CONGENITAL HEART DISEASE in Down Syndrome

Program Goals

- Review cardiac structure and function
- Describe health implications for DS
- Explain common heart defects
- Provide resource information
- Identify associated symptoms
- Manage and establish therapeutic goals
- Improve knowledge of cardiac interventions
- Advocate for preventative care

No Disclosures

CHD

To provide high quality care, nurses need to understand that long term consequences can be related either to Congenital Heart Disease or to the specific repair done.

—American Journal of Nursing 2015

Cardiac Structure & Function

CHAMBERS AND VALVES

One way valves allow blood to flow in one direction

Four Chambers

Ascending Aorta
Right Atrium
Right Ventricle
Left Atrium
Left Ventricle
Forms week 5 in embryo
Two endocardial tubes merge to form heart tube
Endocardial cushions: cells that eventually develop septum
FGF8 Fibroblast growth factor 8: a protein coding gene
Completely formed at 8 weeks
First functional organ to develop
Starts to beat and pump blood around day 21

Electrical Function

HEART DEVELOPMENT

Trisomy 21: 47 chromosomes in each cell instead of 46
Major cause of Congenital Heart Disease
Additional genetic material alters the course of development
Physical traits: low muscle tone; small stature; eye slant
UNIQUE individuals

CAUSES OF CHD

Down Syndrome is the most frequent known cause of atrioventricular septal defects
Characteristic heart defect derives from abnormal development of the endocardial cushions
DSCAM Down Syndrome Cell adhesion molecule overexpression during heart development may play role in CHD
Causes of CHD other than DS
- Alcohol consumption or Drug abuse
- Antiepileptic medications (Valproate)
- Genetic conditions (Turner Syndrome; Noonan Syndrome; PKU)
- Viral infection (Flu; Rubella in First trimester)
- Medications (Lithium; isotretinoin)
- Maternal Diabetes types I and II, not gestational
- Organic Solvent exposure

Common heart defects in Down Syndrome
- ENDOCARDIAL CUSHION DEFECT
  - A-V SEPTAL DEFECT
  - VENTRICULAR SEPTAL DEFECT
  - TETRALOGY OF FALLOT

CONGENITAL HEART DISEASE
- Requires Long Term Monitoring
- Regular Cardiology follow up
- Endocarditis Prophylaxis
- Understanding of sequela (ie: CHD may result in seizures caused by cyanotic rt. to lt. shunts)

ENDOCARDIAL CUSHION DEFECT
More commonly known as AV canal or Septal Defects
- ASD, VSD & improperly formed mitral and/or tricuspid valves
- Abnormal development of endocardial cushions (2 areas of thickening that eventually develop septum or wall, that separates the 4 chambers)
  - Left to Right Shunts
- CHF, Respiratory infections, poor weight gain, adults develop atrial arrhythmias

Mr. B at 2 months old. On lasix, formula and breast milk to gain weight for surgery. Adorable but blue and couldn’t tolerate altitude of home. Gained weight for surgery at 5 months. Residual mitral leakage. No meds or limitations now!
Treatment and Outlook

- Surgery for complete ECD in 1st year of life
- Early surgery important in DS to decrease lung damage
- May require more than 1 surgery
- Diuretics and contractility meds
- Overall health for positive outcomes
- Complications include CHF, Eisenmenger syndrome, pulmonary hypertension, leaky valves, arrhythmia, endocarditis risk

Eisenmenger Syndrome

Refers to any untreated congenital cardiac defect with intra cardiac communication that leads to pulmonary hypertension, reversal of flow, and cyanosis

- Medscape - emedicine

Symptoms: bluish lips, fingers and toes; tingling; clubbing; chest pain; vertigo; syncope; fatigue; S.O.B.; palpitations; stroke; gout

Requires surgical correction

PATENT DUCTUS ARTERIOSUS

Channel between PA and Aorta
Usually closes 1st day of life
Causes high blood flow to lungs
During fetal life, diverts blood from lungs (blood already oxygenated by mom)

PDA's and ASD's can close on their own with growth
PDA non closure causes rapid breathing/ poor weight gain/ fluid in lungs

PDA and prematurity

- Prostaglandin inhibitors (indomethacin or ibuprofen)
- Paracetamol (Tylenol) used in low birth weight and prematurity
- Surgical treatment

-Cochrane Neonatal Review Group

ATRIAL SEPTAL DEFECTS

- Permit shunting of blood left to right
- Abnormally persistent opening between atria
- Undiagnosed adults with ASD may present with arrhythmias.
- Four types of ASD's
- Long term outcomes of transcatheter closure unknown
- Practice endocarditis prophylaxis
1. Secundum – center of atrial septum. 75% of ASD’s. Repair by surgical patching or transcatheter occlusion.

2. Primum – low in atrial septum, cleft in anterior leaflet of mitral valve. 15–20% of ASD’s.


4. Coronary sinus ASD – defective roof of coronary sinus and often associated with left sided SVC. Less than 1% of ASD’s.

ASD closure devices are the Gore Helix septal occluder and the Amplatzer septal occluder transcatheter. 
http://links.lww.com/AJN/A63

ASD CLOSURE DEVICES

- New technology
- Femoral approach threaded to RA, across ASD and into left atrium
- Defect size assessed with balloon sizing
- Collapsed device deployed
- Device becomes endothelialized
- Low dose ASA 3–6 months

POTENTIAL COMPLICATIONS

- Erosion of atrial or aortic wall due to pressure of the device (chest pain, dyspnea, dizziness)
- Residual leaking (require echocardiogram follow up)
- Infective endocarditis
- Long term outcomes unknown
- Atrial arrhythmias

VENTRICULAR SEPTAL DEFECTS

- Hole in ventricular septum (wall between lower chambers)
- Blood from LV flows back into RV due to high LV pressure
- Increases blood volume into lungs by RV and creates congestion in lungs

VSD’s

- Wall between chambers usually closes before birth, but VSD causes mixing of oxygenated blood with unoxgenated blood
- Lungs work harder
- Reduces oxygen to body
- Difficulty eating and slower growth
- Damages lung and blood vessels
**VSD’s Treatment and Outlook**

- Small VSD’s close on their own
- Loud heart murmur
- Pulmonary artery banding or transcutaneous repair
- Larger requires OHS patching
- Limit activity to endurance
- AHA doesn’t require endocarditis prophylaxis

- 4 abnormalities: VSD; Outflow tract stenosis; Enlarged RV; Overriding aorta (aorta lies directly over VSD)
- VSD allows blood to pass from RV to LV without going through the lungs
- Blue skin color and mucous membranes due to lack of oxygen

**TOF Tet Spells**

- Squatting: Increases pulmonary blood flow; increases PVR and decreases magnitude of left shunt across the VSD
- Difficult feeding
- Pale blue during crying/feeding
- Unpredictable
- Hypoxic dyspnea
- Clubbing fingers and toes after 3–6 mo. / old
- Hematocrit elevated

*Spor of infundibular septum worsens RVOT obstruction – Medscape*

**TOF treatment and outlook**

- OHS intra cardiac repair. Timing dependent on symptoms, with goals to close VSD, resect infundibular (base of PA) stenosis, and relieve RV outflow tract obstruction
- Oxygen of limited value as primary abnormality is decreased pulmonary blood flow
- Careful use of meds pre op
- Revision/re operation for residual VSD or RVOT obstruction
- Cannot tolerate volume overload
Danny’s story

One year after successful correction of TOF, Danny was admitted to UCSF with a Clinical picture of Septic Shock, Disseminated intravascular coagulation and severe metabolic acidosis. He had petechiae and purpura on face and extremities, ecchymosis of lower extremities, lethargy. WBC = 1.2 with large left shift. Echo showed poor cardiac function. Had paracentesis for enlarged liver compressing RV. Per physician notes, “It is unfortunate that he received so many antibiotics prior to his hospitalization here because it has clouded the picture for us”.

Subsequently, his spleen infarcted and was no longer functional. He recovered and has done well for many years. He is now 27 years old. In 2016, he became more fatigued with frequent respiratory infections and recommendations for Echocardiograms to be done every 6 months.

Cardiac conferences were held at CPMC and Stanford. Cardiac MRI done (gold standard for diagnostics).

As a result of TOF repair as a child, he now has severe Pulmonary Regurgitation, severe RV dilation and “technically challenging” anatomy.

Interventionalist performed Heart Catheterization and attempted percutaneous Pulmonary Valve Replacement in Cath Lab, rt. Femoral approach under anesthesia.

TOF goals for treatment

Heart and Lungs to pump more efficiently

Protect Ventricle

Decrease Arrhythmias

Reduce Right Heart Failure

Feel Better
Percutaneous transcatheter Interventional Cardiac Procedures are new and promising for those who may not be candidates for OHS.

There is no long term outcome data.

There are 8 sites in the country doing clinical trials on a variety of outflow tracts.

Only since 2008, AHA Guidelines, were PCP’s focused on ongoing adult CHD.

He recovered relatively well and spent the night on telemetry floor. Discharged next day.

The intervention was aborted and unsuccessful.

No available valve to fit the now, severely enlarged, RVOT. Stent used as a landing zone remains, and he is prescribed daily Aspirin 81 mg. lifetime. Decision point?

Serious and sometimes fatal

Bacteria in bloodstream common with daily activities (tooth brushing/flossing; using wooden toothpicks; water picks; chewing food)

Increased risk with prosthetic heart valves, CHD; history of endocarditis

Infection of lining of heart’s chambers or valves.

Group A Strep
Inflammation

Fever
Sweats
Unexplained rashes
Nailbed hemorrhages
Painful sores (Osler’s nodes)
Malaise
Muscle aches
Red spots on palms/feet (Janeway lesions)

Can totally damage valves and require OHS.
Vegetations can break off causing emboli.

Lifelong Care – follow up; carry card
Blood Cultures BEFORE antibiotics
Antibiotic prophylaxis prior to dental or invasive procedures

- ACC/AHA 2008 Guidelines management of adults with CHD
RESOURCES for Lifelong Heart Health

ACHA  Adult Congenital Heart Association:  
info@achaheart.org  www.achaheart.org

AHA  American Heart Association:  
www.heart.org  heartinsight.org (magazine)

ACC/AHA Guidelines for Management of Adults with Congenital Heart Disease (pdf web link)  
www.cdc.gov/heartdefects

RESOURCES

http://emedicine.medscape  Endocardial Cushion Defects
JACC Journal of American College of Cardiology "Timing of PVR in TOF using MRI" 2012 Holmes, K. M.D.
JACC July 2011 Vol. 58 Issue 2 Transcatheter Valve Technology
JACC Sept. 2012 Vol. 60 Issue 11
PubMed Jan-Feb. 2010 "Adults with Down Syndrome: Safety and long term outcome of cardiac operation"

http://link.springer.com/article  Current Pediatrics Reports  

RESOURCES

- ACC/AHA 2008 Guidelines for the management of adults with CHD
- American Journal of Nursing, January 2015, volume 115, No. 1 p24–35
- Genetics in Medicine 2001 Down Syndrome CHD: a narrowed region and a candidate gene. Barlow, G.
- McElhinney, Doff (numerous publications transcatheter procedures)
- National Down Syndrome Society NDSS The Heart and Down Syndrome
- Stanfordchildrens.org Congenital Heart Disease

A very special Thank–You to the mother’s Facebook support group  
"H.E.A.R.T. 21”  
Who shared your stories and tears