Overview

- Embryology of CHD
- Fetal Cardiology
- Select Congenital Heart Defects
- Cardiac Interventions
- Acquired Heart Disease
- Sports Clearance
THE EMBRYOLOGY OF CONGENITAL HEART DISEASE
Aorticopulmonary Spiraling
FETAL ECHOCARDIOGRAPHY

- **Goals:**
  - Achieve early detection of majority of infants with cardiac disease
  - Provide support to families through knowledgeable counseling
  - Offer fetal intervention strategies for intrauterine heart disorders
The concern is raised for a major cardiac defect in the fetus of a 24 yo pregnant woman. The best gestational age at which to refer this individual for the performance of a fetal echocardiogram is:

A. 13 weeks
B. 18 weeks
C. 26 weeks
D. 32 weeks
FETAL ECHOCARDIOGRAPHY

- Timing of Obstetrical evaluation
  - 12-18 weeks

- Timing for Cardiac referral
  - 17 wks, and onward
REASONS FOR FETAL ECHO

- Maternal Risks
- Familial Risks
- Fetal Risks
Maternal Risk Factors

- maternal metabolic disorders
  - diabetes
- maternal exposure to cardiac teratogens
- maternal autoantibodies:
  - SLE: heart block
Maternal Risk Factors

Maternal exposures:

» rubella
» parvovirus
» coxsackie

» ETOH
» anticonvulsants
» retinoic acid
» ? lithium (Ebstein’s)
» indomethacin (NSAIDS)
Familial Risk Factors

Examples:

- Isolated parental CHD: 3-15% risk
- Autosomal dominant condition
  - (parent with Marfan’s): 50% risk
Fetal Risk Factors

Fetal abnormalities associated w/ CHD

- omphalocoele (30% risk)
- diaphragmatic hernia (10% risk)
- duodenal atresia (Downs)
- Increased nuchal translucency (Downs/CHD)
Fetal Intervention

- Fetal Surgery:
  - Congenital diaphragmatic hernia
  - Urinary tract obstruction
  - Sacrococcygeal teratoma
  - Myelomeningocele
Fetal Cardiac Intervention
Select Congenital Heart Defects
A 4-week-old infant has a large ventricular septal defect. He is developing tachypnea but is not yet receiving medications. You accurately inform the family that:

A. diuretic therapy may help to relieve tachypnea and diaphoresis
B. feeding difficulties and problems with poor weight gain are rare
C. most affected children do not develop excess pulmonary blood flow
D. symptoms of congestive heart failure are unlikely
Ventricular Septal Defect (VSD)

- Most common congenital heart defect
  - Symptoms:
    - L → R shunt causing CHF
Ventricular Septal Defect
Management

- **Medical**
  - Spontaneous closure in 30% - 40%
    - Small defects
    - First year of life
  - Anti-congestive medicines

- **Surgical**
  - Patch closure of defect
    - Typically during 1st yr
Congestive Heart Failure

- Poor feeding
- Diaphoresis with feeding
- Tachypnea; respiratory distress
- Tachycardia
- Hepatomegaly
Valve Stenosis
Incidence

- Two of most common congenital cardiac defects
  - Pulmonic stenosis ~ 10%
  - Aortic stenosis ~ 11%
Aortic Stenosis

- Typical morphology: bicuspid aortic valve
- Variable severity / symptoms
- Most common diagnosis in the adult requiring aortic valve replacement
A 4-year-old girl has a bicuspid aortic valve with moderate aortic stenosis but no insufficiency. No intervention is required at present. The parents ask you about prognosis; you inform them that their daughter:

A. is unlikely to develop aortic insufficiency
B. eventually will require surgery as a result of increasing stenosis
C. requires antibiotic prophylaxis to prevent subacute bacterial endocarditis
D. does not have an inheritable condition

**Question 3**
Valve Stenosis
Evaluation / Management

- **Echocardiogram**
  - Diagnostic (non-invasive)
  - Predict severity of obstruction

- **Catheterization**
  - Diagnostic
  - Therapeutic
    - Balloon dilation

- **Surgery**
  - Valve replacement
    - Short longevity
    - Anticoagulation
    - Problems with prosthetic valves
    - Restrict physical activity

- **Anticoagulation**
- **Restrict physical activity**
Cardiac Interventions

- Tetralogy of Fallot
- Hypoplastic Left Heart Syndrome
- Atrial Septal Defect
Tetralogy of Fallot

- Most common cyanotic congenital cardiac abnormality
  - RVOT obstruction
  - Ventricular septal defect
  - Overriding aorta
  - Right ventricular hypertrophy
TETRALOGY OF FALLOT

- Natural History
  - Mortality:
    - 50% by 3yr
    - 94% by 30yr
TETRALOGY OF FALLOT
Complete Repair

- History:
  - Lillelei (1954) - first intracardiac repair using cross-circulation
TETRALOGY OF FALLOT
Surgical Results

- Operative mortality 1-5%
- Long term survival ~ 86% (32 yr)
- Long-term complications:
  - Exercise intolerance
  - RV dysfunction
At 60 minutes of age, a term infant is cyanotic but is otherwise well. Her oxygen saturation is 75%, with a faint low-pitched murmur. The remainder of her PE is normal. Oxygen results in no change in saturation.

Of the following, the MOST likely cause of this child’s findings is:

A. ventricular septal defect
B. hypoplastic left heart syndrome
C. atrioventricular canal defect
D. neonatal pneumonia
Hypoplastic Left Heart Syndrome

- mitral stenosis
- aortic stenosis
- hypoplastic left ventricle
- hypoplastic aorta
- coarctation of the aorta
HLHS
~75% of cases diagnosed prenatally

Prenatal Options

- Termination of pregnancy
- Staged surgical palliation
- Cardiac transplantation
- “Comfort measures”: 95+% mortality in 1\textsuperscript{st} month
Hypoplastic Left Heart Syndrome: Clinical Presentation

Variable:

- No symptoms in 1st days of life
- Cyanosis
- Evolving tachypnea
- CHF, poor perfusion, shock
HLHS

Norwood: Stage I

- Aortic arch reconstruction
- Systemic to PA shunt
- Atrial septectomy

<2 weeks of age
Single Ventricle Procedures

- Glenn (6 months of age)
- Fontan (2 ½ - 3 ½ yrs)
HLHS

Glenn (SVC to PA) Shunt: Stage II

» RPA to SVC anastomosis

6 months of age
HLHS
Fontan Completion: Stage III

**Indication:** Separate systemic and pulmonary venous blood flow

3-5 years of age
HLHS: Surgical Outcomes

Survival Rate:
- Norwood: 75-85%
- Glenn: ~95%
- Fontan: ~95%

Overall 5 year survival 70-75%
Pediatric Cardiac Surgery:

- Occasionally palliative procedures needed
- Modern surgery: perform definitive procedure as early as possible
- In present era, expectation is for an overall success rate >95%
Atrial Septal Defect

- Failure of complete closure of a portion of the interatrial septum
- Left to right shunt
Treatment for ASD

- Surgery
  - Primary closure
  - Patch closure
- Trans-catheter closure
  - ASD Occlusion Device
Acquired Heart Disease

- Rheumatic Heart Disease
- Kawasaki Disease
Kawasaki Disease

- described in 1967:
  - Fever ≥ 5 days
  - and 4 of 5 additional criteria:
    - Conjunctivitis
    - Cervical lymphadenopathy
    - Rash
    - Changes in lips or oral mucosa
    - Extremity edema
In developed countries, KD has replaced acute rheumatic fever as the most common cause of acquired heart disease in children.
Kawasaki Disease: Incidence / Diagnosis

- 6 months – 5 years old
- Peak incidence in ~2 y/o
- No diagnostic test: diagnosis is based on clinical criteria
Kawasaki Disease

- Pink eye
- Oral mucosal change
- Enlarged lymph nodes
- Patchy rash
- Peeling skin

Coronary artery aneurysms (in 20% of cases)

Inflammation within the heart muscle (in 20% of cases)
Non-purulent bilateral bulbar conjunctivitis

“Strawberry” tongue

Polymorphous non-vesicular rash
Edema, erythema of hands

Coronary Aneurysm
Outcome

- Determined by cardiac involvement
- Untreated, ~20% develop coronary artery aneurysm:
  - 50% of these regress over 5 years
  - Regression less likely with giant aneurysms
- Treated (IVIG): 3% develop CAA
- Myocardial infarction can occur secondary to thrombosis of CAA
SBE Prophylaxis
2007 AHA consensus guidelines:

- For “low risk” patients antibiotic prophylaxis is no longer deemed necessary for dental or other procedures.
2007 AHA consensus guidelines:
- "high risk" patient groups (should receive prophylaxis):
  - artificial heart valves
  - history of endocarditis
  - cyanotic CHD
  - completely repaired CHD for 1st 6 mo after the procedure
  - repaired CHD with residual defect at/adjacent to prosthetic material
  - cardiac transplant with valve dysfunction
Antibiotic prophylaxis is recommended for:

- manipulation of:
  - oral mucosa
- procedures on:
  - respiratory tract
  - infected skin
  - musculoskeletal tissue

Antibiotic prophylaxis solely to prevent infective endocarditis is no longer recommended for GU or GI tract procedures.

Circulation. 2007 Apr 19
A 10 yr old with congenital heart disease is scheduled to undergo a dental extraction. The cardiac lesion most likely to require SBE prophylaxis is:

A. repaired Ventricular Septal Defect (VSD) with tiny residual VSD
B. VSD awaiting surgical repair
C. bicuspid aortic valve with moderate regurgitation
D. one year post successful transcatheter ASD device closure
Sports Clearance
A 14-year-old boy loses consciousness while playing basketball. He regains consciousness in 30 seconds and is transported to an emergency department. Head CT, EEG, and echo are normal. ECG is abnormal (sinus rhythm, HR 70 bpm, PR interval 130 msec, QTc 560).

Of the following, the MOST likely explanation for this patient’s syncopal episode is:

A. complete atrioventricular block  
B. hypertrophic cardiomyopathy  
C. long QT syndrome  
D. supraventricular tachycardia due to Wolff-Parkinson-White syndrome
Prevalence of Sudden Death

- Sudden death of the athlete on the field is predominantly cardiac-related.
- Death due to cardiovascular disease occurs once per 200,000 athletes/year.
- 5 million high school athletes compete at high school level each year: 25 high school athletes will die yearly of cardiac causes.
Hank Gathers

“In 1989, his junior season at Loyola Marymount he was the NCAA's leading scorer and rebounder. But early in the season, he blacked out during a game.”
“In his senior season, against Portland in the semifinals of the West Coast Conference tournament, slams home a dunk, enthusiastically high fives a teammate and trots back to midcourt. Suddenly, he puts his hands on his knees and crumples to the floor at Loyola's gym.”
AHA Consensus Panel Recommendations
Maron Circulation 1996 94:850

HISTORY
exertional CP
exertional syncope
SOB with exercise
fatigue with exercise
murmur
systemic HTN
FH of early cardiac death/disability
FH of HCM/DCM
FH of long QT, important arrhythmias, Marfans

PHYSICAL
auscultation (supine/standing)
assessment of femoral pulses
brachial BP
physical stigmata of Marfans
Causes of Sudden Death

- HCM (36%)
- Coronary anomalies (19%)
- Cardiac mass (10%)
- Ruptured Ao (5%)
- Tunneled LAD (5%)
- Myocarditis (4%)
- Dilated C-M (3%)
- ARVD (3%)
- MVP (2%)
- CAD (6%)
- Other (6%)

*Note: The percentages are approximate and may vary depending on the study. HCM refers to Hypertrophic Cardiomyopathy.*
Hypertrophic Obstructive Cardiomyopathy (HOCM)
Coronary Artery Malformations

Left Main Coronary from Right Coronary Cusp

Right Coronary Artery from Left Coronary Cusp
Marfan Syndrome

Dilated Aortic Root
LONG QT

Corrected QT interval: $\frac{Q~T}{\sqrt{RR}} = \frac{360}{700} = 514$

Ventricular Tachycardia $\rightarrow$ Fibrillation $\rightarrow$ Sudden death
WA State protocol

- Completion of Pre-participation H & P Exam
- State form available as sample to school districts, but use not required
- History may be completed by student and/or parent
- Form is valid for 13 months; student must have a current H & P on file
Summary

Pediatric cardiology is a continuum:

- begins with fetal diagnosis in preparation for neonatal cardiac care
- childhood management strategies
- eventual transition to specialists who manage the adult with congenital heart disease.