Congenital Heart Disease
An Approach for Simple and Complex Anomalies

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Disclosures

• Consultant to Fuji Medical Imaging
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 19%
2. Transposition of the Great Arteries 20%
3. Aortic Stenosis 22%
4. Coarctation of the Aorta 19%
5. Atrioventricular Canal 19%
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

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<th>% of CHD</th>
<th>M:F Ratio</th>
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<td>Truncus Arteriosus</td>
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<tr>
<td>Tot. Anom Pulm Veins</td>
<td>1-2</td>
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## Congenital Heart Disease

### Common Syndromes/Chromosomal Anomalies

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<th>Associations</th>
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<td>Trisomy 13</td>
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<td>Supravalvar AS, Peripheral PS</td>
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<td>Holt-Oram</td>
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<tr>
<td>Marfan</td>
<td>Aortic root dilation, MVP</td>
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<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
Case 1 - 3 month old with a murmur, tachypnea and failure to thrive
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):

0% 1. **Left atrium, Left ventricle**
0% 2. Right ventricle
0% 3. Right ventricle, pulmonary artery
0% 4. Aorta
0% 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 should be left to right
• Symptoms depend on the size of the hole
  → 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

RIGHT VENTRICULAR VIEW
LEFT VENTRICULAR VIEW

SVC, MPA, RA, IVC, RV, AO, LA, LV
Ventricular Septal Defect
Parasternal Short Axis - Base

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

Muscular (trabecular) VSD's can be seen anywhere along the septum from this view.
Ventricular Septal Defect
Parasternal Short-Axis "Sweep"
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.
Ventricular Septal Defect
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:

0% 1. Pulmonary valve endocarditis
0% 2. Aortic regurgitation
0% 3. Aortic dissection
0% 4. Tricuspid regurgitation
0% 5. Right ventricular enlargement
Case 2 - Review
Supracristal VSD
Aortic Cusp Prolapse

RV
AO
LV
LA
Ventricular Septal Defect

Case 3 – No questions
Ventricular Septal Defect

Case 3 – No questions
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
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

Innom. Vein → PDA

AO → MPA → LPA → Desc AO
Patent Ductus Arteriosus

Ductal View
Parasternal Ductal View
(High Left Parasternal)
Patent Ductus Arteriosus

Doppler Flow Pattern

Systolic PA pressure = SBP – 4V_{PDA}^2
Patent Ductus Arteriosus

Clinical management

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

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

Peak Gradient = 104 mmHg
Case 5 – Large PDA
Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur
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

Mitral-Aortic Continuity

RV
LV
LA
AO
CS
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
The right ventricular outflow obstruction is seen in this view. This is often the best angle for Doppler interrogation of the RVOT.
Case 6 - Review
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:

0% 1. Perimembranous VSD
0% 2. Truncus arteriosus
0% 3. Corrected transposition of the great arteries (L-TGA)
0% 4. Complete transposition of the great arteries (D-TGA)
0% 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-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

RA

AO

PA

RA
Case 7-Review
D-Transposition of the Great Arteries
Surgical Options

- Arterial switch (Jatene)
  - within first 1-2 weeks of life
- Atrial switch (Mustard/Senning)
  - Has been largely abandoned
- VSD closure/ RV-PA conduit (Rastelli)
  - within first few months of life
  - Used in the setting of d-TGA with PS/sub-PS
  - requires conduit replacement/ future surgery
D-TGA

Intervention-Rastelli Procedure
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

Normal

AV Canal

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

- RA
- LA
- RV
- LV

Complete

- RA
- LA
- RV
- LV

Primum ASD

AV Valves at same level

Inlet VSD
Atrioventricular Canal Defects

Subcostal Views

LA

RA

LV

RV

AO

"Gooseneck" deformity

Primum ASD and Inlet VSD
Atrioventricular Canal Defects

Subcostal Short Axis Views

Diastole

Systole

RVOT

RV

LV

Cleft Mitral Valve

Wide Antero-septal Tricuspid Commissure

"Common" AV Valve

"Common" AV Valve

Systole
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° 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 age 2-4 years
  ➔ Complicating features (AVV regurg., LVOTO) may necessitate earlier intervention

• Complete AVSD
  ➔ Usually repaired by 6 months of age (earlier in trisomy 21) to prevent pulmonary vascular obstructive disease
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

Anatomy

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 sign 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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