUPRACRISTAL
3 MUSCULAR
4 INLET

Defects seen in this region are perimembranous
Supracristal VSD's are seen here
Ventricular Septal Defect
Parasternal Short-Axis - Mid-Ventricle

Muscular (trabecular) VSD's can be seen anywhere along the septum from this view.

Ventricular Septal Defect
Parasternal Short-Axis "Sweep"

VSD
**Ventricular Septal Defect**

**Apical Four-Chamber View**

- Apical muscular VSD's can be seen from this view. Size may vary from tiny to large.
- Inlet VSD's are best seen from this view.

**Apical Five-Chamber View**

- This view nicely demonstrates a perimembranous VSD and its relationship to the aortic and tricuspid valves.
Case 1 - Review

Ventricular Septal Defect
Case 2 – 8 y.o. with asymptomatic murmur
The defect shown in this example is:

1. Secundum VSD
2. Sinus Venosus VSD
3. Perimembranous VSD
4. Inlet VSD
5. Supracristal VSD

Question 10 - A common complication of this defect is:

1. Pulmonary valve endocarditis
2. Aortic regurgitation
3. Aortic dissection
4. Tricuspid regurgitation
5. Right ventricular enlargement
Case 2 - Review

Supracristal VSD
Aortic Cusp Prolapse
Ventricular Septal Defect
Case 3 – No questions
Ventricular Septal Defect
Case 3 – No questions
Case 4 – 6 y.o. with continuous murmur
Case 4 – 6 y.o. with continuous murmur

Peak Gradient = 104 mmHg
The Doppler tracing in this case implies:

1. Severe pulmonary hypertension
2. Severe systemic hypertension
3. Severe coarctation of the aorta
4. Normal pulmonary artery pressure
5. Severe pulmonary artery stenosis

Patent Ductus Arteriosus
Clinical
• Continuous murmur in older patients
• Bounding pulses, wide pulse pressure, respiratory symptoms in neonates with a large PDA
• Large PDA will act much like a large VSD, producing pulmonary over-circulation and signs/symptoms of congestive heart failure
• A small PDA is generally hemodynamically insignificant but is at risk for endarteritis
Patent Ductus Arteriosus

Anatomy

Patent Ductus Arteriosus

Ductal View
Parasternal Ductal View
(High Left Parasternal)
Patent Ductus Arteriosus
Doppler Flow Pattern

Systolic PA pressure = SBP − 4V_{PDA}^2

Clinical management

- Large PDAs in preterm infants
  - Pharmacologic closure – indomethacin or ibuprofen
  - Surgical closure – left lateral thoracotomy

- Small PDA in older infants and children
  - Catheter closure – device or coil
Case 4 - Review

Peak Gradient = 104 mmHg
Case 5 – Large PDA
Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur
The defect shown in this example is:

1. Single ventricle
2. Transposition of the great arteries
3. Perimembranous VSD
4. Tetralogy of Fallot
5. Complete atrioventricular canal

Tetralogy of Fallot

Background

- Accounts for 10-13% of congenital heart disease
- Most common cyanotic CHD
- Usually present as asymptomatic murmur
  - Cyanosis usually develops/progresses with time

Anatomy

- Ventricular Septal Defect
- Overriding Aorta
- RV outflow obstruction
- RV hypertrophy

Conal Septum Malalignment
Tetralogy of Fallot
Anatomic Variables

“Pink Tetralogy”
Mild Pulmonary Stenosis

Classic Tetralogy

Severe Tetralogy or Pulmonary Atresia

Tetralogy of Fallot
Associated Anomalies

- Valvular pulmonary stenosis - 50-60%
- Right aortic arch - 25%
- Atrial septal defect - 15%
- Coronary anomalies - 5%
- Muscular VSD - 2%
Tetralogy of Fallot
Parasternal Long Axis View

"Malalignment" VSD with Aortic Override
RV Hypertrophy

Tetralogy of Fallot
Parasternal Short Axis - Base

Outflow VSD "Malalignment" type
RV Hypertrophy
Infundibular & Valvular Stenosis
Position of "Overriding" Aorta
Tetralogy of Fallot
Apical Five-Chamber

Outlet VSD with Aortic Override

Subcostal RV Inflow/Outflow View

The right ventricular outflow obstruction is seen in this view. This is often the best angle for Doppler interrogation of the RVOT.
Tetralogy of Fallot
Surgical Intervention

- Timing – usually during first 6 months
- VSD closure, relief of RVOTO obstruction
- Many repairs require a trans annular RV outflow patch with results in chronic severe pulmonary regurgitation
  → Likely need for late pulmonary valve replacement
- Rastelli type repair (VSD closure + RV to pulmonary artery conduit) may be required for complex anatomy – pulmonary atresia, coronary anomalies
Case 7 – 1 day old infant with tachypnea and SaO2 of 76%
What congenital heart defect is shown:

1. Perimembranous VSD
2. Truncus arteriosus
3. Corrected transposition of the great arteries (L-TGA)
4. Complete transposition of the great arteries (D-TGA)
5. Tetralogy of Fallot

Which of the following is the preferred surgical palliation of this defect?

1. Rastelli operation
2. Mustard operation
3. Jatene operation
4. Konno operation
5. Fontan operation
D-Transposition of the Great Arteries

- Most common cyanotic CHD presenting in the newborn nursery
- 4-8% of CHD
- Very high mortality without intervention (90% at 1 year of life)

D-TGA Intact Septum

D-TGA w/ VSD

D-Transposition of the Great Arteries

Associated Anomalies

- VSD 40-45%
- Coronary anomalies ~ 40%
- Pulmonary stenosis (valve or sub valve) - 25%
- ASD
- PDA
- Coarctation - 5%
D-Transposition of the Great Arteries
Parasternal Long Axis View

D-Transposition of the Great Arteries
Parasternal Short Axis - Base
D-Transposition of the Great Arteries
High Parasternal Short Axis - Base

Case 7-Review
D-Transposition of the Great Arteries

Interventions

- Atrial Balloon Septostomy (Rashkind)
  - ♥ Increases saturations by improving atrial mixing
- Arterial switch (Jatene)
  - ♥ Performed within first 1-2 weeks of life
- Atrial switch (Mustard/Senning)
  - ♥ Has been largely abandoned
- VSD closure/ RV-PA conduit (Rastelli)
  - ♥ Performed within the first few months of life
  - ♥ Used in the setting of d-TGA with PS/sub-PS
  - ♥ Requires conduit replacement/ future surgery
D-TGA
Atrial Switch (Mustard/Senning) Operation

TGA
Jatene Arterial Switch Operation

http://radiology.rsna.org
Case 8 – 12 year old with asymptomatic murmur
Case 8 – 12 year old with asymptomatic murmur

The defect shown in this example is:

1. Secundum ASD
2. Sinus Venosus ASD
3. Perimembranous ASD
4. Primum ASD
5. Coronary sinus ASD
A common associated defect with this anomaly, shown in this case, is:

1. Bicuspid aortic valve
2. Perimembranous VSD
3. Patent ductus arteriosus
4. Coarctation of the aorta
5. Cleft mitral valve

Atrioventricular Septal Defects

-3-5% of CHD
-High incidence in Down Syndrome
-Physiology depends on which anatomic defects are present

Complete AVSD
1. Primum ASD
2. Inlet VSD
3. Common AV Valve

Partial AVSD
1. Primum ASD
2. No VSD
3. Cleft Mitral Valve
Complete Atrioventricular Canal
Associated Anomalies

- Patent ductus arteriosus
- Hypoplasia of one ventricle
- AV valve problems - regurgitation
- LVOT obstruction

Atrioventricular Canal
Long Axis View

Mitral valve has abnormal orientation and often has abnormal chordal attachments across the LV outflow area
Cleft Mitral Valve
Parasternal Short-Axis View

Normal
Inlet VSD's occur in this location
Cleft

Atrioventricular Canal
Apical 4-Chamber View

Partial
Primum ASD
AV Valves at same level

Complete
Inlet VSD
Atrioventricular Canal Defects

Subcostal Views

Primum ASD and Inlet VSD

"Gooseneck" deformity

Subcostal Short Axis Views

Wide Antero-septal Tricuspid Commissure

"Common" AV Valve

Gleef Mitral Valve
Case 8- Review
Case 9 – Complete AV Canal

Case 9 – Complete AV Canal
AV Septal Defects

Physiology

- Physiology dependent on which components of AV septal defect are present
- If $1^\circ$ ASD and no VSD - physiology similar to isolated ASD (right sided volume overload)
- Complete AVSD - marked volume and pressure overload (VSD shunt physiology)
- AV valve regurgitation may exacerbate volume overload and symptoms of heart failure

AV Septal Defects

Surgical Intervention

- Partial AVSD
  - Usually electively repaired at age 2-4 years
  - Complicating features (AVV regurgitation, LVOTO) may necessitate earlier intervention
- Complete AVSD
  - Usually repaired by 6 months of age (earlier in trisomy 21) to prevent pulmonary vascular obstructive disease
• Surgical Outcomes
  → 3-4% operative mortality for complete AVSD
  → <1% operative mortality for partial AVSD
• Late reoperation in 10-15%
  → Left AV valve regurgitation
  → Left ventricular outflow tract obstruction
    ♥ Hypoplasia of the outflow tract
    ♥ Accessory AV valve tissue
    ♥ Discrete subaortic membrane
Truncus Arteriosus
Anatomy

• Characteristic anatomy characterized by:
  → Single arterial vessel that arises from the base of the heart and gives origin to:
    ♥ Systemic arteries
    ♥ Pulmonary arteries
    ♥ Coronary arteries
  → Single semilunar valve

Truncus Arteriosus
Associated Defects

• Abnormal coronaries (37-49%)
• Right aortic arch (30%)
• Abnormal truncal valve
• Absent pulmonary artery (16%)
• Interrupted aortic arch (15%)
• Left SVC (12%)
• Secundum ASD (9-20%)
Truncus Arteriosus
Clinical Aspects

- Patients usually present due to the presence of a cardiac murmur
- Complete mixing of systemic and pulmonary venous blood results in cyanosis
- Excessive pulmonary blood flow causes signs and symptoms of congestive heart failure
- The cyanosis is generally mild

Truncus Arteriosus
Treatment

- Requires surgical repair in the first weeks of life
- Median sternotomy requiring bypass
- Palliative PA band rarely used in the current era
- Rastelli type repair
  - Close VSD to truncus
  - Disconnect PAs
  - RV to PA conduit
Case 10
Truncus Arteriosus

Case 10
Truncus Arteriosus
Anomalous Left Coronary Artery
From the Pulmonary Artery - ALCAPA

• Rare congenital anomaly
• Usually presents at 2-3 m of age
• Results in severe LV ischemia
• Present as dilated CM, CHF
• ECG often diagnostic
• Patients survive w/ collateral flow
• Surgery done to re-implant vessel
Anomalous Left Coronary Artery
Echocardiographic Clues

• Left ventricular dysfunction (usually severe)
• Mitral insufficiency – due to LV dilation/dysfunction, papillary muscle infarction
• Endocardial fibroelastosis of LV and/or papillary muscles
• Failure to identify proximal LCA from aorta
• Unusual flow into main pulmonary artery

Case 11
ALCAPA
Total Anomalous Pulmonary Venous Return
Anatomic Types

- Supra cardiac
- Infracardiac
- Cardiac
- Coronary sinus
Total Anomalous Pulmonary Venous Return
Echo Clues

- Enlarged right heart
- Right to left atrial shunting
- Unusual “membranes” in left atrium
- Abnormal flow in systemic venous system
- Obstruction may occur at different levels
  → Most common - infracardiac
- May be remarkably asymptomatic (in absence of obstruction)

Case 12
Supra cardiac TAPVR
Good Luck On Your Exam

Questions?
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