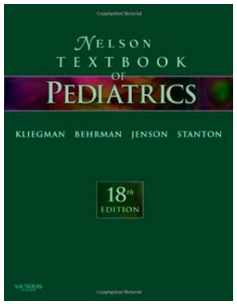
# Congenital Heart Disease: Physiology and Common Defects

Jamie S. Sutherell, M.D, M.Ed.
Associate Professor, Pediatrics
Division of Cardiology
Director, Medical Student Education in Pediatrics
Director, Pediatric Cardiology Fellowship Program

## Congenital Heart Disease




- Pages on congenital heart disease: 145
- Pages on acquired heart disease: 23
- Incidence of congenital heart disease: ~1%

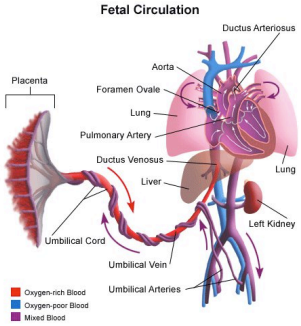
## Congenital Heart Disease

- Objectives:
  1. Describe transitional physiology related to the cardiovascular system: fetus, newborn, child
  2. Describe common congenital heart defects and explain what symptoms will occur and when they will occur
  3. Explain the treatment rationale for common congenital heart defects, including medications and interventions

## Congenital Heart Disease

1. Physiology, physiology, physiology
2. Physical exam
3. Then. . . Pathophysiology
4. Understanding over memorization

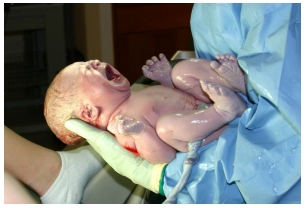
## Dynamic Transitions in Physiology




a) Fetal Circulation
- "Low" Systemic Vascular Resistance (SVR) (placenta)
- "High" Pulmonary Vascular Resistance (PVR) (deflated lungs)
- SVR = PVR
- Fetal structures:
  - Ductus venosus (DV)
  - Foramen ovale (FO)
  - Ductus arteriosus (DA)

## Dynamic Transitions in Physiology

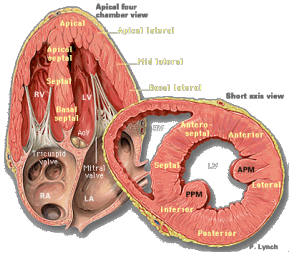
b) Newborn Physiology
- Constriction of umbilical vessels = increasing SVR
- Inflation of lungs = decreasing PVR
- Closure of ductus arteriosus and foramen ovale
- Changes occur in hours-days



## Dynamic Transitions in Physiology

c) Infant Physiology
- Higher SVR = thickening of LV
- Lower PVR = thinning of RV

## Dynamic Infant Physiology

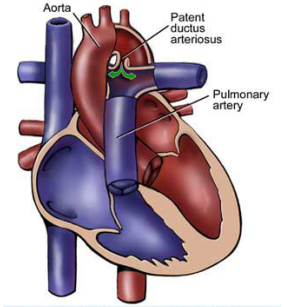



- LV:RV Weight Ratio

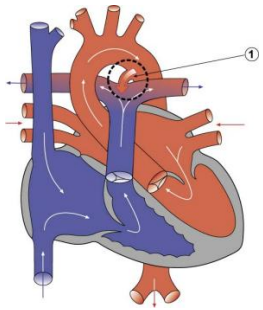
| Age | Ratio |
|---|---|
| Birth | 0.8:1 |
| 1 Month | 1.5:1 |
| 6 Months | 2:1 |
| Adult | 2.5:1 |

## Failure in Transition

- What if Ductus Arteriosus does not close ("Patent")?
  - PVR vs. SVR
  - Aorta vs. pulmonary pressure
  - Symptoms? When?



## Patent Ductus Arteriosus




- Continuous murmur, subclavicular area
- Diastolic "spilling" of blood from aorta to PA
  - Drops diastolic pressure in aorta
  - "Bounding" pulses
- Small PDA: diastolic component may be very quiet

## Patent Ductus Arteriosus

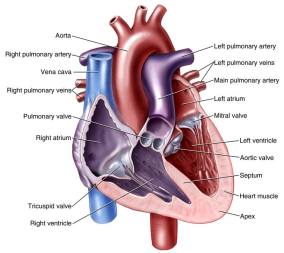
- Symptoms:
  - Tachypnea, diaphoresis
  - Poor feeding
  - Poor weight gain
- Treatment:
  - Indomethacin
  - Diuretics, digoxin, ACE-I
  - Surgical ligation
  - Catheter occlusion

## Cardiac Physical Exam

- The cardiac exam DOES NOT start with listening for murmurs. . .
  - Inspection
  - Palpation
  - Auscultation
    - Heart sounds (S1, S2; possibly S3, S4)
    - Clicks
    - ***Murmurs***

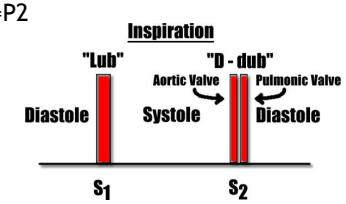
## Cardiac Physical Exam

- S1: closure of AV valves (mitral, tricuspid)
- S2: closure of semilunar valves (aortic, pulmonic)
- Systole: between S1 and S2
- Diastole: between S2 and next S1



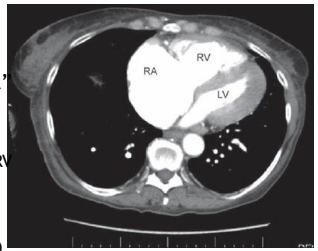

## Second Heart Sound

- "Physiologic" Split S2
  - Inspiration: P2 delayed
  - Expiration (aka dead): No heart sounds
  - Exhalation: A2=P2



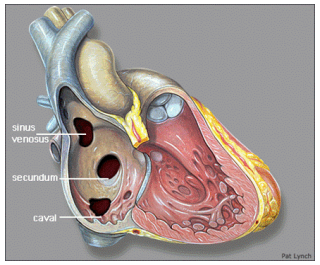

## Second Heart Sound

- What if S2 is always split?
  - No physiologic variability="fixed split"
  - DDx:
    - Volume load to RV
    - Delayed electricity to RV
      - RBBB
    - "Early" closure of P2
      - Pulmonary HTN ("paradoxic" split S2)

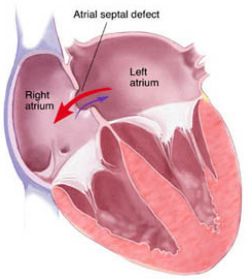


## Atrial Septal Defect

- 10% of all CHD
- 3 distinct types:
  1. Ostium secundum (60%)
     - Involves fossa ovalis
  2. Ostium primum (30%)
     - "AV canal-type"
     - Often with cleft mitral valve
  3. Sinus venosus (10%)
     - Often with RUPV PAPVR



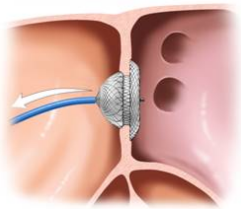

## Atrial Septal Defect




- Early-mid systolic ejection murmur at LUSB
  - = pulmonary flow murmur
- *"Fixed split S2"*
- Symptoms: ~lower side of growth curve
- CXR: cardiomegaly, increased PVM's
- ~40% secundum ASD close by age 4 years

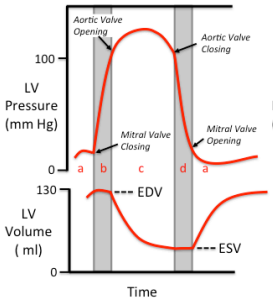
## Atrial Septal Defect

- Treatment:
  - Surgery (primum, sinus venosus, some secundums)
  - Interventional cath



## Cardiac Timing

- Timing: Cardiac Cycle
  - Systole: S1, isovolumic contraction, semilunar valves open
  - Diastole: S2, isovolumic relaxation, atrioventricular valves open

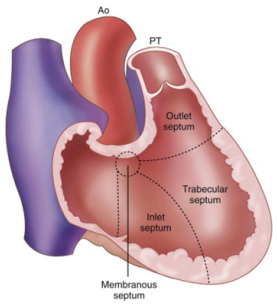



## Murmurs

- Isovolumic Contraction
  - Which valves are open?
  - Can you hear valve stenosis?
  - Regurgitation?
  - ="Holosystolic" murmur
  - DDx:
    - Mitral regurgitation
    - Tricuspid regurgitation
    - Ventricular septal defect

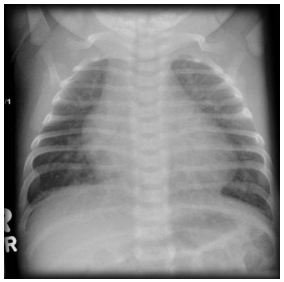

## Ventricular Septal Defect

- Most common CHD
  - 20% of all CHD
- Comprised of 3 embryologic structures:
  1. Trabecular (muscular) septum
  2. Inlet septum (endocardial cushion)
  3. Outlet (conal) septum
- Fusion of septum at membranous septum

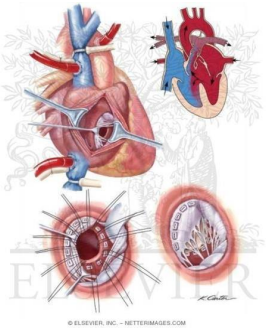


## Ventricular Septal Defect

- "Holosystolic" murmur
  - Pitch/volume varies based on pressure difference from LV to RV
- Symptoms: pulmonary over-circulation
  - Tachypnea, tachycardia
  - Poor feeding
  - Poor weight gain
- CXR: cardiomegaly, increased PVM's

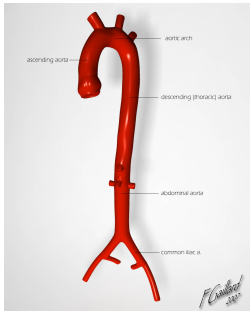


## Ventricular Septal Defect




- Treatment:
  - Diuretics, digoxin, ACE-I
  - Augment nutrition
  - Surgery (versus cath occlusion)

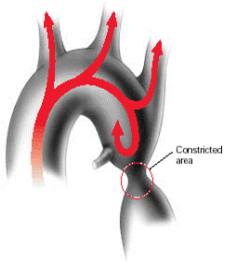
## Cardiac Physical Exam



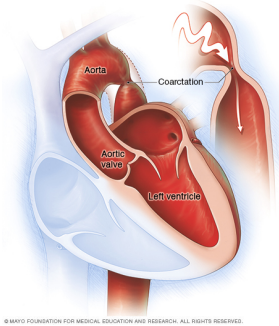

- Palpation of Pulses
  - Bounding?
    - Wide pulse pressure (i.e. PDA, aortic regurgitation)
  - Diminished?
    - Diffusely?
    - Some normal, some diminished?

## Coarctation of the Aorta

- 5-7% CHD
- Typically discrete stenosis of thoracic aorta
  - "Juxtaductal" location
- Embryology: migration of ductal tissue into DAo




## Coarctation of the Aorta

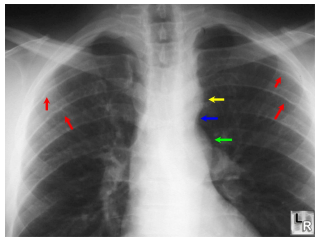


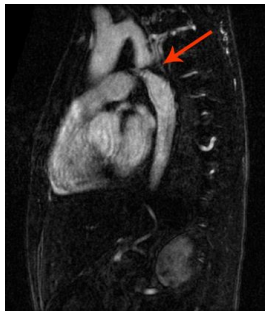



- Symptoms:
  - Relates to increased LV pressure: Tachypnea
- Diagnosis:
  - Weak femoral pulses
  - Brachiofemoral delay
  - Poor LE perfusion
- Age at repair: ~ 1 week
  - Elective if diagnosed older

## Coarctation of the Aorta

- Older Children:
  - Asymptomatic
  - Right arm HTN

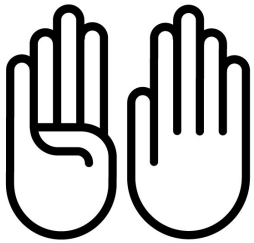




## Cyanosis: The Five "Terrible T's"

- Truncus Arteriosus
- Transposition of the Great Arteries (TGA)
- Tricuspid Atresia
- Tetralogy of Fallot (TOF)
- Total Anomalous Pulmonary Venous Return (TAPVR)

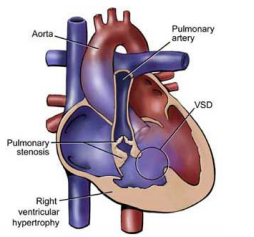
## Cyanosis: The Five "Terrible T's"



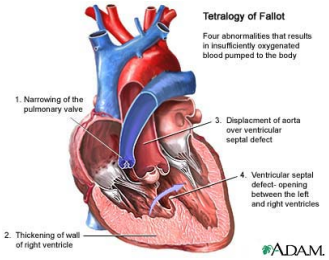
- Only part of the story. . .
  - Pulmonary atresia with VSD
  - Pulmonary atresia with intact ventricular septum
  - Critical pulmonic stenosis
  - Ebstein's anomaly
- Physiology:
  - Can't get blue blood to lungs ----OR----
  - Complete mixing

## Tetralogy of Fallot

- Most common cyanotic defect
- Tetralogy:
  1. VSD
  2. Overriding Aorta
  3. RVOT Obstruction
  4. RVH
- Variable degree of RVOTO
  - = variable cyanosis (pink to blue)



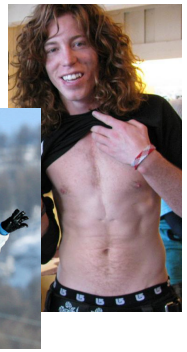
## Tetralogy of Fallot




- Cyanosis: reduced pulmonary blood flow (PBF) (seen on CXR)
- Cyanotic: loud murmur at ULSB (RVOT obstruction)
- Acyanotic: loud holosystolic murmur at LLSB (VSD)

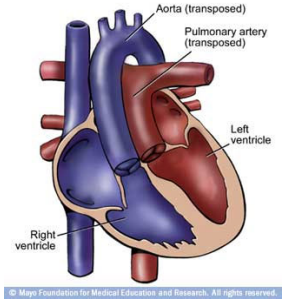
## Tetralogy of Fallot

- Treatment:
  - Beta blocker
  - "Tet" spell=emergency
    - Extreme cyanosis, no murmur
- OR: ~4-6 months depending on cyanosis



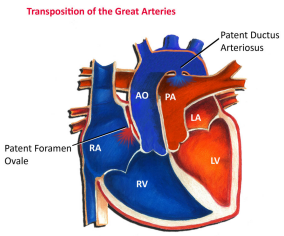
## Transposition of the Great Arteries

- "Ventriculo-arterial discordance"
  - Aka: incorrect ventricle-great vessel connection
- Parallel circulations
  - Hypoxic blood to body; hyperoxic blood to lungs
- Most common cyanotic defect to present in newborn

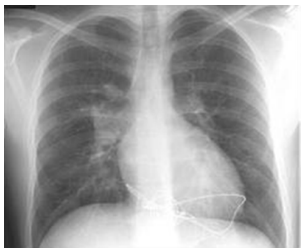


## Transposition of the Great Arteries

- Parallel circulations
  - Arterial desaturation in systemic bed
- Survival depends on ability to mix
  - ASD
  - VSD
  - PDA
- Spectrum of presentation
  - No mixing = profound desaturation



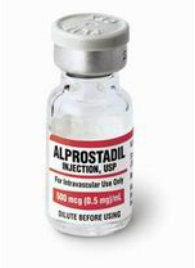

## Transposition of the Great Arteries



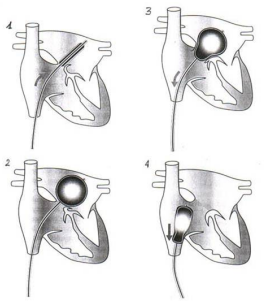
- No "characteristic" exam, ECG, or CXR finding
  - CXR: Narrow superior mediastinum "egg on a string"

## Transposition of the Great Arteries

- Treatment:
  - Enhance mixing: PGE1
- Surgery:
  - ~1 week of life
  - Arterial switch operation



## Transposition of the Great Arteries



- Cardiac catheterization: balloon atrial septostomy (BAS)
  - "Rashkind procedure"
  - Bedside or in catheterization lab
  - Through UVC or femoral vein

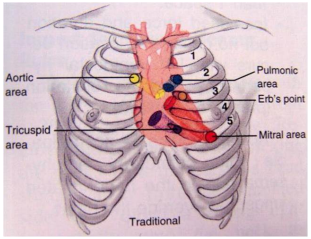
"All it requires is a big jerk on the other end of the catheter"
-Bill Rashkind

## Summary: Congenital Heart Disease

- Remember: the names tell you what the defect is (except. . .tetralogy of Fallot and Ebstein's anomaly)
- Predicting symptoms:
  1. Understand physiology
  2. Be a plumber
  3. Remember your exam

## Pearls: Physiology

- Dynamic newborn transitions: SVR, PVR; PDA, PFO; RV vs. LV
- Location of heart sounds
- Timing of cardiac cycle
  - Isovolumic contraction
- Physiologic split S2



## Pearls: Acyanotic CHD

1. **VSD:** most common CHD. Holosystolic murmur. Symptoms of pulmonary over-circulation.
2. **ASD:** 2nd most common CHD. Wide split S2. Few symptoms.

## Pearls: Acyanotic CHD

3. **PDA:** 3rd most common CHD. Continuous murmur, bounding pulses. Symptoms of pulmonary over-circulation.
4. **COA:** 4th most common CHD. Poor femoral pulses, BF delay. Infancy: tachypnea, shock; child: HTN

## Pearls: Cyanotic CHD

1. **TOF:** Most common CCHD. Variable cyanosis based on variable RVOTO.
2. **TGA:** Most common CCHD in infancy. Need mixing! May be extremely cyanotic, ductal dependent.

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