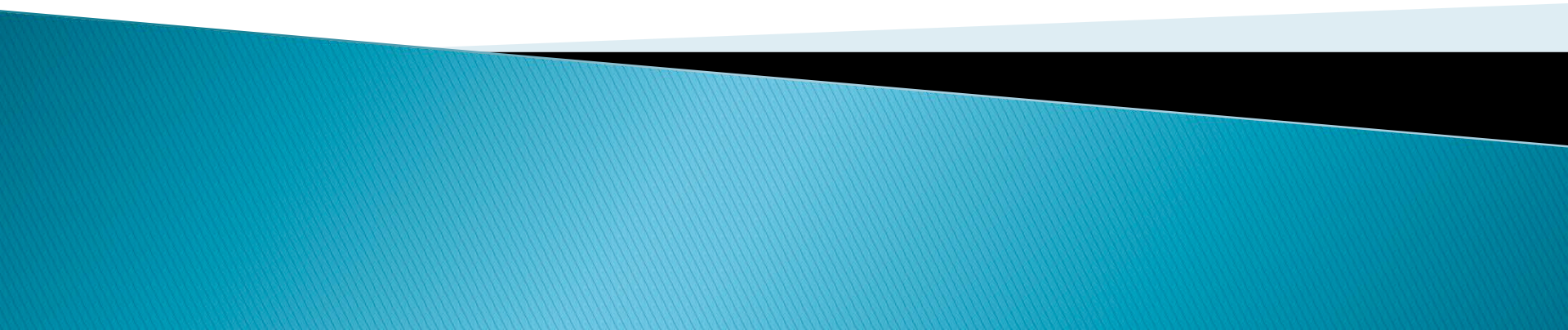
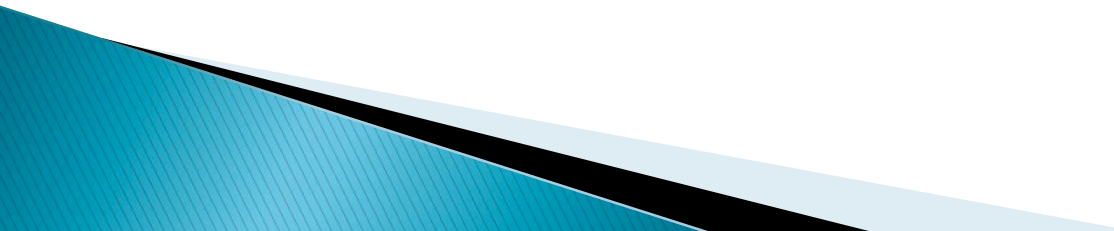


# CONGENITAL HEART DISEASE in Down Syndrome

Doreen Bestolarides R.N.  
(CCRN)



## 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

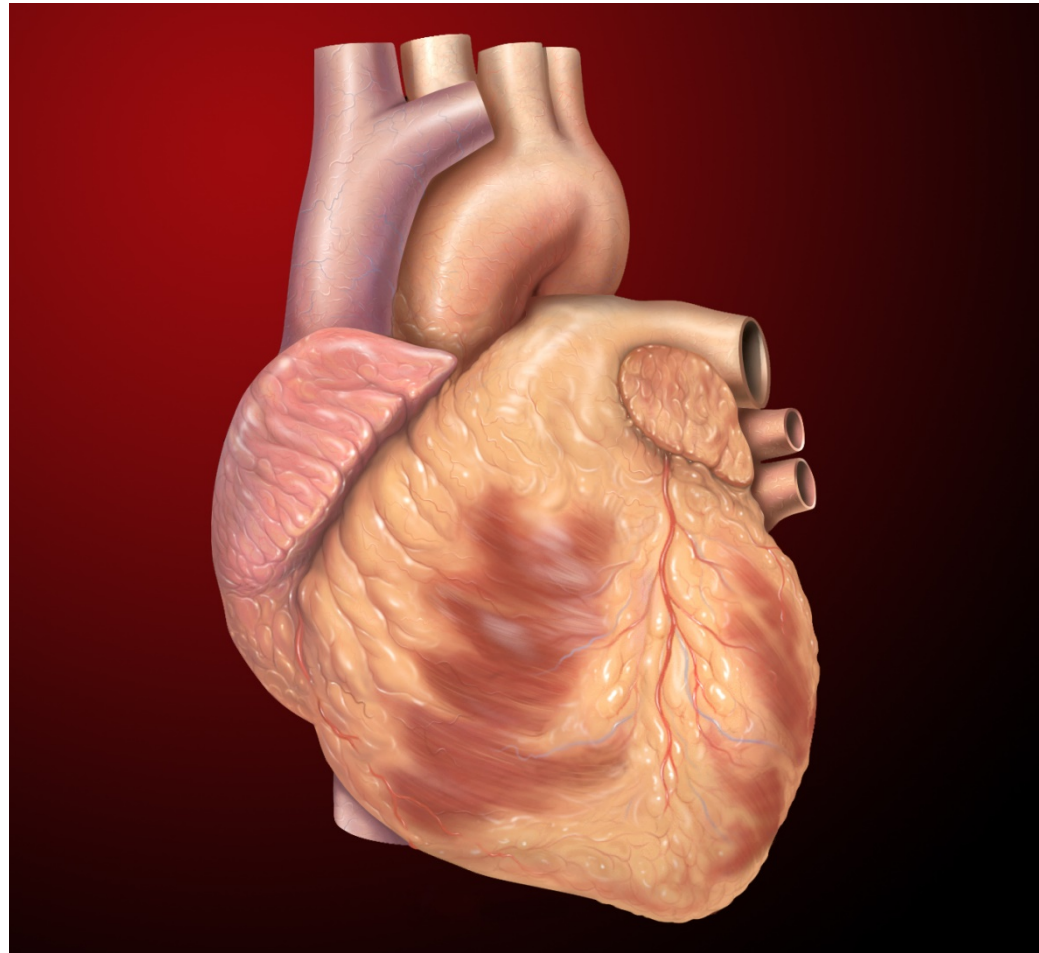
To provide high quality care, nurses need to understand that long term consequences can be related either to Congenital Heart Disease or to the specific repair done.

–American Journal of Nursing 2015



# CARDIAC STRUCTURE & FUNCTION

*the heart is electrical and mechanical*

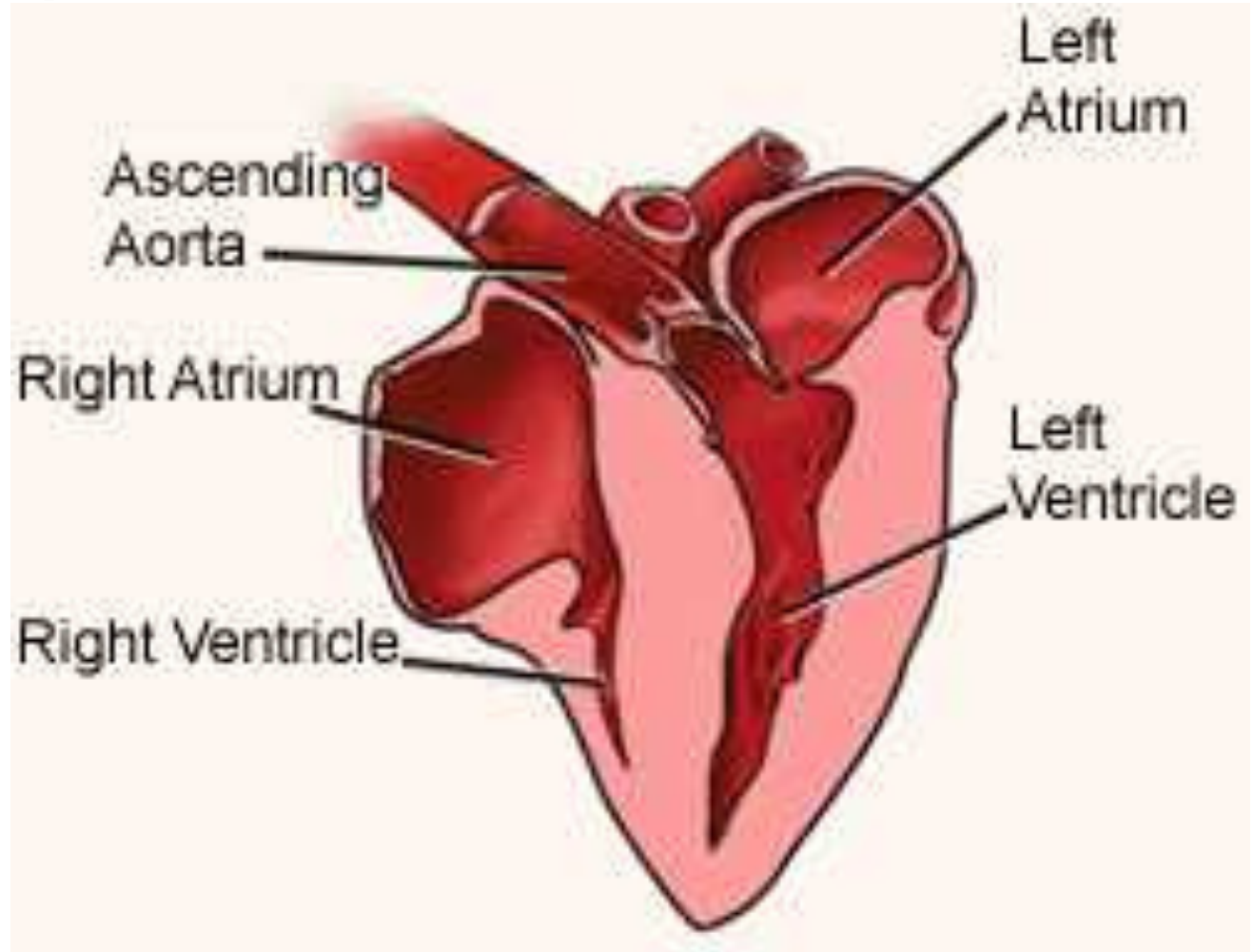


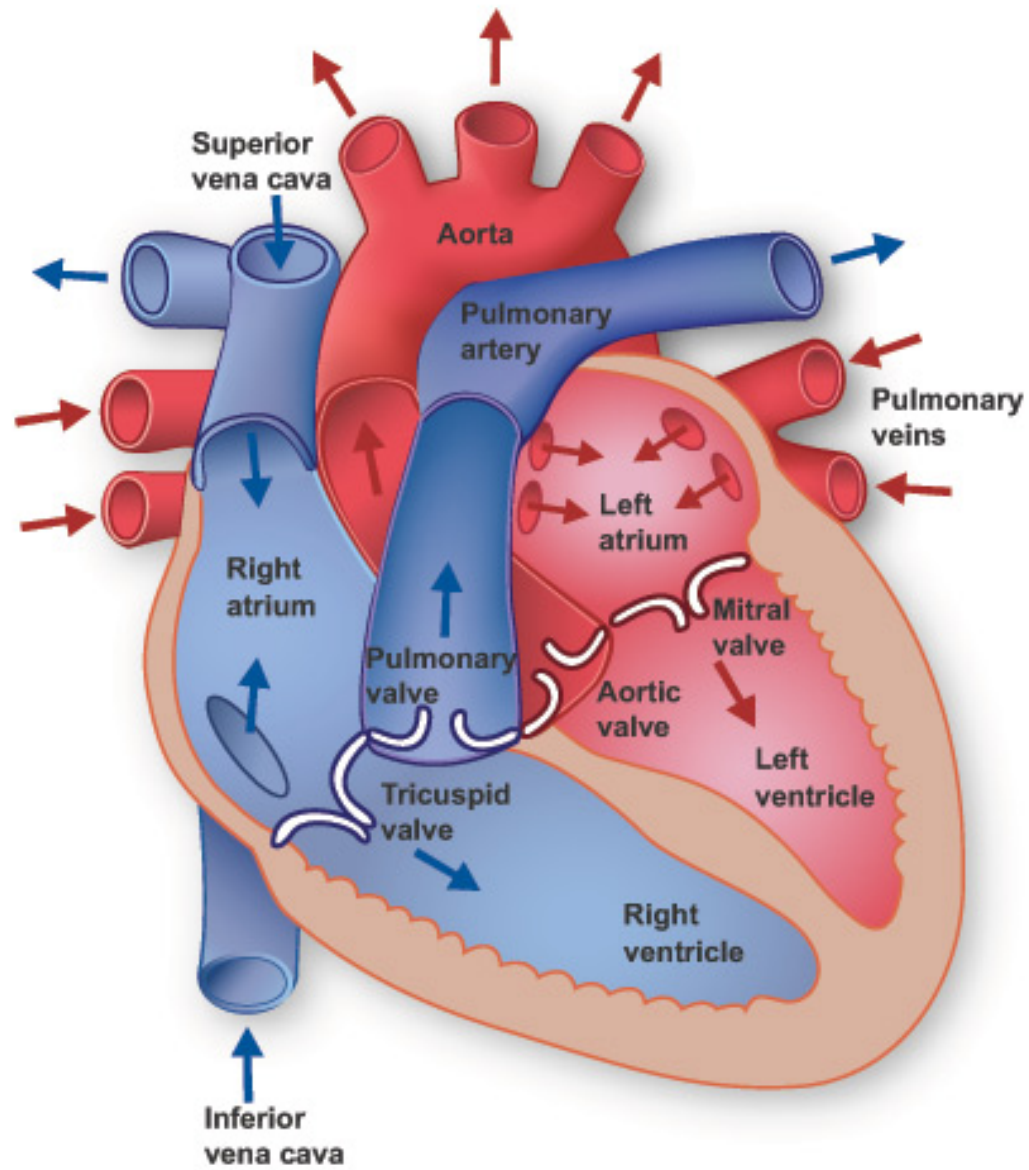


# CHAMBERS AND VALVES

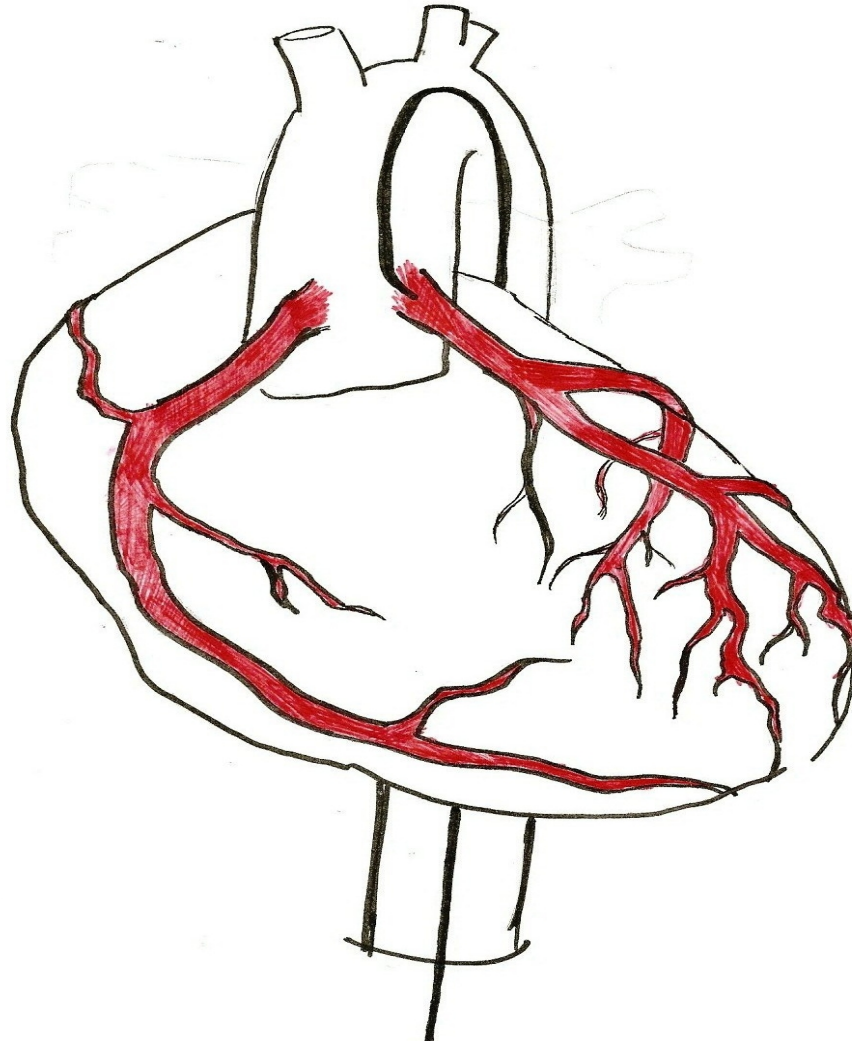
*One way valves allow blood to flow in one direction*

Four Chambers



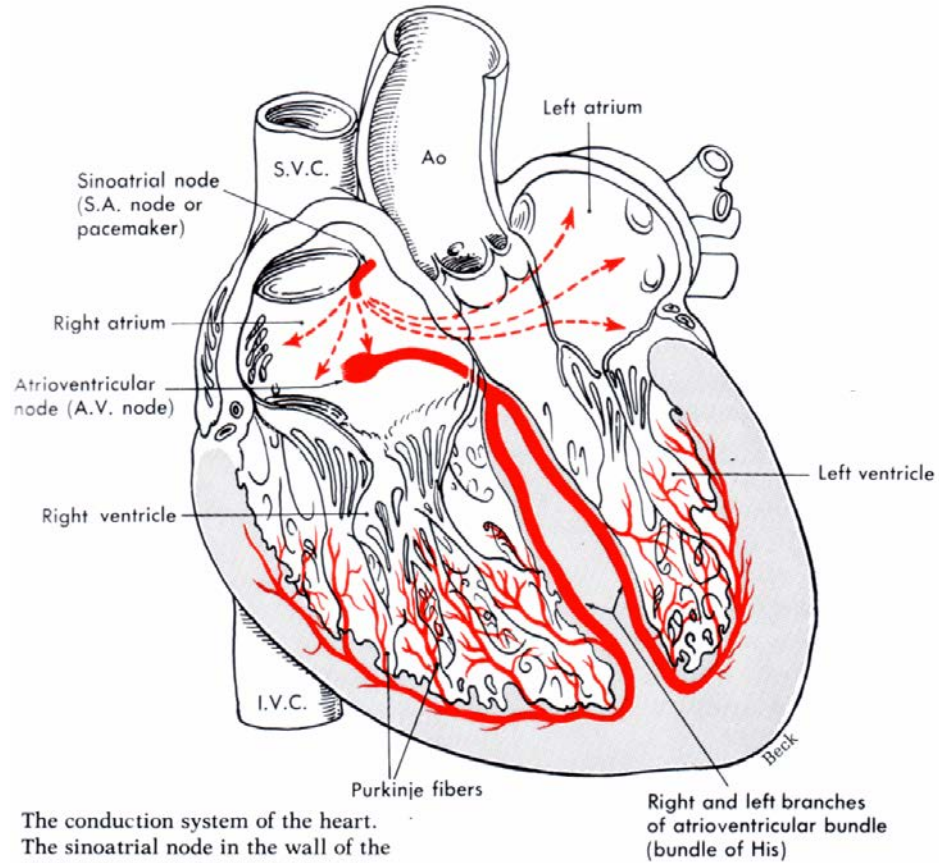


Coronary Arteries lie on the surface of the Heart

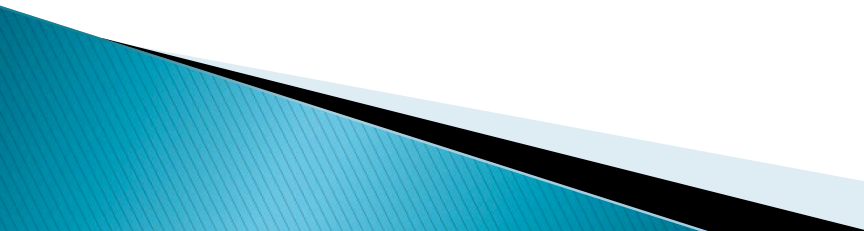




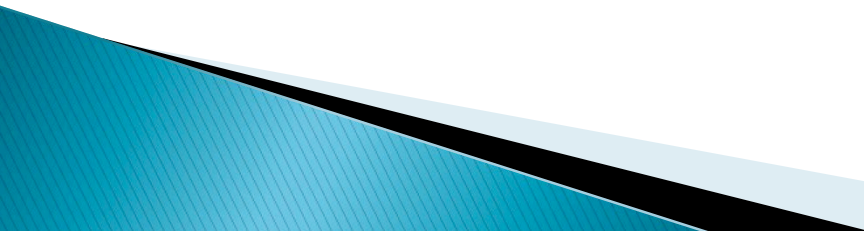
# Electrical Function



# HEART DEVELOPMENT

- ▶ 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
- 

# DOWN SYNDROME

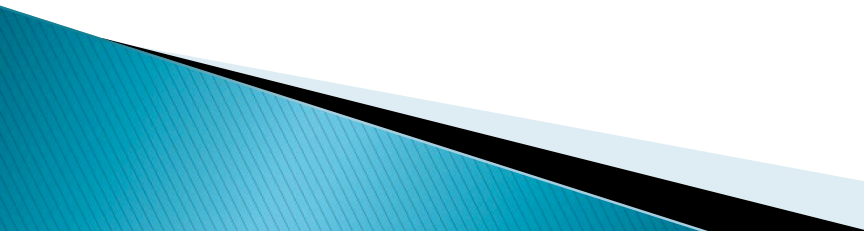
- ▶ Trisomy 21 : 47 chromosomes in each cell instead of 46
  - ▶ Major cause of Congenital Heart Disease
  - ▶ Additional genetic material alters the course of development
  - ▶ Physical traits : low muscle tone; small stature; eye slant
  - ▶ UNIQUE individuals
- 

## 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**

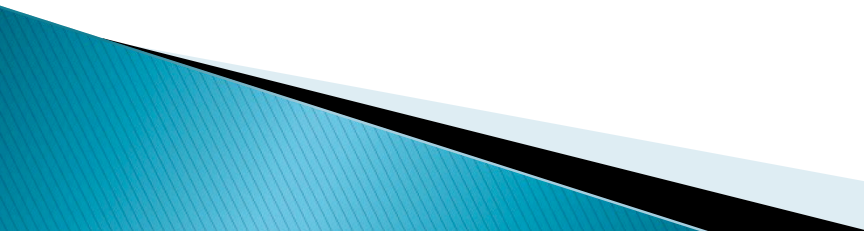
**A-V SEPTAL DEFECT**

**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 !

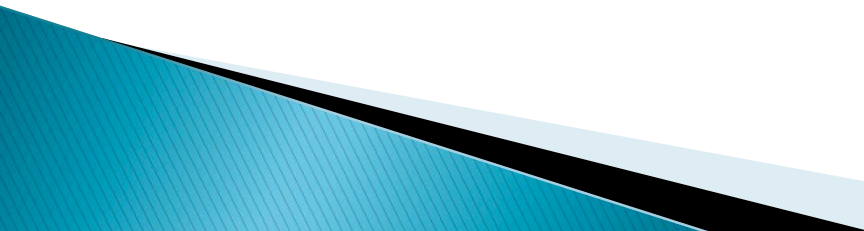


NICU for 5 days post op  
(*no more blue!*)





## Treatment and Outlook

- ▶ Surgery for complete ECD in 1<sup>st</sup> 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



*Micah*



# PATENT DUCTUS ARTERIOSIS

Channel between PA and Aorta

Usually closes 1<sup>st</sup> 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

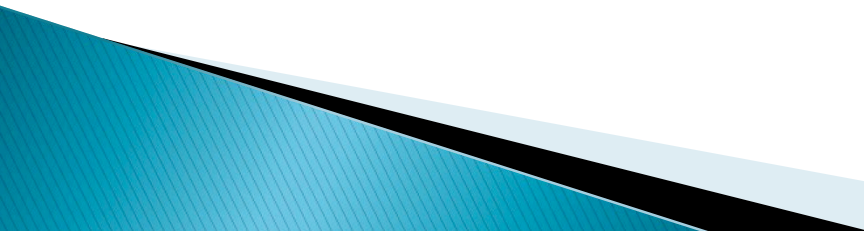
- ▶ Prostaglandin inhibitors (indomethacin or ibuprofen)
- ▶ Paracetamol (Tylenol) used in low birth weight and prematurity
- ▶ Surgical treatment

-Cochrane Neonatal Review Group

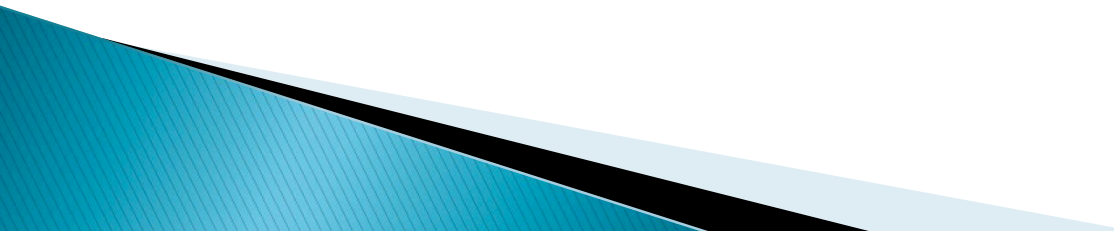




## ATRIAL SEPTAL DEFECTS

- ▶ 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
- 

## 4 Types of ASD's

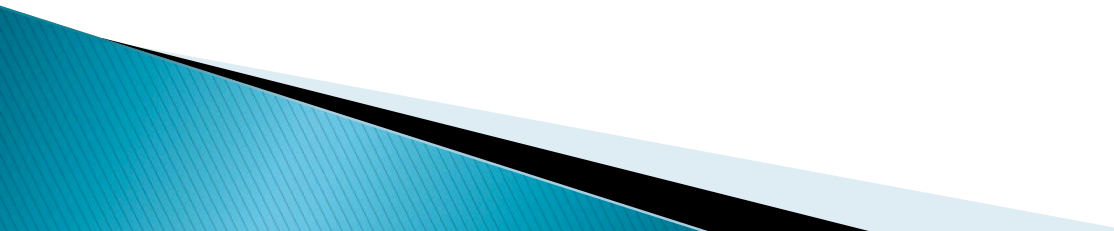
1. Secundum – center of atrial septum. 75% of ASD's. Repair by surgical patching or transcatheter occlusion.
  2. Primum – low in atrial septum, cleft in anterior leaflet of mitral valve. 15–20% of ASD's.
- 

## ASD's continued

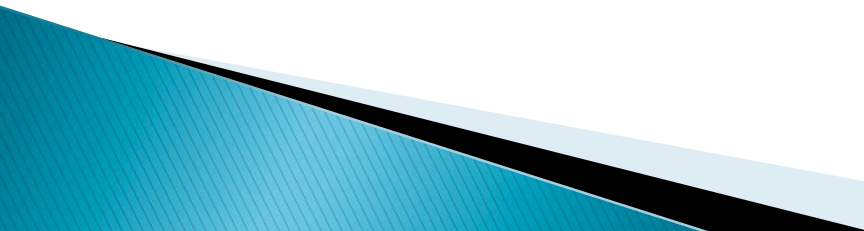
3. Sinus venosis ASD – high in right atrium near SVC. 5–10% of VSD's. May involve pulmonary vein. Requires OHS repair. Redirect drainage via patch. Outcomes excellent.
4. Coronary sinus ASD – defective roof of coronary sinus and often associated with left sided SVC. Less than 1% of ASD's.

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

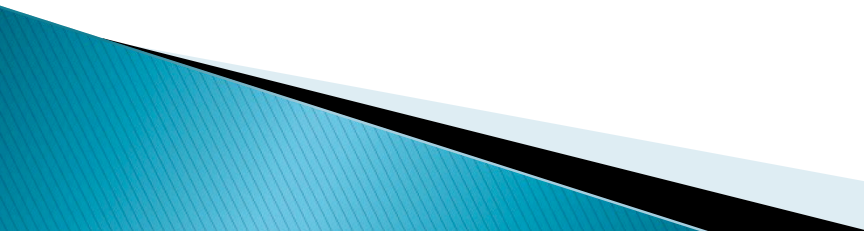
## ASD CLOSURE DEVICES

- ▶ 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

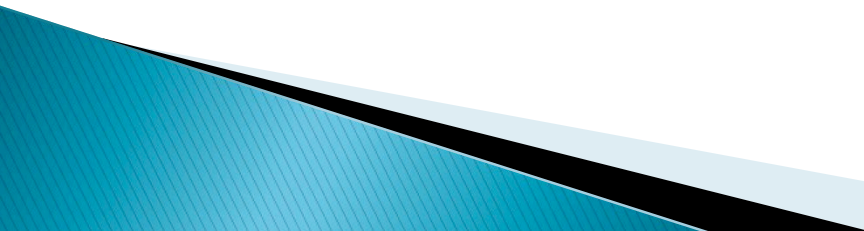
- ❖ Erosion of atrial or aortic wall due to pressure of the device (chest pain, dyspnea, dizziness)
  - ❖ Residual leaking (require echocardiogram follow up)
  - ❖ Infective endocarditis
  - ❖ Long term outcomes unknown
  - ❖ Atrial arrhythmias
- 

## 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

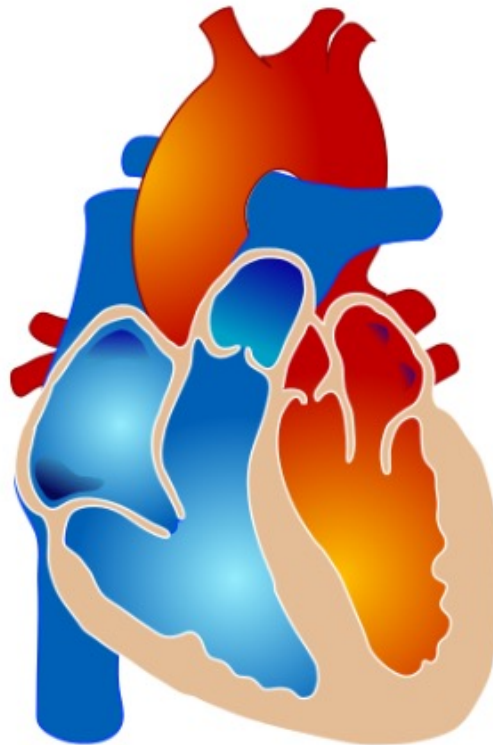
- ▶ Wall between chambers usually closes before birth, but VSD causes mixing of oxygenated blood with unoxygenated blood
  - ▶ Lungs work harder
  - ▶ Reduces oxygen to body
  - ▶ Difficulty eating and slower growth
  - ▶ Damages lung and blood vessels
- 

# VSD's Treatment and Outlook

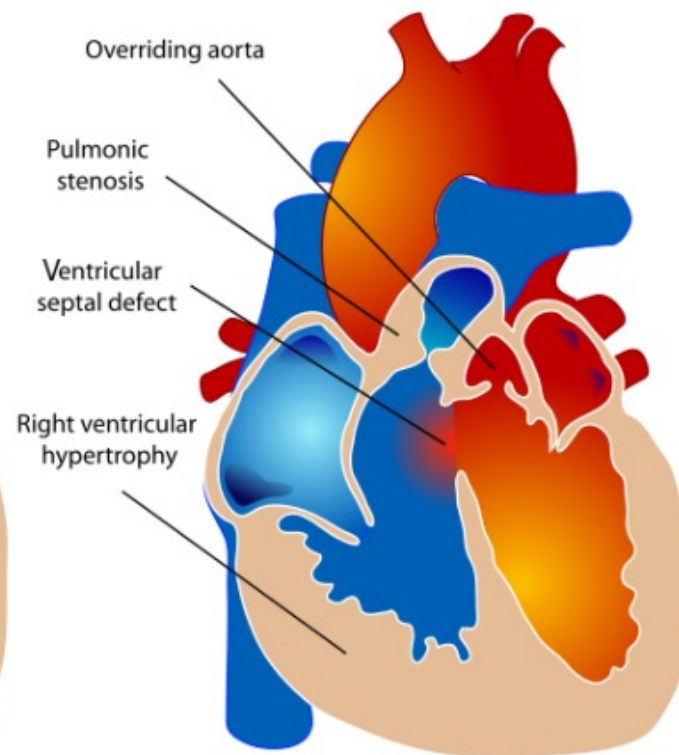
- ▶ Small VSD's close on their own
- ▶ Loud heart murmur
- ▶ Pulmonary artery banding or transcatheter repair
- ▶ Larger requires OHS patching
- ▶ Limit activity to endurance
- ▶ AHA doesn't require endocarditis prophylaxis

# TETRALOGY OF FALLOT

Normal heart



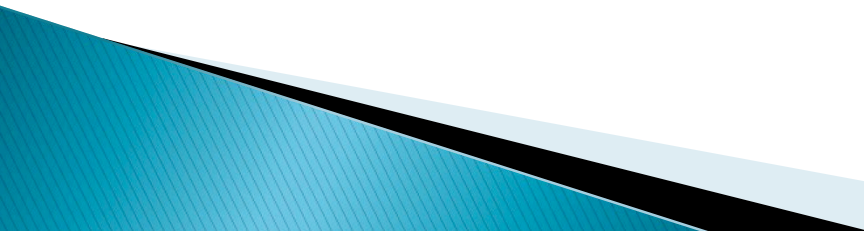
**Tetralogy of Fallot**



Danny was born with Trisomy 21 and Tetralogy of Fallot. At age 2 he had syncopal cyanotic spells. OHS consisted of VSD closure with Dacron patch, Gore-Tex membrane patch used to enlarge pulmonary valve annulus and PFO closed with a suture. The infundibulum of RV incised through pulmonary valve annulus and hypertrophied muscle resected.



## TETRALOGY OF FALLOT

- ▶ 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


*Spasm 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

# TOF repair outlook

- ▶ Pulmonary regurgitation
  - ▶ Right heart failure
  - ▶ Ventricular arrhythmias
  - ▶ Endocarditis risk
  - ▶ Pulmonary Valve Replacement – Percutaneous technology
  - ▶ Pulmonary Valve Replacement – OHS
  - ▶ Weigh technical challenges with Quality of Life
- 

TOF goals for treatment

Heart and Lungs to pump more efficiently

Protect Ventricle

Decrease Arrhythmias

Reduce Right Heart Failure

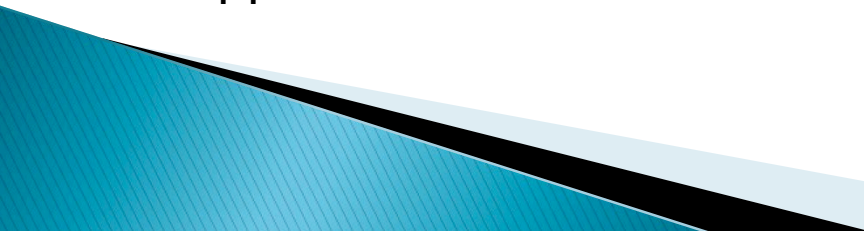
Feel Better



## 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".

## Danny's story

- ▶ 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.
- 

# Percutaneous Interventional Cardiology

May be an alternative to OHS *criteria driven*  
Less invasive *faster recovery*  
Performed in larger Medical Centers






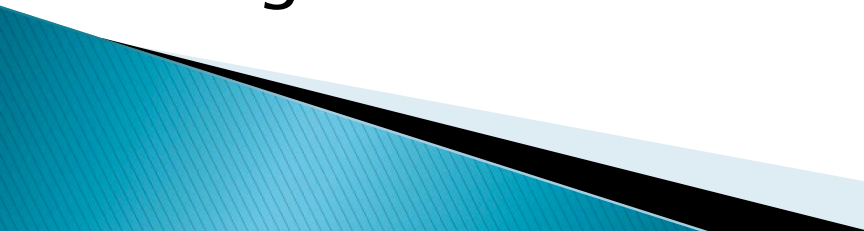
# Percutaneous Interventional Cardiology




## Points to Ponder

- ▶ 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.
- 

## Danny's Story

- ▶ 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 ?*
- 

# BACTERIAL ENDOCARDITIS

- ▶ 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
- 

# ENDOCARDITIS

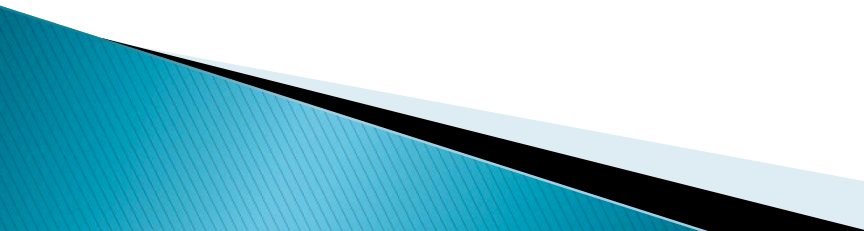
Infection of lining of heart's chambers or valves.

Group A Strep

Inflammation



## Endocarditis symptoms

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



# Endocarditis

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](mailto:info@achaheart.org) [www.achaheart.org](http://www.achaheart.org)

AHA American Heart Association:  
[www.heart.org](http://www.heart.org) [heartinsight.org](http://heartinsight.org) (magazine)

ACC/AHA Guidelines for Management of Adults  
with Congenital Heart Disease (pdf web link)

[www.cdc.gov/heartdefects](http://www.cdc.gov/heartdefects)



# RESOURCES

- ▶ ACC/AHA 2008 Guidelines for the management of adults with CHD
- ▶ American Journal of Nursing, January 2015, volume 115, No. 1 p24–35
- ▶ “Babies with Down Syndrome: A New Parent’s Guide”, p.76–78 Skallerup, Susan J. 2008 Woodbine House, Third Ed.
- ▶ Genetics in Medicine 2001 Down Syndrome CHD: a narrowed region and a candidate gene. Barlow, G.
- ▶ McElhinney, Doff (numerous publications transcatheter procedures)
- ▶ “The Heart of a Child: what families need to know about heart disorders in children” Johns Hopkins University Press 2001 Neill
- ▶ National Down Syndrome Society NDSS The Heart and Down Syndrome
- ▶ Stanfordchildrens.org Congenital Heart Disease
- ▶ <http://www.uptodate.com/management-and-outcome-of-tetralogy-of-fallot>

# 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  
“Transcatheter Pulmonary Valve Replacement: A current review”, June 2013, Volume 1, Issue 2, pp.83–91. Gillespie, M., McElhinney, D.

## RESOURCES

*A very special Thank-You to the mother's  
Facebook support group*

*“H.E.A.R.T. 21”*

*Who shared your stories and tears*

