# **Overview of Congenital Heart Disease**



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# **Outline / Objectives**

- Introduction / epidemiology
- Brief history of CHD
- Anatomy / physiology review
- CHD classification / types
  - Left-to-right
  - Cyanotic
  - Obstructive
- Key points to remember







## Epidemiology

- CHD is most common birth defect (~ 1% births)
- More common with certain genetic syndromes
  - Turner = ~20-45%; T21 = ~ 50%; T18 = ~ 90%; T13 = ~ 80%
  - Others: DiGeorge (22q11), CHARGE, Alagille, Holt-Oram, Noonan
- Most common = VSD (then ASD, TOF, PDA, coarctation, PS)
- Less common: TGA, AVSD, HLHS, truncus arteriosus, TAPVR, tricuspid atresia, DORV, DILV, heterotaxy syndrome...







- Leonardo daVinci: a sketch of partial anomalous pulmonary venous return (PAPVR) of the right pulmonary veins to the right atrium
- Before 20th century, CHD was autopsy diagnosis with no treatment
- Digoxin (*Digitalis lanata* "Foxglove") to treat "dropsy"





- August 8, 1938: first successful operation for an extracardiac lesion
- Robert Gross, MD (Boston) ligated a patent ductus arteriosus of a 7 yo female when he was a chief surgical resident
- His attending Dr. Ladd was on summer vacation!





 Six years later (October 10, 1944) Crafoord and Nylin (Sweden) successfully repaired another CHD: Coarctation of the Aorta



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#### Alfred Blalock





#### Helen Taussig



 November 29<sup>th</sup>, 1944 the first subclavian to pulmonary artery anastomosis was performed (BT *aka BTT* shunt) as a palliation for Tetralogy of Fallot





- Problem remained that surgeons could not get access to the internal structures of the heart
- In 1907, Haecker showed that occluding the superior vena cava and inferior vena cava in dogs longer than 3 minutes caused death or severe CNS damage
- Later, basic science labs found that cooling animals lowered the metabolic demands and could possibly lengthen the occlusion of the vessels longer during surgery







- On September 2, 1952, Dr. John Lewis (Minnesota) successfully closed an ASD with hypothermia
- The technique was soon used by others, but was far from ideal
- In 1953, Dr. John Gibbon (Mass General) attempted to close an atrial septal defect in a young lady using an oxygenator and a roller pump.





- The results were terrible 1 patient out of 18 from that era!!
- Many gave up...

Table 1. Early Attempts at Open-Heart Surgery With a Heart-Lung Machine

Name	Year	No.	Outcome
Dennis	1952	2/2	Died
Gibbon	1953	5/6	Died
Helmsworth	1953	1/1	Died
Dodrill	1953	2/2	Died
Clowes	1954	2/2	Died
Mustard	1954	5/5	Died
	Total	17/18 (94.5%)	Died



- Forunately, C. Walton Lillehei did not!
- Dr. Lillehei (Minnesota) developed a crosscirculation technique that ultimately led to the cardiopulmonary bypass concept that is used today
- There are now many repairs and palliations to wide spectrum of CHD
- Many surgical options for neonates with complex CHD
- Today, >90% with CHD survive to adulthood
  - (more adults with CHD than children!)







# ANATOMY / PHYSIOLOGY









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## Embryology (...briefly)

- Most cardiac development occurs early in pregnancy ~3-8 weeks gestation
- Fetal heart tube present ~ 3 weeks gestation
- Series of looping, septation to form circulation the Atrium in series
  Weeks Days Event

Weeks	Days	Event	
1-2	0-20	No heart or vessel	
3	20	Cardiogenic plate	Sinus Venosus
3	21	Endocardial tubes	
4	22	Fusion of endocardial tubes	(12) State
4	23	Single median cardiac tube, 1st contraction	Day 23
4*	25	Cardiogenic loop	191
4	26	Single atrium	
5	29	Bi-lobed atrium	
5*	30	Beginning of circulation	
5	31	Septum primum	
5	35	A-V orifice, 3 chambered heart	
6	36	Septum secundum	
6	39	Complete inferior septum	
6	40	Septation of bulbus and ventricle	
6	42	Divided truncus arteriosus	
7*	49	4-chambered heart, absorption of pulm, veins	

Fruncus Arteriosus

Bulbus Cordis (cor

Ventricle

Left Atrium





*Deoxygenated* blood: right atrium -> right ventricle -> pulmonary arteries -> GAS EXCHANGE -> pulmonary veins -> left atrium -> left ventricle -> aorta -> tissues

(rinse and repeat)





#### FETAL CIRCULATION

Oxygenated blood: umbilical vein -> ductus venosus -> right atrium -> CHOOSE YOUR OWN ADVENTURE

> ~1/3<sup>rd</sup> PFO (brain) ~2/3<sup>rd</sup> MPA (<10% to lungs!)

#### **CLASSIFICATION OF CHD**



#### First, a few critical concepts...





#### BLOOD FOLLOWS THE PATH OF LEAST RESISTANCE

- some wise person













# LEFT TO RIGHT (ACYANOTIC)







#### Left-to-right CHD

• Ventricular Septal Defect (VSD)

• Atrial Septal Defect (ASD)

• Patent Ductus Arteriosus (PDA)

• Atrioventricular Canal (AV Canal)





#### Ventricular septal defect (VSD)

- "Hole between bottom chambers"
- Many types and locations can occur
- Types and location determine it's fate
- If CHF develops (typically at 6-8 weeks), VSD repaired ~3-6 months

#### **LEFT** HEART DILATION = SIGNIFICANT



## Atrial septal defect (ASD)

- "Hole between top chambers of the heart"
- Many types and locations (secundum, primum, sinus venosus, coronary sinus)
- CHF usually doesn't develop
- Can be asymptomatic until 4<sup>th</sup>-6<sup>th</sup> decade
- Typically repair 3-5 years of age; surgical or cath based, but only if...



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#### **RIGHT** HEART DILATION = SIGNIFICANT







#### Patent ductus arteriosus (PDA)

- Persistence of fetal structure
- Can be asymptomatic if small or symptomatic if large
- Major concern for premature babies
- Pharmacologic closure, surgical closure, cath closure

**LEFT** HEART DILATION = SIGNIFICANT

dren's Mercv



#### **Atrioventricular septal defect (AVSD)**

- "AV canal"
- Primum ASD and Inlet VSD
- Highly associated with Trisomy 21
- LOTS of variety (shunting, valves, "dominance")
- Like VSD, CHF at 6-8 weeks

ildren's Mercy

• Surgical correction at 4-8 months of age

#### **LEFT +/- RIGHT HEART DILATION**





# RIGHT TO LEFT (CYANOTIC)







## **Cyanotic CHD**

- Lower oxygen saturation, typically immediately after birth
- Dependent on the PDA (+/- ASD)
- The 5 Ts

Truncus Arteriosus Transposition of the Great Vessels Tricuspid/Pulmonary Atresia Transposition of the Great Vessel Tetralogy of Fallot Total anomalous pulmonary venous return





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### **Truncus Arteriosus**



- Failure of the aortopulmonary septum to divide aorta and pulmonary arteries
- Typically a mixed picture of oxygen desaturation + CHF
- Significant pulmonary overcirculation
- Variable anatomy of "Truncus valve"
- Typically, repaired 1-2 weeks of age





# Transposition of the Great Arteries (TGA)

A0

RA

PA

RV

- Circulations in parallel
- Cyanosis at birth, "comfortably blue"
- Need ASD (PDA nice too) for blood mixing
- Balloon atrial septostomy?
- Arterial switch operation in the 1<sup>st</sup> week of life



# Tricuspid Atresia

- Agenesis of tricuspid valve → severe right ventricular hypoplasia
- If well balanced, can avoid surgery in the neonatal period.
- Others may require BTT shunt for pulmonary blood flow
  - NRGA vs TGA









#### Fontan @ 3-5 years



## **Tetralogy of Fallot**

- Most common cyanotic\* CHD
- VSD + overriding Ao + PS + RVH
- "Monology" of Fallot?
- Surgical repair varies by institution but typically occurs 3-9 months of age
- Repair: VSD closure, RVOT+/- PA reconstruction
  - "Valve sparing" vs transannular patch
- Many require RV-PA conduit later in life







- Pulmonary veins drain (ultimately) into *right* atrium
- 1. Supracardiac (most common)
- 2. Cardiac (best outcome)
- 3. Infracardiac (worst outcome, most likely to obstruct)
- 4. Mixed...
- 5. Surgery in the first 1-2 weeks OR emergent at birth if veins obstructed





# **OBSTRUCTIVE** (LEFT-SIDED)







#### Left-sided Obstructive CHD

- Present at 2-3 days of life\* with feeding intolerance, poor cardiac output, and cardiogenic shock (gray/shock)
- Hypoplastic left heart syndrome (HLHS)
- Aortic valve stenosis
- Coarctation of the aorta
- Interrupted aortic arch
- Numerous others...
  - Tricuspid atresia w/ TGA, posterior malalignment VSD, DORV (some), sub AS





#### **Aortic Valve Stenosis**

- Severely dysplastic aortic valve
- Poor cardiac output
- Present 2-3 days of life
- Stable until PDA closes!
- PGE until balloon angioplasty
- Normal size of rest of the left heart? *(important question)*





#### **Coarctation of the Aorta**

- Narrowing of the distal\* aortic arch
- PDA closure -> "declares itself"
- Need PGE until surgical correction
- Surgery within the 1<sup>st</sup> week of life
  - thoracotomy vs sternotomy\*
- Femoral pulse exam!
- Most extreme coarctation = IAA





#### Hypoplastic Left Heart Syndrome

- Among most serious CHD, but outcomes have improved
- Staged palliation: Norwood (neonatal), Glenn (4-8 months), Fontan (3-5 years)
- Now able to survive beyond the first decade of life
- 15-year survival of 48% (significant mortality during the first year of life)





#### Some key takeaways

- About 1% births have CHD (VSD most common)
- Blood follows the path of least resistance!
- If equally large/unrestrictive: PDA -> ASD -> VSD
- Important time points: PDA closure (1-3 days), minimum PVR (4-6wks)
- ASD: right heart enlargement
- VSD & PDA: left heart enlargement
- Femoral pulse exam crucial for obstructive CHD
- Major improvement in survivability and morbidity in CHD
- New approaches constantly being developed



# **THANK YOU!**





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