Screening for Critical Congenital Heart Disease in the Apparently Healthy Newborn

A presentation of Texas Pulse Oximetry Project:
A Joint Educational Initiative of The University of Texas Health Science Center at San Antonio/Department of Pediatrics, Baylor College of Medicine/Department of Pediatrics and Texas Department of State Health Services

Disclosure

Alice K. Gong, M.D. has no relationships with commercial companies to disclose.

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Objectives

- Explain the rationale for screening for Critical Congenital Heart Disease (CCHD) in newborns
- Examine the evidence supporting the routine use of pulse oximetry in the Newborn Nursery to detect CCHD
- Discuss evidence-based recommendations for implementation of CCHD screening

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Congenital Heart Disease

- Incidence: 8-9/1000 births
- 2/1000 potentially lethal - “critical”
  - Requiring expert cardiac care and intervention in the immediate NB period or early infancy.
- In the US, about 4800 babies are born each year with CRITICAL CHD
- Leading cause of death in infants < 1 year old

Congenital Heart Disease

- Advances in surgical and interventional cardiology has improved survival over the past 30 years.
  - There are an estimated 800,000 adults living with CHD.
  - Survivors who present late are at greater risk for neurologic injury and subsequent development delay.
  - Focus now has shifted from increasing survival to reducing morbidity.
Critical Congenital Heart Disease

- Those CHD's that will require cardiac intervention in the newborn period or within the first year of life.
  - Ductal dependent systemic circulation
    - HLHS, Coarctation, IAA, Critical AS
  - Ductal dependent pulmonary circulation
    - PA, PS and variants, TOF
  - Complex critical CHD
    - TGA, Truncus Arteriosus, TAPVR, Single ventricle

Critical Congenital Heart Disease

- Physiologic changes may occur after hospital discharge corresponding to changes in the pulmonary vascular resistance and closure of the patent ductus arteriosus.
- Present in extremis with low cardiac output and acidosis, multi-organ failure, hypoxic ischemic brain injury.
- Early detection and timely intervention can thus decrease morbidity and lead to better outcomes.

Why screening?

- Incidence is sufficient in the population
- Therapy provided before onset of clinical manifestations results in an improved outcome
- Screening identifies disease before symptoms
- Test has acceptable sensitivity and false positive rates
- Cost effective

Wilson and Junger WHO 1968 Public Health Paper
Diagnosis vs. Screening

<table>
<thead>
<tr>
<th></th>
<th>Diagnostic</th>
<th>Screening</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pros</td>
<td>Fewer resources needed</td>
<td>Higher detection rate</td>
</tr>
<tr>
<td>Cons</td>
<td>Identification may be too late</td>
<td>High resource use</td>
</tr>
<tr>
<td></td>
<td>Application may be spotty</td>
<td>Adverse impact of false positives</td>
</tr>
</tbody>
</table>

CCHD detection – diagnostic

- Fetal echocardiography
  - >50% detection rates for single ventricle lesions
  - <30% for 2-ventricle
    - Highly variable, limited access
- Newborn physical exam
  (in nursery and in clinic)
  - 4-5 grams of deoxygenated Hgb is needed to detect cyanosis
  - Most CCHD have mild desaturation to 80-95%
  - Harder in darker skinned babies

Diagnostic Process

Newborn presents in shock with murmur

Exam suggestive of CHD

Hypoplastic Left Heart
Perspective on Importance: Timing of Diagnosis of CCHD

Age of initial diagnosis among term infants with CCHD, Arkansas, 2000 - 2008

Missed Diagnosis

- Some babies can appear healthy at first
  - Some have no murmurs or cyanosis

- PE alone failed to identify 50% of CHD’s that were not detected by prenatal U/S
  - Failure to diagnose CCHD may lead to critical events, cardiogenic shock or death

- Estimated 30% of infant deaths from CCHD occur before diagnosis

Chain of Detection
**Missed Diagnosis of CCHD**

Table 2. Total Number of Patients in Each Group by Diagnosis

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Group 1</th>
<th>Group 2</th>
<th>Group 3</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypoplastic left heart syndrome</td>
<td>368 (22.1)</td>
<td>36 (21.1)</td>
<td>36 (21.1)</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>368 (22.1)</td>
<td>36 (21.1)</td>
<td>36 (21.1)</td>
</tr>
<tr>
<td>Pulmonary atresia</td>
<td>368 (22.1)</td>
<td>36 (21.1)</td>
<td>36 (21.1)</td>
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<tr>
<td>Transposition of great vessels</td>
<td>368 (22.1)</td>
<td>36 (21.1)</td>
<td>36 (21.1)</td>
</tr>
<tr>
<td>TGA</td>
<td>368 (22.1)</td>
<td>36 (21.1)</td>
<td>36 (21.1)</td>
</tr>
<tr>
<td>Heterotaxy with hypoplastic left heart syndrome</td>
<td>368 (22.1)</td>
<td>36 (21.1)</td>
<td>36 (21.1)</td>
</tr>
<tr>
<td>Transposition of great vessels</td>
<td>368 (22.1)</td>
<td>36 (21.1)</td>
<td>36 (21.1)</td>
</tr>
<tr>
<td>Heterotaxy with pulmonary atresia</td>
<td>368 (22.1)</td>
<td>36 (21.1)</td>
<td>36 (21.1)</td>
</tr>
<tr>
<td>Median age</td>
<td>13.5 days</td>
<td>13.5 days</td>
<td>13.5 days</td>
</tr>
</tbody>
</table>

*Abbreviations: BVR: branch vein right ventricle, TAPVR, total anomalous pulmonary venous return.*

*Excludes of reading, percentages may not total 100.*

*Small number of patients enrolled to the study selection criteria specified in the “Patient Selection” section of the “Methods” section.

Chang et al, Arch Pediatr Adolesc Med, 2018

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**CCHD Screening**

- **Pulse Oximetry**
  - Indirectly monitors the oxygen saturation of a patient’s blood and changes in blood flow in the skin
  - Can detect mild hypoxemia without obvious cyanosis
  - Can provide continuous and immediate values
  - Non-invasive
  - Easy to use and widely available
  - Cost-effective and widely used

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**Pulse Oximetry Screening - Evidence**

- Using a cut-off of 95% in the LE, Hoke et al identified 81% of infants with CCHD
- Many investigators have since investigated the use of pulse oximetry as a screening tool in newborns NOT known to have CCHD
  - Most studies were small, with different protocols and cut-offs, at low altitude
  - Low false positive rate < 1%, sensitivity <80%
    - Likely because hypoxemia is not present in all CCHD

Hoke et al, Oxygen saturation as a screening test for critical CHD. Ped Cardiol. 2002; 23:203-409

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Pulse Oximetry Screening Program Saxony, Germany


Pulse Oximetry Screening - Evidence

- 2 separate large prospective screening of 40,000 newborns in Sweden and nearly 40,000 in Germany.
- Sensitivity 62%, Specificity 99.8%
- A meta-analysis of pulse ox screening for CCHD in asymptomatic newborns
  - Over 220,000 NB's
  - Overall sensitivity was 76.5%, specificity was 99.9% with a false positive rate of 0.14%


Cost of Routine Pulse Oximetry

- Includes both the direct cost of the pulse oximetry and the follow-up costs of any additional examinations and transfers.
  - At experienced centers, it took technicians only 2 minutes on average to perform screen.
  - Calculation of time in New Jersey 9 min per child
    - No new nursing or medical technician FTEs added
  - Cost of approximately $3-6 per asymptomatic newborn
    - Assumes reusable probe
    - Single use probes up to $30
### Current Status of Recommendations

- US Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (HHS-SACHDNC)
  - In 2010, recommended that CCHD be added to the newborn uniform screening panel
  - Identify newborn with structural heart defects associated with hypoxia that could have significant morbidity or mortality early in life with closing of the patent ductus arteriosus or other physiologic changes
  - 2011, Endorsed by Secretary of Health Kathleen Sibelius

### Texas

- HB 740 passed the Senate on 5/13/2013 and was signed into law – effective September 1, 2013.
  - Taryn Kennedy, Nash Sievers & Rex Van de Putte act. Taryn & Nash are two babies who died from CCHD and their moms worked on the legislation. Rex is the grandson of Sen. Leticia Van de Putte who passed away at 6 months of SIDS.
  - Requires all newborns of a birthing facility be screened
    - Exceptions – parental refusal, transfer prior to screening, screening had previously been completed, discharge before 10 hours and referral made.

### Barriers to implementation

- Reporting/Tracking/ QI
- Inadequate resources – few Pediatric cardiologist
- Resistance from some in the medical community
- Screener
  - Additional work load
  - Education
- Equipment
  - Probe, pulse oximeters (2 are FDA approved)
- Patient/Parent
  - False positives, false negatives
  - Delay in discharge
- Potential transfer to another center
- Costs and reimbursement
AAP/CDC Algorithm

CCHD Screening Protocol
- 7 primary targets
  - Hypoplastic Left Heart Syndrome
  - Pulmonary Atresia (with intact atrial septum)
  - Tetralogy of Fallot
  - Total Anomalous Pulmonary Venous Return
  - Transposition of the Great Arteries
  - Tricuspid Atresia
  - Truncus arteriosus
- 17-31% of all CHD's

CCHD Screening Protocol
- Secondary screening targets
  - Can be just as severe but not consistently detected
  - Aortic arch atresia/hypoplasia
  - Interrupted aortic arch
  - Coarctation
  - DORV
  - Ebstein's anomaly
  - PS, PA, AVCD
  - Other Single ventricle defects
How to Perform Screening

- Screen after 24 hours of age
- Conduct when infant is calm and awake
- Perform in preductal (RIGHT hand) and postductal (one FOOT), in parallel or one after the other
- If < 90% - positive screen, refer
- If ≥ 95% in EITHER extremity with ≤ 3% difference: PASS
- If 90 - 94% in BOTH or difference > 3%: REPEAT in 1 hour up to 2 times, then refer

How is it done?

CCHD Screening Algorithm

Pulse ox on right hand and foot after 24 hours

- > 95% in right hand (RH) or foot and ≤ 3% difference between RH and foot
  - Positive (FAIL)
  - Notify MD/NNP

- 90-94% in RH and foot
  - ≥ 3% difference between RH and foot
    - Repeat in 1 hour

- < 90% in RH or foot
  - Positive (FAIL)
  - Notify MD/NNP

Remind parents that CCHD newborn screening may not find all types of problems in a baby’s heart.
Evaluation for Positive Screen

- Clinical Assessment – transfer to NICU
- Infectious or Pulmonary pathology should be excluded
- Complete echocardiogram
- Pediatric Cardiology referral as indicated

Managing the Positive Screen

“In the absence of other findings to explain hypoxemia, CCHD needs to be excluded on the basis of a diagnostic echocardiogram (which would involve an echocardiogram within the hospital or birthing center or transport to another institution)....”
Kemper et al Pediatrics 2011

- Alternative strategies
  - Keep child until evaluation can be performed
  - Transfer to advanced nursery (without cardiac inpatient service)
  - Transfer to center with advanced cardiac care

Screening in the Real World

- Feasibility of implementing pulse oximetry screening for CHD in a community hospital
- 6745 eligible infants screened at average age 42h
  - 9 positive – 1 had CCHD
- Barriers (1.4%):
  - screening equipment 54%
  - staff 23%
  - infant 20%
  - family 4%
- Physician and Nurse “champions” important to successful implementation
TxPOP

- Texas Pulse Oximetry Project: A Joint Educational Initiative.
- Goal: Develop an appropriate implementation strategy for screening of CCHD using pulse oximetry as a potential public health mandate.
  - Develop and provide educational programs and materials
- Funding: Texas Department of State Health Services’ Children’s Outreach Heart Program

Project Timeline

Quality Improvement – Feb-July

- 12,946 births in the 13 facilities
- 11,713 newborn nursery admissions
- 11,289 CCHD newborn screens
- 96% of babies admitted to the newborn nursery received a CCHD screen during the recommended time frame (between 24 hours and discharge).
- Babies not admitted to the newborn nursery after birth (approximately 1,235)
- Transfers out of newborn nursery prior to CCHD screen (249)
- Screens performed prior to 24 hours (38)
Positive Screens – 11 – all had ECHOs
False Positive rate of 0.097%

- 2 were in the <90% group – 1 had severe CCHD from secondary target; other had subclinical seizures
- 7 were the indeterminate of 90-95% X 3
- 1 had >5% difference

- 32 babies from the 13 facilities had ECHOs
- Only 3 transfers from initial facility, all within same zipcode

TAPVR

pneumonia
Toolkits
- http://txpeds.org/txpop
- http://www.dshs.state.tx.gov

CCHD learning module
- www.txhealthsteps.com
  - 1 hr CME, CNE, CES (social workers), NCHEC (certified health education specialists).

Example 1
- A term newborn at 24 hours has the following:
  - Right hand SpO2 of 99%
  - Right leg SpO2 of 94%

Pass
Fail
Repeat
Example

- A term newborn at 24 hours has the following:
  - Right hand SpO2 of 99%
  - Right leg SpO2 of 94%
  - Pass
  - Fail
  - Repeat

Example 2

- A newborn at 36 hours has the following:
  - Right hand SpO2 of 95%
  - Left leg SpO2 of 94%
  - Pass
  - Fail
  - Repeat
References


References


References

7) Congenital heart disease (CHD) in the newborn: Presentation and screening for critical CHD. Carolyn A. Altman, MD; Wolters Kluwer Health, Official reprint from UpToDate; Literature review current through 2012
