Overview of Congenital Heart Disease

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1

Outline / Objectives

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

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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…

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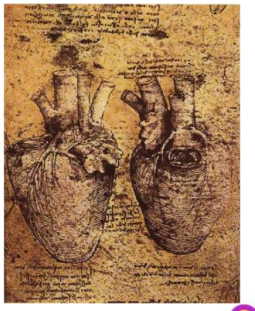
History of CHD

- 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"



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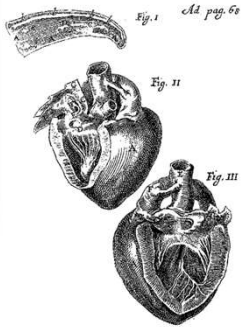
History of CHD

- 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!



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History of CHD

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

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History of CHD

Alfred Blalock

Helen Taussig

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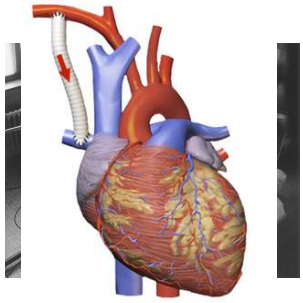
History of CHD

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



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History of CHD

- 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

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History of CHD

- 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.

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History of CHD

- 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

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History of CHD

- Forunately, C. Walton Lillehei did not!
- Dr. Lillehei (Minnesota) developed a cross-circulation 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!)**

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ANATOMY / PHYSIOLOGY

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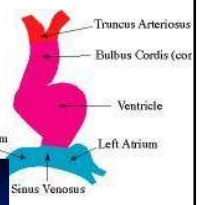
Embryology (…briefly)

- 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 in series

Weeks	Days	Event
1-2	0-20	No heart or vessel
3	20	Cardiogenic plate
3	21	Endocardial tubes
4	22	Fusion of endocardial tubes
4	23	Single median cardiac tube, 1st contraction
4*	25	Cardiogenic loop
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



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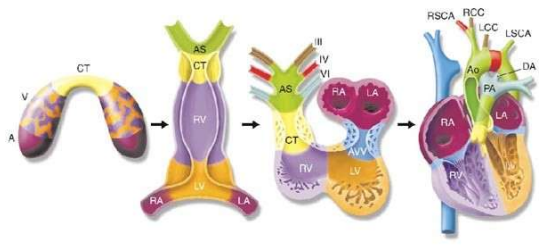

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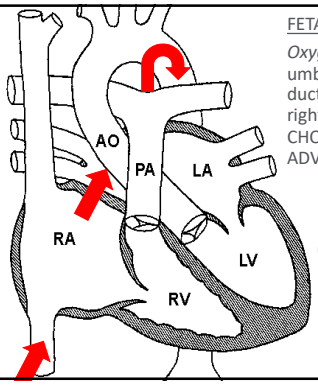
15

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

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

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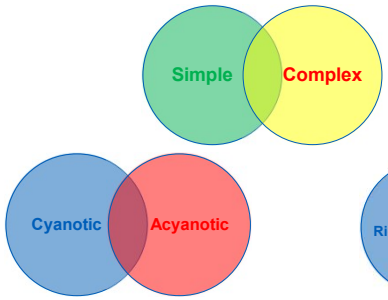
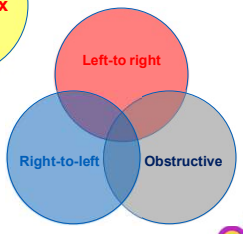
CLASSIFICATION OF CHD





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First, a few critical concepts…

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BLOOD FOLLOWS THE PATH OF LEAST RESISTANCE
- some wise person

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lungs body

Qp Qs

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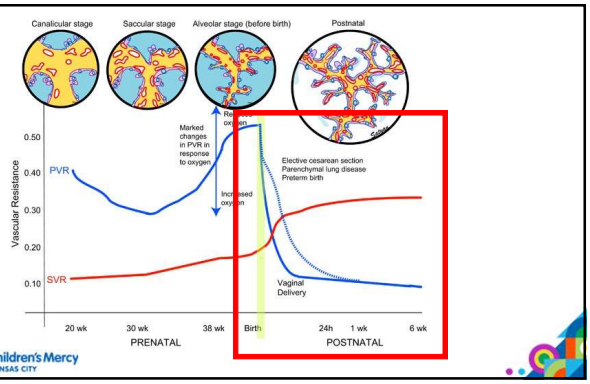
20



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LEFT TO RIGHT
(ACYANOTIC)

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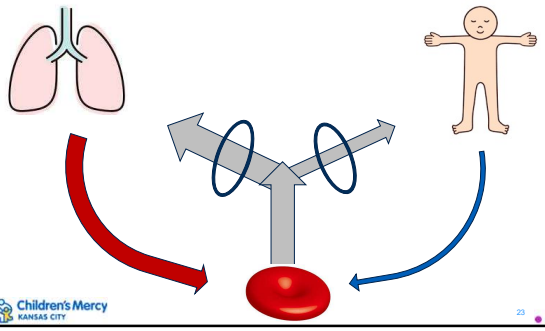

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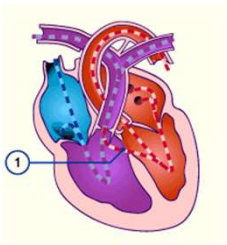
Left-to-right CHD

- Ventricular Septal Defect (VSD)
- Atrial Septal Defect (ASD)
- Patent Ductus Arteriosus (PDA)
- Atrioventricular Canal (AV Canal)



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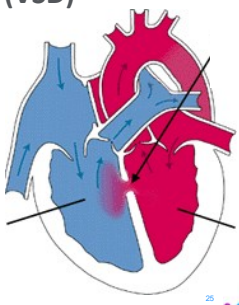
Ventricular septal defect (VSD)

- "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



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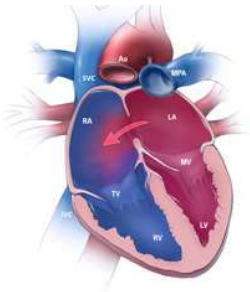
Atrial septal defect (ASD)

- "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 4th-6th decade
- Typically repair 3-5 years of age; surgical or cath based, but only if…

RIGHT HEART DILATION = SIGNIFICANT



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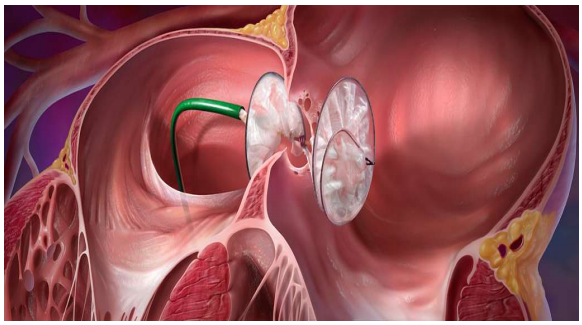

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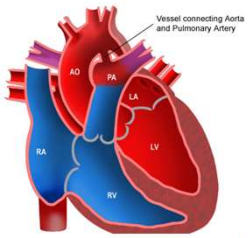
Patent ductus arteriosus (PDA)

- 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

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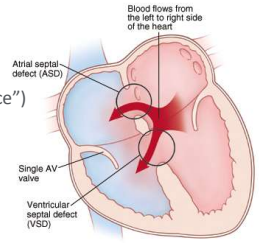
Atrioventricular septal defect (AVSD)

- "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
- Surgical correction at 4-8 months of age



LEFT +/- RIGHT HEART DILATION

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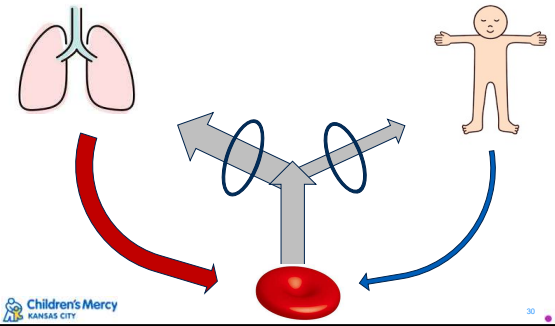

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RIGHT TO LEFT
(CYANOTIC)

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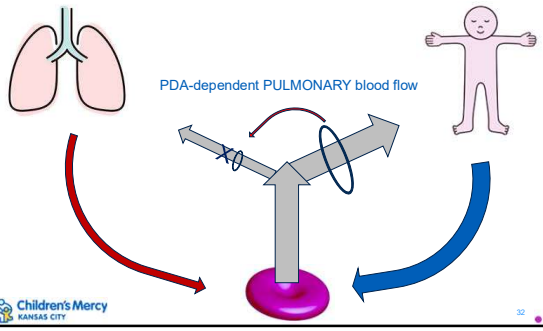

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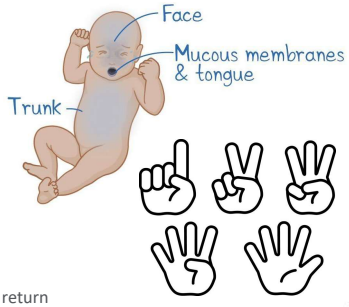
Cyanotic CHD

- 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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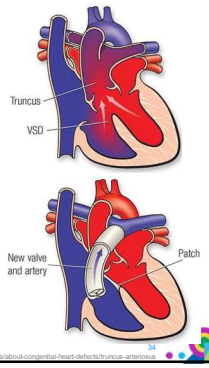
Truncus Arteriosus

- 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



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https://www.heart.org/en/health-topics/congenital-heart-defects/about-congenital-heart-defects/truncus-arteriosus

34

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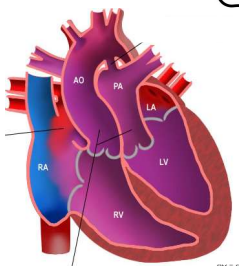
Transposition of the Great Arteries (TGA)

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



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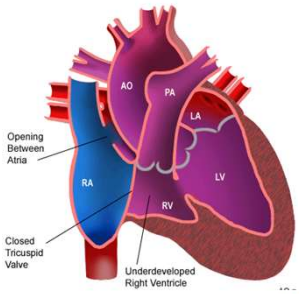
Tricuspid Atresia

- 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



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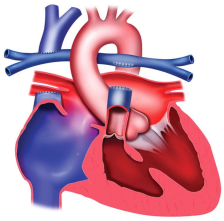
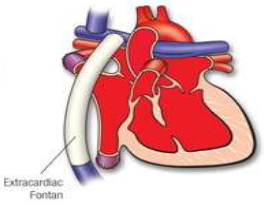
Tricuspid Atresia

Glenn @ 4-8 months



Fontan @ 3-5 years



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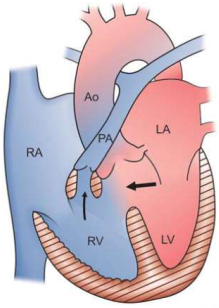
Tetralogy of Fallot

- 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



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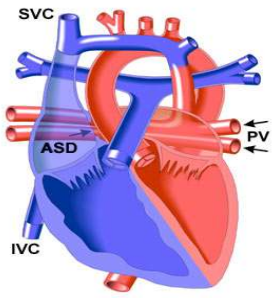
TAPVR

- 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



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OBSTRUCTIVE
(LEFT-SIDED)

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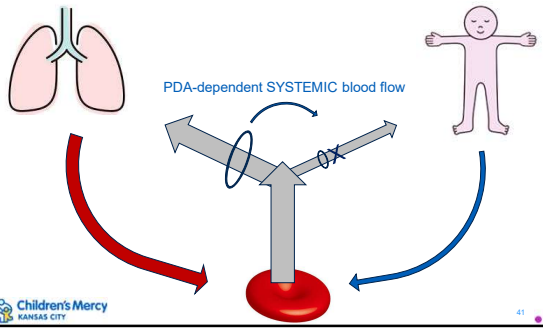

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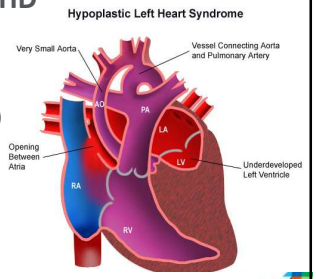
Left-sided Obstructive CHD

- 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



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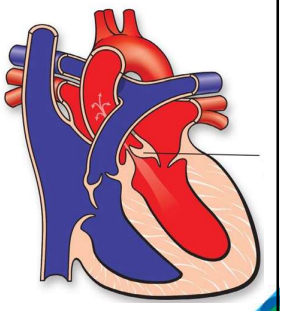
Aortic Valve Stenosis

- 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)*



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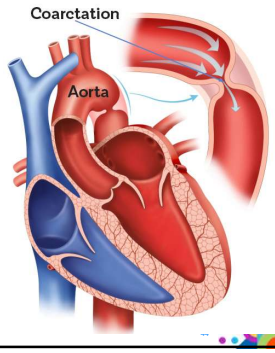
Coarctation of the Aorta

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



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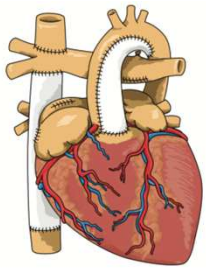
Hypoplastic Left Heart Syndrome

- 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)



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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

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THANK YOU!

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