Updates in Congenital Heart Disease

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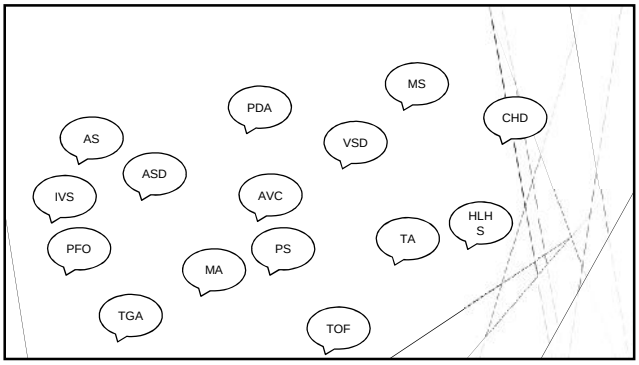
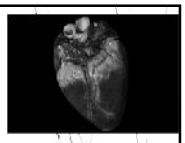
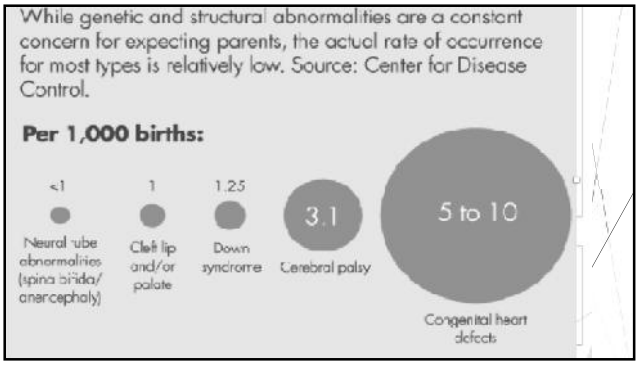


Congenital Heart Defects (CHD)



- CHDs are the most common birth defects in the United States
- The incidence of CHD is 1 in 100 births per year
  - 25% of these infants will require surgery prior to their first birthday
- In 2009, direct hospital costs for children & adults with CHD was over $2 billion
- Medical advances have lead to improved survival for children born with CHD



Current trends

- Survival depends on the severity of defect
  - 97% of infants with non-critical CHD survive to 1 year of age (1993-83%)
  - 75% of infants with critical CHD survive to 1 year of age (1993-67%)
- Significant number of adults now living with Congenital Heart Disease

Changes that have led to improved survival & outcomes

- Prenatal diagnosis
- Improved genetics testing
- Improved surgical techniques
- Improved diagnostic testing
- Better medications
- Awareness of the need for developmental screening & follow-up
- Improved care of adults with CHD

Summary of Updates

- Hereditary Conditions & Genetic testing
- Prenatal Diagnosis
- Imaging (MRI/CT)
- Interventions
- Developmental Screening
- Transplant
- Adults with CHD
- Endocarditis
- Newborn Screening
- Activity
- Technology/ Telehealth
- Preventive Cardiology

Hereditary Cardiac Conditions

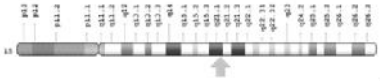
- **Arrhythmias**
  - Long QT Syndrome
  - Catecholaminergic Polymorphic VT
  - Brugada
- **Cardiomyopathies**
  - Hypertrophic
  - Familial Dilated
- **Aortic Disease**
  - Familial thoracic aortic aneurysm and dissection
  - Marfan Syndrome
- **Congenital Heart Disease**
  - Left sided lesions

Genetic Counseling

- Goals:
  1. Identify the gene responsible for a patient's heart condition through genetic testing
  2. Identify family members who are at risk for developing the same heart disease and have them screened as well
  3. Improve patient outcomes with early diagnosis and management of condition
     - The first sign of disease may be sudden cardiac arrest.

Genetic Testing Limitations

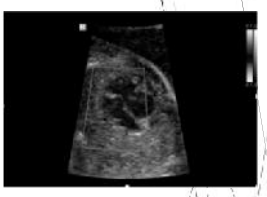
- Not available for all cardiac conditions
- Does not detect 100% of disease causing mutations
- Results are not always straightforward
  - Many gene variants that are not understood yet



Risk Factors for CHD

- Maternal Infection
- Diabetes
- Medications
- Alcohol and Illicit Drug Use
- Family history of CHD
- Chromosomal and Genetic Abnormalities
- Maternal Lupus
- Phenylketonuria

Prenatal Testing



- Fetal Echocardiography
  - Any risk factors for CHD
  - Increased nuchal thickness
  - Abnormal fetal cardiac exam
  - Hydrops
  - Fetal dysrhythmia
  - Major extracardiac abnormalities
- Amniocentesis

Importance of Prenatal Diagnosis

- Family education and counseling
- Possible fetal intervention
- Planned delivery at cardiac center
- Reduction in morbidity and mortality
  - Faster medical and/or surgical intervention
  - Trained personnel familiar with congenital heart disease

Fetal Intervention

Possible Candidates

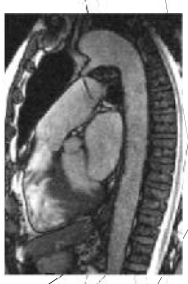
- Critical Aortic Stenosis
- Pulmonary Atresia with intact ventricular septum
- Hypoplastic left heart syndrome with intact atrial septum
- Heart block

Possible Interventions

- Aortic valvuloplasty
- Pulmonary valvuloplasty
- Creation of atrial communication
- Fetal cardiac pacing

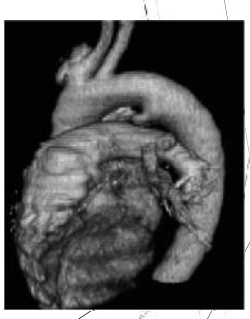
Cardiac MRI

- Provides anatomical and physiological information
  - Heart function and myocardial perfusion
  - Valvular heart disease and regurgitation
  - Diseases of the aorta and pulmonary arteries
  - Degree of shunting in different types of CHDs



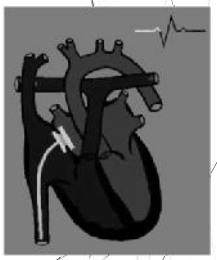
Cardiac CT

- Excellent for defining anatomy
  - Extracardiac great vessel anomalies
  - Aortic arch abnormalities
  - Size of pulmonary arteries
  - Size of intracardiac shunts
  - Small structures such as coronary arteries



Cardiac Cath

- Diagnostic:
  - Evaluate structures and assess hemodynamics
- Interventional:
  - Atrial septostomy
  - Device closure of septal defects
  - Balloon dilation of valves
  - Pulmonary valve replacement (Melody Valve)
  - Stent placement
  - Hybrid approach to complex single ventricle lesions
  - Biopsy



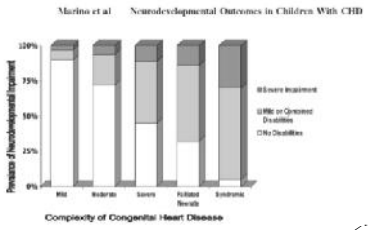
Neurodevelopment

- Developmental concerns are more apparent as survival among children with CHDs improves
- Early detection and intervention of developmental concerns is crucial.
- Regardless of the child's higher risk for developmental delays, any delay should not be considered normal.

Factors Associated with CHD that can Impact the Developing Brain

- In Utero Environment
- Hypoxia
- Genetic Syndromes
- Nutrition
- Pre-operative factors
- Bypass
- Surgery Risks
- Post-operative morbidity
- Vulnerable Child

Neurodevelopmental Outcomes



Neurodevelopmental Outcomes

American Academy of Pediatrics

American Heart Association. Learn and Live

AHA Scientific Statement

Neurodevelopmental Outcomes in Children With Congenital Heart Disease: Evaluation and Management

A Scientific Statement From the American Heart Association

Circulation

Targeted Patients

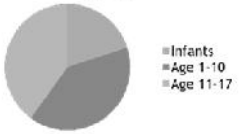
- Neonates or infants (< 12 mos) with open heart surgery
- Any combination of Congenital Heart Disease and:
  - Prematurity < 37 weeks
  - Known developmental delay
  - Genetic abnormality
  - Mechanical support
  - Heart transplantation
  - CPR at any time
  - Hospitalization > 2 weeks
  - Perioperative brain injury

Neuro-Cardiac Clinic

1. Provide neurodevelopmental evaluations
2. Educate families regarding their child's long-term outcomes and needs
3. Help families find programs, services and resources in their communities based on their child's needs
4. Assess the neurodevelopmental outcomes of children over time

Heart Transplant Demographics

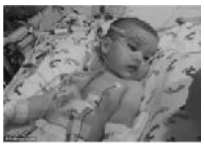
400-450 children receive a heart transplant every year in the US



172 centers in the US perform pediatric heart transplants

Advances with Transplant

- Medications
- Ventricular Assist Devices (VADs)
- Allomap
- Median survival years post-transplant have increased
- ABO incompatible (ABOi) transplants for infants



Why consider ABO Incompatible transplant?

- Infants historically had long wait-list times due to blood type, but at the same time many organs were going unused
- Between 1999-2006, 17% of children died while on the waitlist
- Infants do not have fully competent immune system

History of ABO Incompatible Heart Transplant

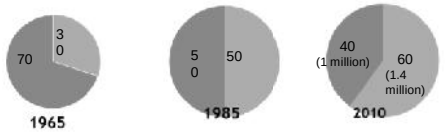
- **1996:** The Hospital for Sick Children in Toronto
- **2001:** US study, compared infants with ABO vs ABOi transplant
  - No hyperacute episodes of rejection or increase in chronic problems
  - Waitlist mortality decreased from 58% to 7% for infants
- **2012** Pediatric Heart Transplant Study: multi-center study
  - 17% of transplants were ABOi
  - 1 year survival and freedom from rejection was equivalent
- **2016:** UNOS pediatric heart allocation change
  - Allows ABOi listing for children < 2 years old

Compatible Blood Groups

TABLE 1. COMPATIBLE AND INCOMPATIBLE COMBINATIONS OF BLOOD TYPE FOR POTENTIAL ORGAN DONORS AND RECIPIENTS.

VARIABLE	COMPATIBLE BLOOD GROUPS	INCOMPATIBLE BLOOD GROUPS
Recipient's blood type		
O	O	A, B, AB
A	A, O	B, AB
B	B, O	A, AB
AB	AB, B, A, O	—
Donor's blood type		
O	O, A, B, AB	—
A	A, AB	O, B
B	B, AB	O, A
AB	AB	O, A, B

Changing Proportion of Pediatric and Adults Living with CHD



Complications in Adults with Congenital Heart Disease

- More adults living with CHD → Growing recognition of cardiac and non-cardiac issues
  - Heart Failure
  - Atrial Arrhythmias
  - Vascular Complications
  - Neurological
  - Restrictive Lung Disease
  - Liver Dysfunction
  - Renal Disease
  - Cancer Risk due to radiation exposure
  - Gynecologic
  - Psychosocial


AHA Scientific Statement

Congenital Heart Disease in the Older Adult:
A Scientific Statement From the American Heart Association

General updates

- Endocarditis Prophylaxis
- Newborn screening for critical CHD
- Activity and Sports Guidelines
- Technology and CHD
- Preventive Cardiology

Endocarditis Prophylaxis Recommendations

1. Prosthetic cardiac valve
2. History of infection endocarditis
3. Cardiac transplant with abnormal heart valve function
4. Congenital heart disease:
   - Unrepaired or not fully repaired CHD
   - Any prosthetic material or a device- for the first 6 months after repair
   - Residual defects

Newborn Screening for CHD

- The AAP recommends that ALL newborns be screened for CHD
- Since 2011, 43 states have mandated pulse ox screening as part of the newborn screen in hospital
- Cost is approximately $6.28/child
- Earlier diagnosis leads to better outcomes

Activity Restriction for Children with CHD

Importance

- Avoid catastrophic event that would not have otherwise occurred
- Avoid progression of disease

Problems

- No evidence it actually works
- May provide false reassurance
- May not change the risk of sudden death
- Not benign
- May still be active, even if not playing on a sports team
- Physicians and patients do not always follow restriction guidelines

Sports Participation for Children with CHD

- 3 recent studies showed that sports participation for children with CHD is associated with improved:
  - Exercise Capacity
  - BMI
  - B-type natiuretic peptide (BNP) levels
  - Quality of life scores
  - Survival

Which CHDs should be restricted?

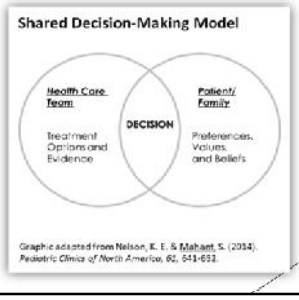
Restriction Not Needed:

- Atrial Septal Defect
- Ventricular Septal Defect
- Patent Ductus Arteriosus
- Mild valve anomalies

Restriction May be Indicated:

- Aortic Disease
- Tetralogy of Fallot
- Single ventricles
- Eisenmenger's Syndrome
- Coronary artery anomalies
- Significant symptoms or arrthymias
- Anticoagulation
- Pacemaker

Activity Guidelines:



Technology and CHD

- Databases
  - Allows for sharing of information among centers across the country
  - Allows for better research and development of best practices
  - Share Seamlessly, Steal Shamelessly

Technology cont'd.

- Telehealth
  - Remote cardiology appointments
  - Remote study readings (echos, ekgs)
- Apps
  - Building HOPE, monitorinig high risk CHD infants from afar with better caregiver engagement
- Jabber
  - HIPPA secure video chatting

Preventive Cardiology

- Goal: to reduce or eliminate risk factors in children that may lead to adult heart disease
- Multidisciplinary team
- Children with risk factors should be referred:
  - Dyslipidemia (fasting)
  - HDL < 40
  - Family history
  - HTN
  - Obesity
  - Type 1 and 2 Diabetes
  - Kidney disease

Case #1

Tyler: 3 month well visit

- History: Full term, healthy. Tyler has not been feeding as well over the last several weeks. Tyler becomes tired with feeds, and often cannot finish his 3oz bottle.
- Exam:
  - Weight: 3rd percentile
  - Alert
  - Pink in color
  - Tachypneic
  - 2/6 systolic murmur
  - Pulses +2 throughout
  - Liver is 2cm below RCM
- EKG: possible right ventricular hypertrophy (RVH)
- Tyler is referred to pediatric cardiology.
- Echo: secundum ASD and mild RVH

Case#2

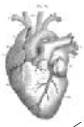
- Liam is 6 month old who had been seen by pcp twice and diagnosed with viral URI.
- Liam was then admitted to UVA with emesis and general malaise. He was found to have
  - Elevated CK, AST, LDH
  - Echo: giant coronary aneurysms and reduced left ventricular systolic function
  - Cardiac CT: numerous coronary artery aneurysms and likely left anterior descending (LAD) thrombus
  - Cardiac Cath: complete occlusion of LAD was found
- Treated with systemic tPA x 12 hours without resolution
- Diagnosis: untreated Kawasaki's disease x 5 weeks
- IVIG and Infliximab for ongoing evidence of inflammation due to Kawasakis. (CRP was elevated, peak troponin 15.7)
- Abciximab given as antiplatelet and to promote vascular remodeling
- ASA and Lovenox
- Enalapril, Aldactone, Lasix and Cavediolol for regional wall motion abnormality and acute MI

Case #3

- Kacy presented for her 20 week fetal ultrasound
- Concerns about fetal heart
- Kacy was referred to fetal cardiology
  - Fetal echo at 21 weeks gestation
  - Diagnosed with hypoplastic left heart syndrome
  - Discussed fetal intervention, but decided against it
  - Couseling and education provided
  - Future clinic visit to meet MFM OB, cardiac surgeon, neonatologist, cardiology NP, social worker, take a tour, etc
  - Birth plan to deliver at UVA
  - Neonatal surgery scheduled before baby was born

What does the future hold?

- CHD: Regenerative therapies; 3-D Printing; Improved imaging techniques; Robotic tools; less invasive cardiac interventions
- Trained workforce to care for growing adult CHD population
- Telehealth resources to rural areas
- Improved bridge to transplant for children



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