OSD
Congenital Heart Disease
Part I

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UMass Chan Medical School September 14, 2021

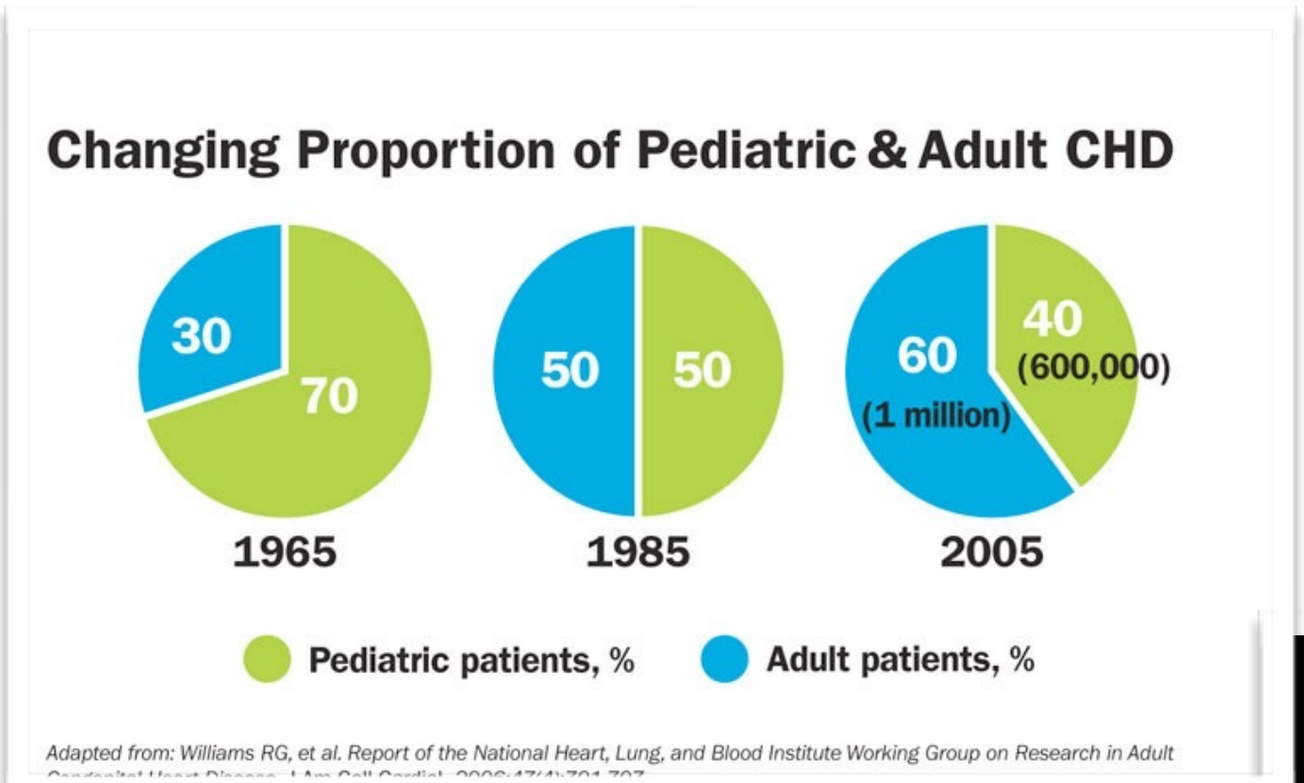
I have no conflicts to share.

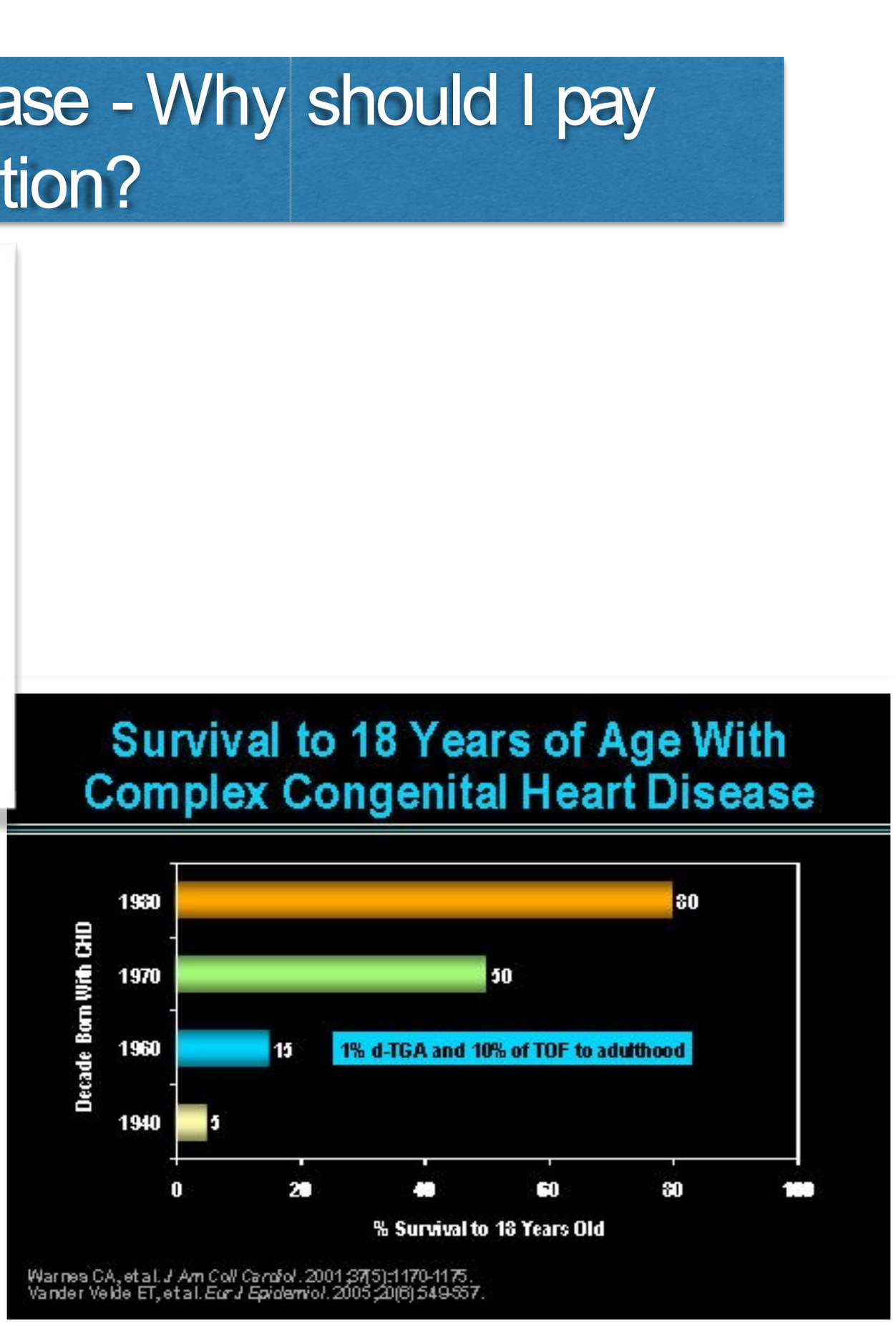
Fahey-OSD-2020

Acknowledgement

These slides were originally developed by Dr. David Kane. They have been edited for use in this presentation with his permission.

Congenital Heart Disease - Why should I pay attention?



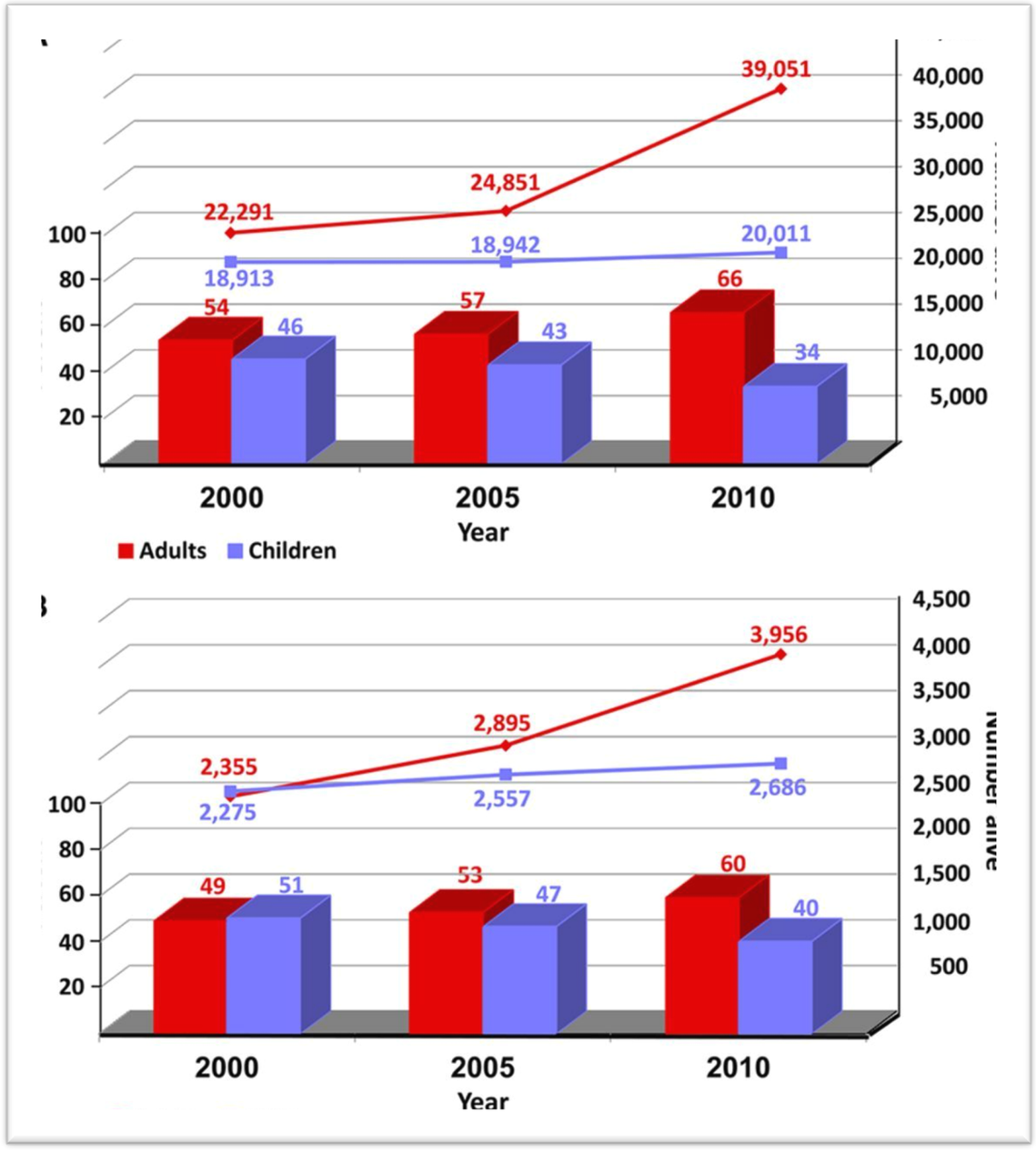


Most common cause of congenital anomalies (8/1000 live births)

2/3 of Patients with Congenital Heart Disease are Adults

Numbers and proportions of adults (red) and children (purple) with any congenital heart disease (top graph) and with severe congenital heart disease (bottom graph)

Data from Quebec, Canada



Marelli AJ, et. al Lifetime Prevalence of Congenital Heart Disease in the General Population from 2000 to 2010. *Circulation*, 2014; 130:749-756.

Objectives

- Describe differences in fetal circulation and post-natal circulation
- Explain how changes in resistance result in changes in flow
- Describe how the heart and vasculature respond to these changes
- Describe 3 forms of acyanotic congenital heart disease:
  - Atrial Septal Defect (ASD)
  - Ventricular Septal Defect (VSD)
  - Patent Ductus Arteriosus (PDA)

Overview

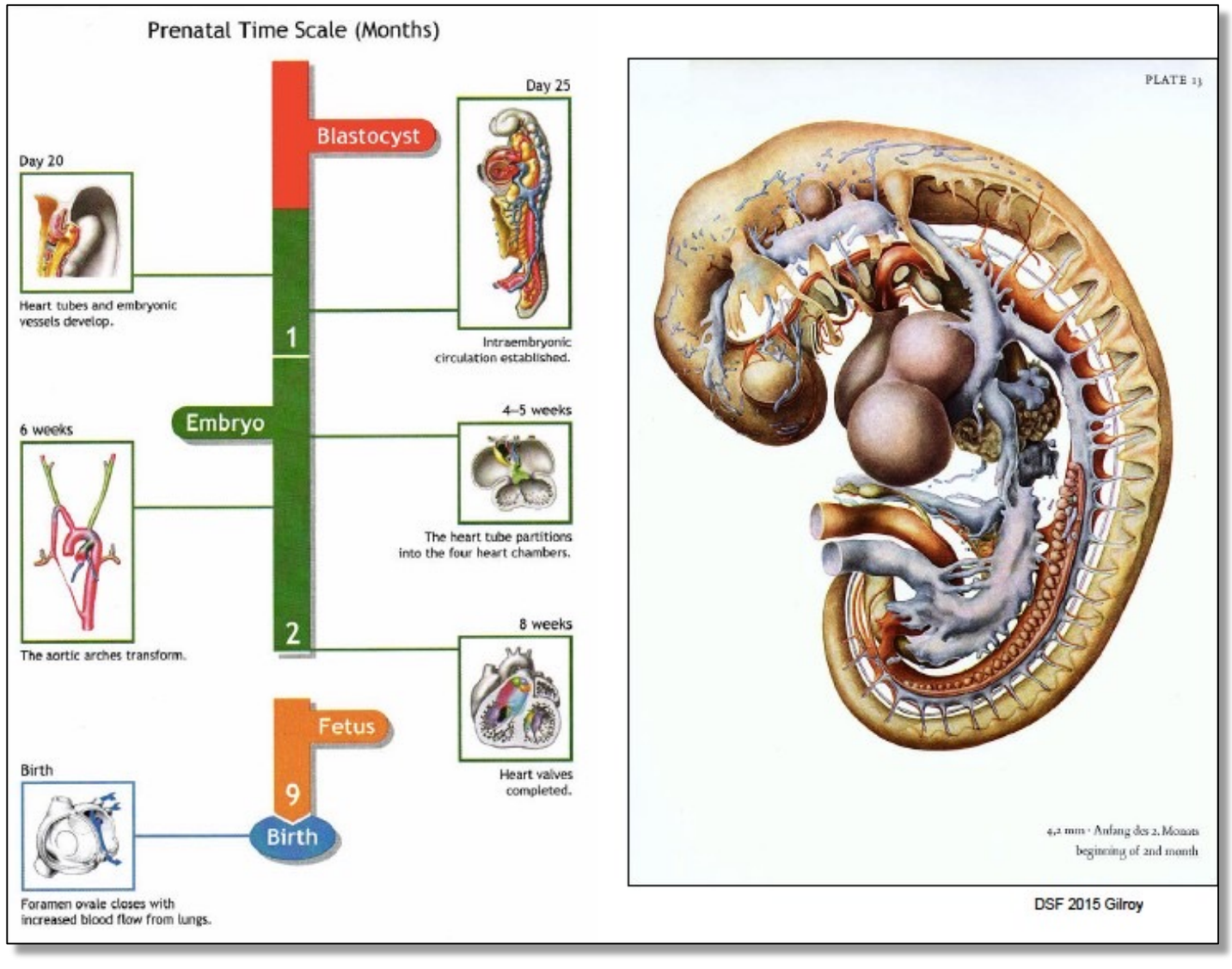
- Act I: Rules of the Game/Fetal Circulation
  - Describe how the fetal circulation works
  - Review concepts of vascular systems that determine flow and resistance

- Act II: Shunts, obstruction, and remodeling
  - Describe the different forms of remodeling and what stimulates them
  - Contrast the underlying mechanism of concentric versus eccentric hypertrophy

- Act III: Acyanotic Congenital Heart Disease
  - Give examples of 3 forms of left-to-right shunting lesions
  - Explain the hemodynamic challenges of these lesions
  - Describe the physiologic response to these challenges and the signs/symptoms they create

Act I: Rules of the Game/Fetal Circulation

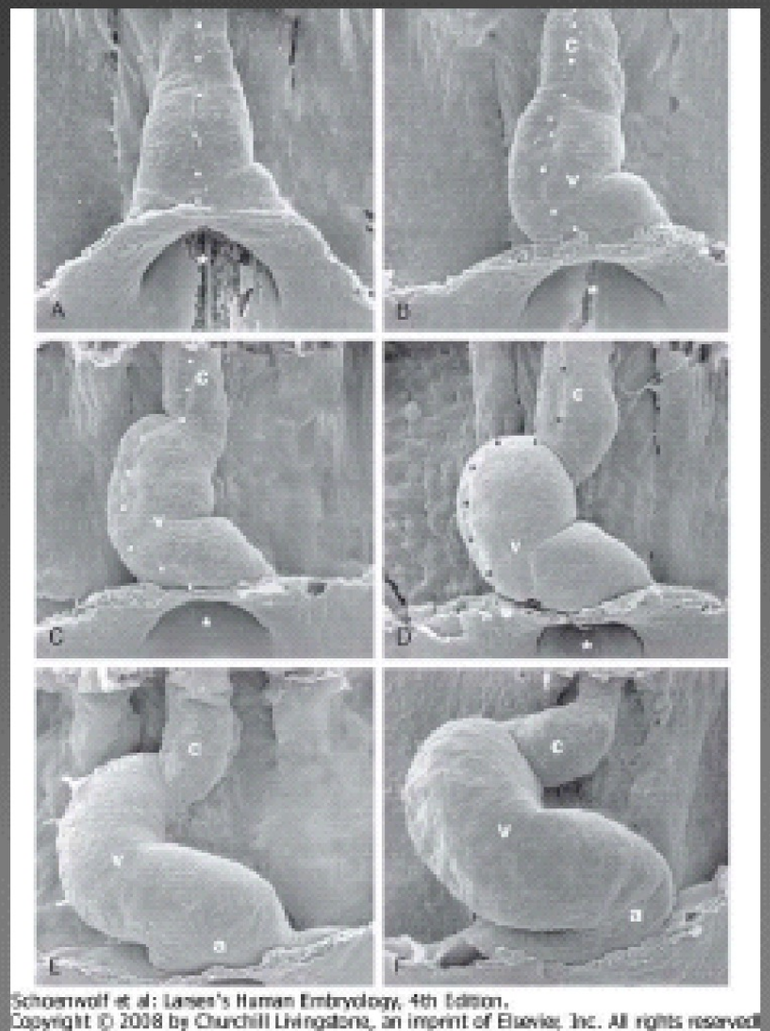
Cardiac Embryology



This material has been covered in more detail in your DSF Lecture on CV Development

There are so many steps that can go wrong in the process of cardiac development...

Picture of looping



Schoenwolf et al: Larsen's Human Embryology, 4th Edition.
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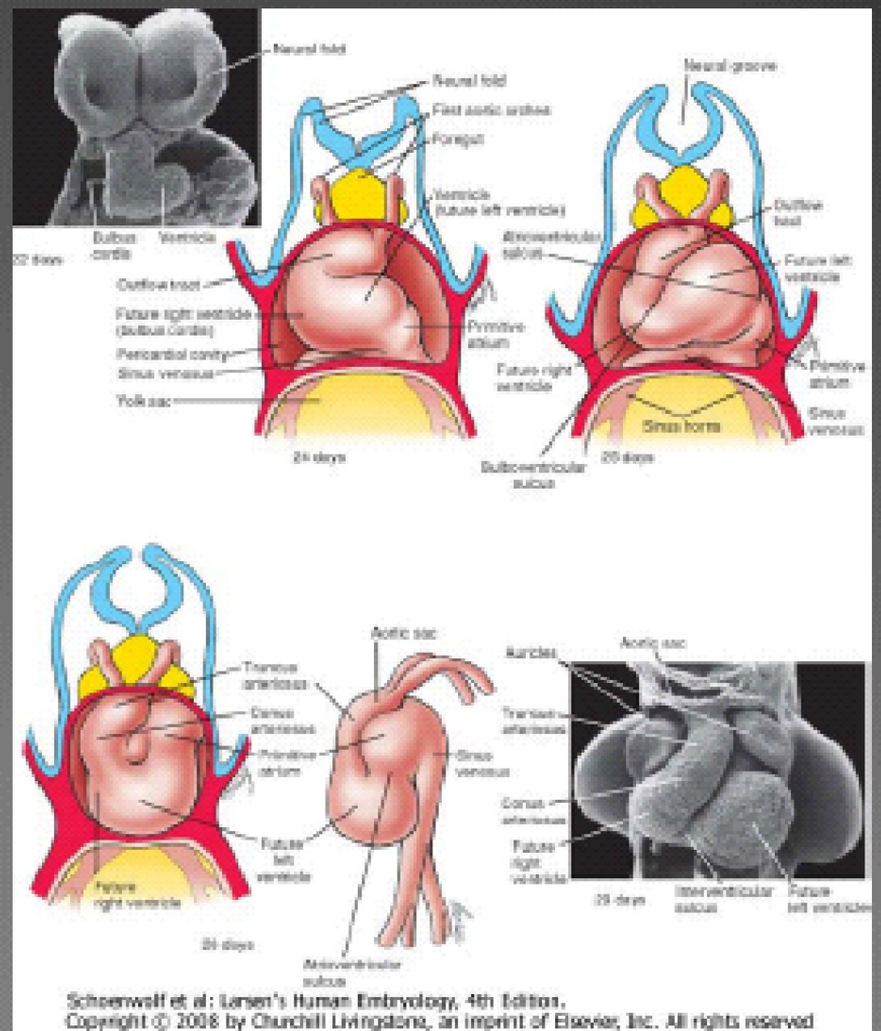

Schoenwolf et al: Larsen's Human Embryology, 4th Edition.
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Daniel M. Schwartz, MD — University of Massachusetts Medical School — DSF, October 2010

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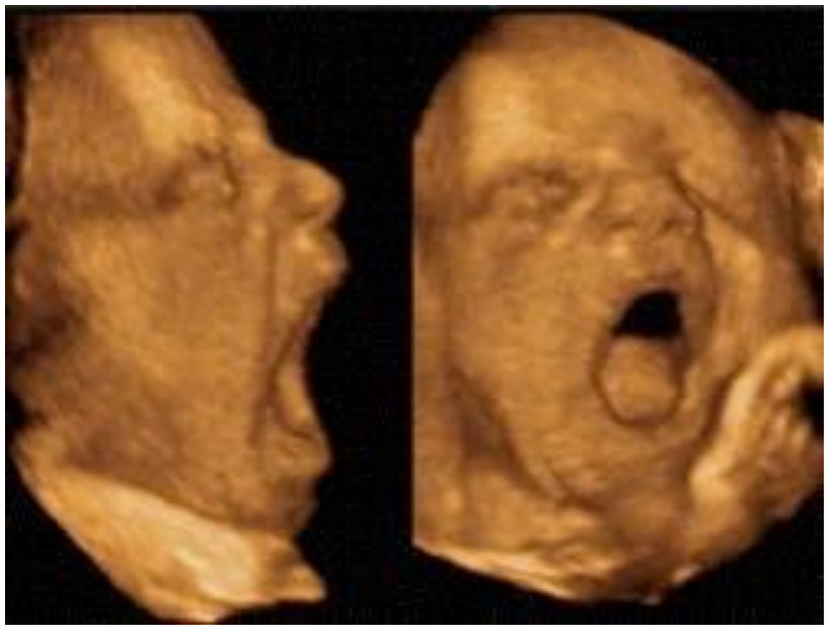
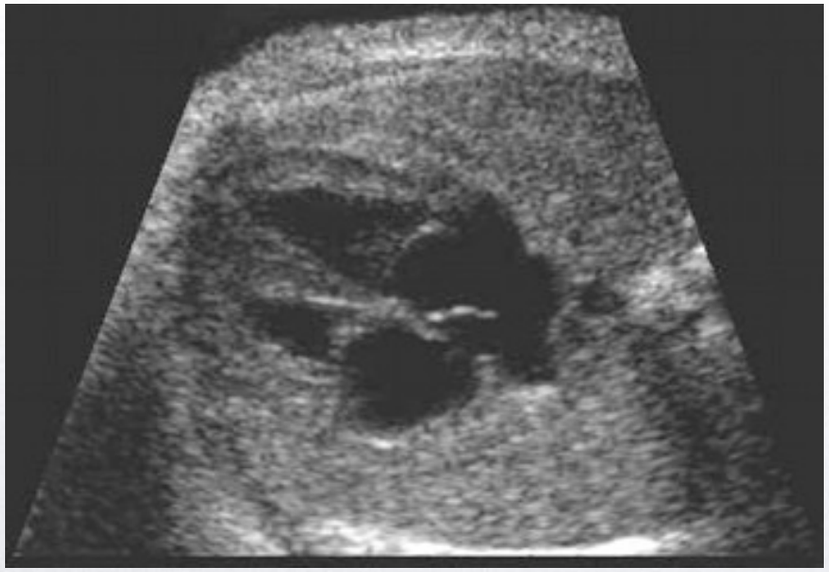
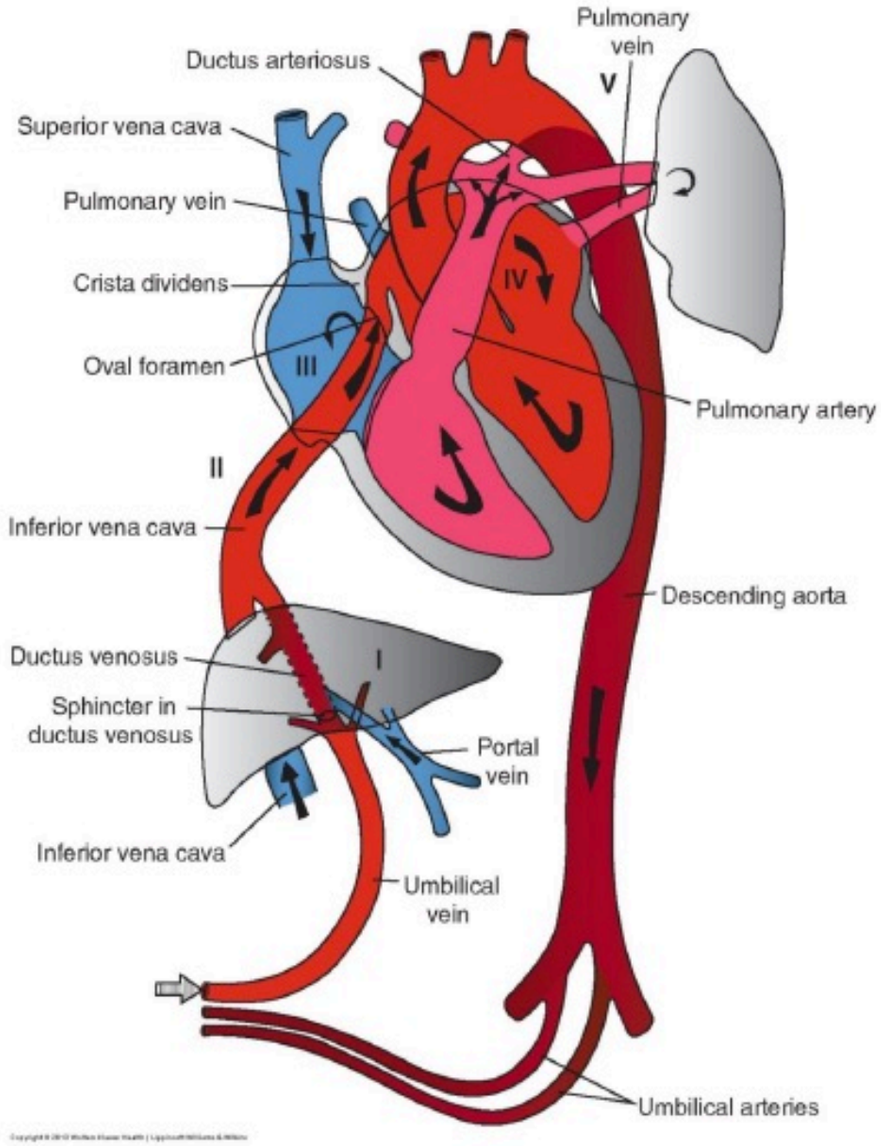
If you understand the factors that dictate **fetal circulation,** you will be able to understand any congenital heart disease





Fetal Circulation

11/27/2018



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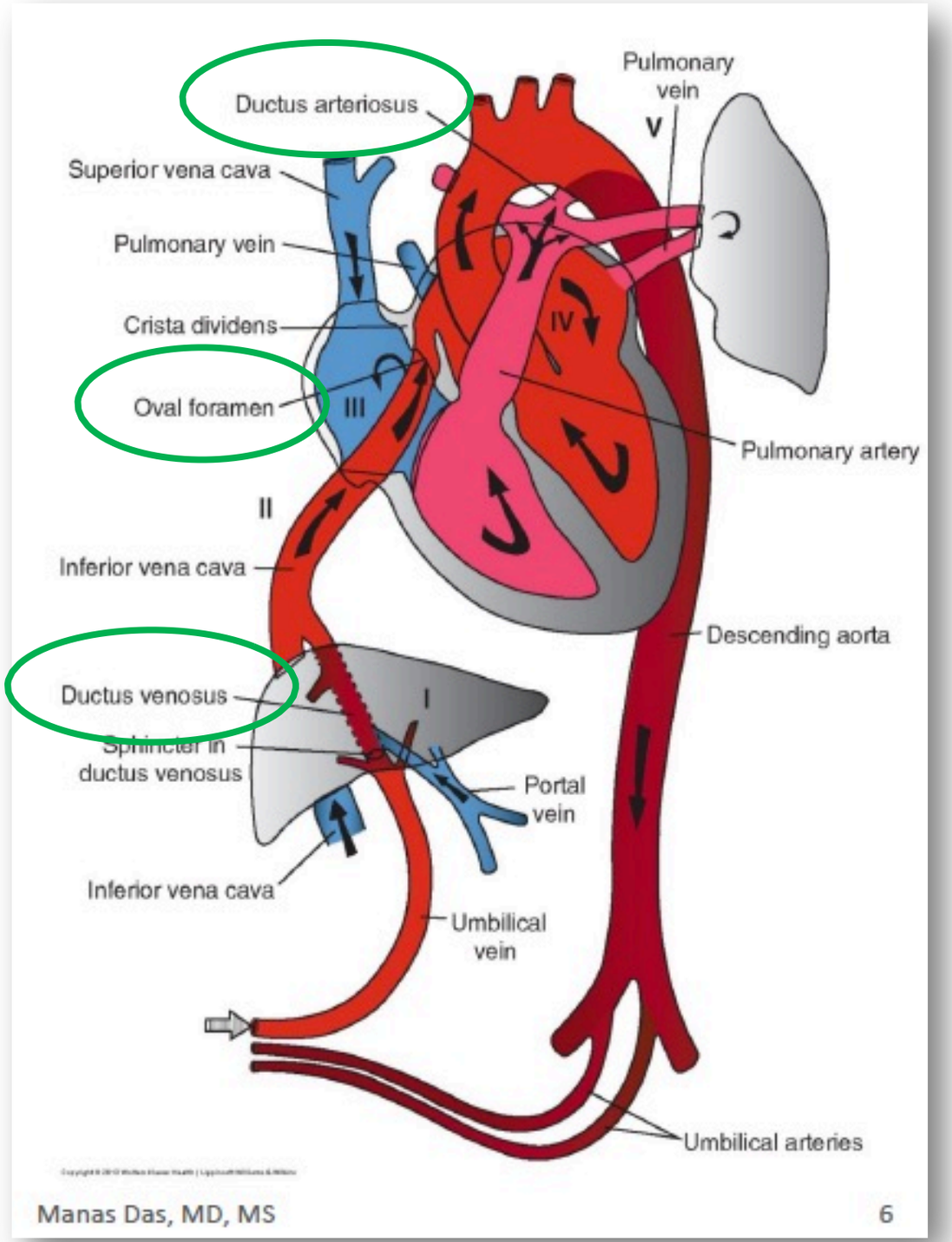
Fetal Circulation characterized by shunts, mixing, and variable resistance

- 3 Important Shunts
  - Ductus arteriosus
  - Ductus venosus
  - Foramen Ovale

- Lungs underwater (collapsed)—High resistance system

- Systemic circulation hooked up to placenta—Low resistance system



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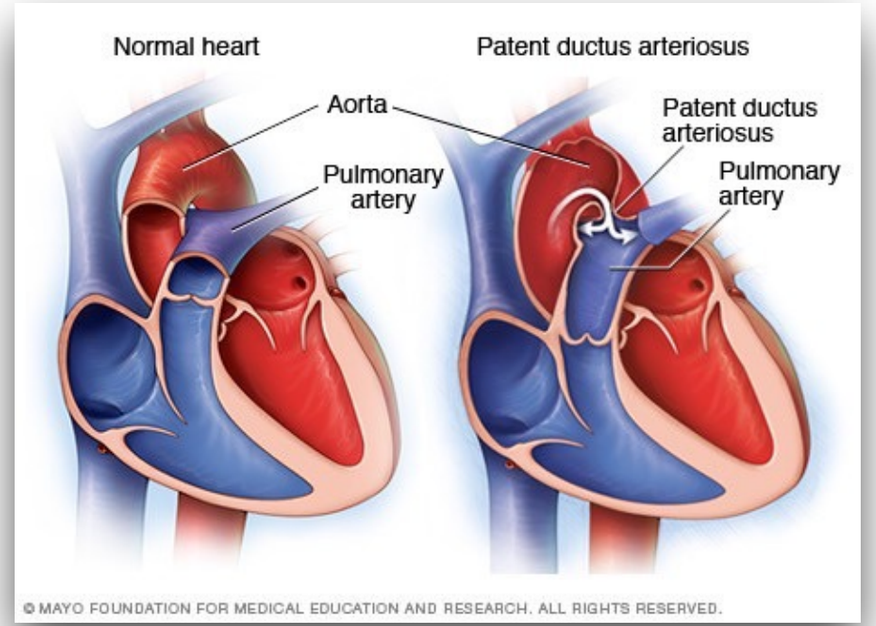
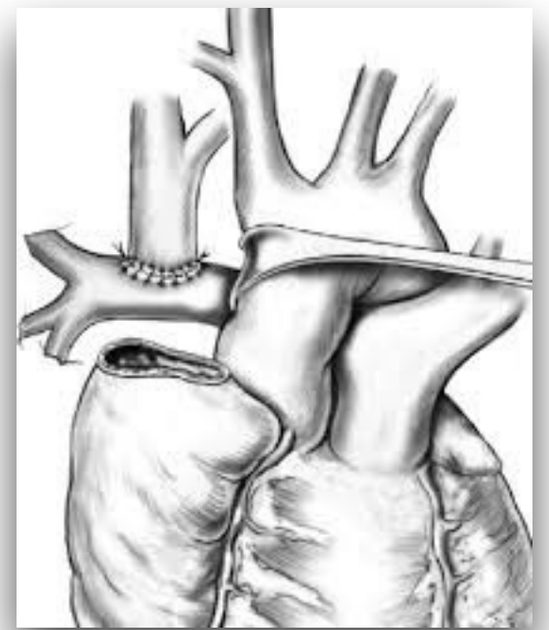
A shunt is hole or small passage that moves blood or fluid from one part of the body to another.

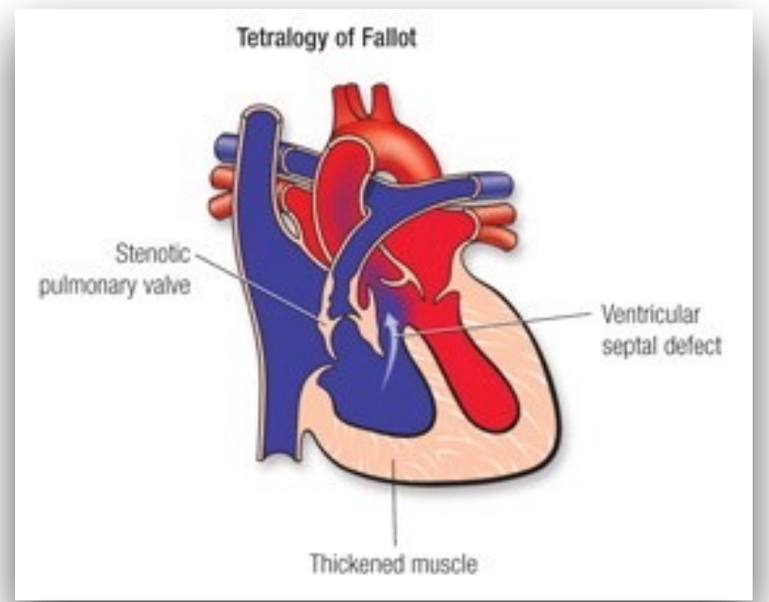
- Congenital (e.g. cardiac) or acquired (surgical)
- Cardiac shunts are often described by the direction of blood flow or in an anatomic description.
  - Left to right (suggests oxygenated blood diverted to pulmonary circulation)
  - Right to left (suggests deoxygenated blood diverted to systemic circulation)
  - Bidirectional
  - Anatomic (cavopulmonary)

Left-to-right shunt





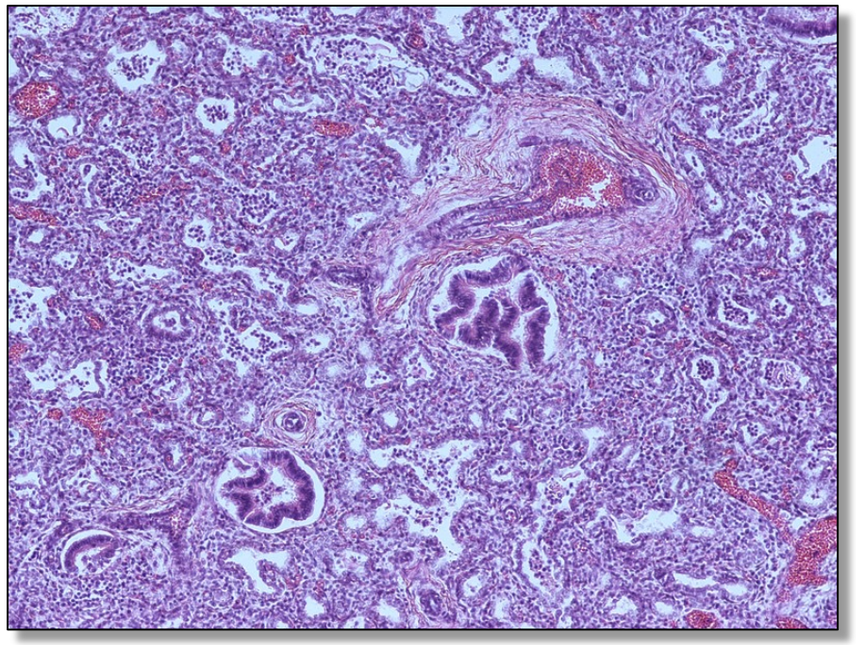
Bidirectional cavopulmonary shunt



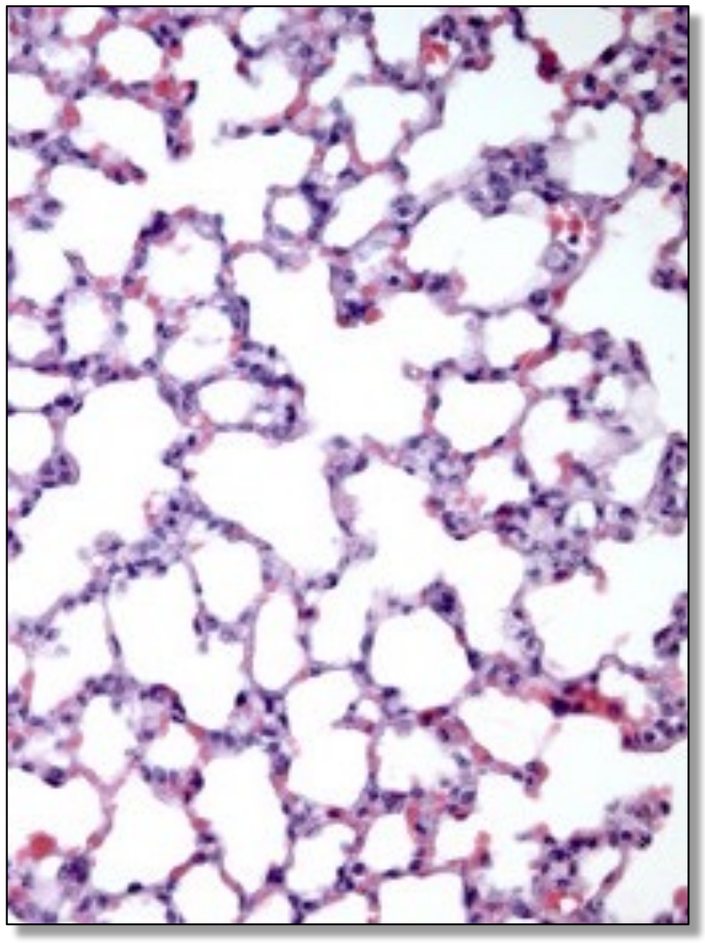
Right-to-left shunt

Fetal lung tissue is compressed, resulting in high pulmonary vascular resistance (PVR)
Placental villi have little resistance, lowering the overall systemic vascular resistance (SVR)

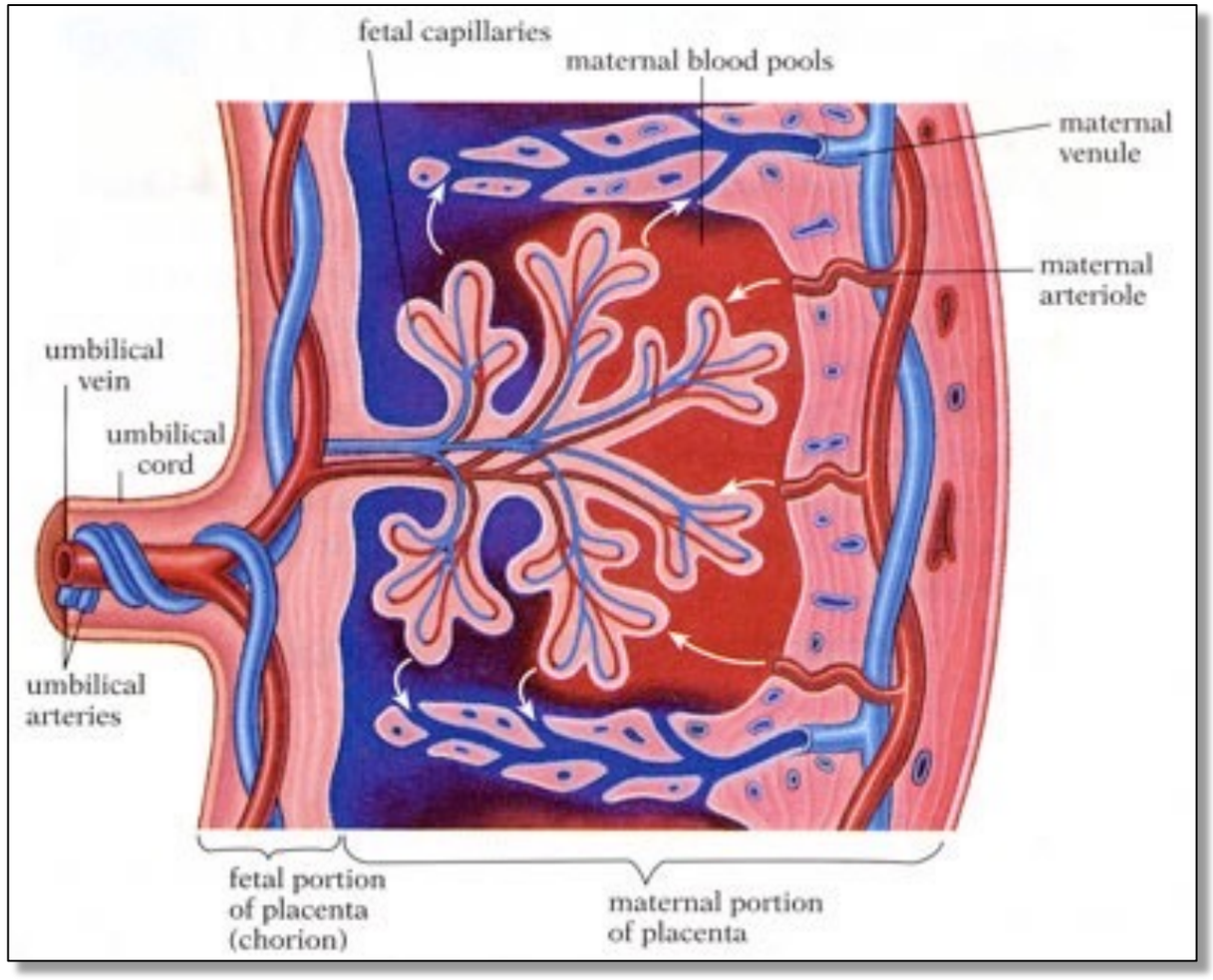
Prenatal Lung



Postnatal Lung



Placental circulation



http://www.biog1105-1106.org/demos/105/unit8/media/placenta.jpg

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$$Q = \frac{\pi(\Delta P)r^4}{8\eta l}$$

Pouiselle's Law says that **Flow (Q)** is:
directly related to **radius^4** and
inversely related to **resistance**

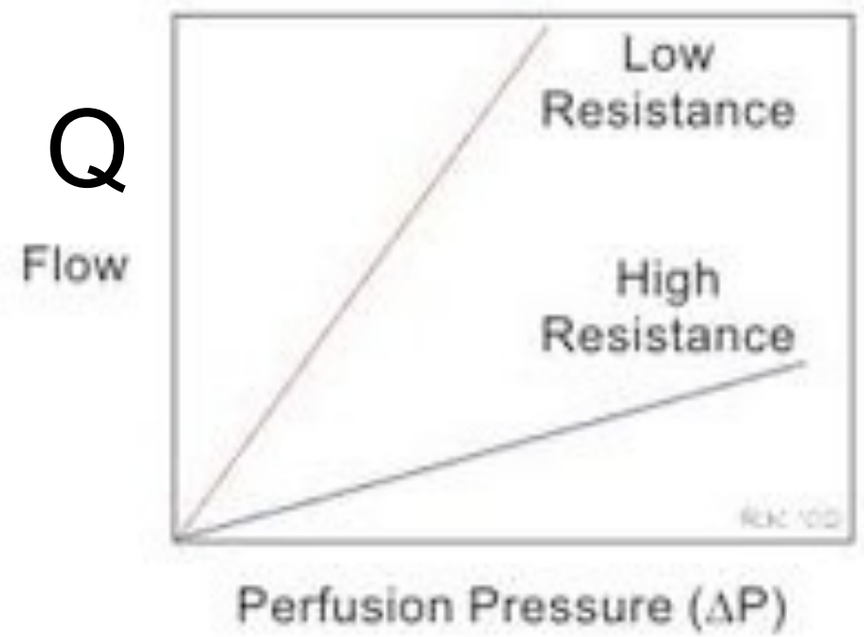
Resistance is therefore **inversely** related to **radius^4**

Part of this equation should look familiar

$$Q = \Delta P/R$$

$$R = \frac{8\eta l}{\pi r^4}$$

Fahey-DSF-2012



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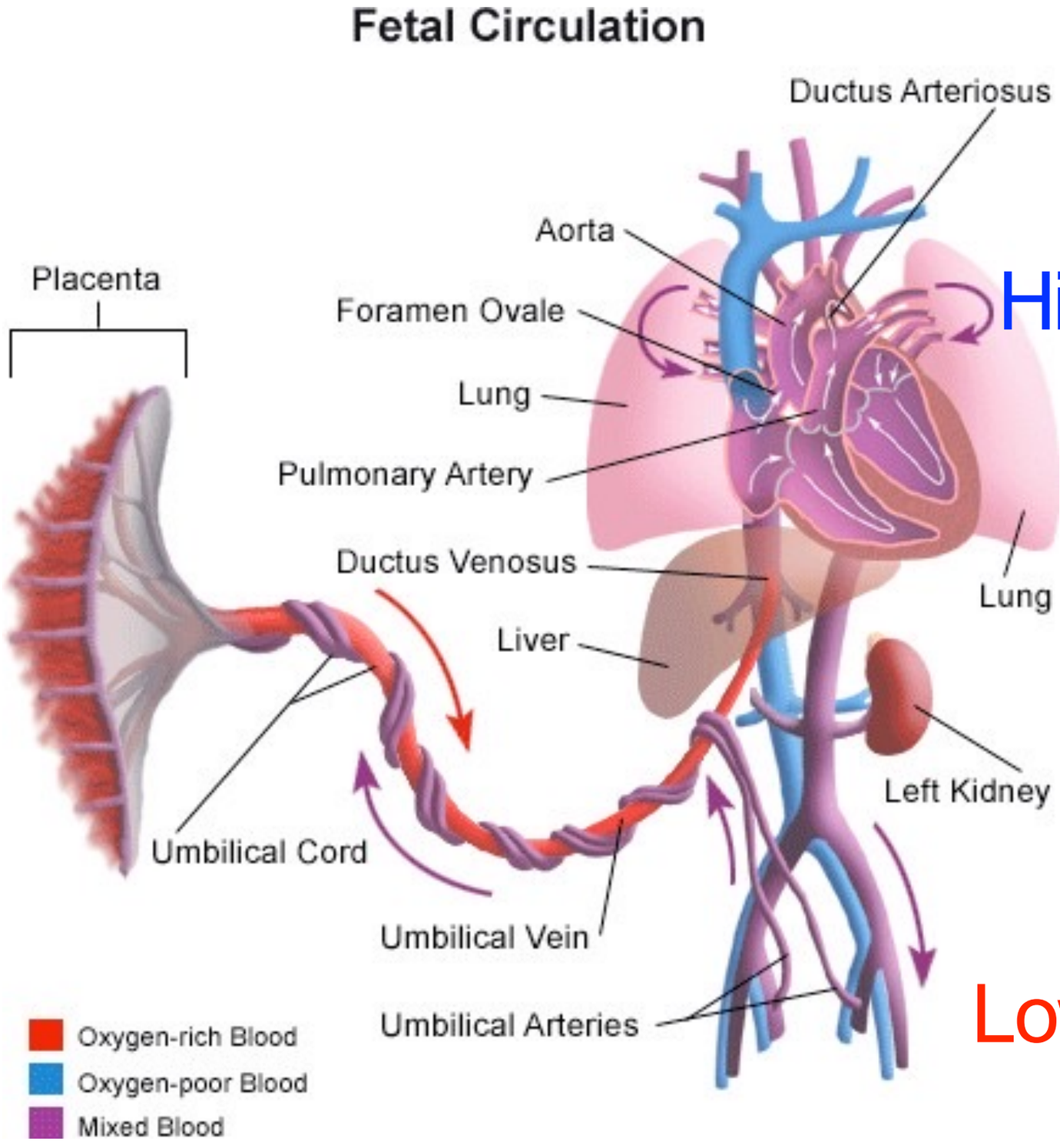
Congenital Heart Disease Rule #1:

Blood generally follows the path of least resistance!

We will use this and a few other rules to determine how all forms of congenital heart disease work!



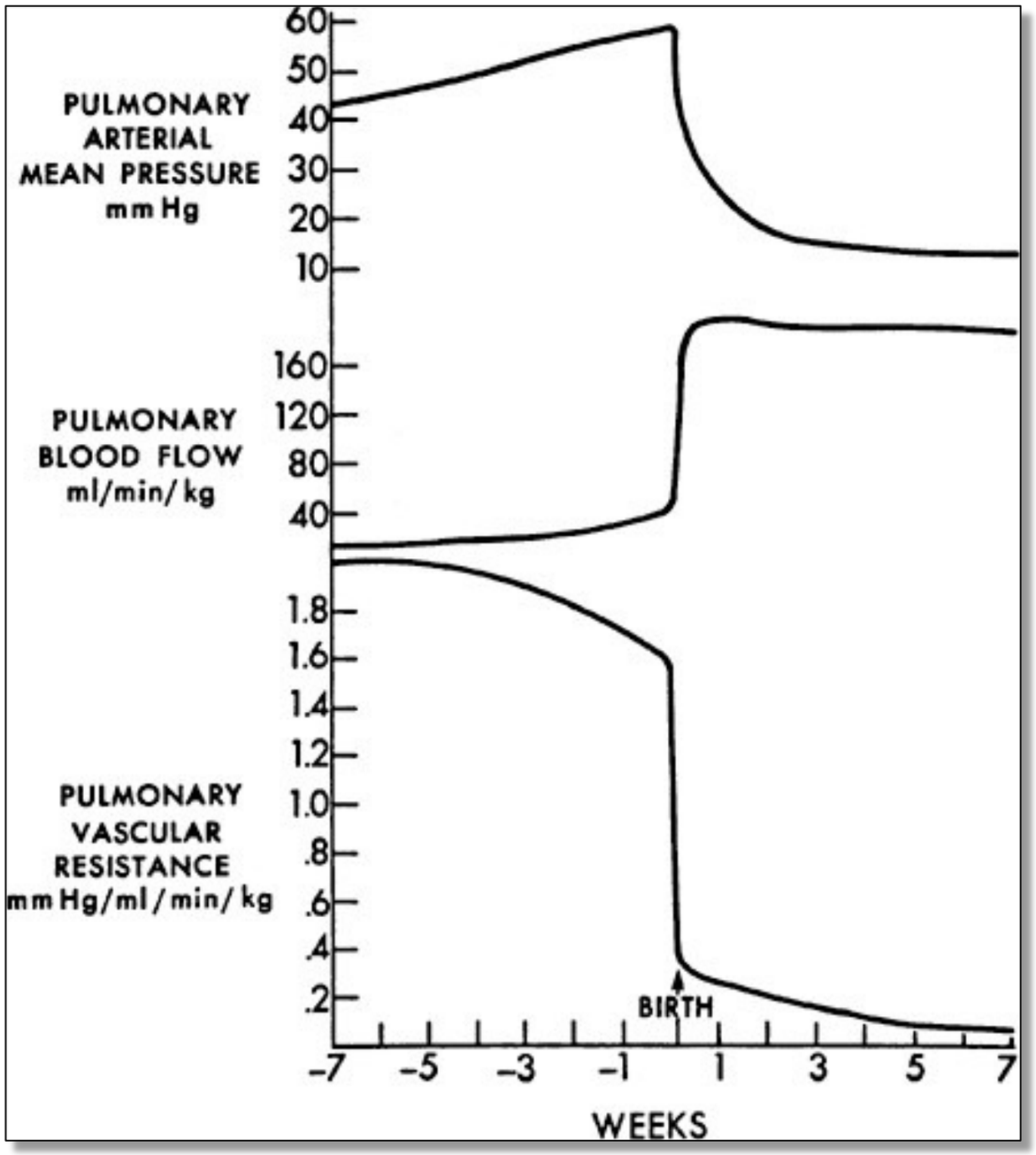
High PVR

Blood will pass through this system down the path of least resistance, i.e. away from the lungs and toward the systemic circulation

Low SVR

Note that this is the opposite scenario compared to post-natal life (i.e. breathing air, no placenta)

What happens after birth?



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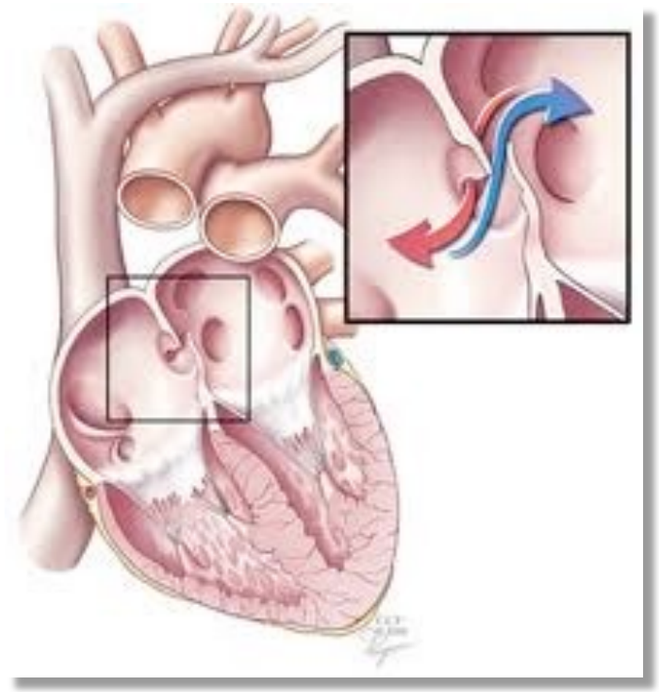
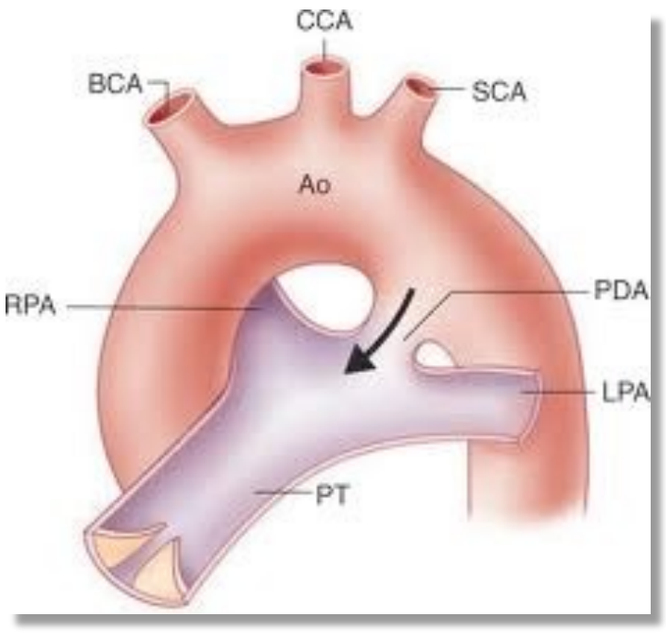
Transitional Circulation

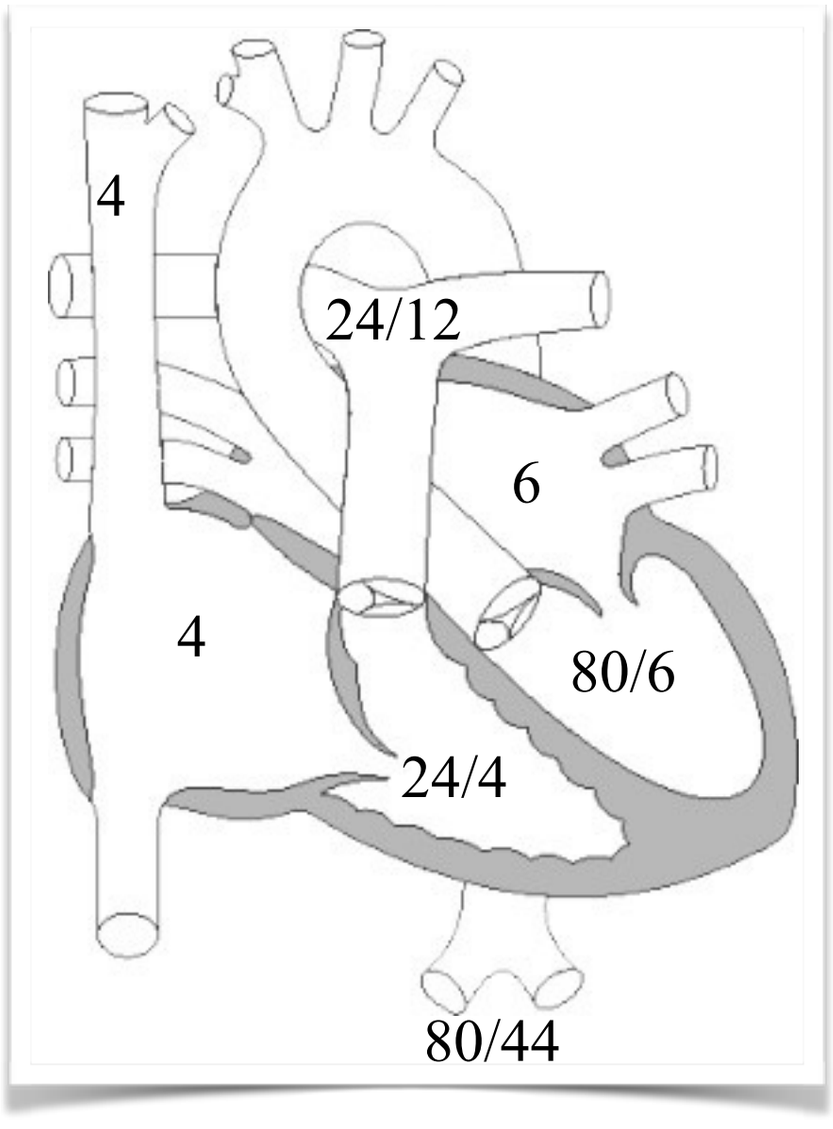
- As the pulmonary vascular resistance drops, **blood flow to the lungs increases**
- This increases blood return to the left atrium, increasing pressure there and **closing the foramen ovale**
- Also, as the pulmonary artery pressure decreases, **flow reverses in the ductus arteriosus**
- Increasing O_2 saturation in the ductus arteriosus, and lack of placental prostaglandin **stimulate ductal constriction and closure**
  - Usually closes within 10-15 hours after birth





Pressure is a byproduct of flow (Q) and resistance (R)

Pressures



This is why pulmonary artery pressures are so much lower than aortic pressures!

Congenital heart diseases generally comprise abnormal shunts, abnormal resistance to flow, or combinations of these two problems

Act I: Rules of the Game/Fetal Circulation Review

- The fetal circulation differs from post-natal circulation because of the presence of shunts, mixing, and vascular beds with variable resistance

- Shunts result in mixing of blood with different oxygen content, and flow through shunts is determined by resistance

- Resistance is inversely related to (vessel radius)^4

- Flow is inversely related to resistance

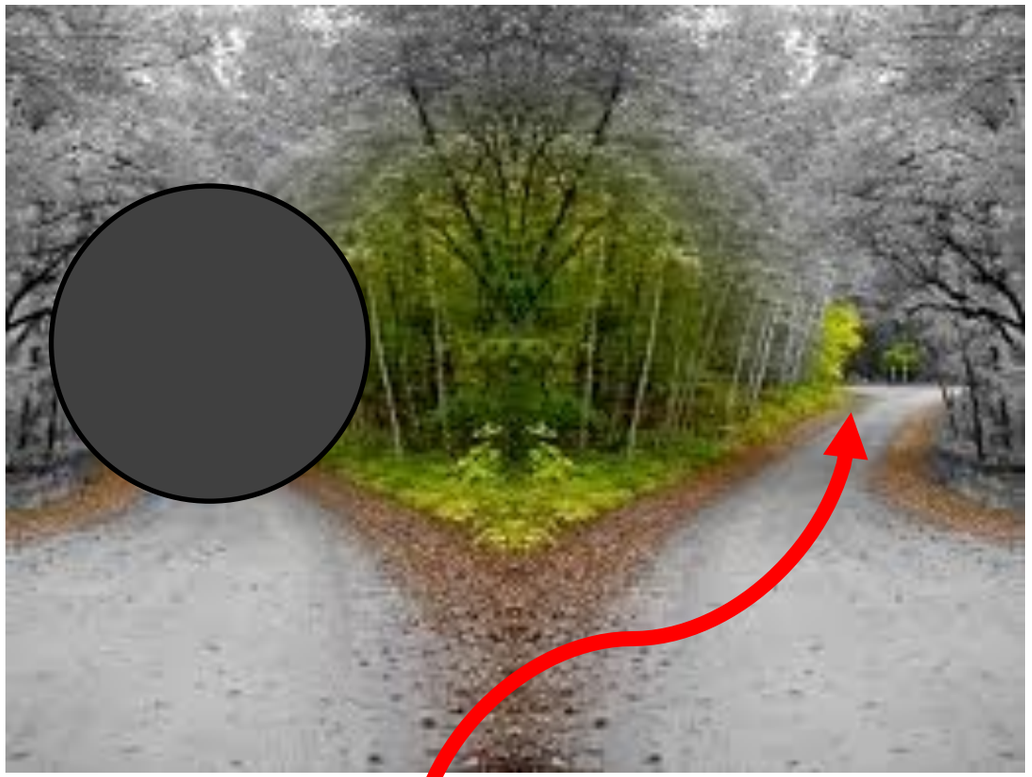
- ***Blood generally follows the path of least resistance***

Act I: Rules of the Game/Fetal Circulation
Review (continued)

- In most vessels, pressure is a byproduct of flow and resistance
- ***Blood generally follows the path of least resistance***

Act II: Shunts, Obstructions and Remodeling

Obstruction will result in higher pre-obstruction pressures



Blood is going to travel in the path of least resistance!

Congenital Heart Disease Rule #2:

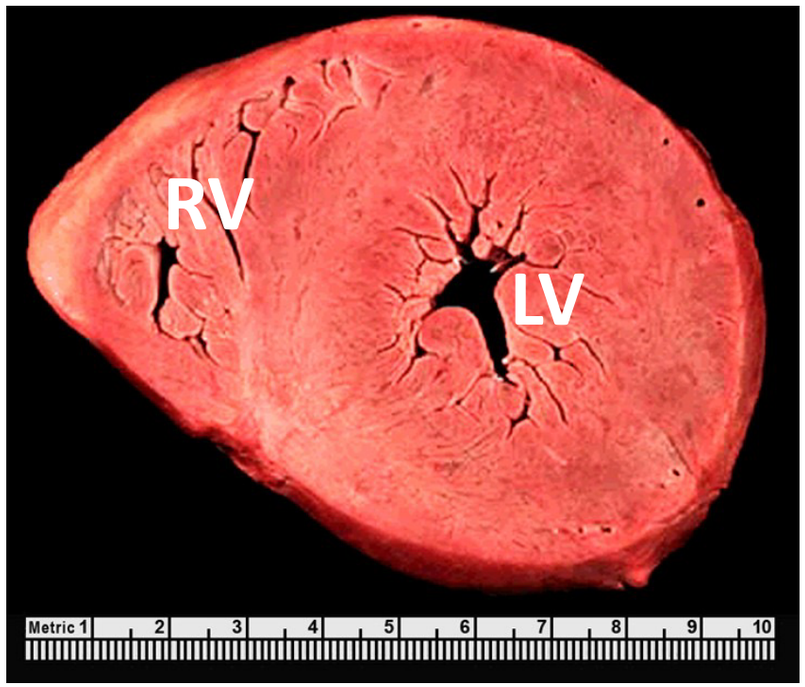
Heart chambers and blood vessels respond to pressure challenges with concentric hypertrophy, and to volume challenges with eccentric hypertrophy

Concentric hypertrophy = increased wall thickness

Eccentric hypertrophy = chamber dilation (often normal thickness)

Changes in chamber thickness or dimensions are referred to as ***remodeling***

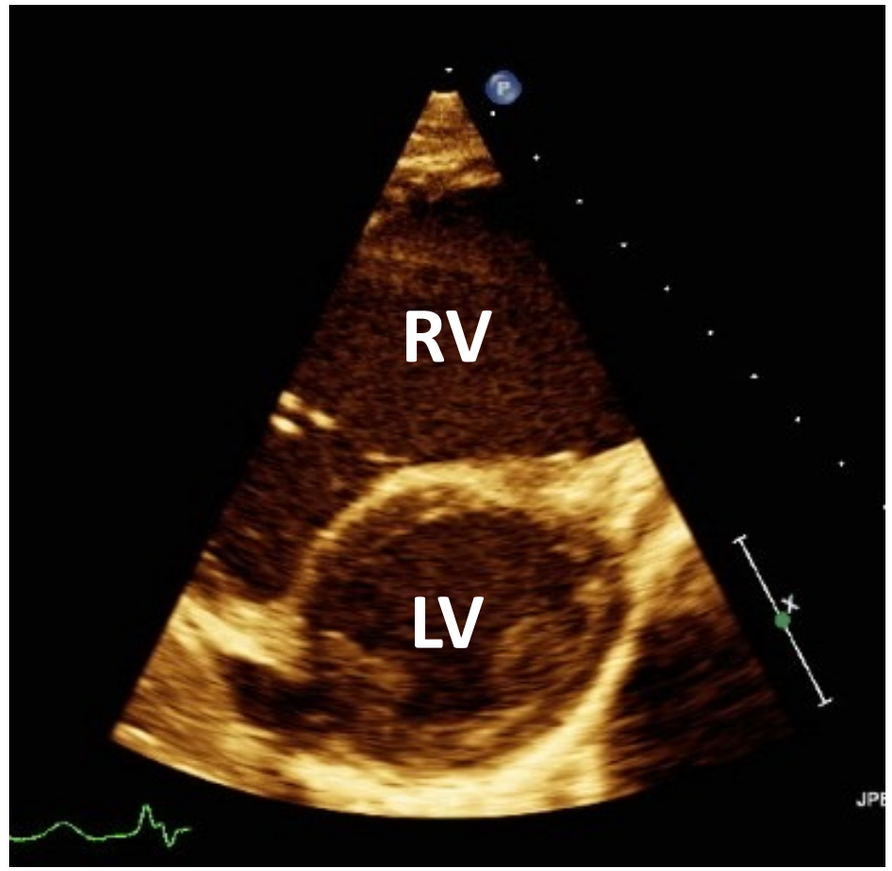
Aortic valve stenosis or systemic hypertension



http://www.aurorahealthcare.org/portals/doctors/education- training/acs-cases/art/sept-figure-5.jpg

Concentric hypertrophy (in this case, of LV)
-addition of myocytes in parallel

Atrial septal defect



Eccentric hypertrophy (in this case, of RV)
-addition of myocytes in series

Remodeling can be adaptive/physiologic or maladaptive/pathologic

Adaptive/physiologic remodeling:

- Pregnancy
- Exercise

Maladaptive/pathologic remodeling:

- Shunts
- Obstructions
- Ischemic heart disease

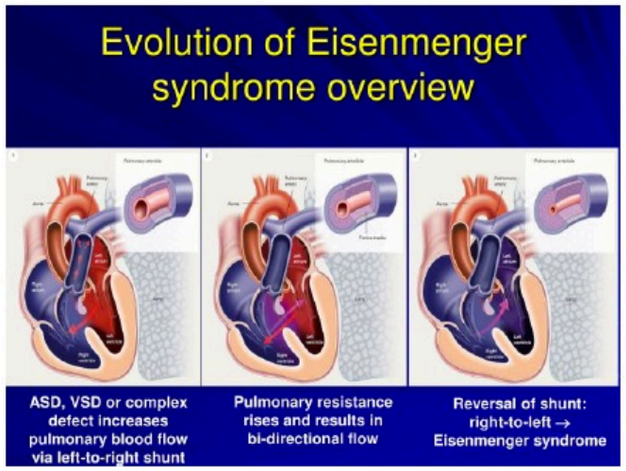
Concentric hypertrophy of pulmonary vascular smooth muscle is a major morbidity associated with many congenital heart defects

- Congenital heart disease that results in excess flow and/or pressure in the pulmonary arteries stimulates smooth muscle hypertrophy, which increases pulmonary vascular resistance

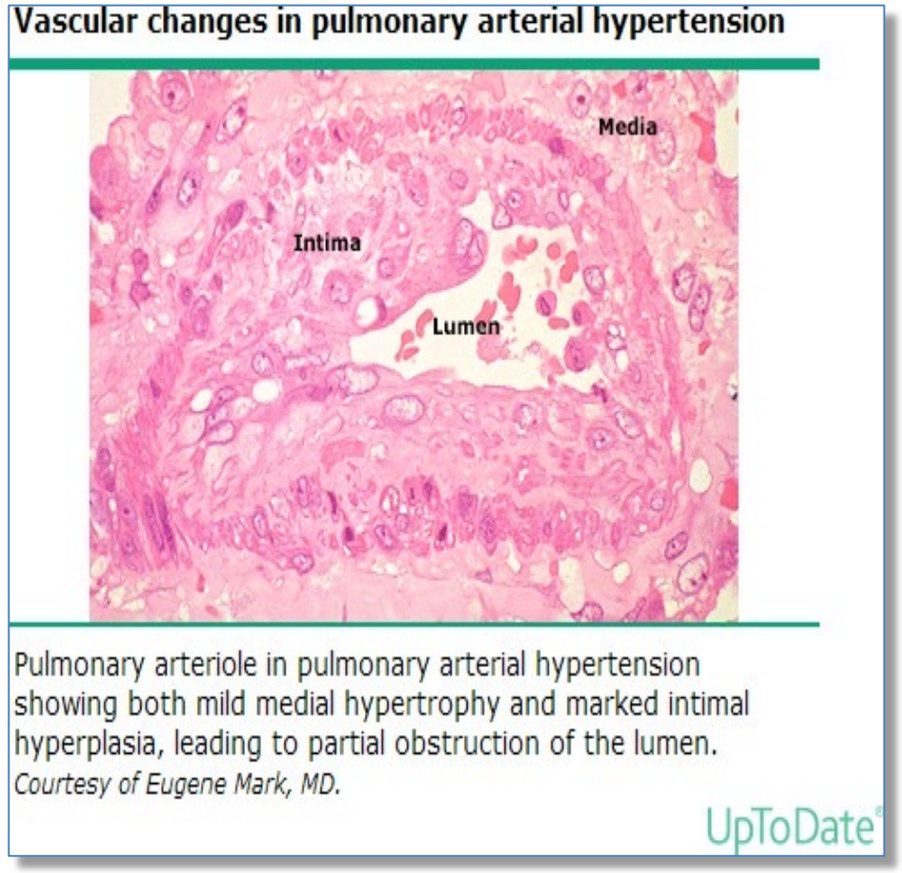
- This is also known as pulmonary vascular obstructive disease, or ***Eisenmenger Syndrome***

Eisenmenger's Syndrome

11/27/2018



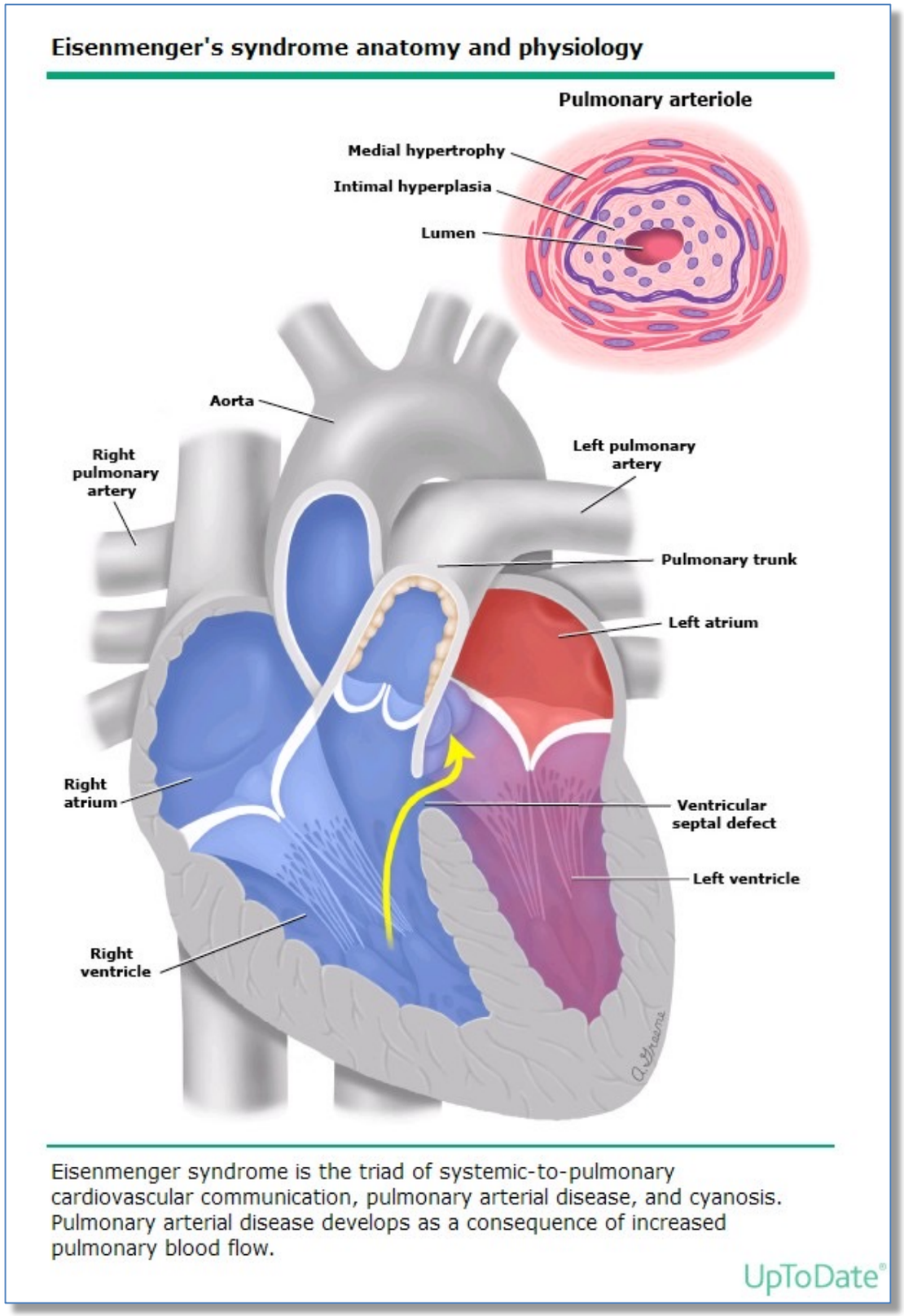
Manas Das, MD, MS 10



Eisenmenger Syndrome develops gradually, and results in a reversal of flow through the original shunt

Progression of Disease

1. Systemic to pulmonary connection (intracardiac, vascular)
2. Net left-to-right shunt resulting in increased pulmonary blood flow and pressure
3. Pulmonary vascular injury and remodeling
4. Increased pulmonary vascular resistance (PVR)
5. Net right-to-left shunt with hypoxemia/cyanosis



Act II: Shunts, Obstructions and Remodeling Review

- Shunts offer choices for blood to follow the path of least resistance

- Pressures build up pre-obstruction

- Increases in flow and/or pressure can result in heart chamber or blood vessel remodeling

- ***Pressure challenges result in concentric hypertrophy, volume challenges result in eccentric hypertrophy***

Act II: Shunts, Obstructions and Remodeling Review (continued)

- Remodeling can be adaptive/physiologic or maladaptive/pathologic
- Pathologic remodeling of pulmonary vasculature, known as **Eisenmenger syndrome**, can occur in a number of congenital heart diseases if left untreated

Act III: Acyanotic Congenital Heart Defects

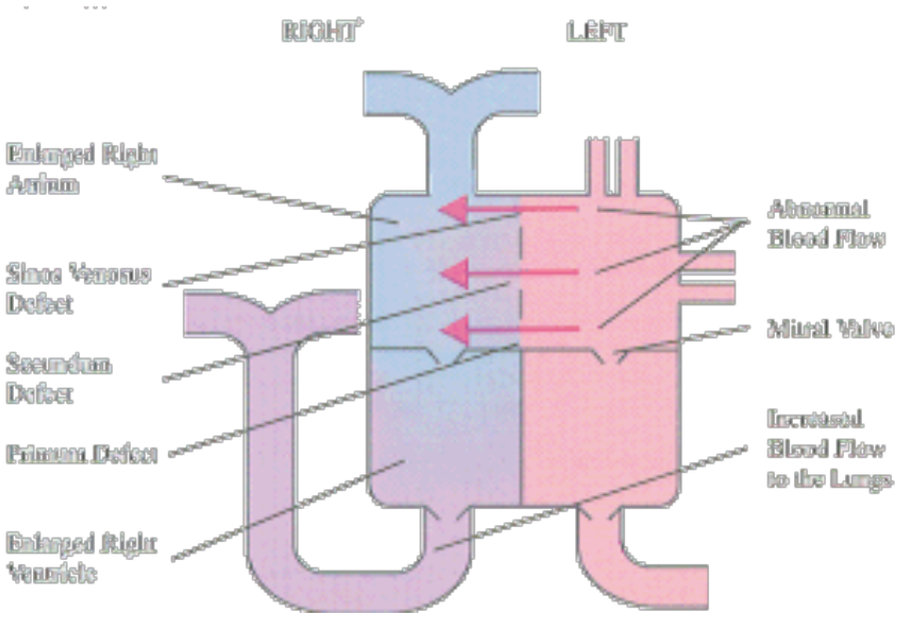
- "Acyanotic" implies there won't be any mixing of blue blood into the systemic circulation

- These defects generally comprise **"left-to-right" shunts**, meaning red blood is shunting toward the pulmonary arteries and lungs

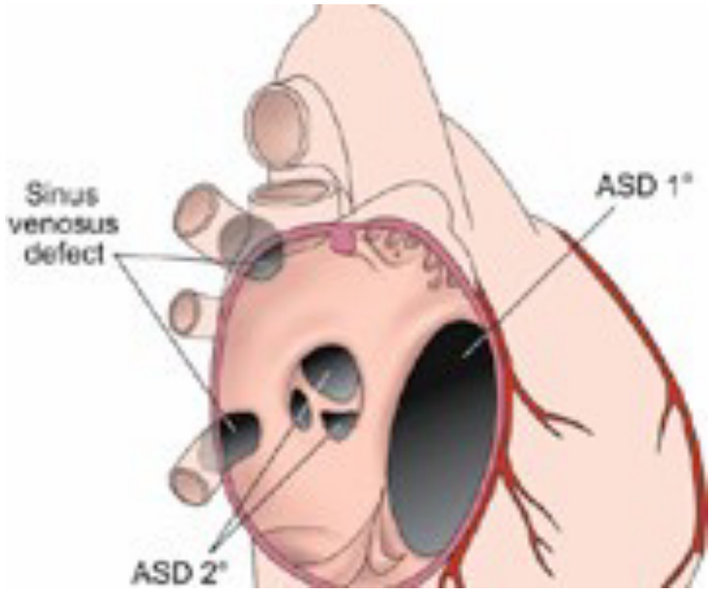
- The 3 main types of acyanotic CHDs are **Atrial Septal Defects**, **Ventricular Septal Defects**, and **Patent Ductus Arteriosus**

Atrial Septal Defects

- There are avariety of different types
  - Secundum ASD (75% of ASDs; 6% of CHD)
  - Primum ASD (15-20% of ASDs)
  - Sinus Venosus defects (5-10%)



http://www.riverbendds.org/asd.gif



http://www.accessmedicine.ca/loadBinary.aspx?name=hurs13&filename=hurs13_c083f007t.gif

- The clinical presentation and complications are based on the left-to-right shunt that results
- The **flow is left-to-right** due to the relative ventricular compliance (diastolic phenomena)
- **Increased pulmonary blood flow** and a "volume-load" on the right atrium and ventricle

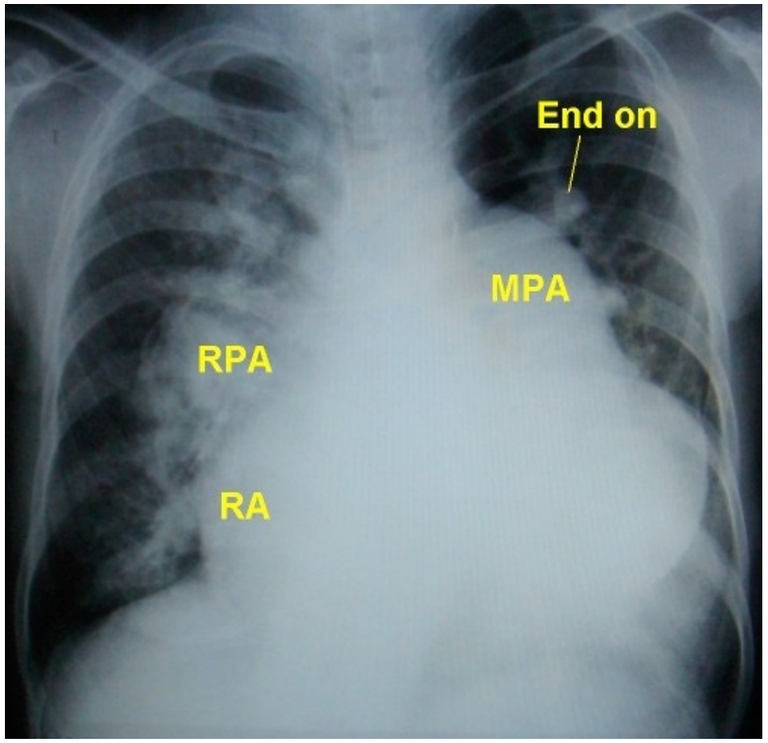
ASDs often do not cause many symptoms

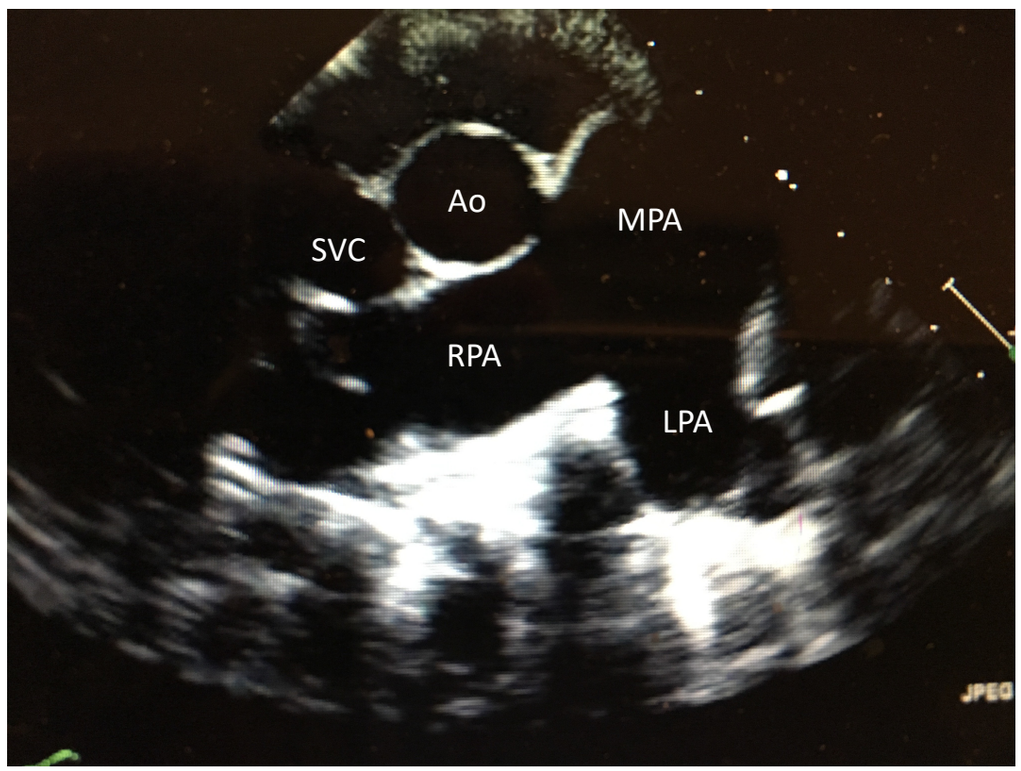
- Despite pulmonary overcirculation, the pulmonary artery pressures do not increase very much, since the ASD offers a “pop-off” for the left atrium

- Therefore, Eisenmenger syndrome develops much later (e.g. 2nd or 3rd decade) in life

- Symptoms may include:
  - Exercise intolerance or dyspnea with exertion
  - Frequent lower respiratory tract infections
  - Palpitations (from atrial arrhythmias

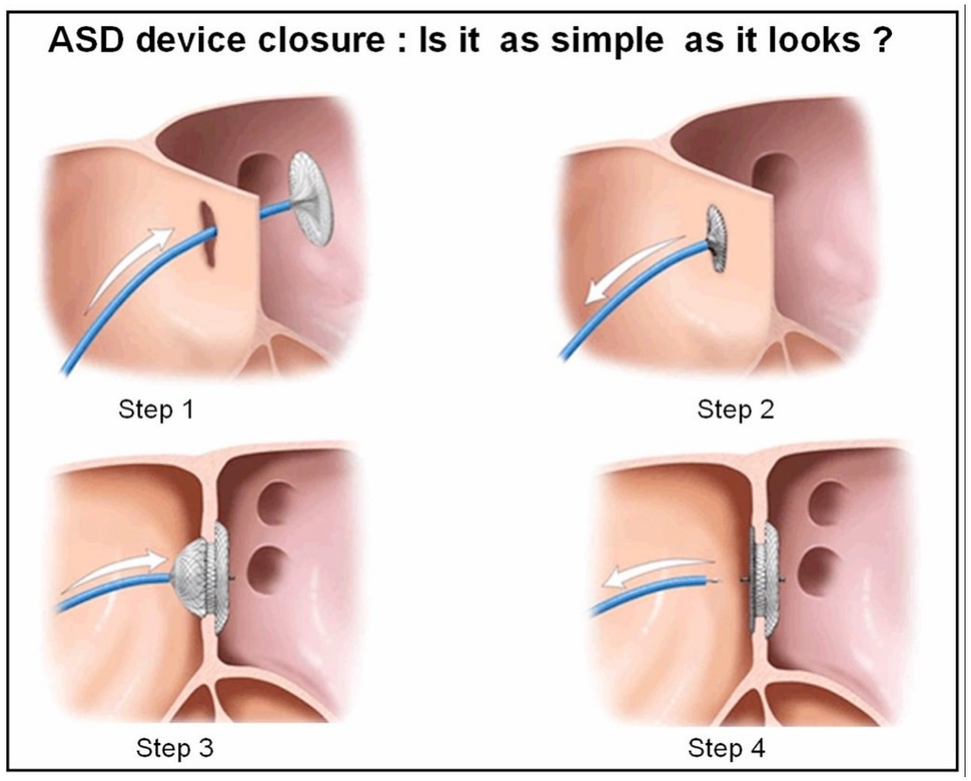
- Physical Exam findings:
  - Systolic ejection murmur: sound of extra blood passing over pulmonary valve
  - Fixed splitting of S2 (pulmonary component comes later from extra volume)
  - Right ventricular heave

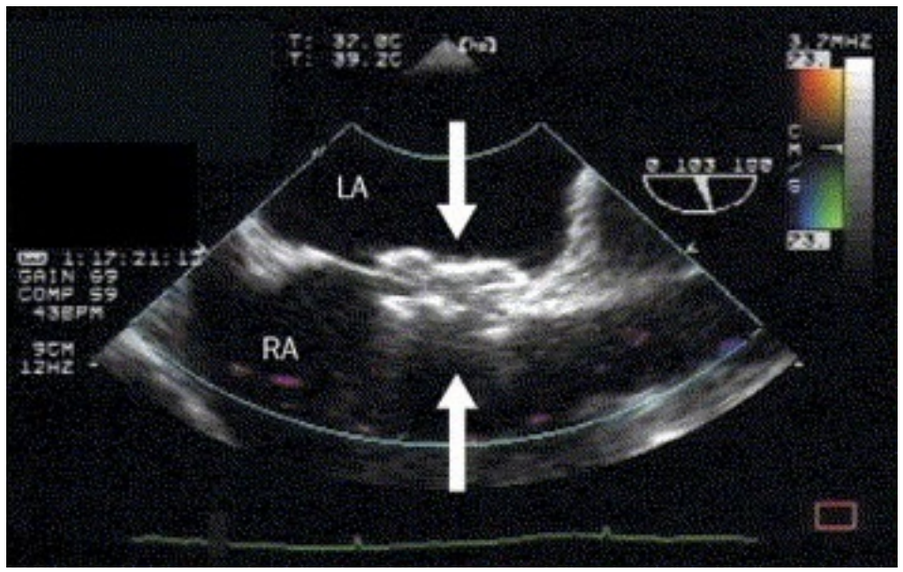




ASD Treatment/Interventions

- Many ASDs will close spontaneously
- If they are small enough, they don't require intervention
- If they require closure, surgical and transcatheter device closure are possible





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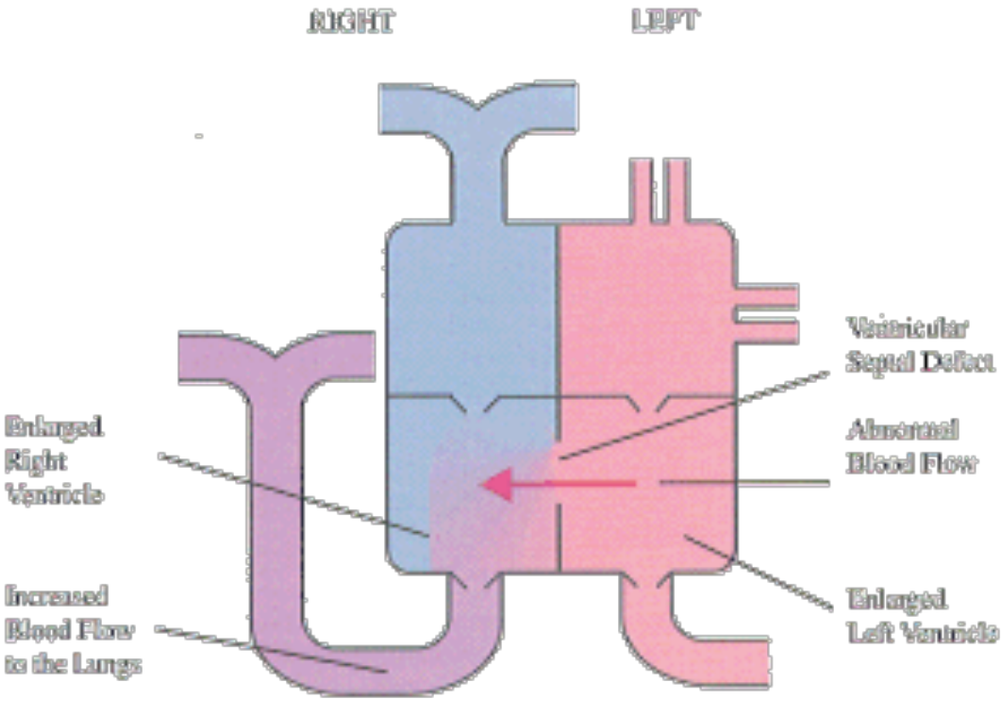
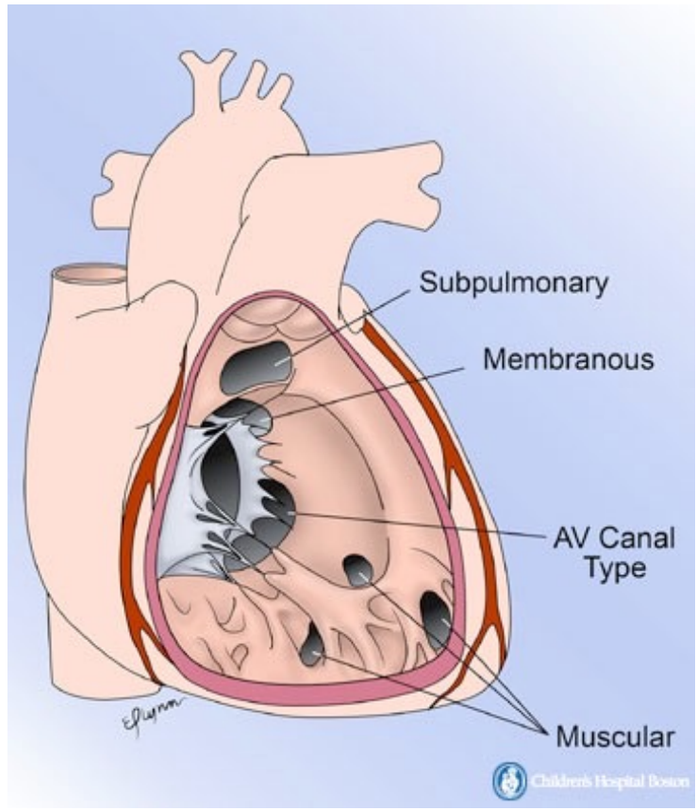
Ventricular septal defects (VSDs) are the most common congenital heart lesion (4.2/1000 live births)*

- There are a variety of different anatomic types of VSDs
- Pathophysiology depends on the quantity of the shunt



http://www.riverbendds.org/vsd.gif



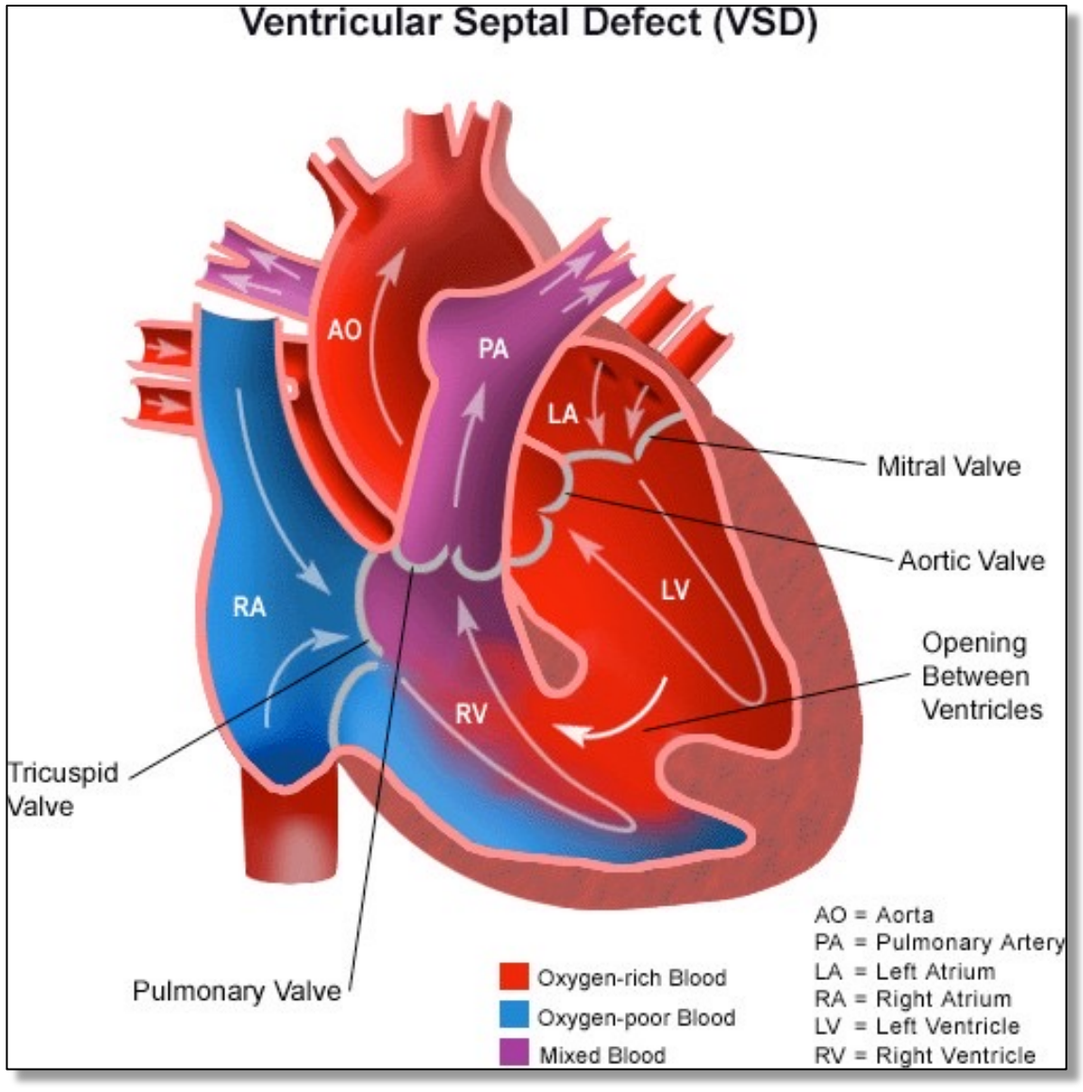
http://www.childrenshospital.org/cfapps/mml/viewBLOB.cfm?MEDIA_ID=356

- Symptoms are a result of too much pulmonary blood flow, and the response to this challenge!

*Unless you count bicuspid aortic valve, which is present in ~2% of the general population

Many Left-to-Right shunts result in Left-sided volume overload!!!

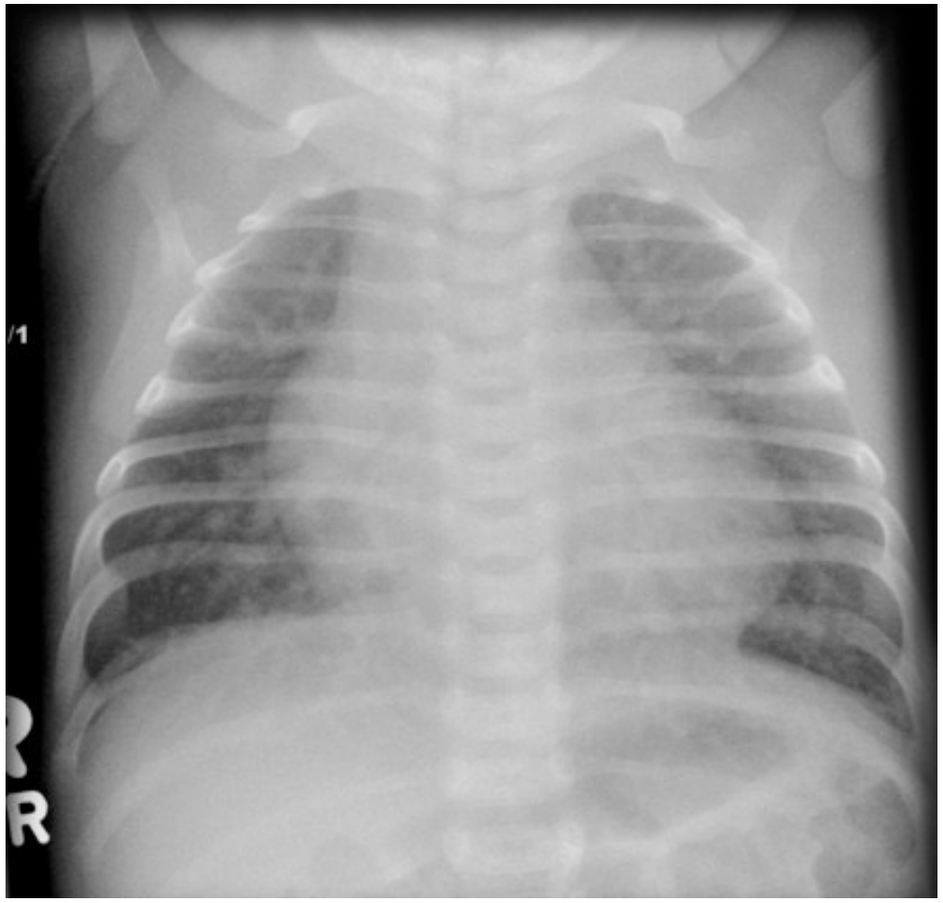
- The degree of shunting depends on the relative resistance to flow during systole
- After birth, the PVR will be less than the SVR resulting in a left-to-right shunt
- This left to right shunt results in increased pulmonary blood flow
  - Increased pulmonary venous return
  - Volume load on the left atrium and ventricle



http://heart.phoenixchildrens.com/sites/default/files/125877_Ventricular%20Septal%20Defect.jpg

"You look like your pulmonary blood flow."

- VSDs result in pulmonary overcirculation
- In addition, the increased volume returning to the left side of the heart jacks up left atrial pressure, which backs up into the lungs
- This results in a "traffic jam," or congestion, in the pulmonary vasculature
- There is also an increased volume of blood the left ventricle needs to circulate
  - CO = HR x SV
  - Sympathetic stimulation to maintain increased output
- Signs/symptoms include:
  - Harsh, holosystolic murmur
  - Tachypnea, increased work of breathing
  - Tachycardia, diaphoresis, peripheral vasoconstriction (color changes)
  - Peripheral edema, hepatosplenomegaly
  - *These signs and symptoms have a name…*



Congenital Heart Disease Rule #3:

Congestive Heart Failure (CHF) occurs whenever there is pulmonary overcirculation plus left-sided volume overload, or problems that cause a pressure backup into the pulmonary vasculature

CHF is a constellation of signs and symptoms that result from the physiologic response to particular derangements in the cardiovascular system

-Most left-to-right shunts (not ASDs)
-Most left-sided obstructive lesions
-Severe left-sided valvar regurgitation
-Moderate/severe LV dysfunction

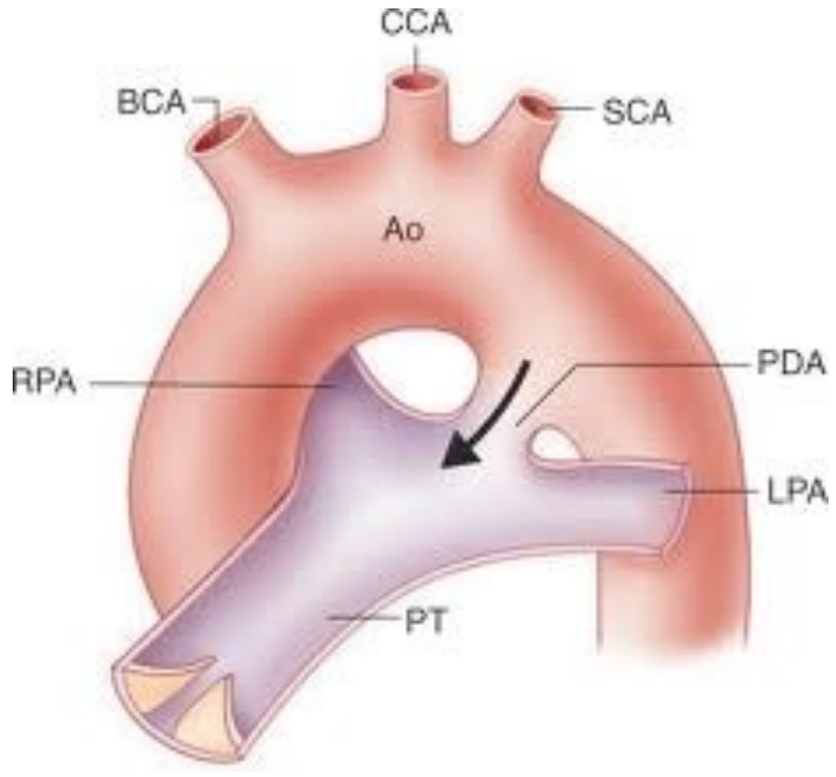
VSD Treatment/Interventions

- The anatomic type of VSD is important; some types are more likely to close spontaneously than other types
- Manage the infant's congestive heart failure
  - Diuretics
  - Afterload Reduction (ACE Inhibitors)
  - Nutrition
- Surgery - Close the shunt that's causing the problem!
  - Close the shunt before you develop Eisenmenger syndrome

Big difference between VSDs and ASDs is that a moderate sized ventricular or arterial level shunt will cause Eisenmenger syndrome much sooner than an ASD:

2-3 years as opposed to 20-30 years

Patent Ductus Arteriosus (PDA)



- The DA remains patent *in utero* due to the low arterial oxygen content and the circulating Prostaglandin E2 produced in part by the placenta

- Functional ductal closure usually occurs within 10-15 hours after birth. Permanent closure may take several months before it converts into the *ligamentum arteriosum.*

- The incidence of PDA is extremely common in premature infants (~30%) and the risk is inversely related to gestational age.

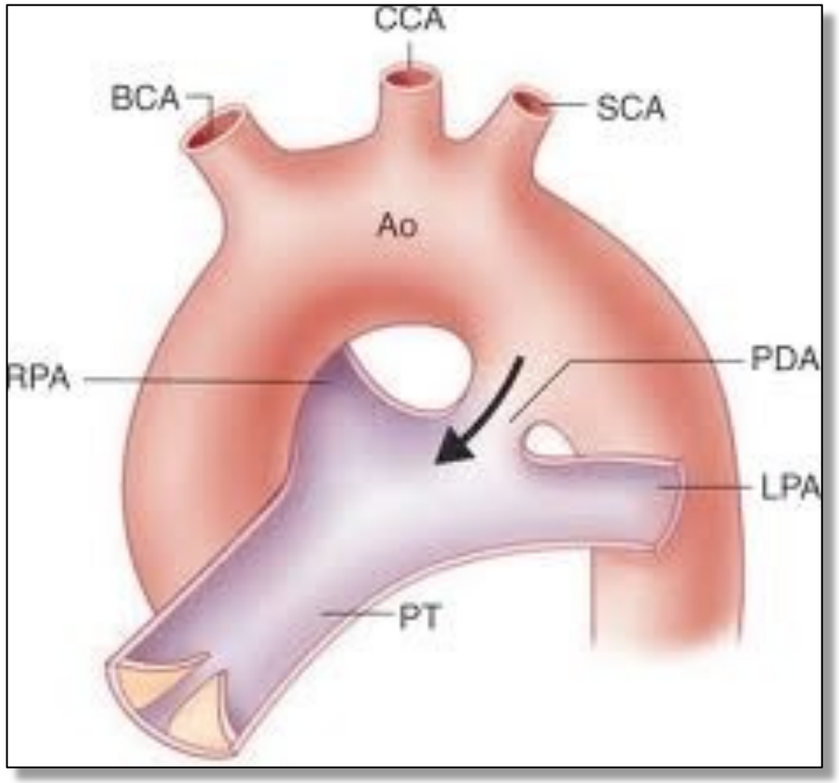
- Infective endarteritis is an uncommon complication of a PDA

"You look like your pulmonary blood flow."

- PDA will continually shunt down the path of least resistance
  - In a sick newborn infant with lung disease, this might be out to the aorta!
  - Generally, after birth the PVR will be less than the SVR, so there will be a left-to-right shunt

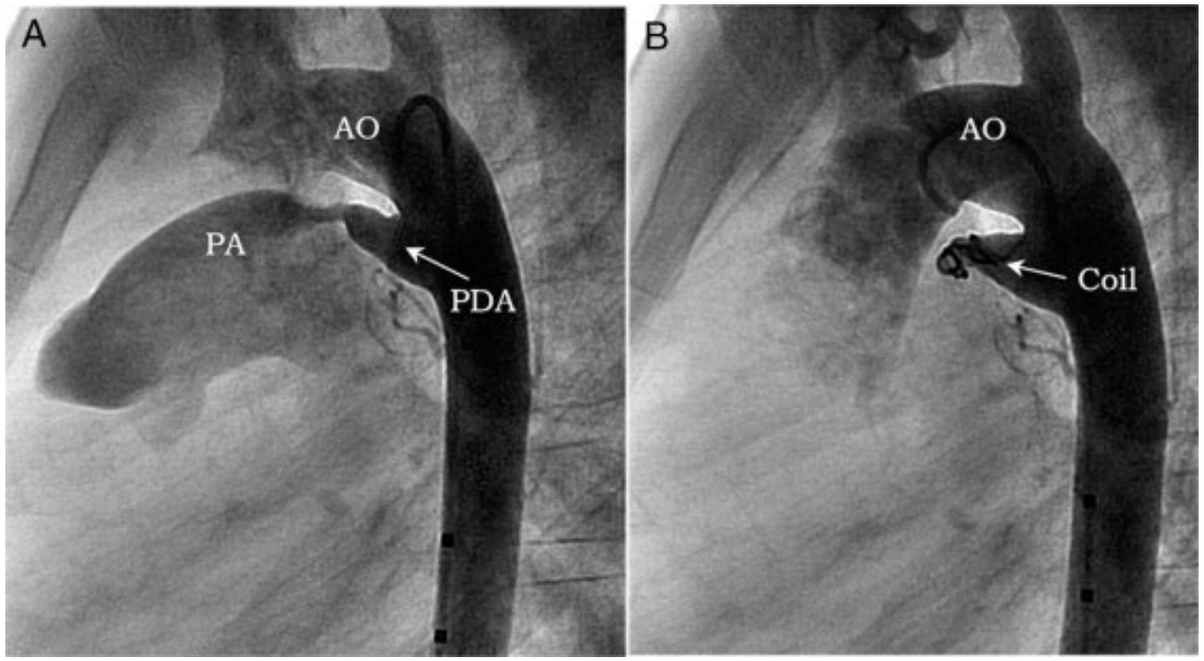
- If the L to R shunt is large, pulmonary overcirculation and left-sided volume overload will occur

- Signs/symptoms include:
  - Continuous murmur*
  - Tachypnea, increased work of breathing
  - Tachycardia, diaphoresis, peripheral vasoconstriction (color changes)
  - Peripheral edema, hepatosplenomegaly
  - *These signs and symptoms have a name…*



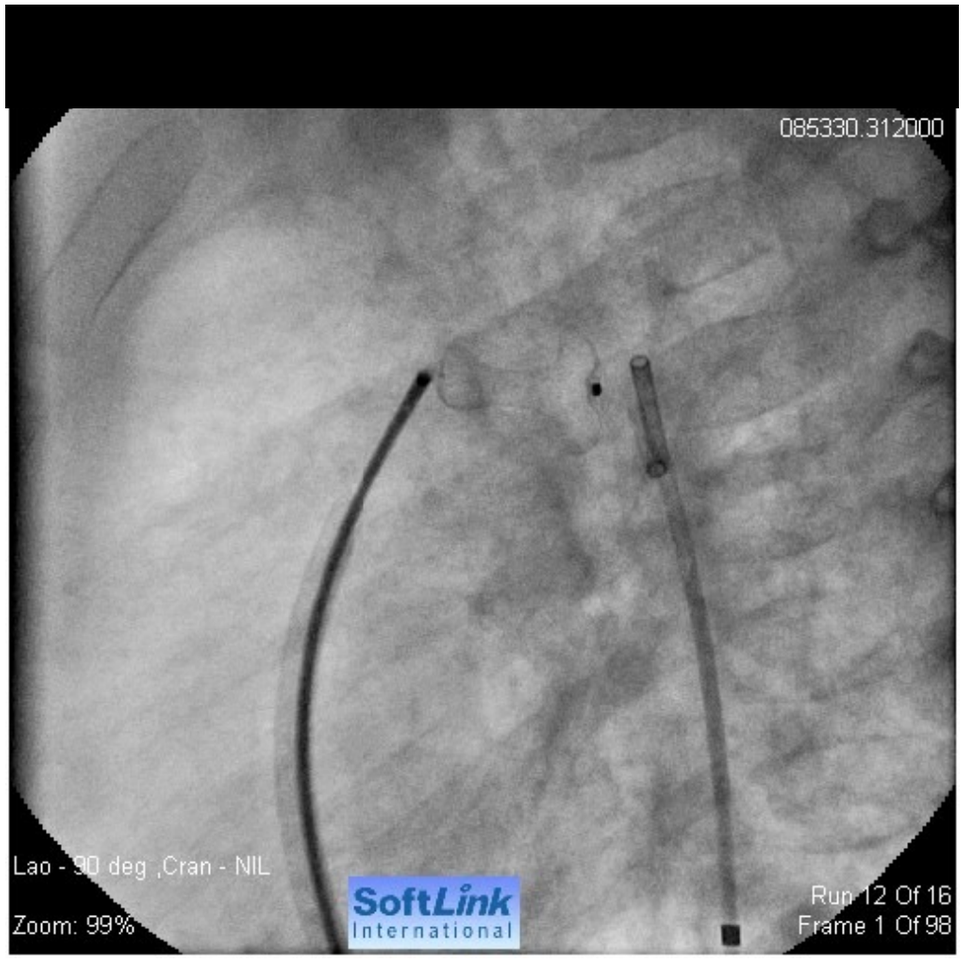
*the murmur associated with a PDA can be pretty variable

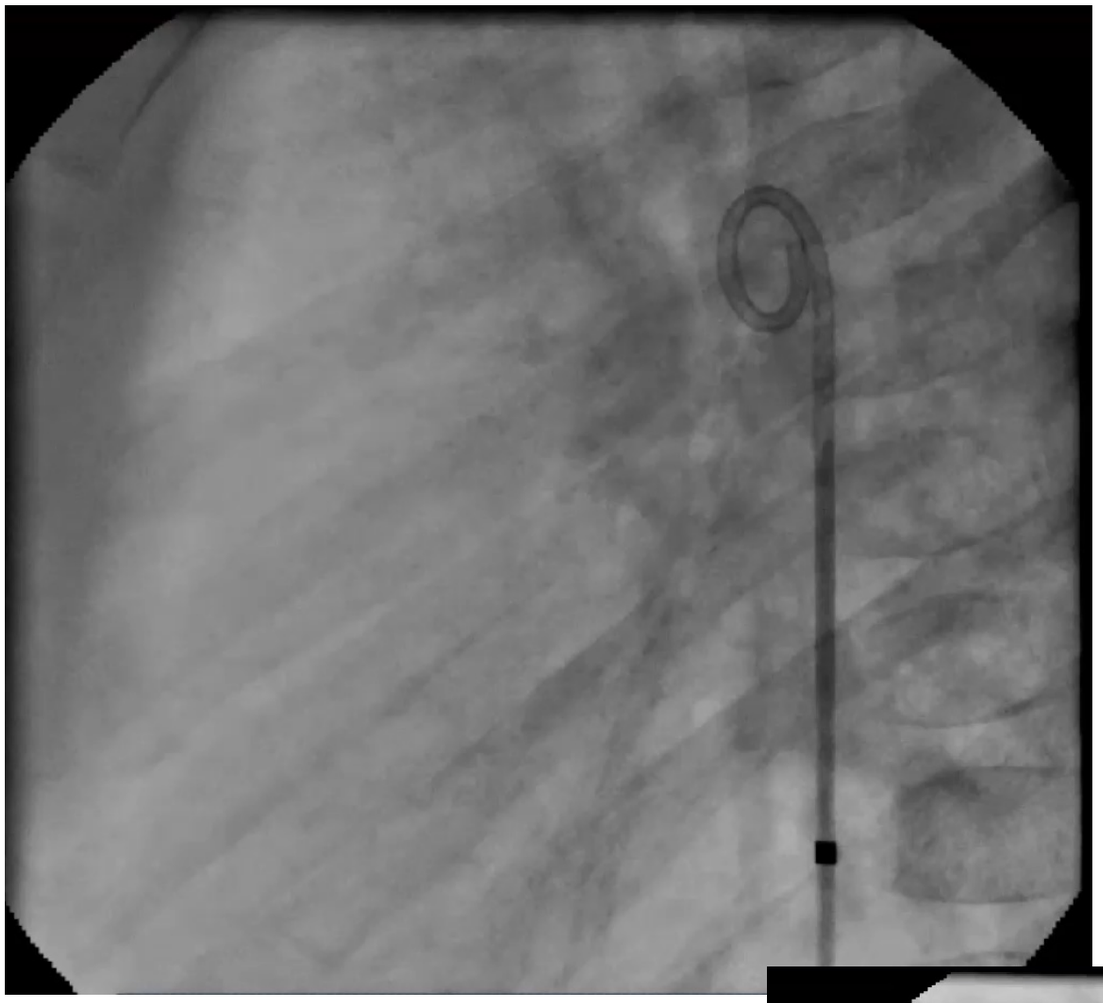
PDA Treatment/Intervention: Close the defect

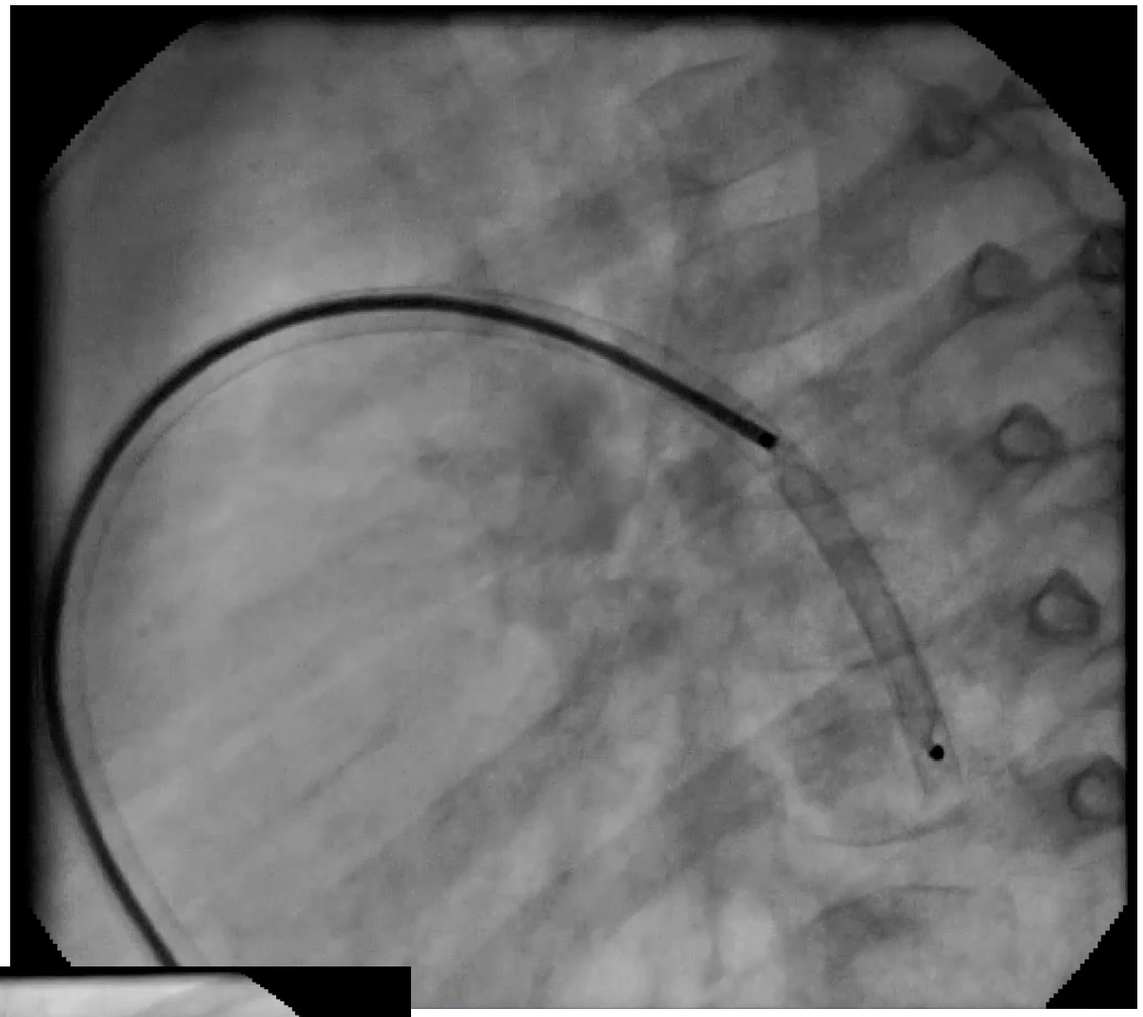


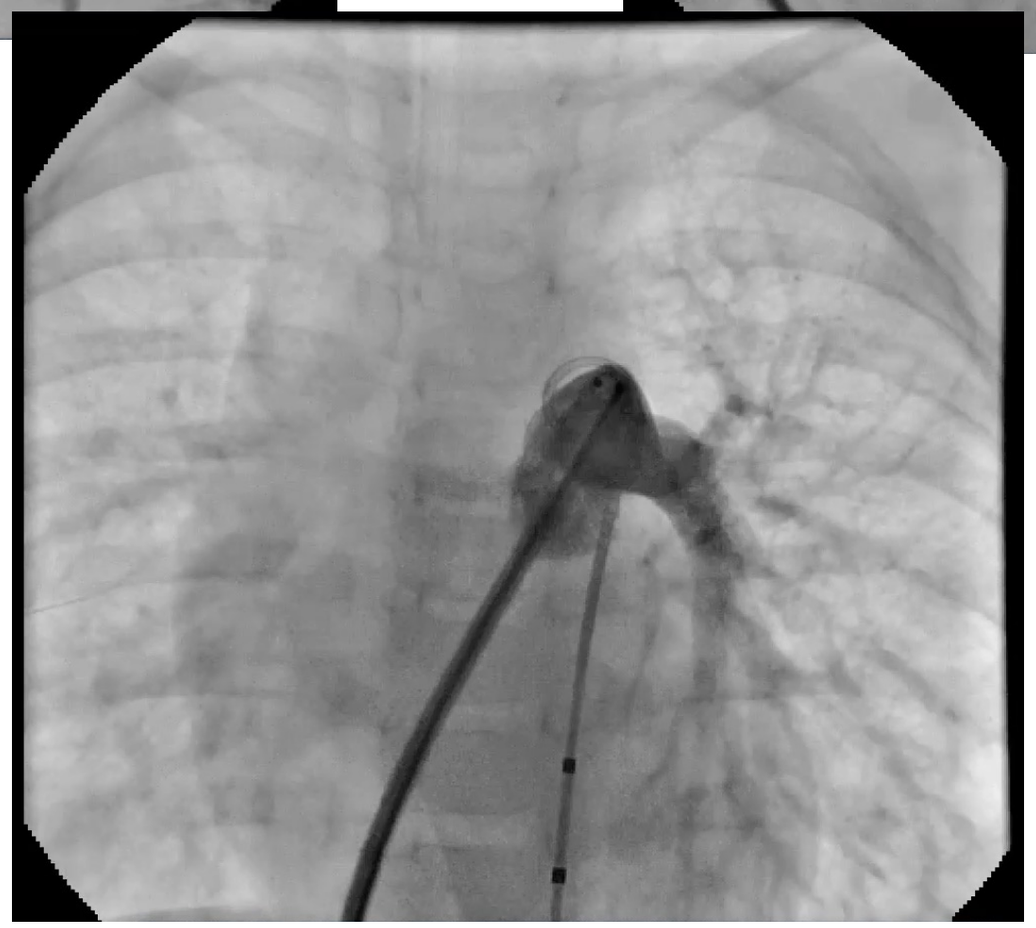
- Premature Infants
  - Prostaglandin synthesis inhibitors
    - Indomethacin & Ibuprofen
  - Surgical ligation

- Infants/children
  - Spontaneous closure by 1 year of age; if not, it is unlikely to close
  - Catheterization based closure
  - Surgical ligation









Act III: Acyanotic Congenital Heart Defects Review

- Acyanotic congenital heart defects usually result in a ”left-to-right” shunt and pulmonary overcirculation

- When pulmonary overcirculation is combined with volume overload of the left heart, signs and symptoms of congestive heart failure ensue

- Significant acyanotic heart defects can result in Eisenmenger Syndrome if not treated appropriately

Act III: Acyanotic Congenital Heart Defects Review (continued)

- Some defects will close on their own, in which case initial treatment is geared to reduce signs and symptoms of CHF

- These anatomically straightforward lesions can be treated with surgery or sometimes with transcatheter device closure

In conclusion

- The fetal circulation is a model for congenital heart disease as it entails shunts, vascular beds with different resistance, and mixing of oxygenated and deoxygenated blood

- Blood generally follows the path of least resistance

- Remodeling occurs in the face of pressure and volume challenges, which can have adaptive or maladaptive effects

- Pulmonary overcirculation plus left-sided volume overload results in signs and symptoms of congestive heart failure

- Unrepaired left to right shunts can result in Eisenmenger syndrome

Thank you for your attention!

Questions??

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