Congenital Heart Disease

This is pat I of a series of 4 lectures that are intended to give us a sense of the basics - how to think about heart disease. Congenital heart disease, our topic for today, is a very diverse group of conditions. Because Duke has a very good pediatric cardiology department, some of the things that you can see here don't even have names yet.

APPROVED

Body & Disease 2011

Keys to Congenital Hearts

- Normal Cardiac Anatomy
- Developmental Embryology
- Pathophysiology
  - Flow direction
  - Volume/Pressure effects

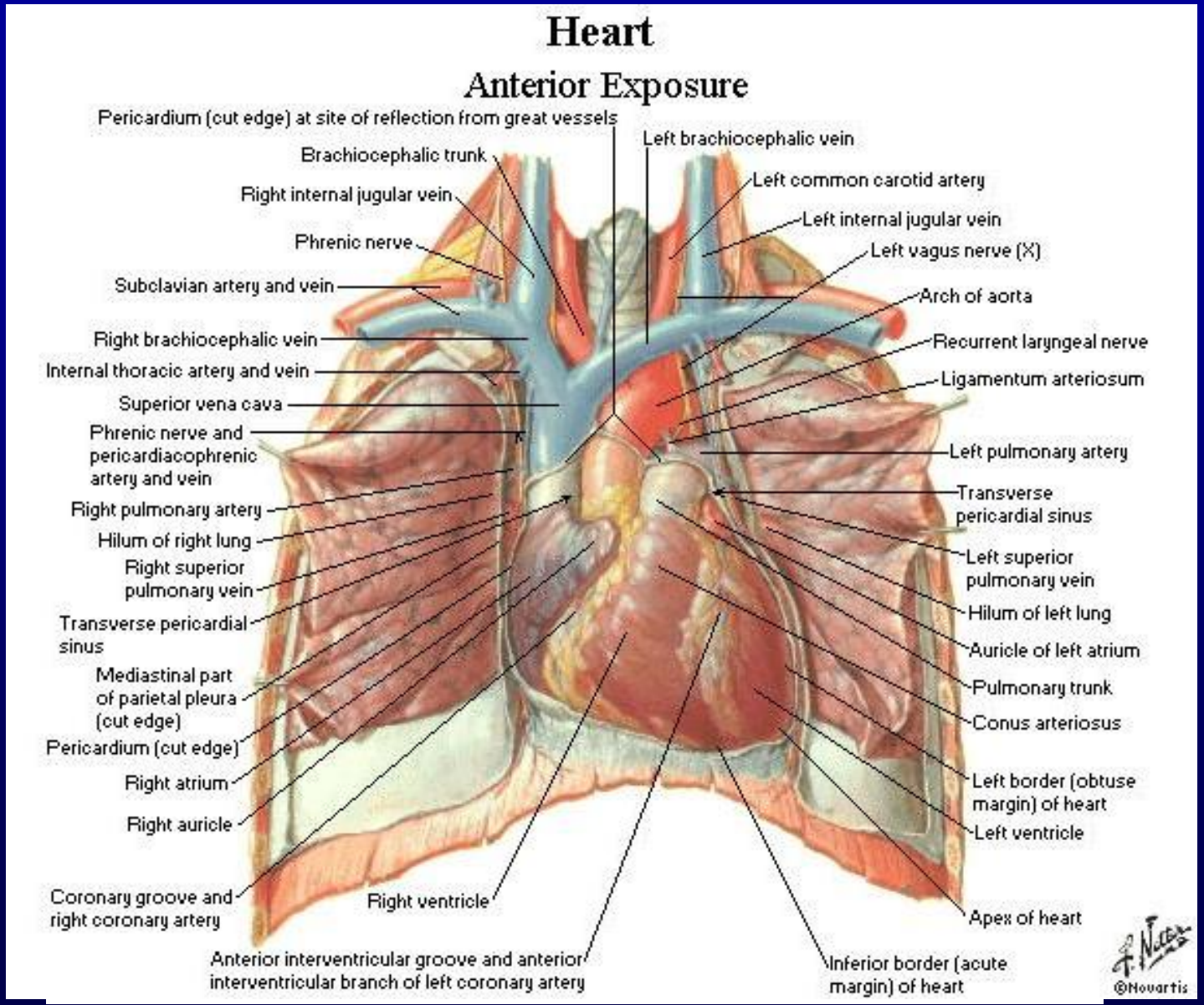
Important for knowing what kinds of things will be clustered together. Thanks, Dr. Velkey!

Or, "How does the patient present, and what are the long-term effects?"

Remember, we talk about these as being present in kids, but adults, too can present with some of these abnormalities that they've had their entire lives. Often, this means that the adult is dealing with a whole set of problems caused by the untreated congenital abnormality.

A Duke Student's Worst Nightmare

Look Familiar?



Cardiac Development

- **Elongation and Folding of Tubular Heart**
  - BMP family – directs cardiac lineage
  - Outstrips growth of embryo as a whole
- **Partitioning into Four Chambers**
  - Endocardial cushions – atrioventricular division
    - BMP's, TGF-β family
  - Ventricular Septum – from apex to base
    - Left and Right Ventricles are specific
    - HAND1, HAND2 – chamber restricted genes
  - Atrial Septum
    - Primary Septum: from roof to cushions
    - Secondary Septum: "closes" fenestration

Form tricuspid and mitral valves

Note the direction here

Complicated by differing fetal & adult blood flow patterns

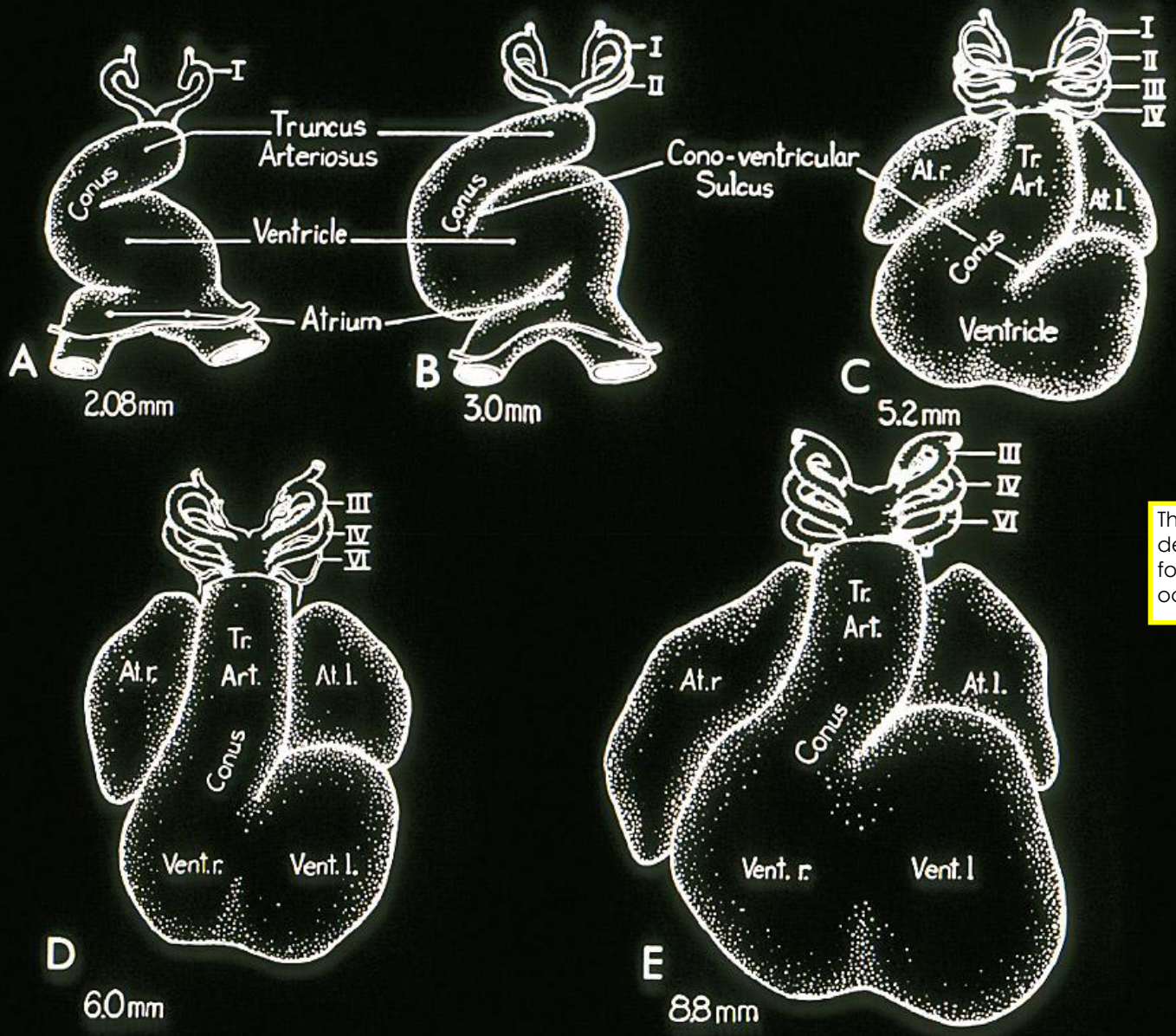
Genetics, not location, determines tissue characteristics

Unlike the ventricular septum, this is two muscular bands

Foramen ovale

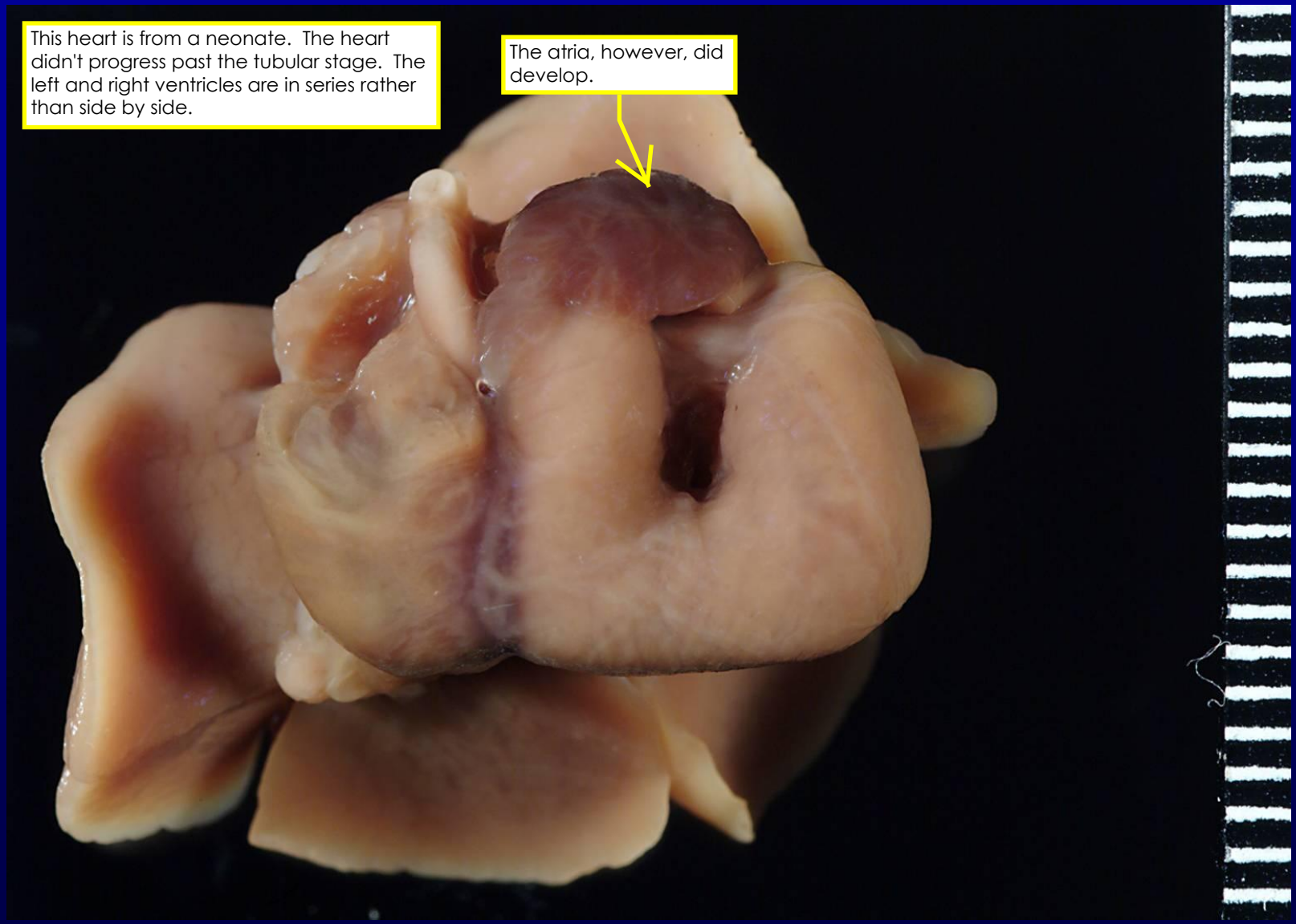
Really quick embryology review: the heart forms from two tubes that fuse around day 19 of gestation; the area of fusion contains (from rostral to caudal): the bulbis cordis (future RV and ports of outflow tracts for aorta and pulmonary trunk), the primitive ventricle (future LV), the primitive atrium (anterior parts of LA and RA), and the right and left horns (not fused - still separate), which form the superior vena cava and part of RA).
Then folding occurs - bulbis cordis moves anteriorly, inferiorly, and to the right, primitive ventricle moves left.

Animation: http://www.indiana.edu/~anat550/cvanim/htube/htube.html



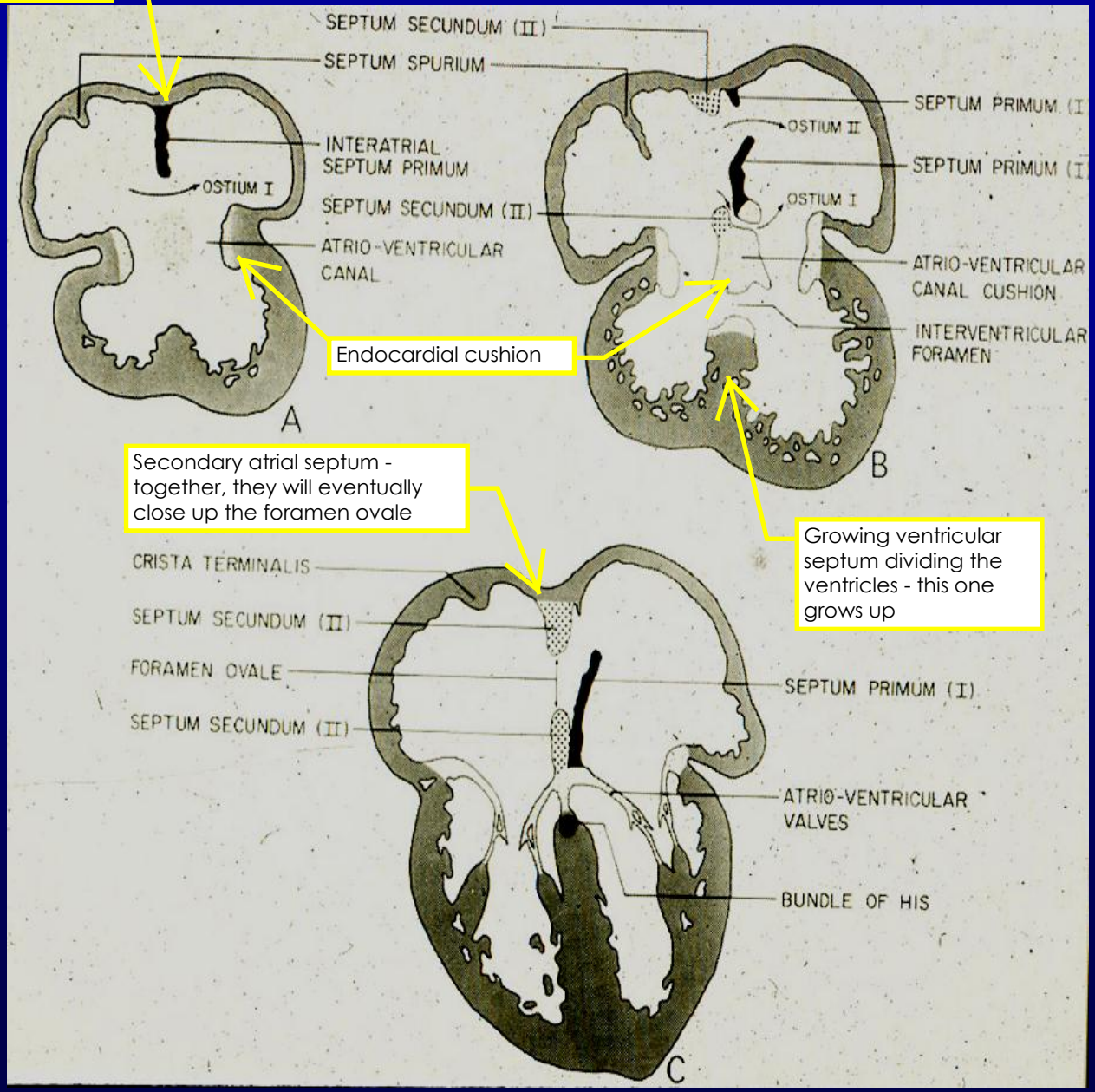
This basically demonstrates that folding and dividing occur together.

Tubular Heart



Atrial septum - this one grows down

A view of how things develop on the inside



Endocardial cushion

Secondary atrial septum - together, they will eventually close up the foramen ovale

Growing ventricular septum dividing the ventricles - this one grows up

Cardiac Development

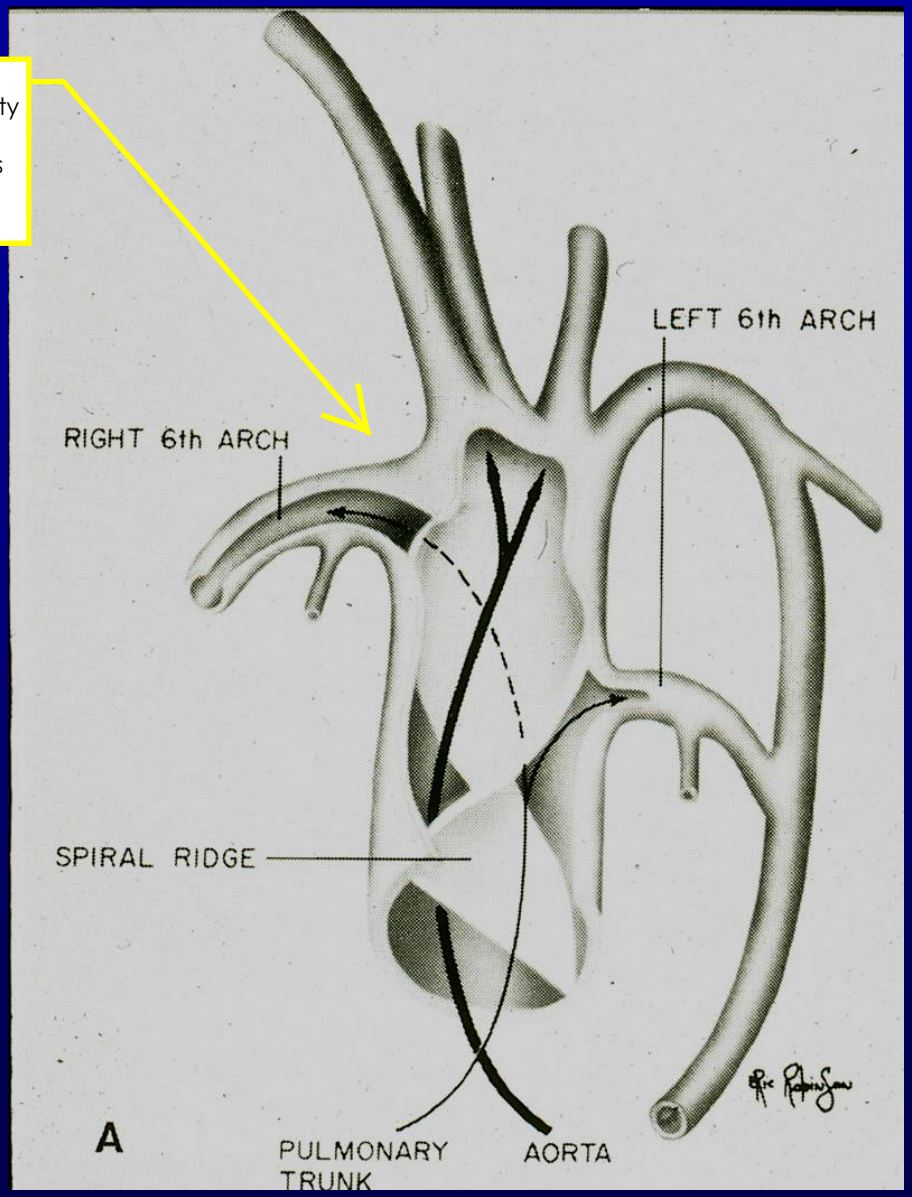
- Great Vessels
  - Common Truncus Arteriosus
  - Spiral septum – forms to separate into aortic and pulmonary trunks
- Aortic Arches
  - Involute or evolve
    - 3rd arches – internal carotids
    - 4th arch – right subclavian; left aorta
    - 6th arch – pulmonary arteries

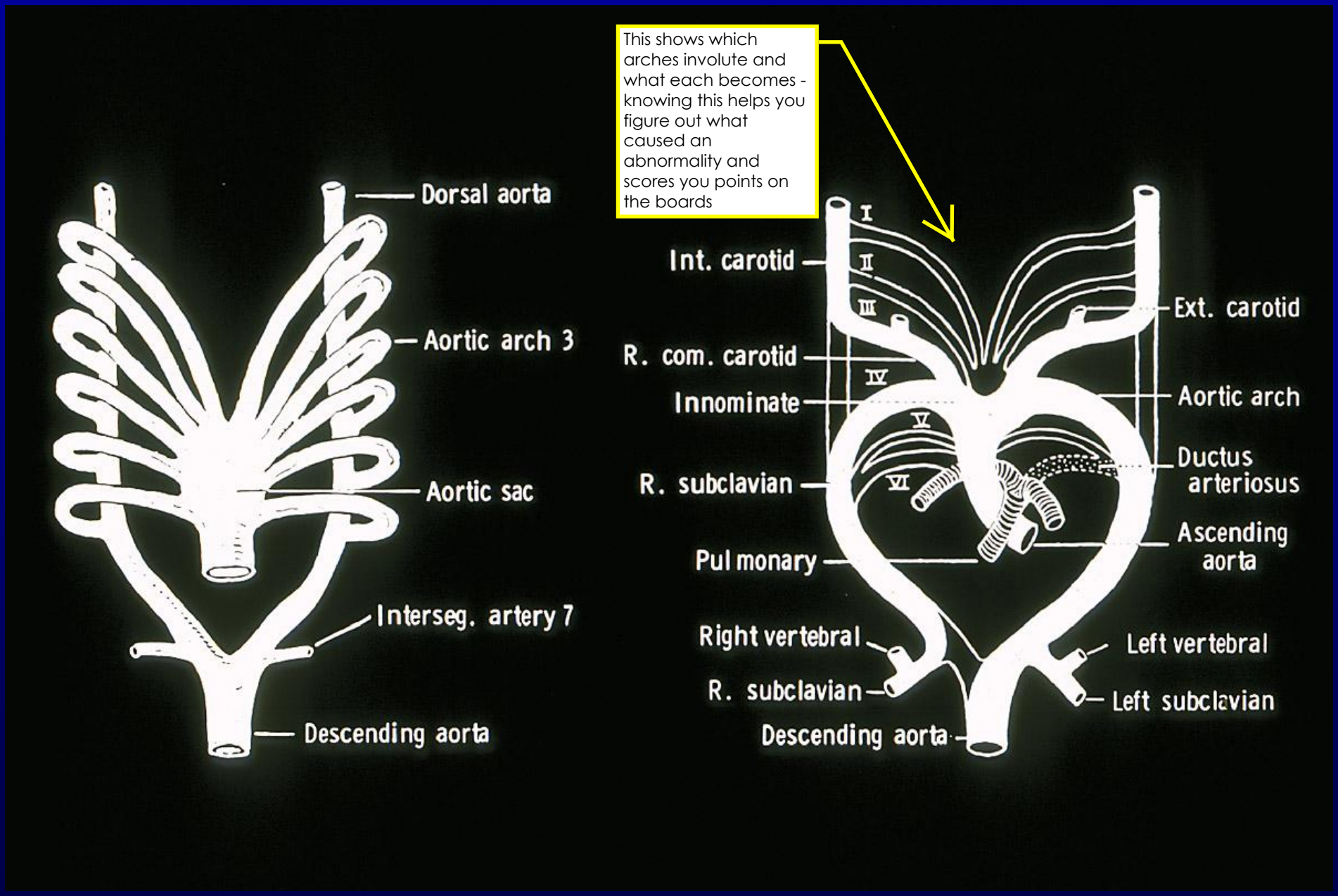
Remember, you start out with one great vessel that grows a spiral septum

Pulmonary and systemic arteries are derived from these

Favorite board question: which ones involute and which ones evolve?

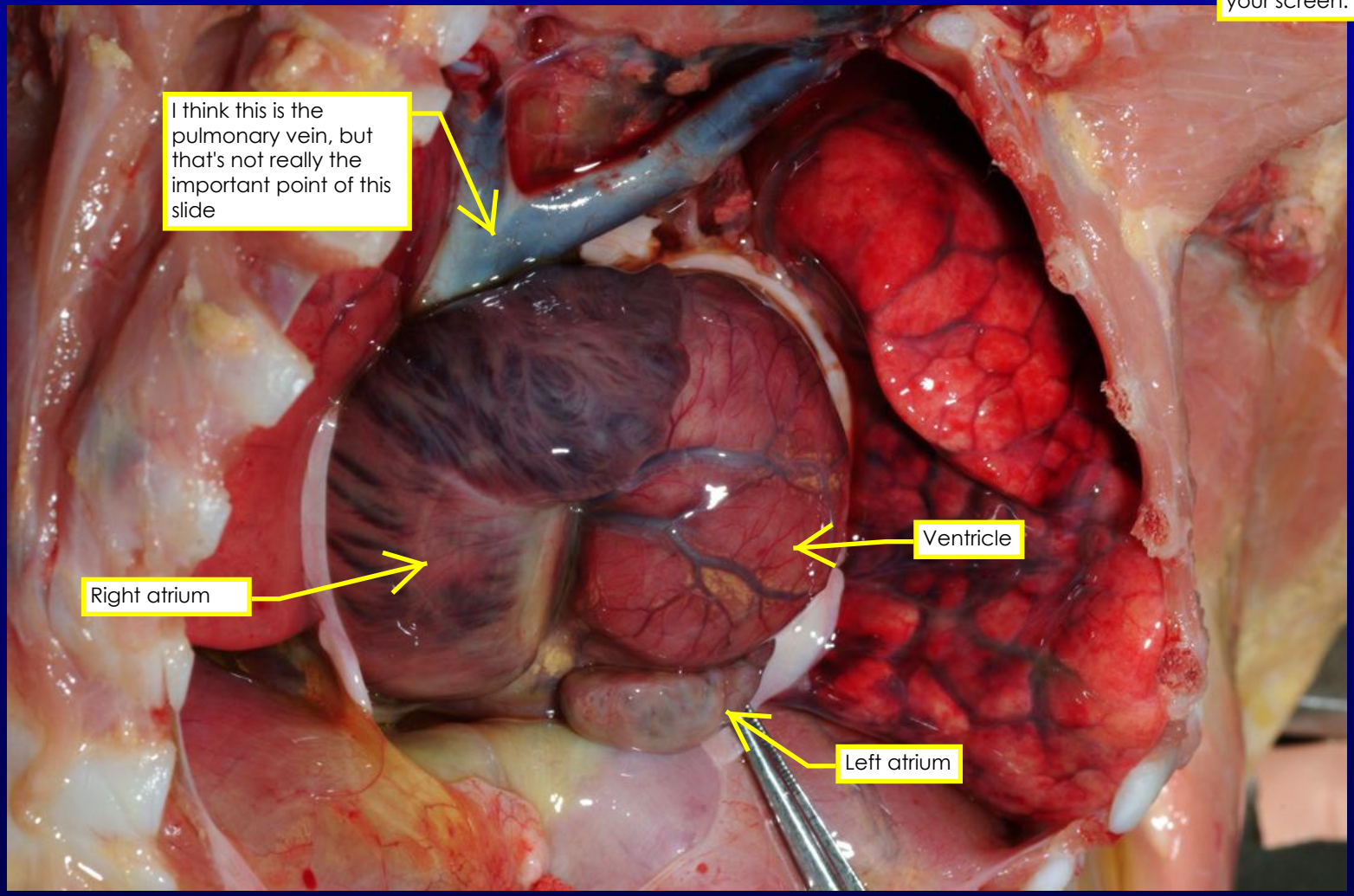
The formation of the spiral septum is a pretty complex event - multiple abnormalities can occur during this process



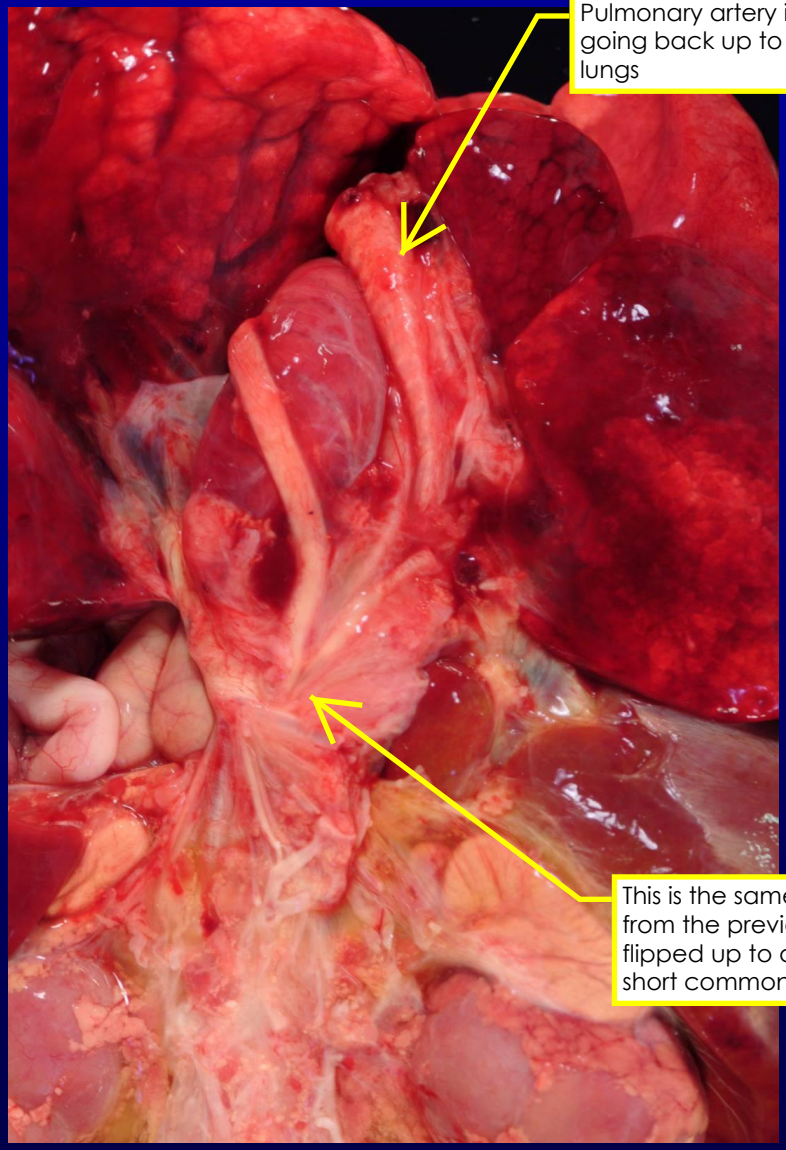

This shows which arches involute and what each becomes - knowing this helps you figure out what caused an abnormality and scores you points on the boards
Dorsal aorta
Aortic arch 3
Aortic sac
Interseg. artery 7
Descending aorta
I
II
III
IV
V
VI
Int. carotid
R. com. carotid
Innominate
R. subclavian
Pulmonary
Right vertebral
R. subclavian
Descending aorta
Ext. carotid
Aortic arch
Ductus arteriosus
Ascending aorta
Left vertebral
Left subclavian

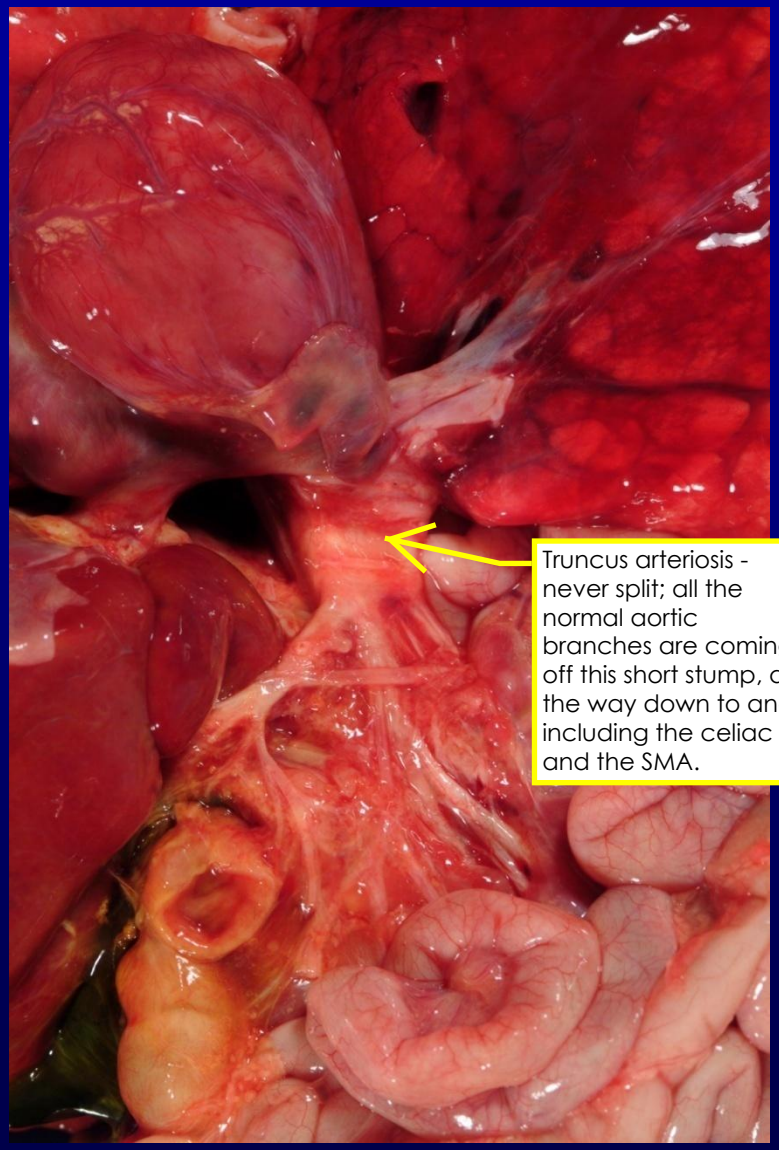
Arch Abnormality?

This heart is oriented the way it actually sat in the chest - the head is at the top of your screen.

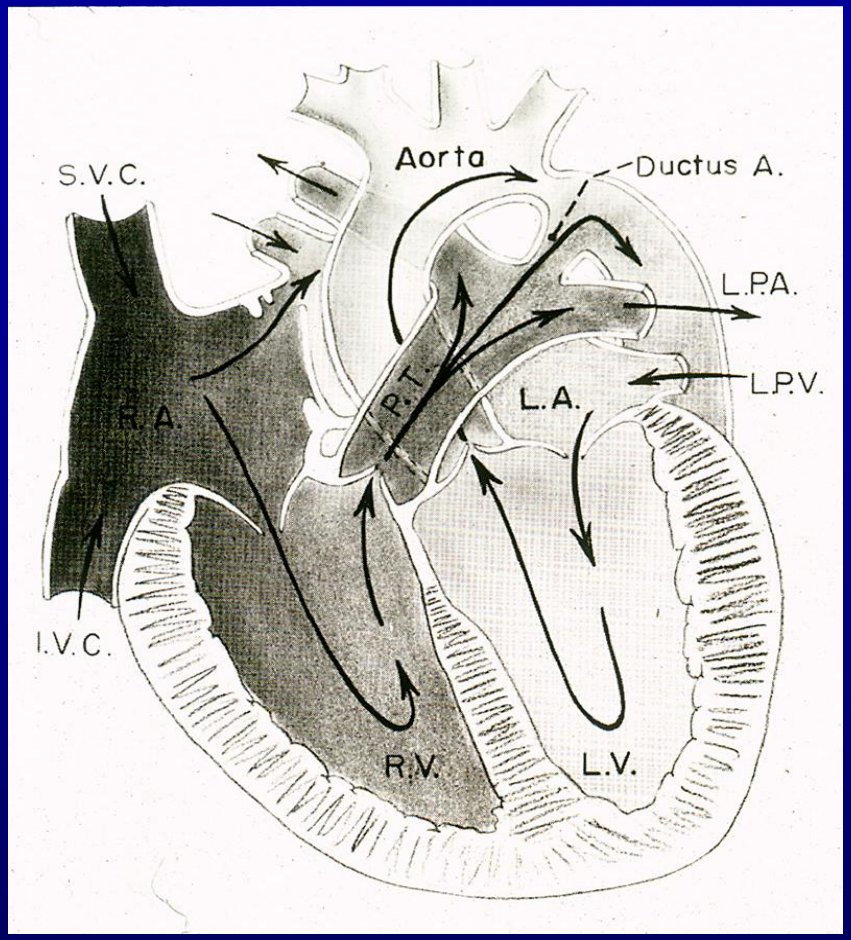


Arch Abnormality





To Dr. Dibernardo's knowledge, this has never been described before or since.



Completion – When?

1st 8 weeks or so

Fetal Flow Pattern?

Changes at Birth?

Pressure change (lower pressure in lungs) closes "flap valve" of foramen ovale

Remember, the lungs are largely excluded from the flow pattern (high pressure compared to systemic flow).

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Unlike diagrams you may have seen, the ductus is not a wimpy little branch. It's as big as, if not bigger than the main pulmonary artery and aorta. A problem with the ductus may result in a small, hypoplastic aorta.

Many named syndromes will encompass heart defects with other problems in the face or other solid organs.

Congenital Heart Disease

- **Incidence:** 0.3 to 1.0% of live births

This number is small and seems to be stable, but because Duke is a tertiary care center for these things, we get a lot of them.

We actually don't know all of it, but this is a partial list of some that we do know

- **Etiology:**
  - Viral – rubella
  - Teratogens
  - Genetic : Down's , Turner's, VATER/VACTERL

These two chromosomal anomalies have a high incidence of congenital heart disease. Downs trisomy 21; Turners XO

Vertebral anomalies
Anal atresia
Cardiac defects
Trachea problems (ex. tracheal esophageal fistula)
Esophageal atresia (blind stump)
Renal defects
Limb defects

Diversity of associated defects shows that some complicated genetic feature ties these together

Pathophysiology

- Flow abnormalities
  - Direction of flow/shunt
    - Determinants
      - Parts ← In other words, blood can only flow through structures you have.
      - Downstream compliance ← Blood takes the path of least resistance (e.g. lungs instead of body in most instances)
      - Generally left to right ← Most shunts flow L->R; having long-term problems can switch this around, though.
  - Obstruction
  - Effect on heart and organs (lungs)
    - **Volume hypertrophy** – enlarged chamber
    - **Pressure hypertrophy** – mass increase
    - **Right to left shunts (cyanosis)** ← Obstruction, physiologic changes can cause this

The chamber getting blood dumped in will hypertrophy/dilate

A chamber having to pump harder will get thicker

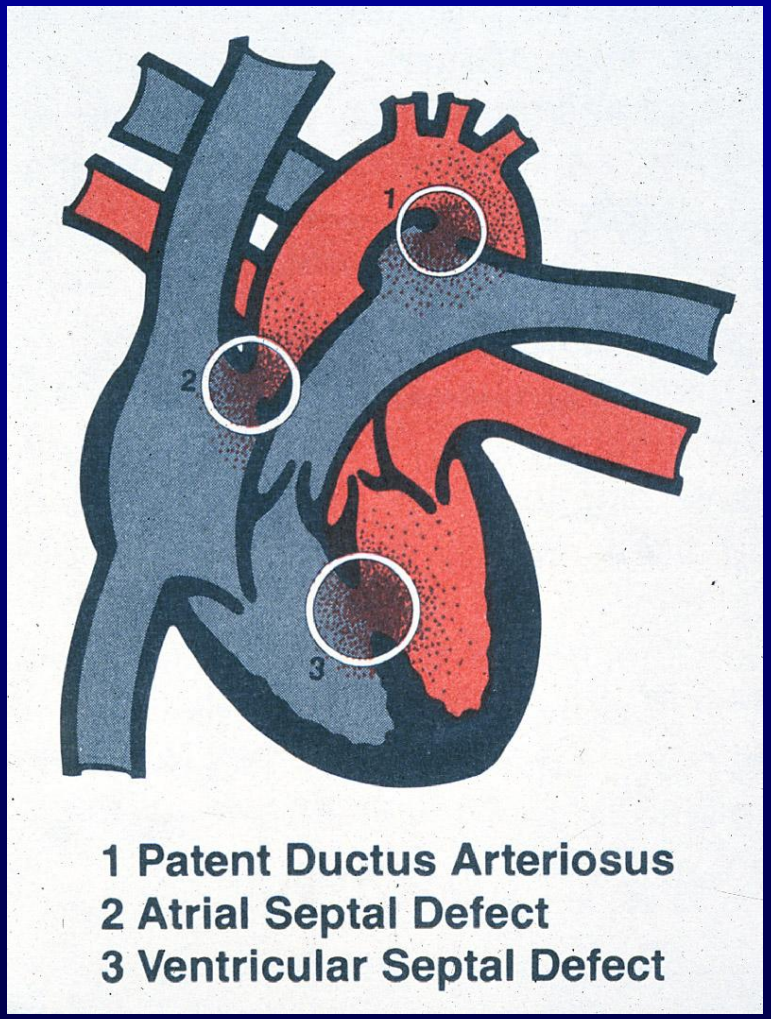
Now onto different categories of defects:

Isolated Shunts

"A hole in the heart" - an opening that shouldn't be there, but is

.

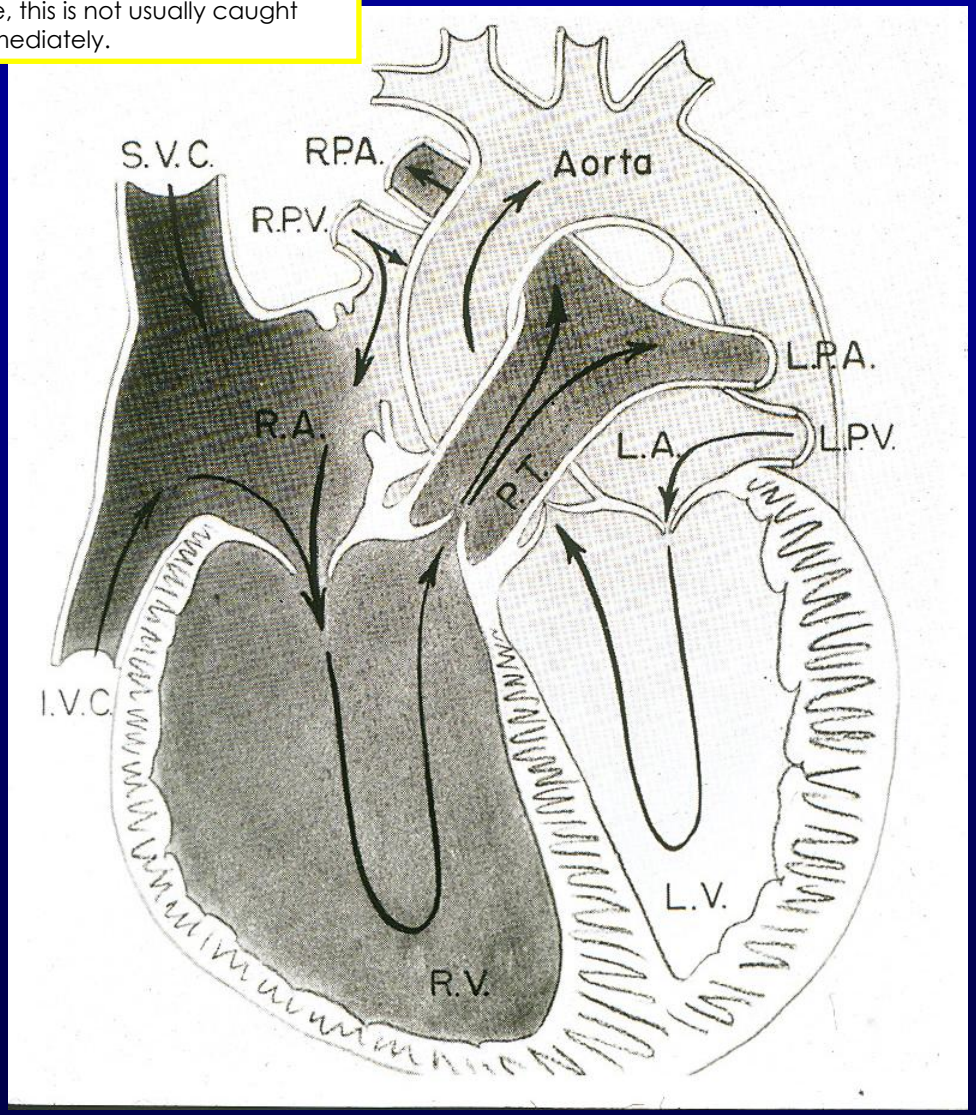
http://www.youtube.com/watch?v=I-h4A7bF8wQ - Tangentially appropriately-themed '90s nostalgia. If I were a pediatric cardiologist, this would be my theme song.



- **Ventricular Septal Defects**
  - **25-30%** Most common
- **Atrial Septal Defect**
  - **10-15%**
- **Persistent PDA**
  - **10-20%**
- **Truncus Arteriosus**
  - **2%**
- **Anomalous Pulmonary Venous Return**
  - **Rare** But you will see this and truncus arteriosus at Duke

Atrial Septal Defect

L->R shunt: first RA hypertrophies, then RV. Because oxygenated blood is being pumped into R side, this is not usually caught immediately.



Defect in septum primum, close to center of heart

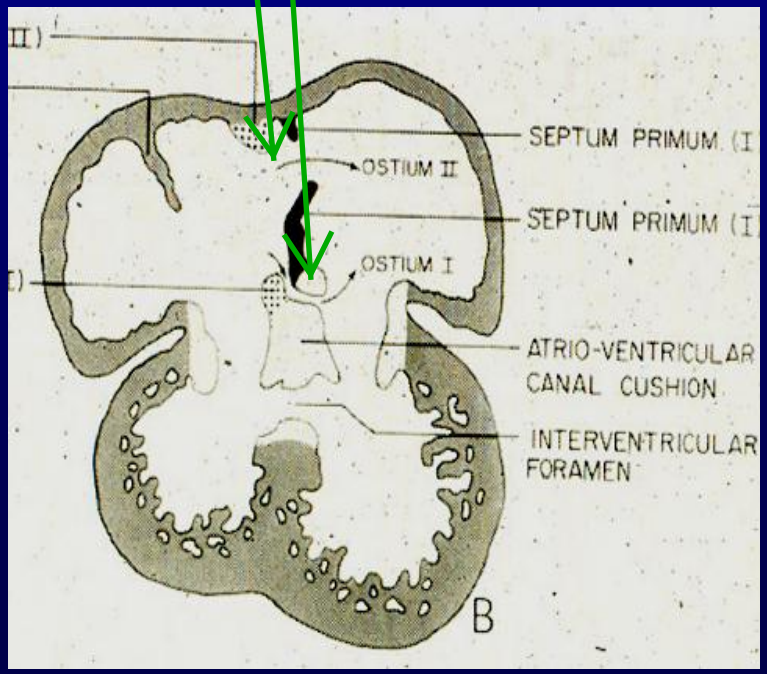
1) Primum Type

More prevalent; larger opening in secundum

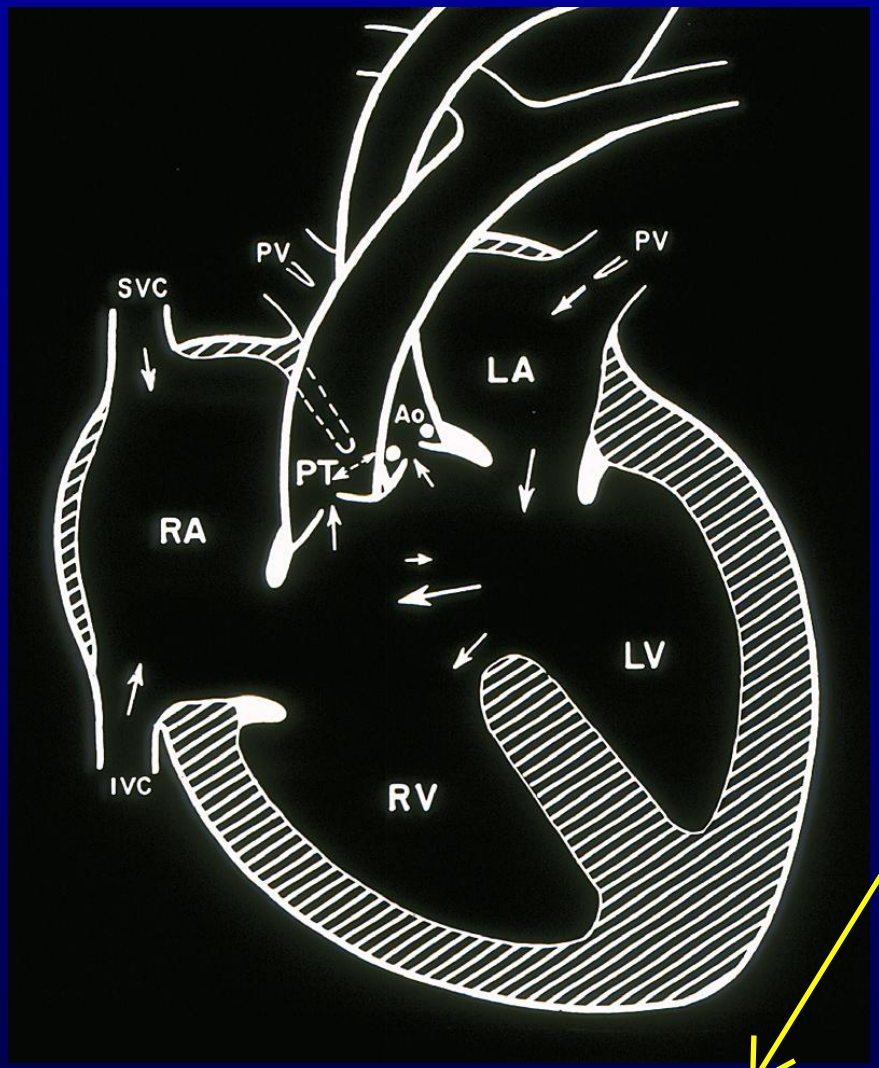
2) Secundum Type

3) Patent Foramen Ovale

-Valve competency



Atrial Septal Defect



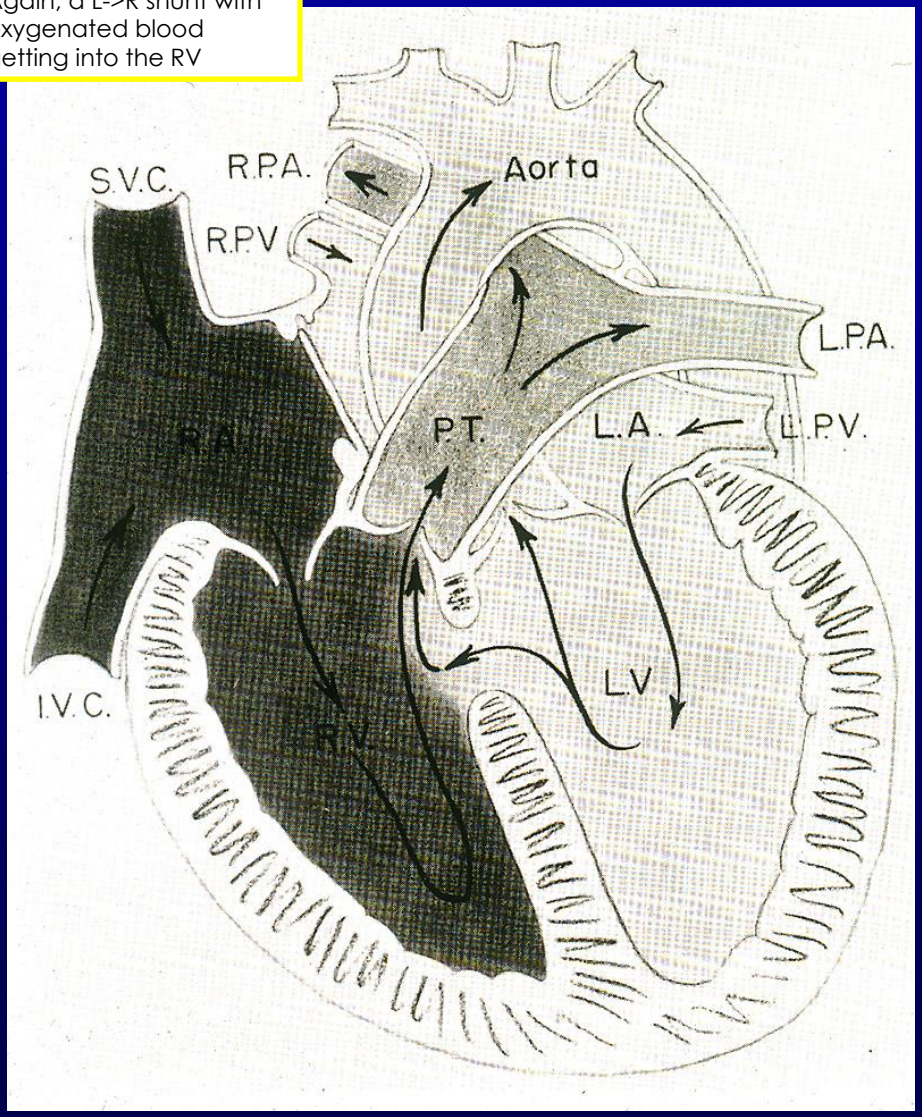
"Super deluxe ASD" - a common opening in the center of the heart due to failure of endocardial cushions to separate chambers - size of opening can vary greatly; the closer to natural anatomy you are, the easier it is to repair

Endocardial Cushion Defect – AV Canal

Dr. Dibernardo read the slide

Ventricular Septal Defect

Again, a L->R shunt with oxygenated blood getting into the RV



- Membranous (high)
- Muscular
  - Anterior
  - "Swiss Cheese"
- Volume increase
- Pressure Increase
- Long Term?

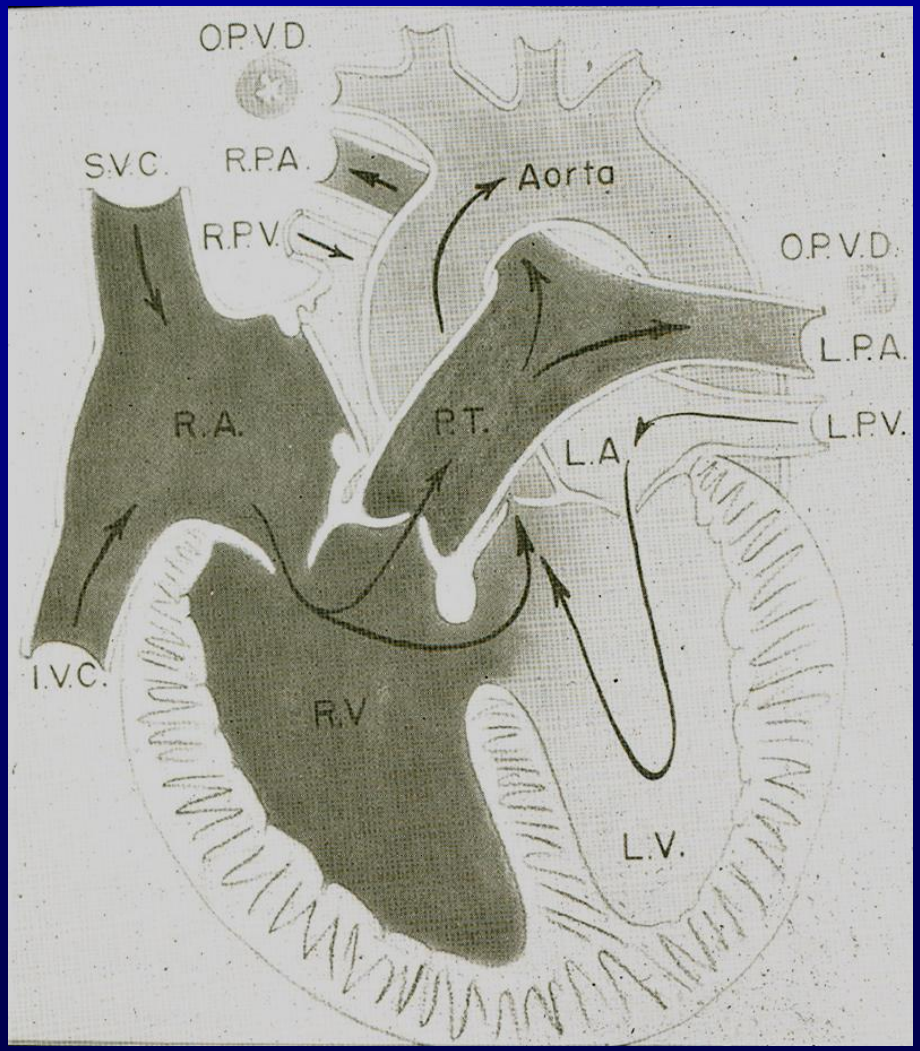
Can be anywhere, tend to be anterior

Multiple holes, hard to close because it's hard to find all of them

In RV, which gets the extra blood

In lung - causes lung damage over time

VSD Endstage



Increased volume into pulmonary vessels

Thickening of vessels causes "obstruction" and pulmonary hypertension

Pressure in right ventricle increases

Up to the pressure of the LV

Right ventricle thickens (already volume hypertrophy)

Shunt direction reverses to bidirectional or right to left

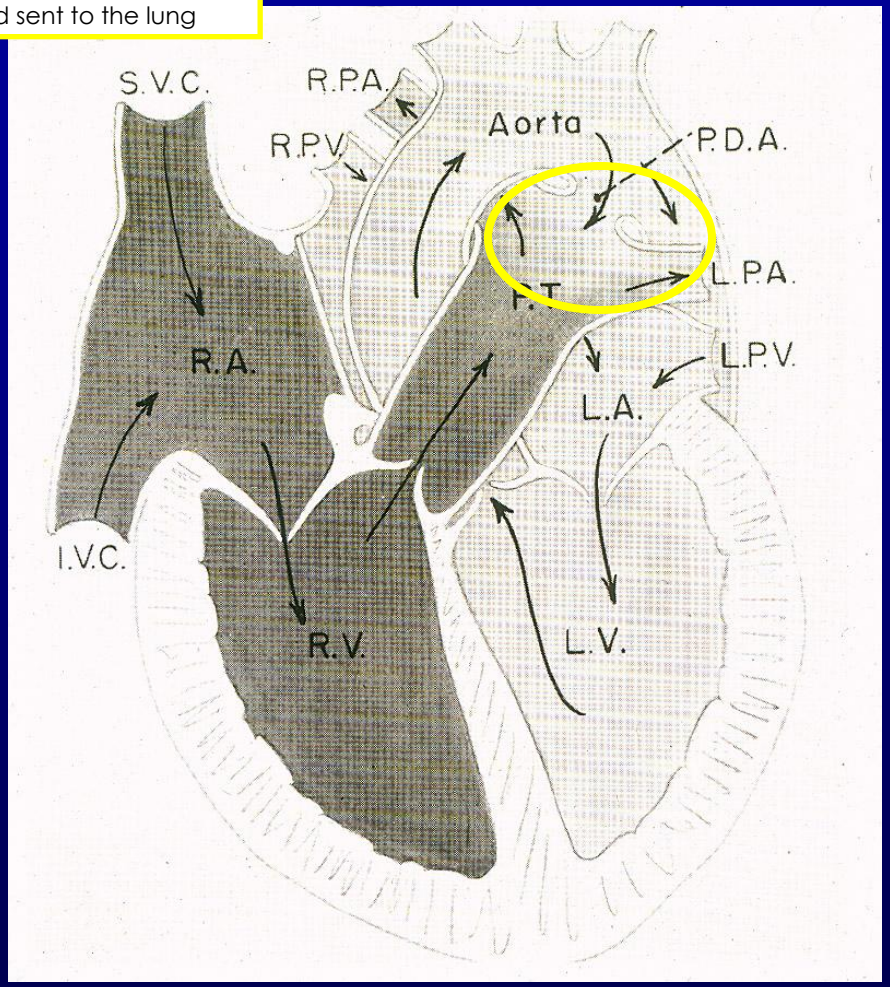
Occurs when RV pressure ≥ LV pressure

PDA

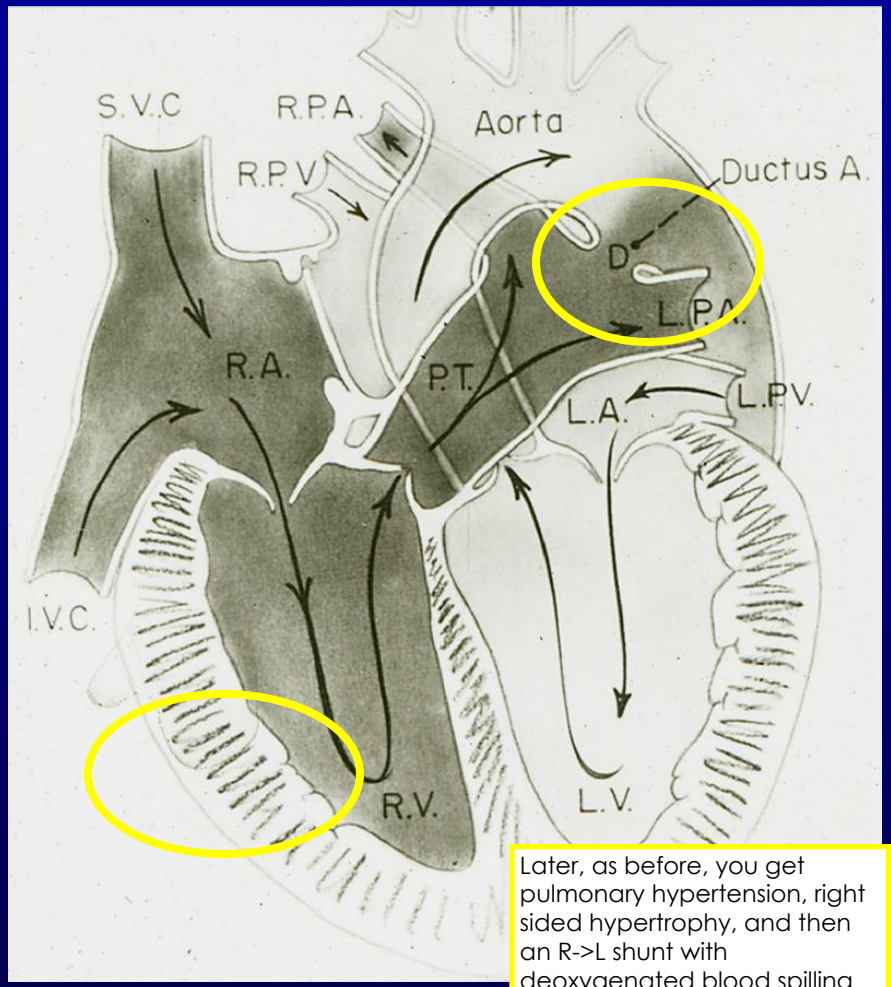
Patent Ductus Arteriosis
(I know, you guessed Public Display of Affection first, then Personal Digital Assistant)

EARLY

At first, the low pressure system to which blood flows is the lung - blood is "stolen" from the aorta and sent to the lung



LATE



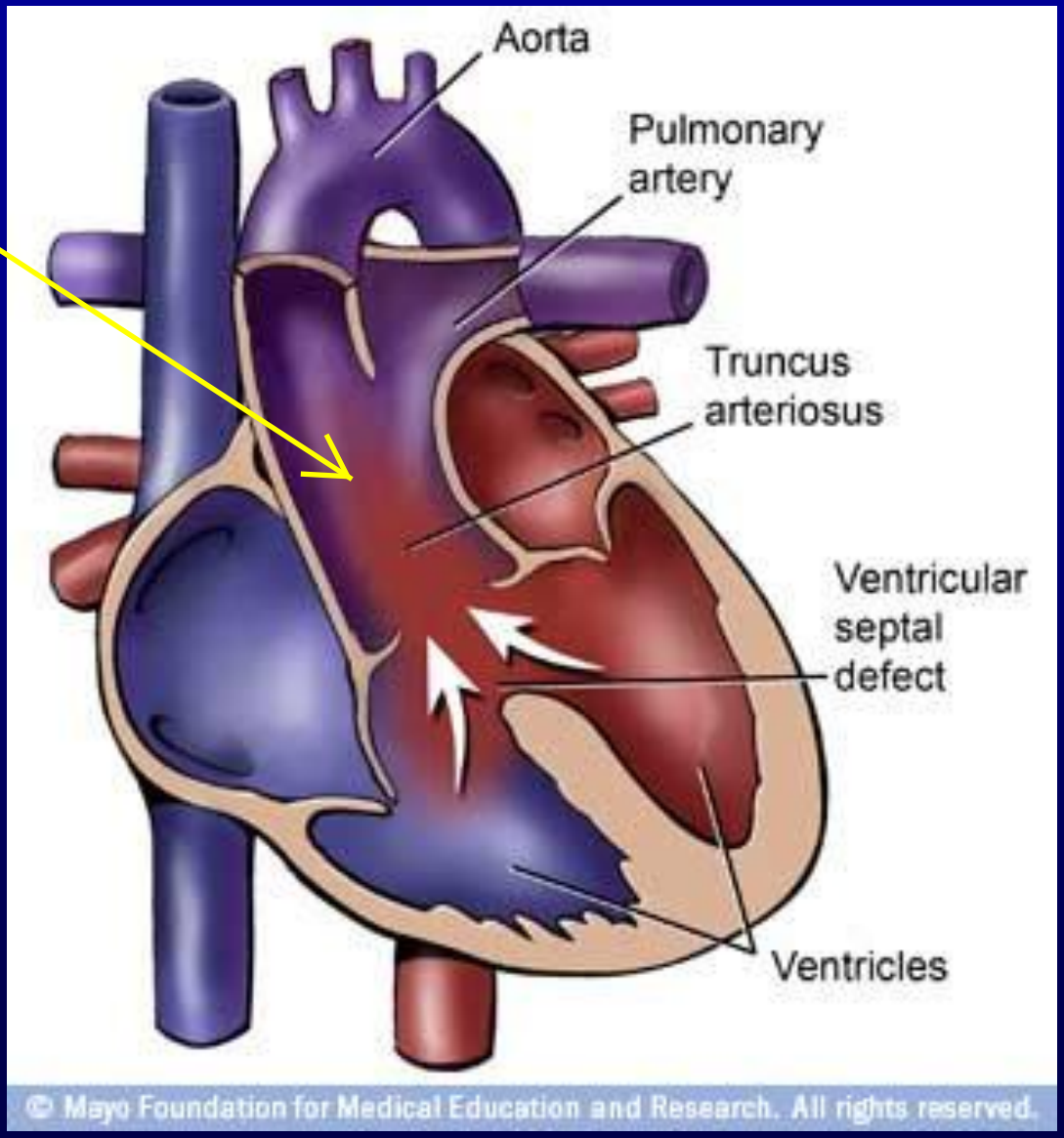
Later, as before, you get pulmonary hypertension, right sided hypertrophy, and then an R->L shunt with deoxygenated blood spilling into the aorta - drop in O_2 sats

Patency maintained by Prostaglandin E, Closure induced with Indomethacin

Why would you *want* a patent ductus arteriosis? If you have an issue with your aortic valve that prevents flow, you may need it to get flow out of the heart. To keep the ductus, give prostaglandin E. To close a PDA (in some cases), you can give indomethacin. Both sometimes require surgery, though

Persistent Truncus Arteriosus

Spiral septum does not form; mixing occurs in the truncus between blood that would normally be in the pulmonary artery and blood that would normally be in the aorta. Remember, blood favors the lungs (more compliance) until you get secondary changes (pulmonary hypertension, etc.)



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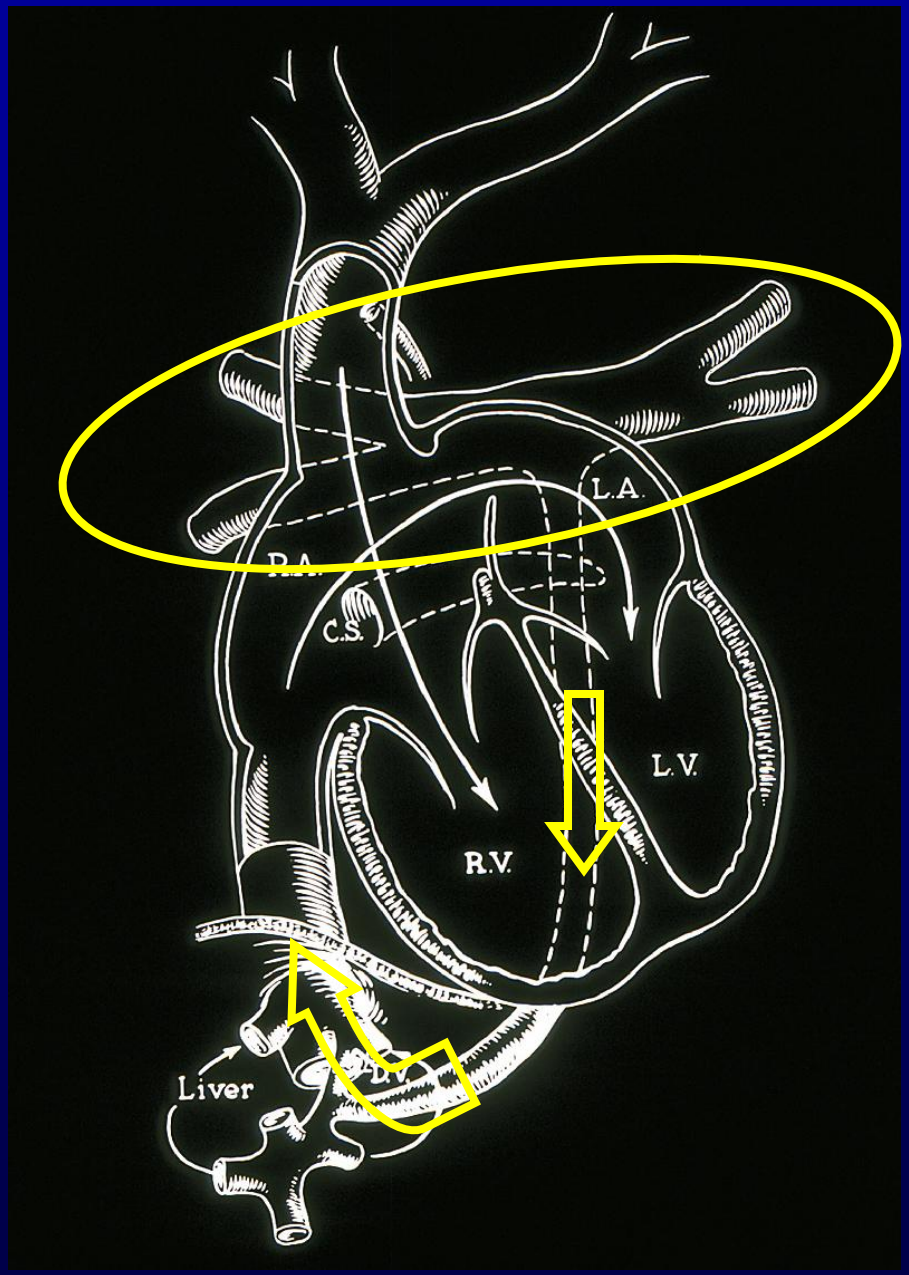
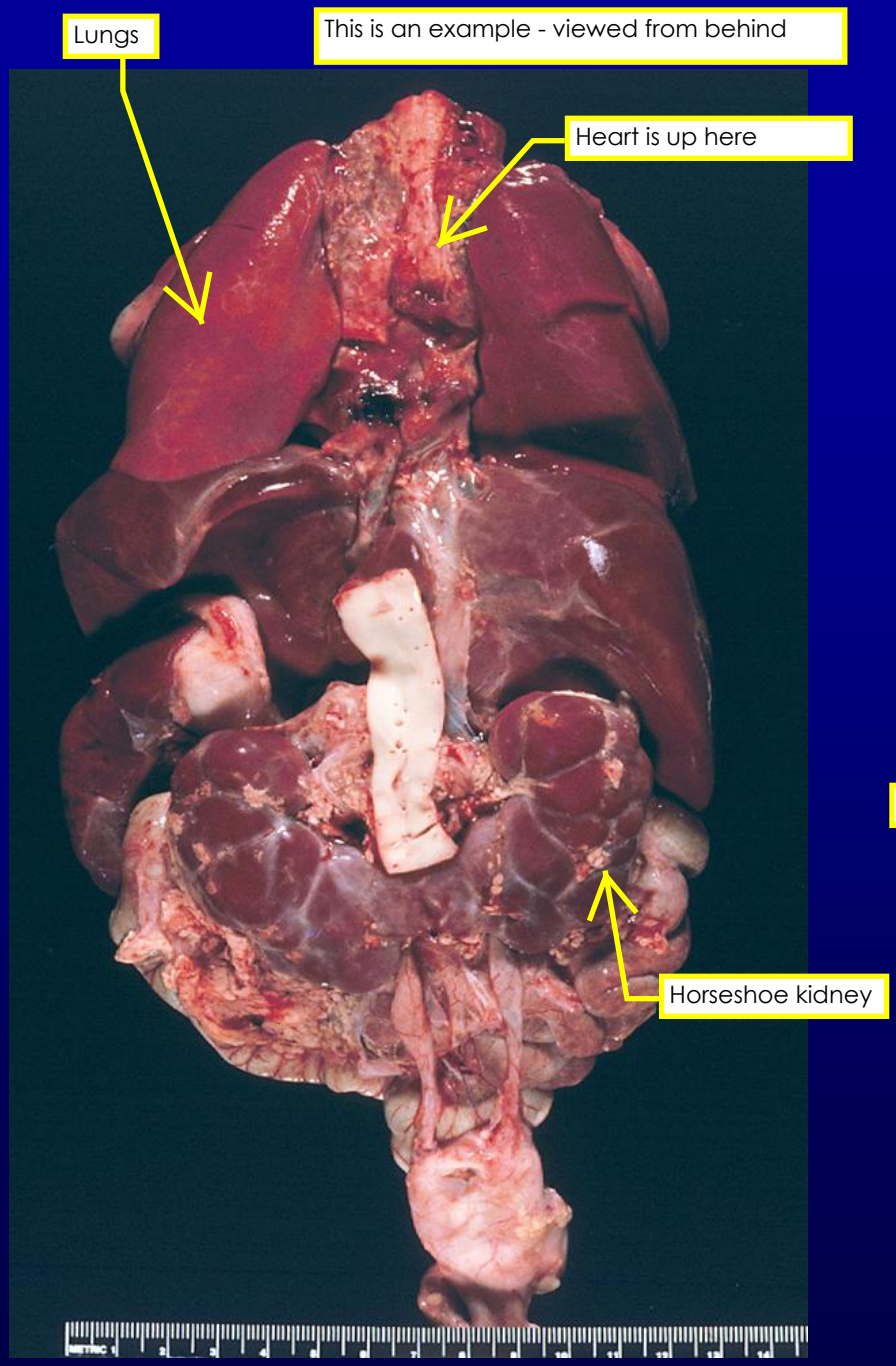


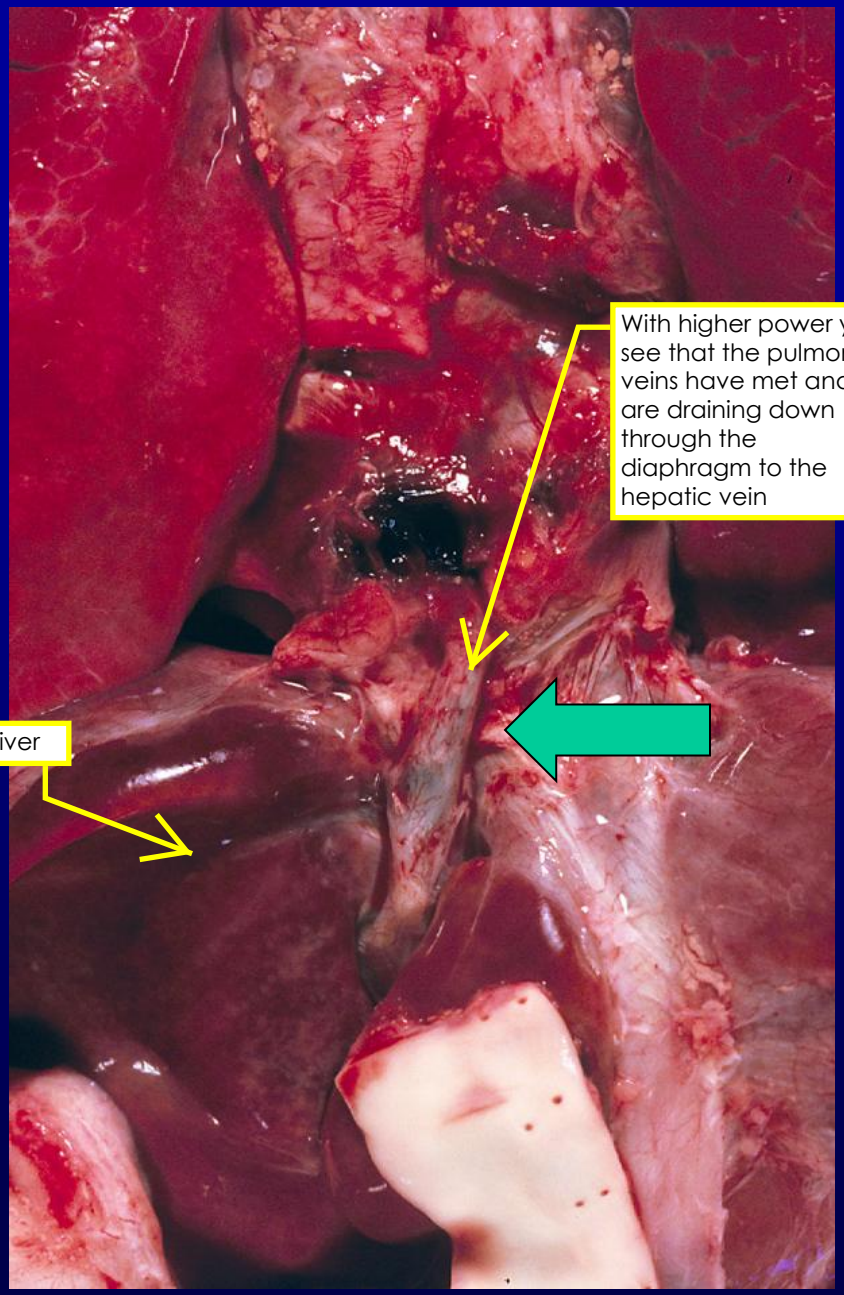
- **Anomalous Pulmonary Venous Return**

Pulmonary veins meet in a separate chamber behind the heart instead of dumping into the LA. "Draining vein" takes this blood down through the diaphragm into the systemic venous system and back to right side

  - **Total or Partial**
  - **Mixing lesion**
    - **Oxygenated and Deoxygenated blood**
  - **Associated shunt**
    - **Allows oxygenated blood to enter systemic circulation**

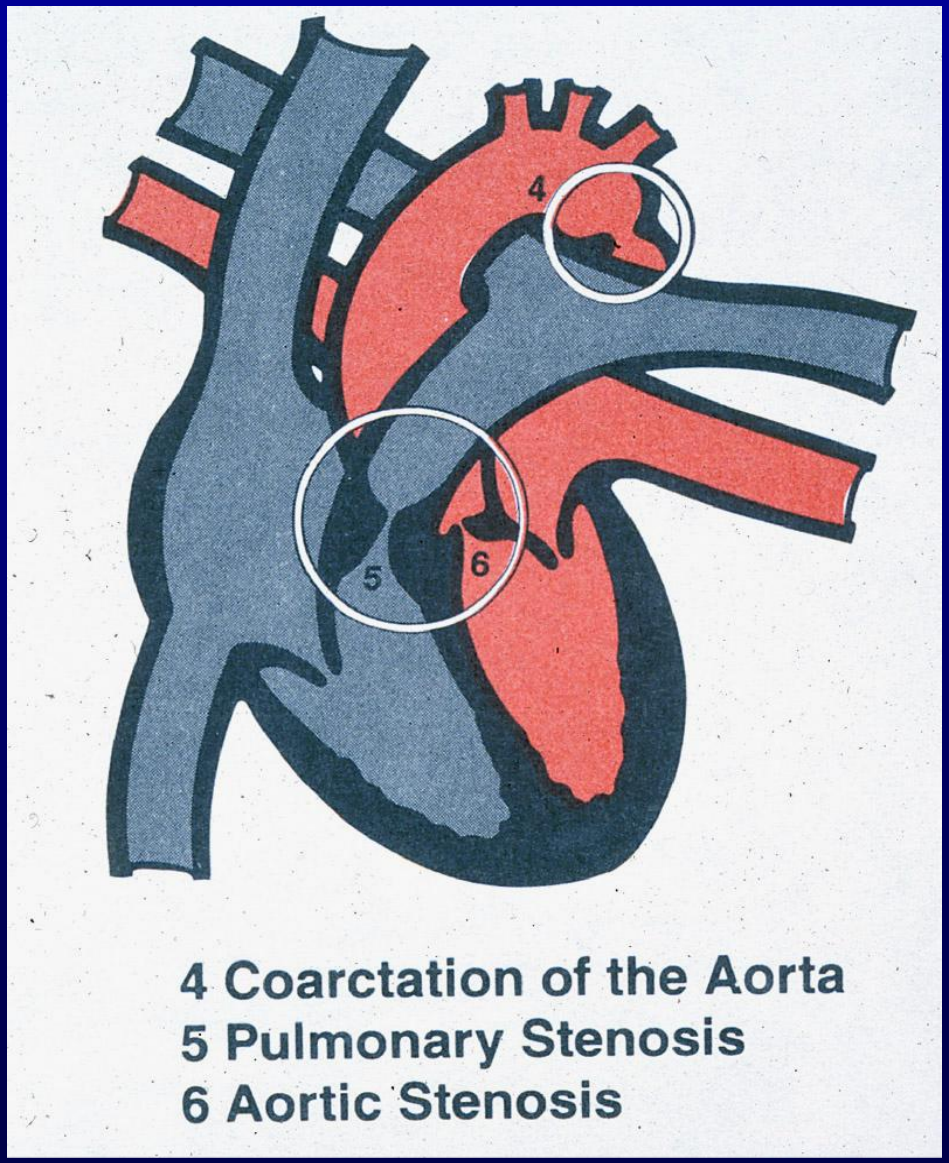
Ductus, ASD, or VSD - if a kid doesn't have one of these, the first step may be creating one until a more complete repair can be done


Lungs
This is an example - viewed from behind
Heart is up here
Horseshoe kidney


With higher power you see that the pulmonary veins have met and are draining down through the diaphragm to the hepatic vein
Liver

Obstruction

Can be muscle or fibrous bands or simply a failure of the structure to form



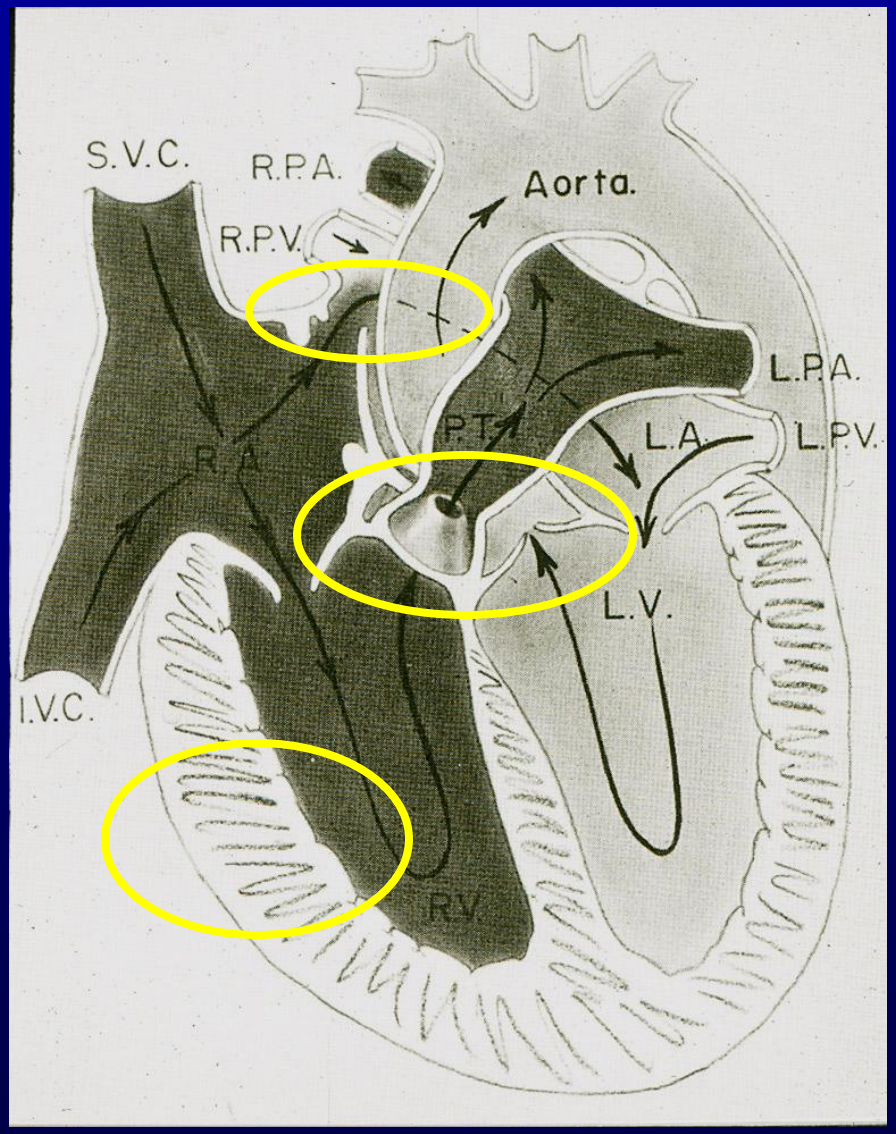
- Pulmonary Stenosis
  - 5-7%
- Aortic Coarctation
  - 4-6%

Can even have an interrupted aortic arch - ascending and descending do not fuse during development

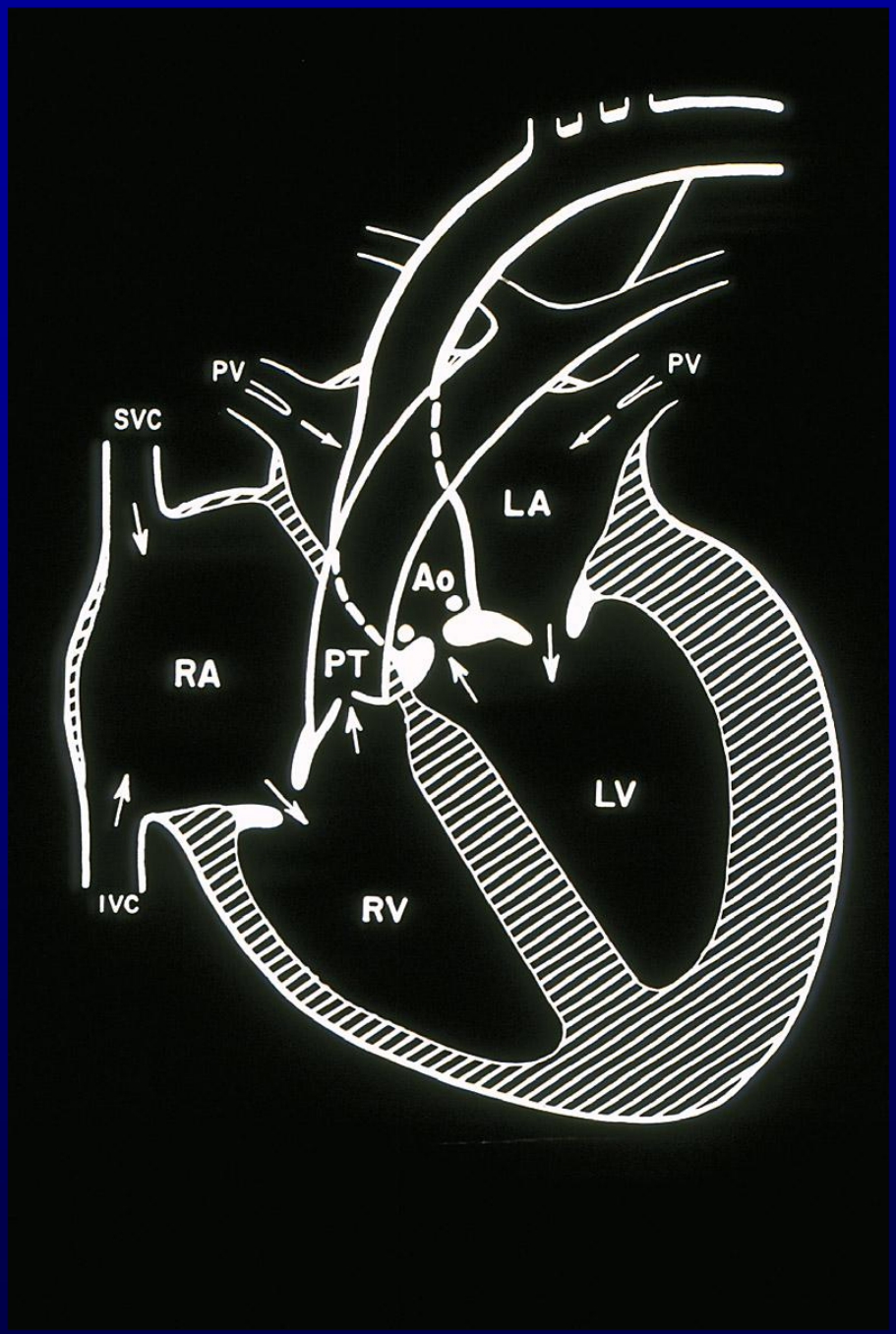
- Aortic Stenosis
  - 5-7%
  - Valvular
  - Sub/supra valvular

Remember, stenosis represents a range of defects - leaflets can be too large, valve can be too small, extra tissue can be present around the valve

Pulmonary Stenosis



Pulmonary stenosis results in early development of the same things you see happening with a VSD: RV has to pump extra hard and hypertrophies, RA pressure increases, and then with RA pressure > LA pressure, foramen ovale stays open - R -> L shunt



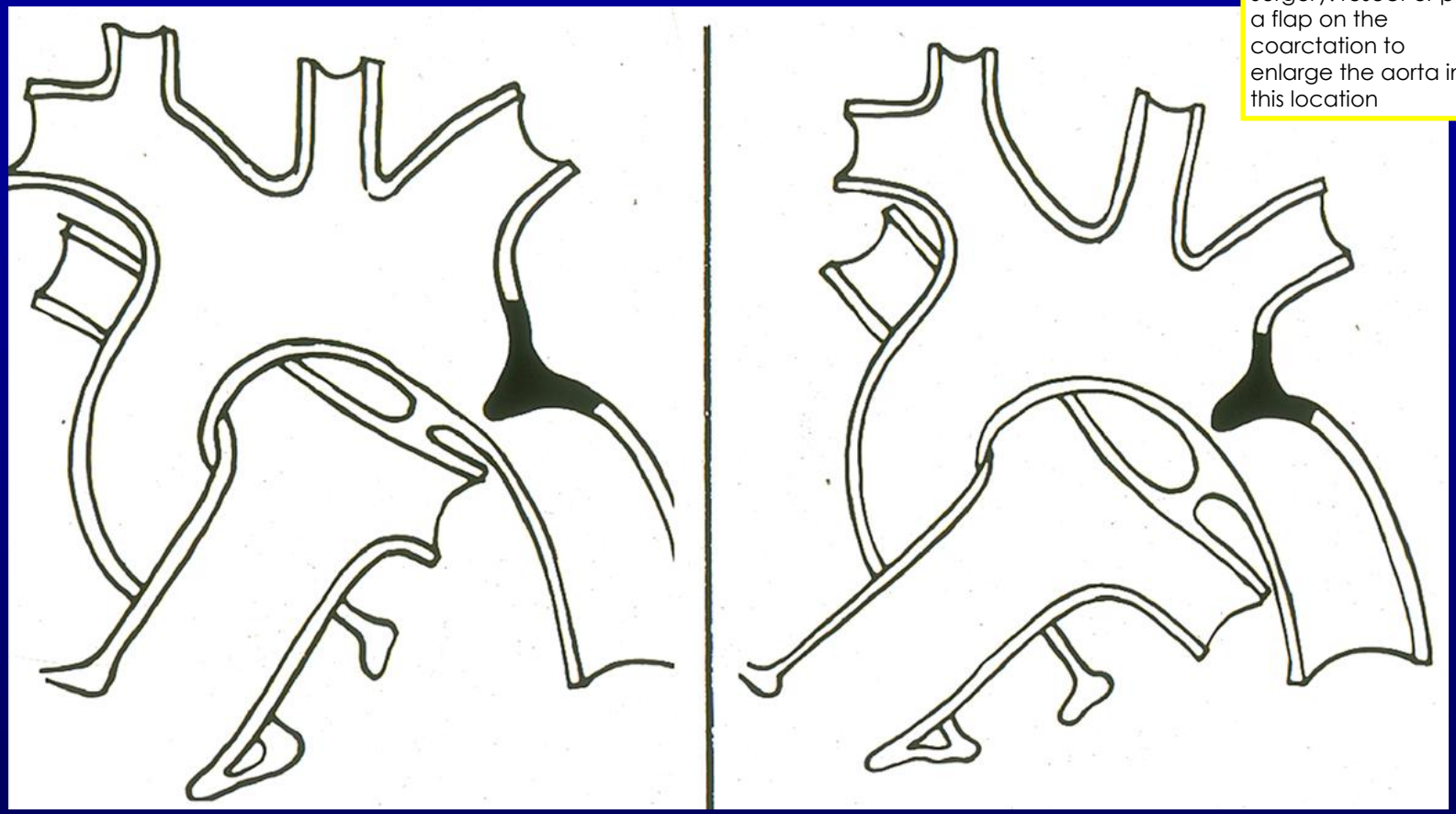
Aortic Stenosis

- Obstructs LV outflow
- LV Hypertrophy
- Aortic Arch Hypoplasia

Won't grow without blood flowing into it

You get backup of flow on the left side, going up into the lungs

Aortic Coarctation



Surgery: resect or put a flap on the coarctation to enlarge the aorta in this location

- **Closely Pre- or Postductal**
  - **Proximal : Hypertensive**
  - **Distal: Hypotensive**
  - **Interrupted aortic arch**
  - **Collaterals: Intercostal arteries**

Or, if right on it, juxtaductal

Can lead to erosions of the rib from enlarged arteries

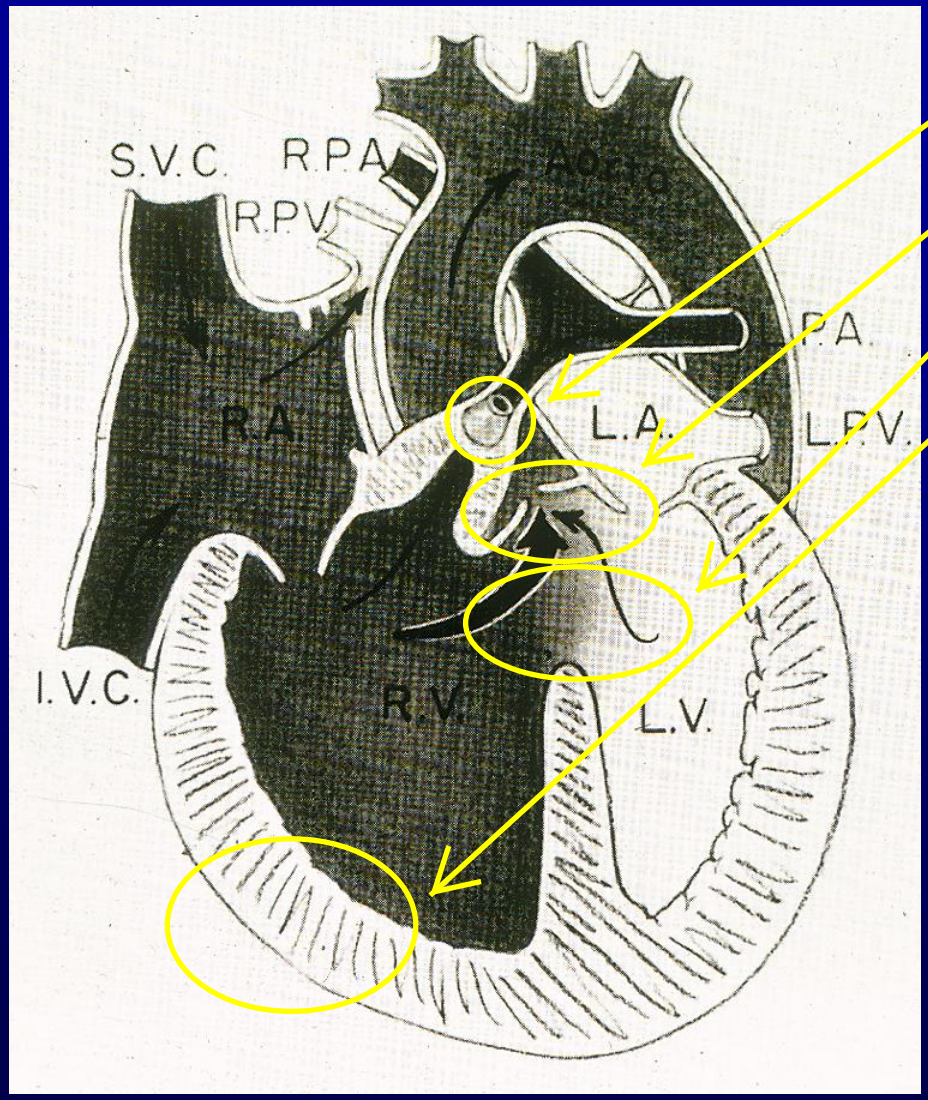
Combination Defects – Cyanotic Heart Disease

- **Tetralogy of Fallot**
  - 6-15%
- **Double Outlet Right Ventricle**
  - Rare
- **Transposition of the Great Vessels**
  - 4-10%
- **Tricuspid Atresia**
  - 1%
- **Hypoplastic Left Heart**
  - rare

Most common.
* Ventricular septal defect
* Narrowing of the pulmonary outflow tract
* Overriding aorta that is shifted over the right ventricle and ventricular septal defect, instead of coming out only from the left ventricle
* Right ventricular hypertrophy

Remember, you'll see even the rare ones here...

Tetralogy of Fallot

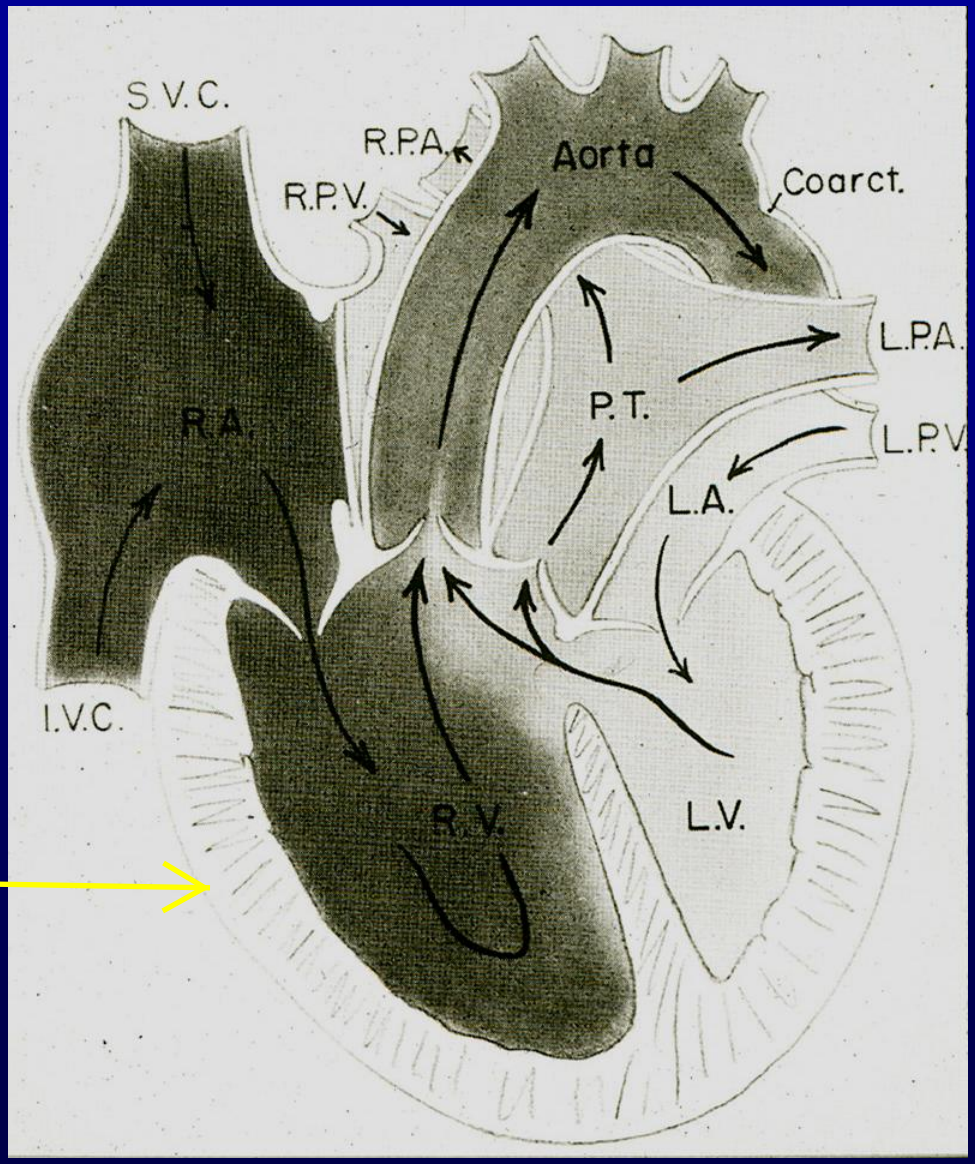


- Pulmonary Stenosis
- Overriding Aorta
- VSD
- Right Ventricular Hypertrophy *
  - Cyanosis from right sided pressure increase - shunting

Kids have "Tet spells" - become cyanotic when they need more blood flow (crying, etc.) Kids figure out quickly that squatting down increases systemic pressure and reverses the shunt

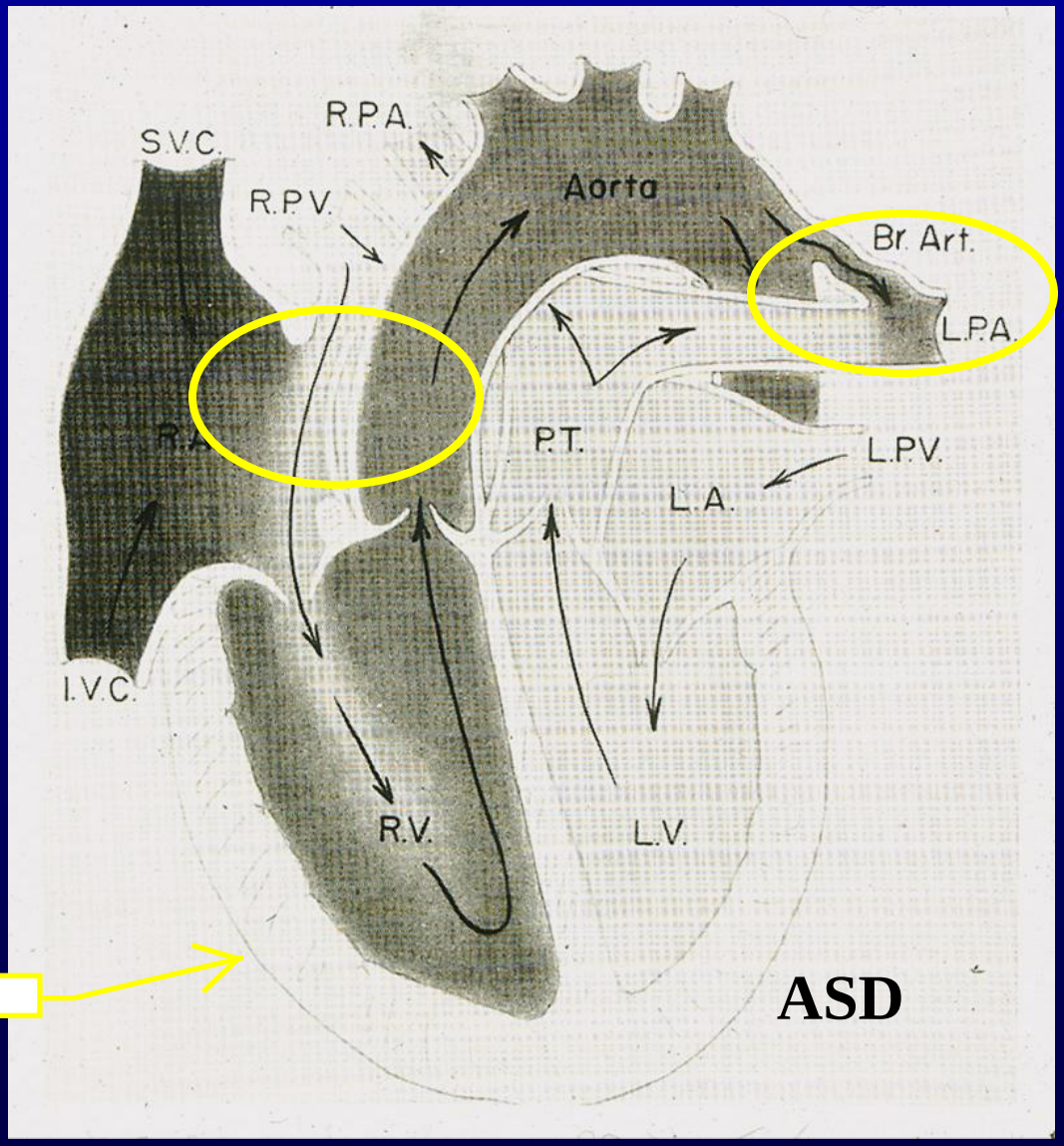
Double Outlet Right Ventricle

Both aorta and pulmonary artery come off RV - there is usually an associated VSD to allow oxygenated blood to get out.



RV hypertrophy

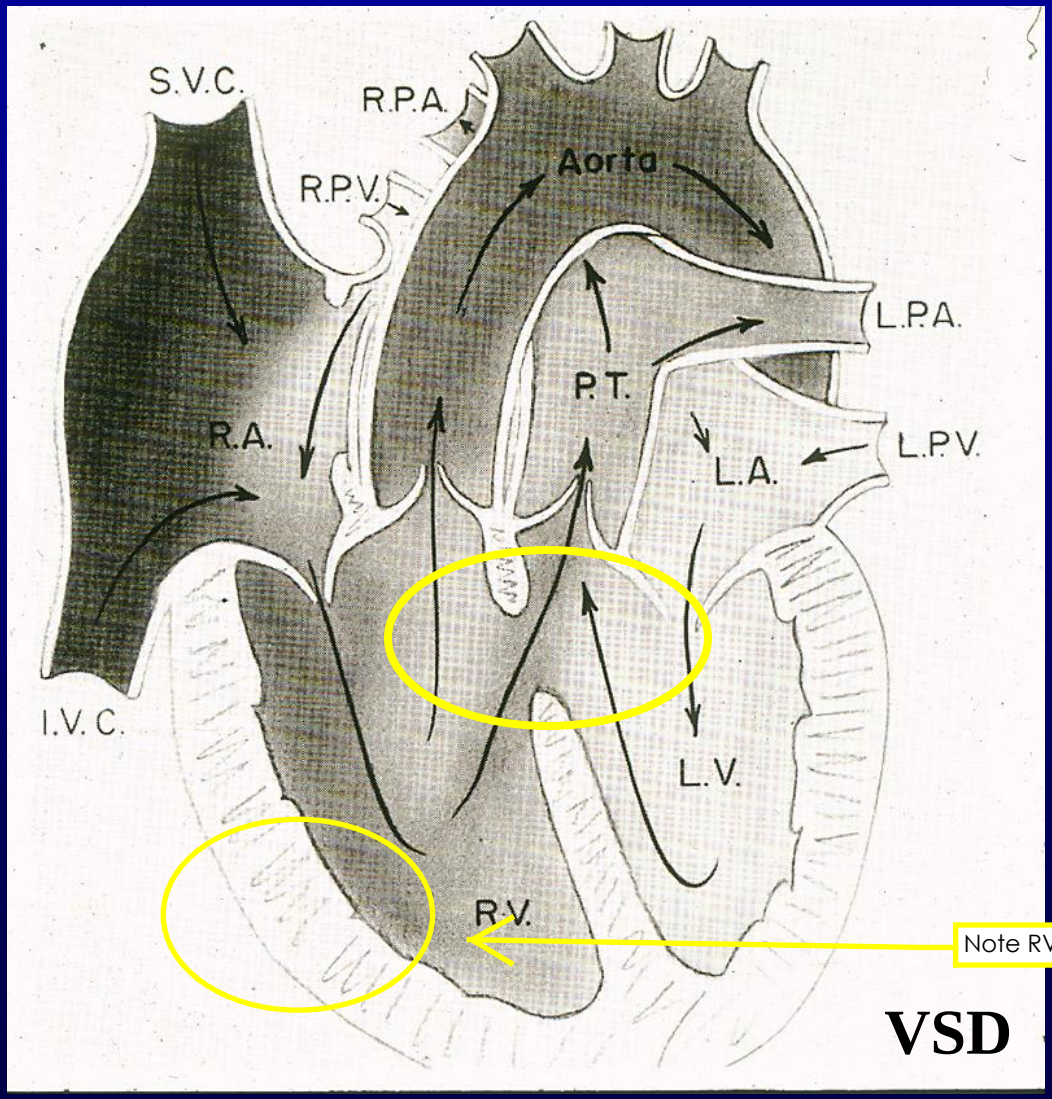
Transposition of the Great Vessels



Without any additional septal defect, you have two separate curcuits - Lv -> PA -> LA -> LV and RV-> Aorta -> Vena Cava -> RA -> RV. You MUST have a hole somewhere - ASD, patent foramen, or brachial arteries can provide collateral flow, or VSD.

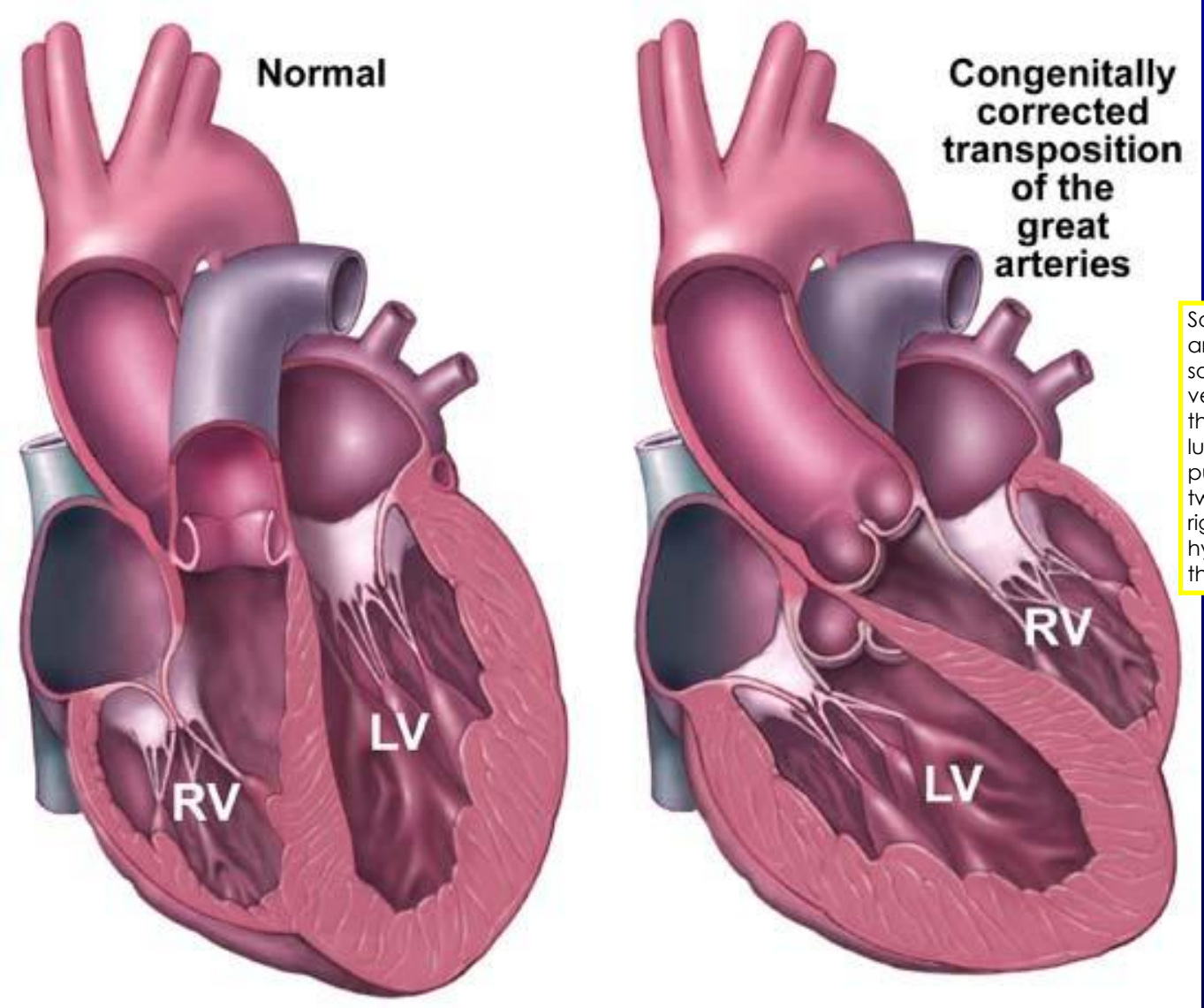
RV hypertrophy

Transposition of the Great Vessels



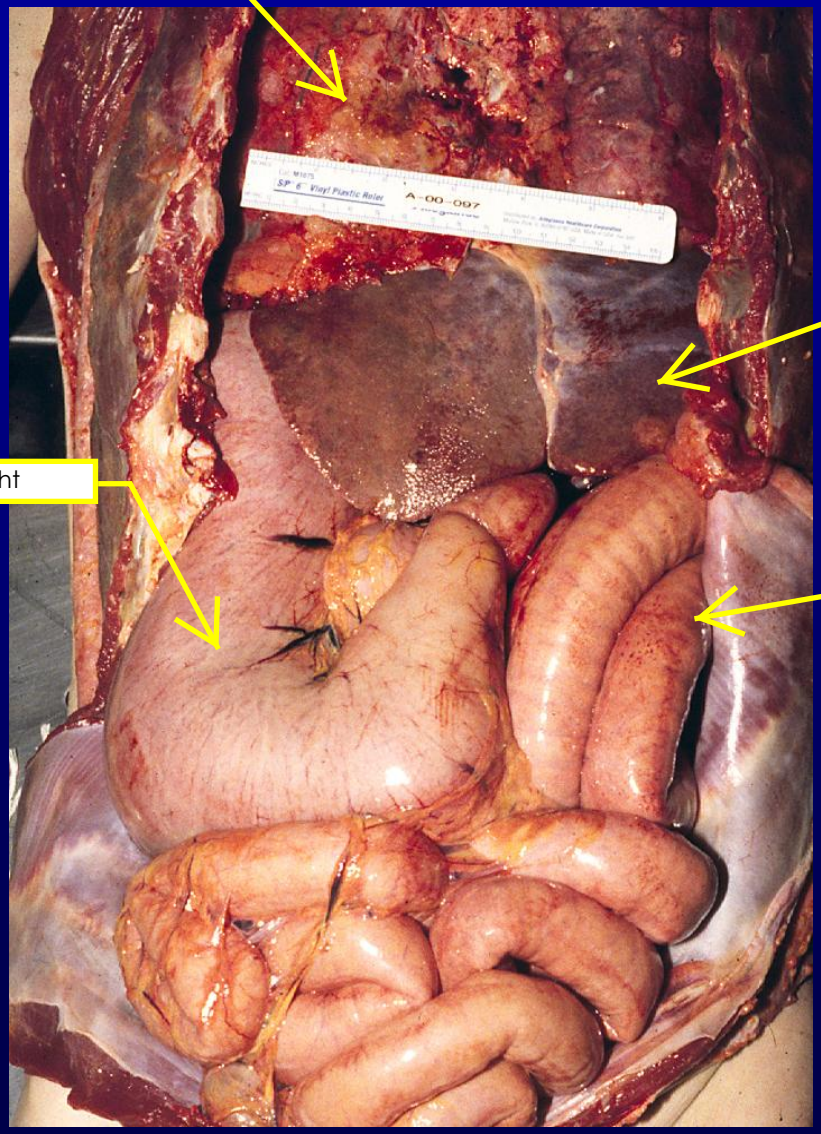
It used to be that these kids were monitored and the defect was repaired when they got worse, but the changes that occur in the RV lead to right-sided heart failure, so those kids did worse. Now these defects are fixed as close to birth as possible.

Corrected Transposition



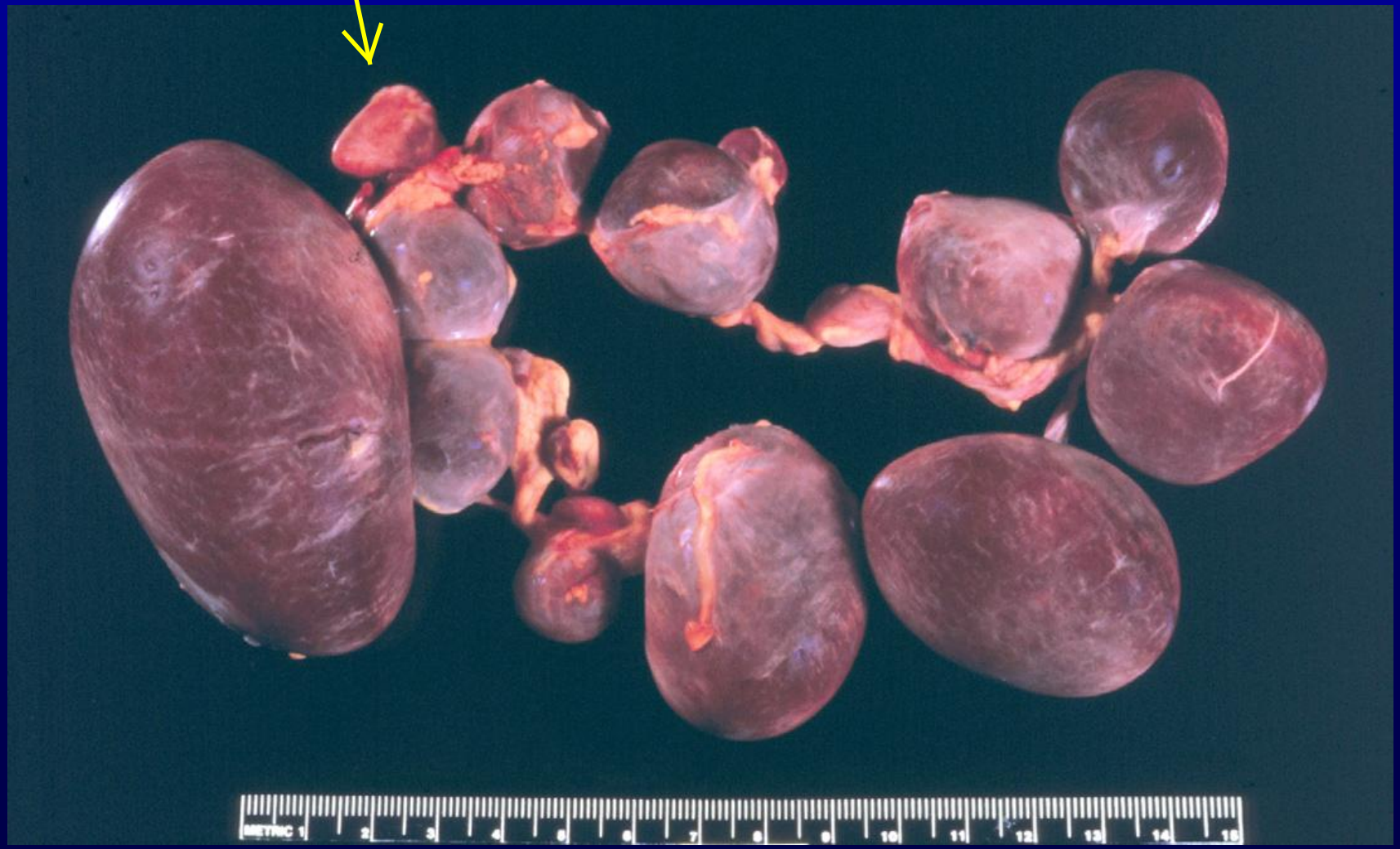
So your great arteries are transposed, and so are your ventricles. Note that the LV pumps to the lungs and the RV pumps to the body - two wrongs make a right... sort of... RV will hypertrophy, unlike this drawing

Situs Inversus

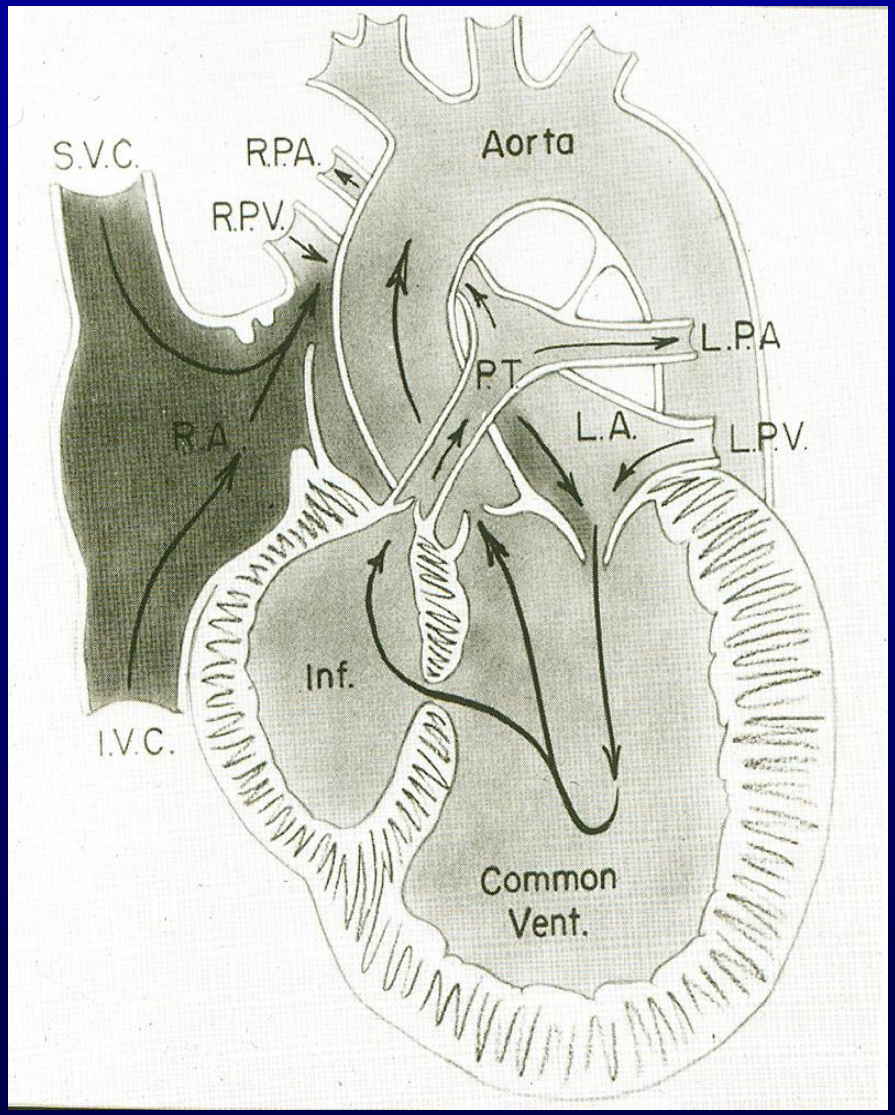


Polysplenia

This dude had 17 spleens. Does that count as splenomegaly, or is it just splenomultiply?



Tricuspid Atresia



No way to get blood to the right side of the heart, pulmonary artery is hypoplastic because of low flow

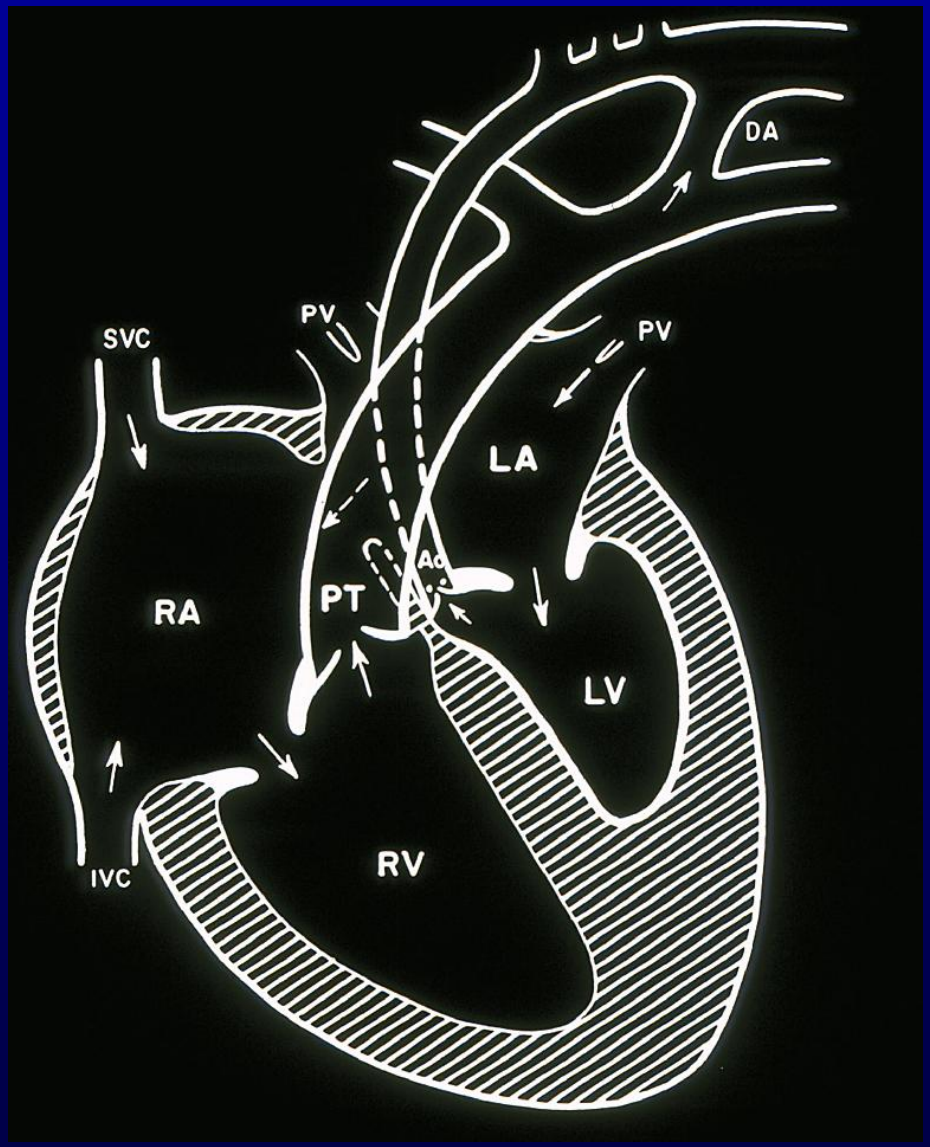
- No tricuspid valve
- RV and PA hypoplasia
- ASD for venous return
- VSD for pulmonary flow

Another case where you might want the ductus to exist

- Mixing and diminished pulmonary flow -- cyanosis

This one isn't easily corrected because you are missing parts

Hypoplastic Left Heart



- LV and aortic hypoplasia
- ASD - left to right flow
- Ductus Dependent for systemic flow

- May be secondary to Mitral Atresia

For these kids, whichever ventricle they have is made the systemic pump, and the lungs get blood through shunts

Implications

- Can it be Fixed?
  - Repair vs transplant
- When should it be fixed?
  - Palliate
  - Definitive
  - other

Problem is not enough hearts; kids waiting for transplant undergo secondary changes, never get a heart, and then are much sicker

The earlier the better

Palliation

Remember, palliation doesn't mean that it won't ever be fixed; it just means that it's a stopgap measure for right now.

- **Shunts: Provide blood flow**
  - **Generally to lungs**
- **Septostomies: Allow mixing of blood**
- **Ductus preservation**

Blalock-Taussig Shunt

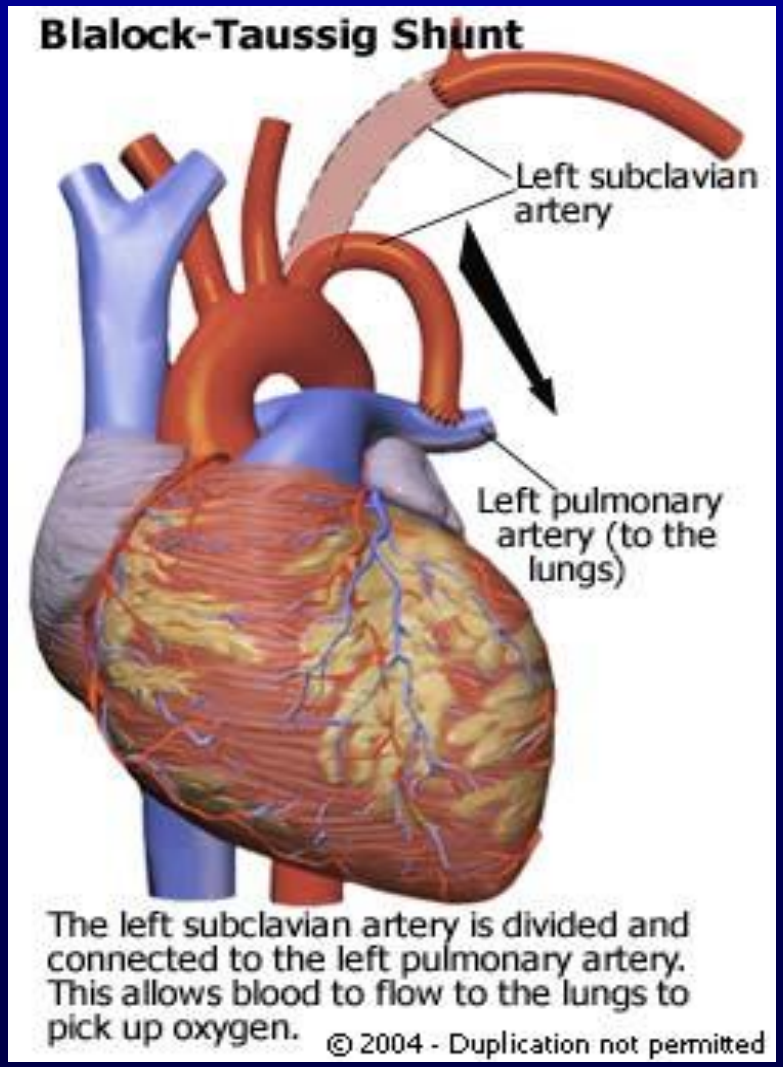
No one wanted to do heart surgery; resident who did anesthesia ended up becoming chair for anesthesiology @ Duke

African American lab assistant figured out how to create the animal model for the tetralogy and how to fix it, but the doctors took all the credit.

Vivien Thomas

Blalock-Taussig Shunt

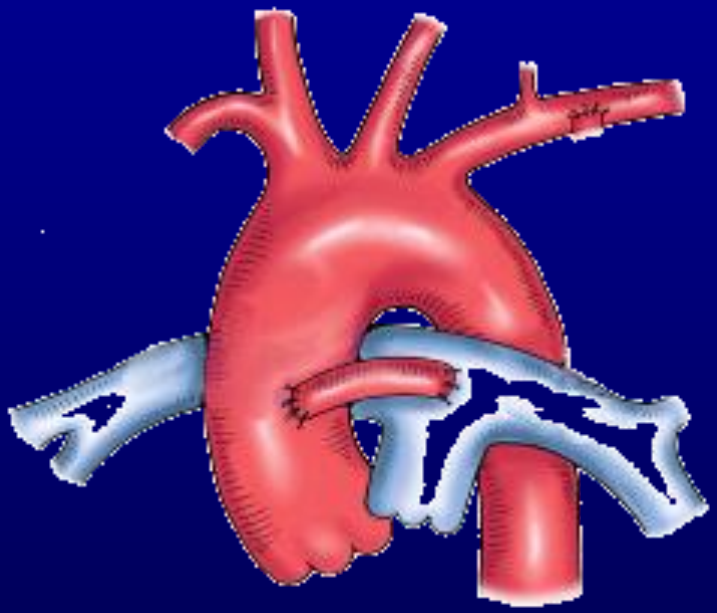
Designed for tetralogy of Fallot



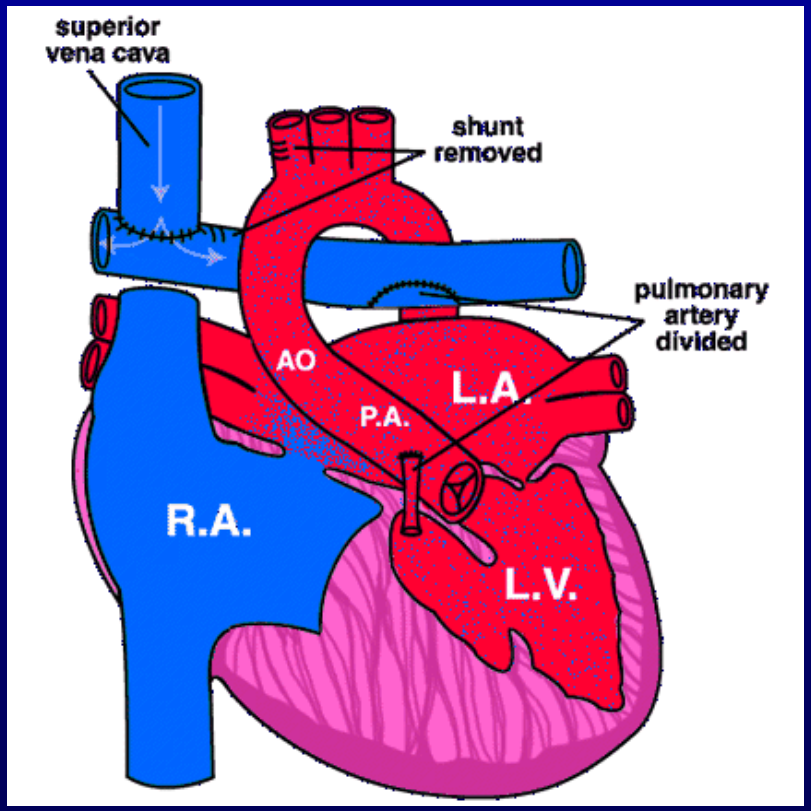
This is the original procedure; these days you just add a tube of Gore-tex between an aortic branch and the pulmonary artery

Shunts

Just examples of shunts that can be used - probably good pimping material for next year, but not likely to be on the test from the emphasis put on it (or lack thereof)

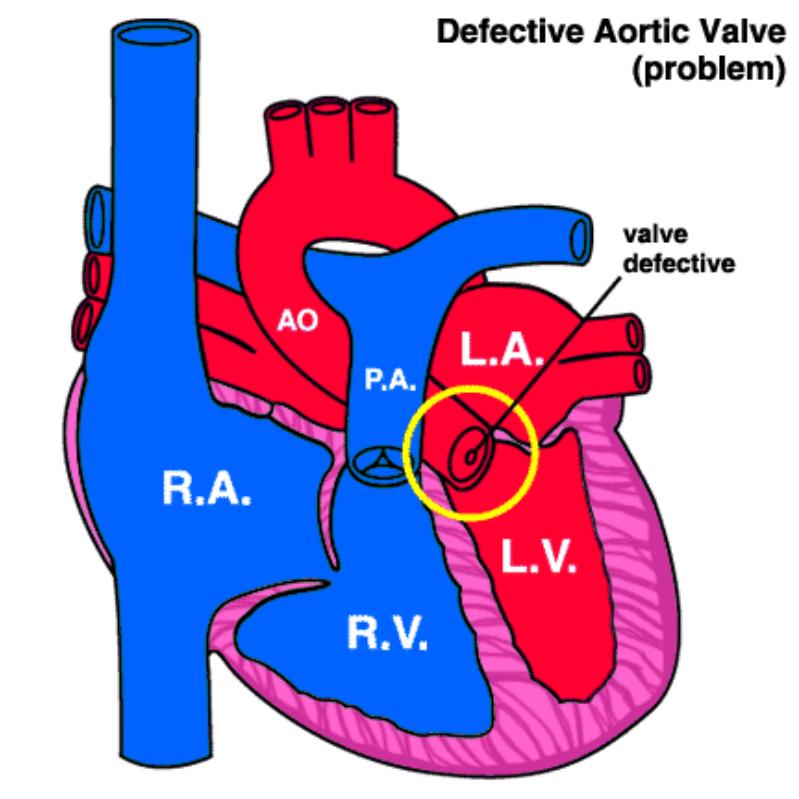


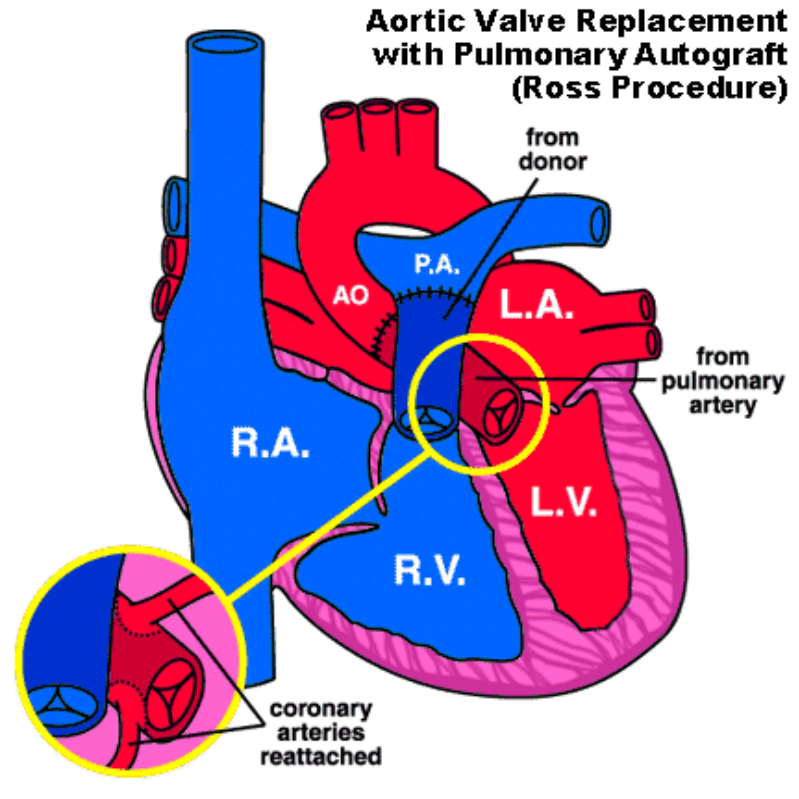
Central Shunt



Glenn Shunt

Repairs: Ross





1) Replace aortic valve with pulmonary valve

2) Donor pulmonary valve homograft

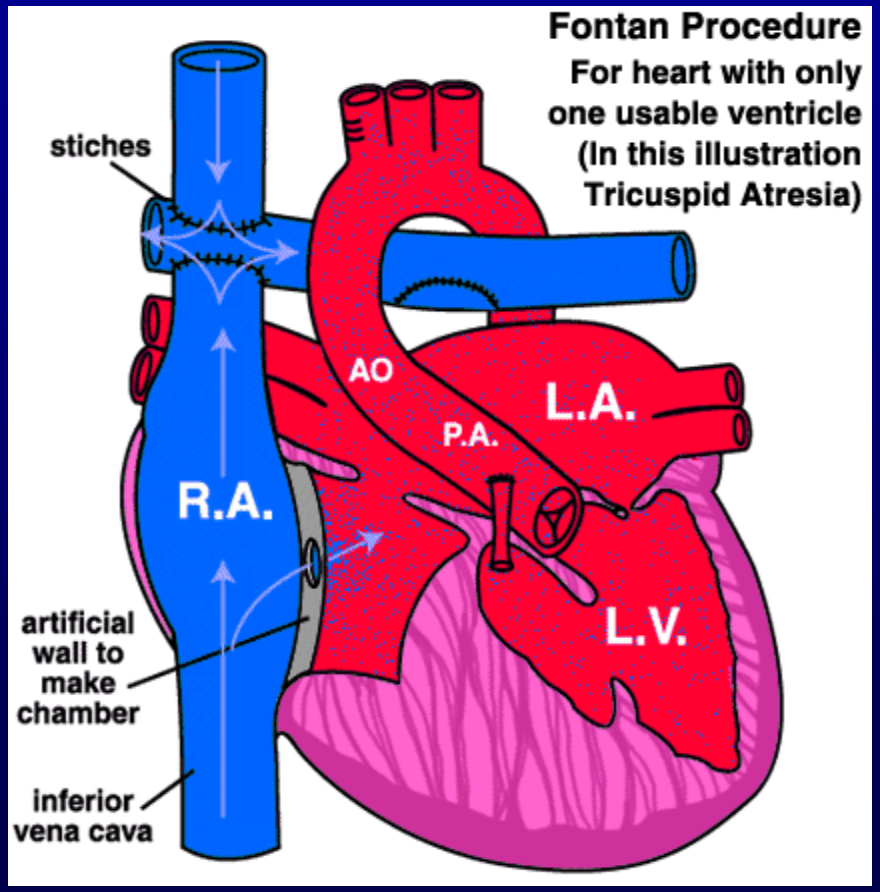
WHY?

Pt, does better with own valve in high-pressure system; prosthetic ok for low pressure

Repairs: Complex & Staged

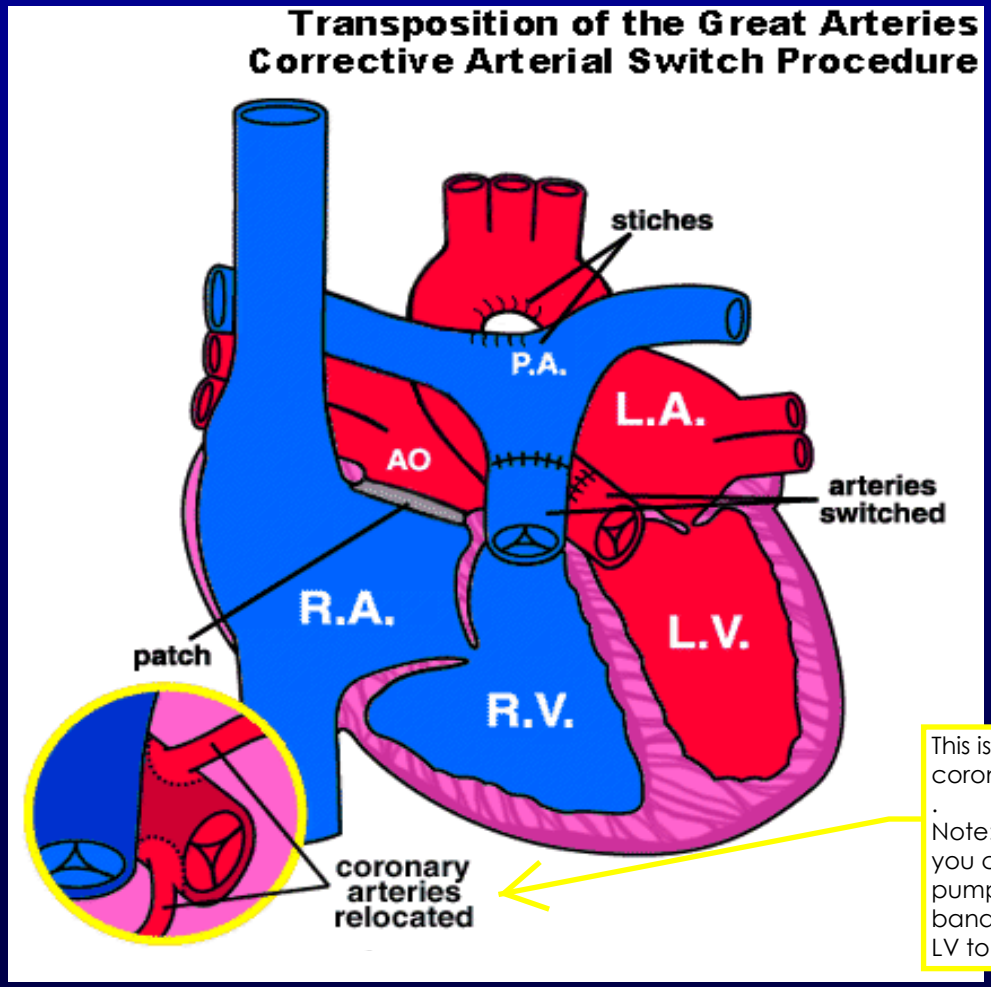
Skipped

Fontan: Tricuspid Atresia



Repairs: Complex & Staged

Transposition: Arterial Switch



This is the catch - you have to re-implant the coronaries after you do the switch.

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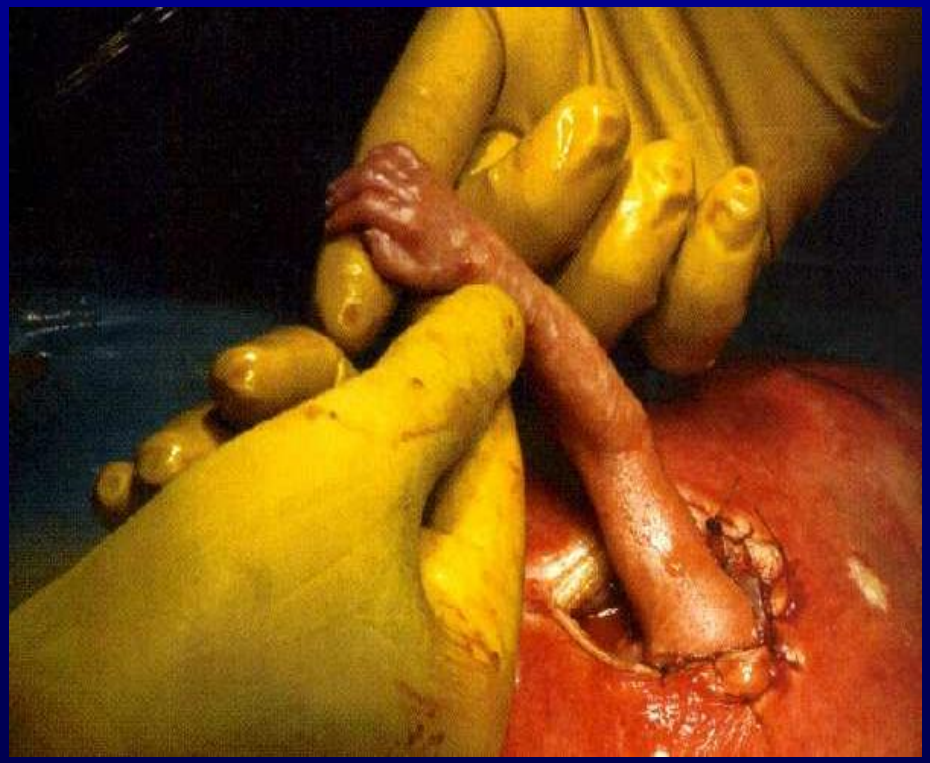
Note: If you don't do this operation early, you can't get the LV to be the systemic pump, but can inc. resistance in PA with a band around it for about a week ("Take the LV to the gym")

Fetal Surgery

Future of surgery; not as well progressed on the heart as on other organs - heart-lung bypass is really tricky on these kids; premature delivery can be stimulated.



The Center *for* Fetal Diagnosis *and* Treatment

1-800-IN-UTERO

The Fetal Treatment Center
UNIVERSITY OF CALIFORNIA SAN FRANCISCO

Prevent volume/pressure hypertrophy

No Scarring

Barriers remain

Advantages; no scarring = plastic surgeon's dream