Moving Forward with Prenatal Congenital Heart Disease

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Epidemiology

• Most common birth defect 1% of births per year (~40,000 neonates/yr)

• 57% of infantile mortality as a result of CHD


### Categories of CHD

<table>
<thead>
<tr>
<th><strong>Mild CHD</strong></th>
<th><strong>Moderate CHD</strong></th>
</tr>
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<tbody>
<tr>
<td>Muscular VSD</td>
<td>Mild aortic stenosis (AS)</td>
</tr>
<tr>
<td>Small PDA</td>
<td>Moderate pulmonary stenosis (PS)</td>
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<tr>
<td>ASD</td>
<td>Non-critical coarctation of the aorta</td>
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<tr>
<td>Mild pulmonary stenosis</td>
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<td>Bicuspid aortic valve</td>
<td>Membranous VSD</td>
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Categories of CHD

**Severe CHD**

d-Transposition of the Great Arteries
Tricuspid and Pulmonary Atresia
Hypoplastic Left Heart Syndrome (HLHS)
Single Ventricle Anatomy
Double Outlet Right Ventricle (DORV)
Categories of CHD

**Severe CHD**

- Truncus Arteriosus
- Total Anomalous Pulmonary Venous Return
- Critical Pulmonary or Aortic Valve Stenosis
- Aortic Arch Abnormalities (IAA, COA)
Optimal Timing of Diagnosis
Sub-Optimal Timing of Diagnosis
Unrecognized CCHD

Morbidity & mortality:

- Metabolic acidosis
- Intracranial hemorrhage
- Hypoxic-ischemic encephalopathy
- Necrotizing enterocolitis
- Cardiac arrest
- Death

Impact on Surgery

HLHS postnatal diagnosis = less stable prior to surgery

Prenatal dx = better surgical survival

Neurocognitive effects

Pre vs. postnatal diagnosis of dTGA

- Remedial school services 45% pre vs 69% post
- Postnatal show poorer executive function
- Deficits more prevalent & severe with postnatal dx
  - Cognitive flexibility
  - Social cognition

Delivery Location

• Neonates with HLHS:
  • Birth <10 minutes from tertiary center = 21% mortality
  • Birth >90 minutes away = 40% mortality

Delivery Location

- Delivery in cardiac OR
- Immediate transfer to cath lab
- Emergent cardiac stent
Parental Benefits

Family preparation

• Emotional
• Financial
• Social
Multidisciplinary Teams

Cardiology

Child Life/Chaplains

Social Work/PACT

MFM/OB

Neonatology

Genetics

CV surgery
Parental Education

“Core curriculum” for families
58 point checklist
Individualized counseling

Fetal Cardiology Program Checklist

- Cardiac anatomy and physiology
  - Normal cardiac anatomy/physiology
  - Fetal cardiac anatomy/physiology
  - Specific fetal cardiac diagnosis with anatomy/physiology
  - Any diagnostic uncertainty
  - Cardiac disease etiology
  - Fetal echocardiography limitations

- Immediate Postnatal management
  - Immediate postnatal stabilization and management
  - Unbilical lines and possible intubation
  - Prostaglandins
  - Expected immediate postnatal cath/surgery interventions

- Neonatal Management
  - Feeding issues (NPO, breastfeeding)
  - NICU parent/visitor policies

- Surgical Planning
  - Details of surgery/cath intervention expected
  - Timing of surgery/intervention
  - Possible complications of surgery/cath
  - Venulator Support
  - Laparoscopic needs
  - Drain
  - Open Chest
  - Possible Blood Transfusions
  - ECMO

- Post Surgical Management
  - Length of hospital stay
  - Other possible medical issues (need for NGT, g tube, malrotation)
  - Likely discharge medications
  - Other anticipated monitors, home nursing/home monitoring

- Longer term Issues
  - Subsequent surgeries/interventions expected
  - Possible Transplant
  - Projected life expectancy

- Extracardiac Issues
  - Other diagnosed extracardiac fetal abnormalities, interaction with CHD
  - Undiagnosed known associated extracardiac anomalies
  - Known genetic associations

- Pregnancy Counseling
  - Location of delivery
  - Timing and mode of delivery
  - Expectations for the rest of gestation
  - Risk for hydrops
  - Possible intrauterine demise
  - Outcome of delivering preterm
  - Hybrid

- Family Issues
  - Acute and long term housing needs
  - Financial effect of child with significant medical needs
  - Effect of special-needs child on family
  - Genetic implications for future family planning
  - Introduction of community family support groups
  - Connection of family to other family with similar CHD

- Consults
  - PACCT
  - Genetics
  - CV Surgeon
  - Social Work
  - Surgery

- Other System Issues
  - ENT/joint/heart dysfunction
  - Endocrinology/obesity/bleeding
  - Neurodevelopmental
Parental Stress

+ time + support + education =
National Prenatal Detection

(Donofrio 2014; Levy et al 2013)
Prenatal Detection Process

- Abnormal OB Screening Ultrasound
- MFM/Perinatal
- Fetal Cardiology

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Fetal Anomaly Screening

Normal

Not Normal
Fetal Anomaly Screening

CHD detection rates vary widely

- 55-65% with four-chamber view alone
- 80-84% with outflow tract view in addition
Barriers to Prenatal Detection

Retrospective cohort study in Utah

- Utah Birth Defect Registry, 1997-2007
- Mandatory prenatal screening

Objectives:

- Determine rate of prenatal detection of CHD
- Identify maternal & encounter risk factors for failed prenatal detection

Results:

- 97% of mothers had ≥ 1 prenatal US
- 39% of CHD was detected prenatally

Barriers to Prenatal Detection

- Abnormal ultrasound:
  - 89% seen by MFM
  - ...but 42% never had fetal echo
- 35% of mothers with FamHx did not receive fetal echo
- Of those with fetal echo, 3% had a missed CHD Dx

National Prenatal Detection

Percent Detection

- Bull 1999 (n=4799)
- Game 2001 (n=2454)
- Tegnander 2006
- Acherman 2007
- Pinto 2007
- Khoo 2008 (n=1474)
- Acharya 2008 (n=200)
- Friedberg 2009 (n=39)
- Marek 2011 (n=1604)
- Levy 2013 (n=93)

74%
Standardized Fetal Echo Screening

- Educational program for sonographers
- Rotate sonographers with Pediatric Cardiologists
- Include 4 chamber view and both outflow tracts on routine screening U/S
- Video Clips (not still-frames)

(Donofrio 2014; Levy et al (2013))
Indications for Fetal Echo

### Table 3. Factors Associated With Increased Risk of CBD in the Fetus

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<th>Timing/Frequency of Evaluation</th>
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<tbody>
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<td>Maternal factors</td>
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<td>Preeclampsia</td>
<td>1-3%</td>
<td>10-12 wk</td>
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Reasons for Referral

- Concern for CHD
- Maternal Risk Factors
  - Maternal disease
  - IVF
  - Family history
- Fetal Risk Factors
  - Genetic abnormality
  - Extracardiac anomaly
  - Multiple gestations
How do we identify neonatal CHD?
Child in well-baby nursery ≥ 24 hours of age or shortly before discharge if < 24 hours of age

Screen

- < 90% in right hand or foot
- 90%–<95% in right hand and foot or >3% difference between right hand and foot
- ≥ 95% in right hand or foot and <3% difference between right hand and foot

Repeat screen in 1 hour

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Positive Screen

Negative Screen
Summary

• 1% of neonates have CHD
• Prenatal diagnosis can save lives
• Many other benefits to prenatal diagnosis
Summary

• Prenatal diagnosis is challenging
• Team effort
• Currently 50% of our regional children receive prenatal dx…

…but we can do better!
Plaza Art Fair tonight
Thank you!
References


References

Ainsworth, S. "Prevalence and clinical significance of cardiac murmurs in neonates." Arch Dis Child Fetal ed. 1999;80:F43-F45

