Congenital Heart Defects

Initial Diagnosis and Management

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Congenital Heart Defects

Congenital heart defects are the most common type of birth defect ~1 out of 100 live births - affecting nearly 40,000 births in the United States each year. They are the leading cause of infant deaths due to birth defects—and birth defects are the leading cause of neonatal deaths!

Ventricular Septal Defect (VSD) is the most common heart anomaly.

At CMH we currently follow 11,000 children with CHD in our clinic, and perform 472 cardiac catheterizations and over 450 surgeries annually.

With the development of our new Fetal Health Center, babies that might normally have been undiagnosed and become very sick now deliver with a full staff of MD’s, NP’s, RT’s, and other staff with a pre-planned action plan.
Review of Cardiac Anatomy and Physiology

Fetal Circulation

In fetal circulation, the placenta accepts the deoxygenated blue blood from the fetus through the umbilical arteries. In the placenta, it picks up oxygen and becomes red and then returns to the fetus via the umbilical vein. The red blood enters the fetus and passes through the fetal liver to the right side of the heart. It then goes across a connection between the atria, the patent foramen ovale (PFO), from the right atrium (RA) to left atrium (LA). It then enters the left ventricle (LV) and goes out the aorta. As a result, the blood with the most oxygen goes to the brain.

Blood coming back from the fetus from the vena cava also enters the RA and is pumped to the RV. Most of the blood that leaves the RV bypasses the lungs through the ductus arteriosus (DA). The DA sends this blue blood to the organs in the lower half of the fetal body. Then this blue blood leaves the fetus through the umbilical arteries to go back to the placenta to pick up oxygen.

Cardiac Anatomy and Circulation

Deoxygenated blood returning through the superior and inferior vena cava goes to the right atrium (RA), passes through the tricuspid valve into the right ventricle (RV) where it is pumped to the lungs through the pulmonary valve, then into the main and branch pulmonary arteries to the capillary level so that carbon dioxide can be dropped off and oxygen picked up through the process of diffusion from the alveoli. Then the oxygenated blood enters via the 4 pulmonary veins to the left atrium (LA), passes through the mitral valve into the left ventricle (LV) where it is pumped to the body thru aortic valve to the aorta. The aorta forks and the blood is divided between major arteries which supply the upper and lower body. The blood travels in the arteries to the arterioles and then to the tiny capillaries that feed each cell. The deoxygenated blood then travels to the venous circulation returning to the heart through the inferior and superior vena cavae and finally back to the RA where the process began.
Outflow Tracts

- RVOT: the infundibular extension of the RV which connects to the pulmonary artery.
- RVOTO may be due to a defect in the pulmonary valve, the supravalvar region, the infundibulum, or the pulmonary artery.
- LVOT: the infundibular extension of the LV which connects to the aorta.
- LVOTO may be due to a defect of the aortic valve, the supravalvular region, the infundibulum, or the aorta.

Review of Cardiovascular Assessment

- Color
- Respiratory Rate and Effort
- Heart Rate and Rhythm
- Blood Pressure
- Pulses
- Capillary Refill
- PMI
- Murmurs
- Liver edge
- O2 sets
- CXR
- ABG
- ECG
- ECHO
- Cath

(next... why do congenital heart defects happen?)
Why do Congenital Heart Defects Occur?

- Most defects are random, occurring during fetal development of the heart at 3-8 weeks gestation
- Risk Factors
  - Maternal factors
    - Chronic illnesses such as diabetes (d-TGA), advanced maternal age (increased risk for birth defects in general), medications (such as anti-seizure meds, accutane, alcohol intake (ASD is associated with FAS)
  - Positive family history (such as PS and AS)
  - Chromosomal abnormalities (next slide)

Chromosomal Abnormalities

- Usual and Usually Obvious:
  - Trisomy 13, 18 (VSD, PDA, ASD)
  - Trisomy 21 (CAVC, ASD, VSD, PDA)
  - Turner's Syndrome, Noonan Syndrome (aortic stenosis, coarctation of the aorta)
  - DiGeorge Syndrome: 22q11 deletion
  - Congenital heart defects—in up to 74%, usually conotruncal, involving the outflow tracts of the great vessels
    - Truncus Arteriosus
    - Tetralogy of Fallot
    - Interrupted aortic arch
    - VSD
    - Right Arch

Pulsox Screening

In September 2011, U.S. Dept of Health and Human services approved adding Critical Congenital Heart Disease (CCHD) to the Recommended Uniform Screening Panel. In the United States, about 4,800 (~12/10,000) babies born every year have CCHDs. Babies with a CCHD are at significant risk for death or disability if their CCHD is not diagnosed and treated soon after birth. Pulseoximetry is the recommended screening method to detect CCHDs in newborns. There are seven defects classified as CCHD: Hypoplastic left heart syndrome, Pulmonary atresia (with intact septum), Tetralogy of Fallot, Total anomalous pulmonary venous return, Transposition of the great arteries, Tricuspid atresia, Truncus arteriosus
Classification of Congenital Heart Defects

Other Ways of Categorizing Lesions

- Ductal dependent lesions
- Lesions needing adequate atrial level shunting
- Lesion with parallel circulation
- Lesions that give rise to pulmonary edema

(next....had to put in principles of oxygen delivery!)
Review of Principles of Oxygen Delivery

Oxygen delivery = systemic cardiac output x arterial oxygen content

Arterial oxygen content = (Hgb x \(SaO_2\) x 1.34) + 0.003(PaO2)

O2 utilization is CO x (arterial 02 content - mixed venous 02 content)

Therefore, oxygen delivery is primarily dependent on:
- Systemic cardiac output
- Arterial oxygen content
- Hgb concentration
- \(O_2\) saturation.

Defects with Increased Pulmonary Blood Flow (acyanotic defects)

Atrial Septal Defect
Ventricular Septal Defect
Patent Ductus Arteriosus
AV Canal

Atrial Septal Defect (ASD)
- Abnormal opening between the atria
- Increased flow of oxygenated blood into the right side of the heart leading to right atrial and ventricular enlargement
- Can have desats with right-to-left shunt when PVR is high
- CHF is unusual in uncomplicated ASD (atrial pressures too low)
- Pulmonary vascular changes occur after several decades if unrepaired
- Usually asymptomatic—may have murmur when older
- The risk of small ASD or PFO is in older age with the risk of clots in heart (like from atrial fibrillation) that pass from the right to the left side of the heart causing a stroke
- Treatment
  - Surgical repair usually before school age with patch closure of moderate-to-large defects
  - Non surgical: ASD device closure during cardiac catheterization
Ventricular Septal Defect (VSD)

- Abnormal opening between the right and left ventricles
- Most common CHD
- Two types of tissue make up the ventricular septum. Muscular and membranous:
  - Around 80% of defects are perimembranous which spontaneously close in 50-75% of cases.
  - Muscular ventricular septal defects have a high rate of spontaneous closure - up to 90% close by 18 months of age.
- Blood flows through the defect into the pulmonary artery causing increased blood volume pumped into lungs (left to right shunting)
- CHF is common after PVR falls (at 2 weeks to 2 months of life), depending on size of lesion.
- Then avoid supplemental oxygen which can increase left-to-right shunting and pulmonary overcirculation; NC flow is ok and good!
- Treatment: early palliative treatment (pulmonary artery banding), complete repair at 4-9 mo., or occlusion during cardiac catheterization
- Can lead to fixed pulmonary hypertension if untreated
Patent Ductus Arteriosus (PDA)

- FT: 1:2000
- 80% in infants <1000
- 45% in infants <1750
- Becomes a problem when PVR falls and a left-to-right shunt occurs, causing pulmonary overcirculation
- May be asymptomatic, acidotic, or show signs of CHF.
- May significantly impact preterm neonate.
- Characteristic machinery-like murmur in older infants, may not have murmur in neonate.
- Wide pulse pressure, bounding pulses.

- (next .... PDA in color)
Clinically significant lesions may need to be closed. These include:

- Large lesions in relation to size of baby (is it bigger than the aorta?)
- ECHO showing LA enlargement
- Acidosis
- Inability to wean from respiratory support
- In the preterm baby with little chance of spontaneous closure

1. Treatment: indomethacin (prostaglandin inhibitor) unless contraindicated: Creatinine > 1.8, Plt count < 80, bleeding tendency (such as IVH), NEC
2. Surgical ligation via left thoracotomy
3. Device closure with a coil to occlude the PDA during cardiac catheterization (usually after age of 1 yr)

Atrioventricular Canal Defect (AVC)

- Associated with Down Syndrome—almost 40% of patients with AV Canal have Trisomy 21
- Low ASD that is continuous with a high VSD with cleft of the mitral and tricuspid valves, creating a large central AV valve that allows blood to flow between all 4 chambers
- Unbalanced AV canal is where one ventricle is hypoplastic
- Causes left-to-right shunting when and IF PVR falls; pulmonary blood flow increases causing pulmonary congestion, cardiomegaly, tachypnea, poor feeding; in some, PHTN continues (sometimes until surgery)
- Usually has murmur after PVR falls, mild cyanosis when crying
- Avoid supplemental O2 unless treating PHTN (keep sats > 85%)
- Treatment: anti-congestive meds, pulmonary artery banding; surgical repair at 3-6 mo of life—patch closure of the septal defects and reconstruction of the AV valves; may need valve reconstruction or replacement later on in life
Coarctation of the Aorta

- An obstructive defect, but is acyanotic & can cause CHF
- Usually narrowed near the isthmus (place where ductus inserts) resulting in increased pressure pre-ductally and decreased BP post-ductally; can also have a narrowed aortic arch
- High BP and bounding pulses in the arms and cool lower extremities with weak or absent pulses and lowered BP. Signs of CHF with rapid hemodynamic deterioration.
- May be ductal dependent if severe or critical
- Need to monitor pulses, perfusion, BP, U.O, ABG for acidosis
- If ductal dependent, avoid supplemental oxygen that may cause systemic steal
- Treatment: surgical resection of the coarcted portion with anastomosis OR enlargement of the constricted section by balloon angiocath in cardiac cath lab (not done in the neonatal period)
Defects of Decreased Pulmonary Blood Flow

Tetralogy of Fallot
Tricuspid Atresia
Pulmonary Atresia/Stenosis

Tetralogy of Fallot (TOF)
• The classic form includes four defects: (1) ventricular septal defect, (2) pulmonic stenosis, (3) overriding aorta, and (4) right ventricular hypertrophy
• All have some amount of infundibular stenosis, which may cause "TET" spells
• Infants may be acutely cyanotic at birth, may have no cyanosis, or have mild cyanosis that progresses over the first year.
• O2 is ok. TOF's rarely overcirculate. Keep sats > 85%
• Associated with DiGeorge Syndrome
• Surgical treatment: early palliative shunt if significant PS or sub PS or complete repair at 6-12 months
TET Spells

- Hypercyanotic spells due to infundibular stenosis (narrowing below the level of the pulmonary valve)
- Prevention: avoid dehydration, undue stress, illness
- Treatment: O2, IV fluids, morphine, knee chest position, beta blockers (then....repair!)

Pulmonic Stenosis (PS)

- Narrowing at the entrance to the pulmonary artery varies from mild to critical to atretic
- Resistance to blood flow causes right ventricular hypertrophy and decreased pulmonary blood flow; may have small PA’s
- May be asymptomatic or have cyanosis. Supplemental oxygen as needed to keep sats > 85%
- Characteristic harsh murmur at LMSB
- May be ductal dependent if very stenotic or atretic
- Treatment: balloon angioplasty in the cardiac cath lab to dilate the valve (usually balloon, stop prostins, assess pulmonary blood flow by sats and ECHO), BTS, surgical valvotomy procedure (to open up valve)
Mixed Defects

D-TGA
Truncus Arteriosus
HLHS

Transposition of the Great Arteries (TGA)

- The pulmonary artery leaves the left ventricle and the aorta exits from the right ventricle, with no communication between the systemic and pulmonary circulation (2 parallel circuits)
- Need mixing of saturated and desaturated blood at ASD level (start PGE1 and perform cath if needed with balloon atrial septostomy-BAS)
- Severely cyanotic and depressed at birth. Can be infant of diabetic mother. More often term, male infant.
- After BAS, can turn off PGE and monitor sats; keep > 70%; can use 02 as needed; may need to turn PGE back on
- Surgical treatment = arterial switch procedure at 7-10 days depending on coronary anatomy
Truncus Arteriosus (TA)

- Failure of normal septation and division of the embryonic bulbar trunk into the pulmonary artery and the aorta (there are several types)
- Blood from both ventricles mixes in the common great artery (trunk), causing desaturation and hypoxemia
- Keep sats > 75%; PVR falls and CHF occurs within 1-2 weeks with variable cyanosis, poor growth, and activity intolerance (no supplemental oxygen then).
- Associated w/ DiGeorge Syndrome
- Repair is in the first 1-2 months, depending on size of infant
Repair of Truncus Arteriosus

TA – truncus arteriosus, VSD – ventricular septal defect
Right (upper arrow) – light blue valved conduit

Hypoplastic Left Heart Syndrome
• Underdevelopment of the left ventricle usually with mitral atresia, aortic atresia, and small aorta, possible coarctation
• Saturated blood returns via the pulmonary veins to the left atrium, crosses to the right atrium via the ASD, mixes with desaturated blood returning from vena cava, goes to RV, leaves the heart via the PA, then crosses to the aorta via the ductus providing blood flow to the body
• Presents with mild cyanosis, tachypnea, acidosis, decreased CO, shock; fortunately, about 90% are prenatally diagnosed
• Ductal dependent lesion!—(symptoms occur when ductus closes)
Management of HLHS

- Immediate therapy for infants with HLHS is initiation of IV infusion of prostaglandin to manipulate the ductus arteriosus and maintain ductal patency. (usually start at 0.02 mcg/kg/min)
- O2 sats of 75% to 85% suggest an adequate balance between systemic and pulmonary blood flow
- Blood flow to the pulmonary and systemic circulations should be nearly balanced (goal Qp:Qs ratio of 1—but rarely manage to achieve this). Need to keep PVR high to prevent systemic steal
- ABG goal: 7.4 40-60 40 -0; lower CO2 can decrease PVR;

HLHS Management Continued:
- Aim for H&H 14-15/40-45% -- higher Hgb has more oxygen carrying capacity and higher Hct increases blood viscosity which increases PVR
- Close monitoring of urine output, upper and lower extremity BP, LE perfusion
- Inotropic support as necessary only after: Adequate fluid volume!!
- Use of subatmospheric oxygen as needed (follow head NIRS!!) some centers manage balance with hypercarbia (pipe in CO2 gas into ventilator circuit)
- Intubation/ventilation/sedation only if can’t support any other way!

Palliative Surgeries

- Norwood at 1-2 weeks
- Bidirectional Glenn at 6 months
- Fontan at 2-4 years
- High mortality
Stage I Norwood

- Provides reliable blood flow to lungs with BT shunt
- Atrial septectomy
- Creation of neoaorta
- Allows time for physiologic drop in PVR until next surgery

(next.....the Glenn!)

Bidirectional Glenn Shunt

- Blood from head/neck/arms returns directly to lungs
- IVC blood still mixes in RA-> thus mixed blood still goes out to body; usually ~88-90%
- LA->RA flow across ASD
- Need low (normal) PVR for this to work or blood backs up in the SVC causing congestion and poor cardiac output

Fontan completion

- All SVC and IVC blood goes directly to the lungs
- Only oxygenated blood goes to the body -> sats are normal
- All blood in the heart goes to the body

(next.....the hybrid!)
Who Gets a Hybrid?

- 'Low risk' babies get Norwoods
- 'High Risk' babies get Hybrids:
  - Low birth weight, pre-term
  - Pulmonary disease
  - Other significant co-morbid conditions (where bypass would not be well-tolerated)
  - History of Intact atrial septum
- What is a Hybrid?
  - It is a combined OR and Cardiac Catheterization procedure (like a crossbred pig or corn plant) (I'm from Iowa)
  - (next, the procedure)

Hybrid Procedure

- Stage I:
- Bilateral PA bands
- PDA stent
- Atrial septostomy

(rem, stage 2)

Stage 2: Glenwood

At the usual time a Glenn would be done, a hybrid patient will undergo the extensive Norwood type of arch reconstruction, atrial septectomy, as well as conversion to the Glenn shunt. The PDA is removed.

(rem, Fontan)
Advantages and Disadvantages to the Hybrid

Advantages:
- No need for cardiopulmonary bypass (although a sternotomy is still performed for band and stent placement)
  - Stent placed in cath lab through the PA, after chest opened, prior to band placement
  - Bands placed on both branch PA's (can tailor size to each PA)
- Can be done with lower weight babies, higher risk babies to get them home

Disadvantages:
- Can be tricky to size the PA bands correctly
- Ductal stent may accidentally occlude the distal aorta, thereby preventing flow from reaching the head/neck/arms/heart (!)
- Can be done in infants that may not be candidates for a Glenn or Fontan down the road (then what?)

How Does the Hybrid change the Game?
- Allows sicker, more complex kids to survive the initial stage, like kids with significant underlying disease (pulmonary, genetic, prematurity)
- Can add complexity to respiratory management, such as in the case of a preemie HLHS patient who may get the ductal stent but require prolonged management with sub-atmospheric O2 to enable growth prior to getting PA bands (you can't put the bands on too early or the baby will get blue too fast with growth!)
- May NOT change the ultimate outcome, as significant lung disease will prevent some of these patients from becoming Glenn or Fontan candidates
- BUT, it may allow some of these kids to live longer and perhaps become a transplant candidate, even if further surgical palliation options are not present.
Diagnostic/Management Summary

- Anticipate needs of the fetally diagnosed CHD infant—at delivery and beyond
- Provide timely and appropriate initial resuscitation and stabilization
- Identify the defect thorough non-invasive (ECHO or CT Angio) means (ask!)
- Provide appropriate respiratory management (know the goals!)
- Appropriate monitoring (sats, gasses, NIRS, fluids, nutrition, perfusion)
- Participate in evaluation and treatment of secondary organ dysfunction, particularly the brain, GI tract, kidneys and liver (they all work together!)
- Individualized POC for co-morbid infants (such as 26 weeks with CAVC)
- Assist with Cardiac Catheterization as needed:
  - Interventional procedure - balloon atrial septostomy or balloon dilation of a valve
  - Diagnostic cath - to define anatomy (cardiologist, PA, RN, NAPCA)
- Know whether the baby needs emergency surgery, surgery before discharge, or surgery in a few months (know why!)
- Remember how important each member of the team is to the future of this baby and family
- Thank you!

The End