The Right Hand and One Foot: A Critical Congenital Heart Disease (CCHD) Screening Update

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Future of Pediatrics
Bethesda, Maryland
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Congenital Heart Disease

- Most common birth defect
- 8:1,000 with CHD
  3:1,000 with CCHD
- Accounts for ~ 40% deaths from congenital anomalies
- Majority of deaths due to CHD occur in first year of life

Hoffman JACC 39:2002
Missed Diagnosis of Critical Congenital Heart Disease

- 15 year retrospective study
- 898 infants died of CCHD in infancy
  - 152 with missed diagnosis
  - 299 with late diagnosis
- >50% of CCHD deaths were attributed to late/missed diagnosis
- 30 babies died each year secondary to late diagnosis

Figure 1. Selection and identification of patients with missed and unknown critical congenital heart disease (CCHD) diagnoses. HLHS indicates hypoplastic left heart syndrome.
Why is Detecting Newborns with CCHD Difficult?

- Complex changes from fetal to normal circulation after birth
- Fetal Ultrasound
- Detection through physical examination may be < 50%

Cyanotic “Blind Spot”

Mean threshold for detection 69%!
Diagnostic Gap

- Death
- Late
- Clinical
- Prenatal

Percentage distribution chart showing the diagnostic gap.
Pulse Oximetry as a Screening Method

♥ Pulse oximetry measures oxygen saturation of hemoglobin in arterial blood
♥ Non-invasive and painless test
Normal Newborn Circulation

Passing Sat 100%
Hypoplastic Left Heart Syndrome

Failing Sat 90%
CCHD Screening Primary Targets

1. Hypoplastic Left Heart Syndrome
2. Pulmonary Atresia (with intact septum)
3. Tetralogy of Fallot
4. Total Anomalous Pulmonary Venous Return
5. Transposition of the Great Arteries
6. Tricuspid Atresia
7. Truncus Arteriosus
Secondary Target: Pneumonia
Impact of pulse oximetry screening on the detection of duct dependent congenital heart disease: a Swedish prospective screening study in 39 821 newborns

Table 2 | The performance of screening methods in the detection of duct dependent circulation in newborn infants in West Götaland (1 July 2004 to 31 March 2007)

<table>
<thead>
<tr>
<th>Performance</th>
<th>Physical examination alone (n=38374)</th>
<th>Pulse oximetry (n=38429)</th>
<th>Physical examination plus pulse oximetry (n=38429)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sensitivity (95% CI, %)</td>
<td>62.50 (35.43 to 84.80)*</td>
<td>62.07 (42.3 to 79.31)</td>
<td>82.76 (64.23 to 94.15)</td>
</tr>
<tr>
<td>Specificity (95% CI, %)</td>
<td>98.07 (97.93 to 98.21)</td>
<td>99.82 (99.77 to 99.86)</td>
<td>97.88 (97.73 to 98.03)</td>
</tr>
<tr>
<td>Positive predictive value (95% CI, %)</td>
<td>1.35 (0.65 to 2.47)</td>
<td>20.69 (12.75 to 30.71)</td>
<td>2.92 (1.88 to 4.31)</td>
</tr>
<tr>
<td>Negative predictive value (95% CI, %)</td>
<td>99.98 (99.96 to 99.99)</td>
<td>99.97 (99.95 to 99.99)</td>
<td>99.99 (99.97 to 100.00)</td>
</tr>
</tbody>
</table>

Table 3 | Pathology found in 69 babies with false positive results from pulse oximetry screening for duct dependent circulation in West Götaland (1 July 2004 to 31 March 2007)

<table>
<thead>
<tr>
<th>Pathology found</th>
<th>No (% of babies)</th>
<th>≥5 days after screening</th>
<th>≤5 after screening</th>
<th>Follow-up only</th>
<th>Surgery</th>
</tr>
</thead>
<tbody>
<tr>
<td>Other critical congenital heart disease*</td>
<td>4 (6)</td>
<td>4/4</td>
<td>0/4</td>
<td>0/4</td>
<td>4/4</td>
</tr>
<tr>
<td>Other milder congenital heart disease</td>
<td>10 (14)</td>
<td>4/10</td>
<td>1/10</td>
<td>5/10</td>
<td>4/10</td>
</tr>
<tr>
<td>Persistent pulmonary hypertension</td>
<td>6 (9)</td>
<td>3/6</td>
<td>0/6</td>
<td>3/6</td>
<td>N/A</td>
</tr>
<tr>
<td>Transitional circulation†</td>
<td>8 (12)</td>
<td>0/8</td>
<td>3/8</td>
<td>2/8</td>
<td>N/A</td>
</tr>
<tr>
<td>Infections</td>
<td>10 (14)</td>
<td>6/10</td>
<td>4/10</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Pulmonary pathology</td>
<td>7 (10)</td>
<td>5/7</td>
<td>1/7</td>
<td>1/7</td>
<td>N/A</td>
</tr>
<tr>
<td>Normal (verified from hospital charts)</td>
<td>24 (35)</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
</tbody>
</table>

*Pulmonary atresia with multiple aorto-pulmonary collaterals (n=2), tricuspid atresia with pulmonary stenosis and ventricular septal defect (n=1), total anomalous pulmonary venous return (n=1).
†Right to left shunting across foramen ovale without pulmonary hypertension.
Is Pulse Oximetry Effective in Detecting CCHD?

Pulse oximetry screening for critical congenital heart defects in asymptomatic newborn babies: a systematic review and meta-analysis

**Sensitivity (95% CI)**

<table>
<thead>
<tr>
<th>Overall estimate</th>
<th>Sensitivity (95% CI)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>76.5 (67.7-83.5)</td>
</tr>
</tbody>
</table>

**False-positive rate % (95% CI)**

<table>
<thead>
<tr>
<th></th>
<th>False-positive rate % (95% CI)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0.14 (0.06-0.33)</td>
</tr>
</tbody>
</table>

- 13 primary studies
- 229,421 infants screened

**Figure 3**: Accuracy estimates based on clinical and test characteristics of pulse oximetry in detection of critical congenital heart defects in newborn babies

**CHD**—congenital heart defects.

**Interpretation** Pulse oximetry is highly specific for detection of critical congenital heart defects with moderate sensitivity, that meets criteria for universal screening.
Research on Populations

- 42,240 infants in 34 German hospitals
- Sensitivity 78%
- Specificity 99%
- PPV 26% NPV 99%

Riede Eur J Peds 169:2010
CCHD Screening Feasibility in Community Hospitals

ORIGINAL ARTICLE

Feasibility of implementing pulse oximetry screening for congenital heart disease in a community hospital

EA Bradshaw¹, S Cuzzi¹,²,³, SC Kiernan², N Nagel², JA Becker¹,² and GR Martin¹,³
¹Children’s National Medical Center, Washington, DC, USA; ²Holy Cross Hospital, Silver Spring, MD, USA and ³The George Washington University School of Medicine, Washington, DC, USA

6,880 Newborns

6,841 Eligible
19 Ineligible

6,745 Screened
96 Not Screened

6,736 Passed
9 Referred

- Avg. Pox Sat
Rt Hand/Foot
100% (90-100%)

Difference
0.2% (0-6%)

-CCHD screening did not lead to a significant increase in echos
United States Efforts

Strategies for Implementing Screening for Critical Congenital Heart Disease

Measurement #1
Pulse Ox on Right Hand (RH) and One Foot After 24 Hours of Age

FAIL
Pulse ox of 89% or less in either the RH or foot
Action: Do Not Repeat for Screening, Refer for Immediate Assessment

RETEST
Pulse ox of 90-94% in BOTH the RH and foot OR a difference of 4% or more between the RH and foot
Action: Repeat pulse ox measurements in 1 hour

PASS
Pulse ox of 95% or more in RH or foot AND difference of 3% or less between the two
Action: Do Not Repeat for Screening, Provide Normal Newborn Care

Measurement #2
Pulse Ox on Right Hand (RH) and One Foot 1 Hr After Measurement #1

FAIL
Pulse ox of 89% or less in either the RH or foot
Action: Do Not Repeat for Screening, Refer for Immediate Assessment

RETEST
Pulse ox of 90-94% in BOTH the RH and foot OR a difference of 4% or more between the RH and foot
Action: Repeat pulse ox measurements in 1 hour

PASS
Pulse ox of 95% or more in RH or foot AND difference of 3% or less between the two
Action: Do Not Repeat for Screening, Provide Normal Newborn Care

Measurement #3
Pulse Ox on Right Hand (RH) and One Foot 1 Hr After Measurement #2

FAIL
Pulse ox of 89% or less in either the RH or foot
Action: Do Not Repeat for Screening, Refer for Immediate Assessment

RETEST
Pulse ox of 90-94% in BOTH the RH and foot OR a difference of 4% or more between the RH and foot
Action: Do Not Repeat, Clinical Assessment

PASS
Pulse ox of 95% or more in RH or foot AND difference of 3% or less between the two
Action: Do Not Repeat for Screening, Provide Normal Newborn Care

Children’s National Heart Institute
Part of the Children’s National Health System
Screening in Your Unit

**REMINDER ALGORITHM FOR SCREENERS**

- **Confirm** that the infant is at least 24 hours of age and eligible for screening.
- **Help** the parent to warm and calm the infant in a quiet and peaceful environment.
- **Describe** the pulse ox test to the parent.
- **Select** a site on the right hand and one foot that is clean and dry.
- **Place** the pulse ox sensor and perform the pulse ox test.
Management of Failed CCHD Screen

Infants in Well Baby Nursery

Symptomatic
- SpO2 < 90%
  - Immediate referral for cardiac eval and transfer to NICU

Asymptomatic
- Murmur or SpO2 90 – 94% or absolute difference of >3%
  - Timely referral for cardiac evaluation

If CCHD is identified:
- Initiate appropriate therapy and, if necessary, arrange transfer.

If CCHD is not identified, initiate sepsis/respiratory evaluation

Cardiac Evaluation:
- Physical Examination
- Pulse Ox
- Perfusion Check
  - BP and Pulses x 4 extremities
- ABG
- ECHO
- EKG

Sepsis and Respiratory Evaluation:
- Physical Examination
- Rectal Temperature
- Blood Culture
- CBC with Differential
- Chest X-Ray
- C-Reactive Protein
- Blood Glucose
- Lumbar Puncture

If CCHD is not identified, initiate sepsis/respiratory evaluation

If CCHD is identified, initiate sepsis/respiratory evaluation
CCHD Screening Using Pulse Oximetry: National and Global Implementation
United States Efforts

September 21, 2011

R. Rodney Howell, M.D.
Committee Chairperson
Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children
5600 Fishers Lane, Room 38A19
Rockville, MD 20857

Dear Dr. Howell:

As indicated in my letter to you on April 26, 2011, I determined that the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children’s (SACHDNC) recommendations pertaining to the addition of Critical Congenital Heart Disease (CCHD) screening to the Recommended Uniform Screening Panel (RUSP) were not yet ready for adoption. Consequently, I referred the SACHDNC’s recommendations to the Interagency Coordinating Committee on Screening in Newborns and Children (ICC) for additional review and input regarding implementation. I asked the ICC to review the evidence gaps described by the SACHDNC and propose a plan of action to address: identification of effective screening technologies, development of diagnostic processes and protocols, education of providers and the public, and strengthening service infrastructure needs for follow-up and surveillance. I have received and reviewed the updated ICC Plans of Action.

As you know, congenital heart disease causes up to 32% of all infant deaths in the first year of life. Heart defects affect about 7 in 1,000 live births, one quarter of which could be detected and potentially treated by measuring blood oxygen saturation. Given this reality and the available information on the effectiveness of screening, I have decided to adopt the SACHDNC’s first recommendation to add CCHD to the RUSP. In addition, I am requesting that the SACHDNC collaborate with the Health Resources and Services Administration (HRSA) to complete a thorough evaluation of the potential public health impact of universal screening for CCHD, as required by the authorizing statute, section 1111 of the Public Health Service Act (42 U.S.C. § 300b-11(a)(1)).
United States Efforts

- 2011: Indiana and Maryland first states to pass CCHD screening legislation. New Jersey first state to implement universal CCHD screening.

- 20 states enacted legislation in the first half of 2013 (rolling implementation dates during 2014 – over 80% of births screened by end of 2014)

- 36 states total with legislation pending in many more

Maryland Bill Signing May 19, 2011
Working Towards a Mandate for the District of Columbia

- Recommendation filed by the Mayor’s Advisory Committee, October 2013
- Support letters from all 7 hospitals, a parent advocate, March of Dimes and Children’s National
- Follow up with Department of Health through rulemaking process
United States Efforts 2014
European Efforts: Strategizing for a Uniform Recommendation

Pulse oximetry screening for congenital heart defects

*Andrew K Ewer, Anne De-Wahl Granelli, Paolo Manzoni, Manuel Sánchez Luna, Gerard R Martin
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• Germany
• Italy
• Netherlands
• Spain
• Sweden
• UK
Right is Right.

congenital heart disease is the #1 killer of infants with birth defects.

you can help
CCHD Screening Using Pulse Oximetry: Next Hurdle - False Negatives
Regional False Negative Surveillance

Maryland CCHD Advisory Council Initiative

Working with:
• Children’s National
• INOVA Health System
• Georgetown University Medical Center

To evaluate why the infant was not identified on birth screening

Dr. Gerard Martin
Pediatric Cardiology, Children’s National Medical Center
111 Michigan Avenue
NW Washington, DC 20010

Dear Dr. Martin,

The Maryland Critical Congenital Heart Disease (CCHD) Newborn Screening Follow Up Program is charged with providing surveillance and quality assurance for CCHD screening. As part of that function, this program needs to be informed of babies who are diagnosed with CCHD after their newborn care is completed. This allows us to evaluate why the infant was not identified on birth screening. Children’s National Medical Center is a major referral center for pediatric cardiology patients from Maryland, and as such, I would like to request that your institution provide us with the following information on infants seen for a new diagnosis of CCHD:
## Regional False Negatives

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Birth Hospital</th>
<th>Screened</th>
<th>Pulse Ox Screen Results</th>
<th>Pulse Ox Value Diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Coarctation of Aorta</td>
<td>A</td>
<td>Yes</td>
<td>98/97</td>
<td>99</td>
</tr>
<tr>
<td>TOF</td>
<td>B</td>
<td>Yes</td>
<td>99/97</td>
<td>97</td>
</tr>
<tr>
<td>TOF/AV Canal</td>
<td>C</td>
<td>No</td>
<td>Not screened at birth hospital</td>
<td>85</td>
</tr>
<tr>
<td>TAPVD</td>
<td>D</td>
<td>Yes</td>
<td>95/95</td>
<td>84</td>
</tr>
<tr>
<td>Coarctation of Aorta</td>
<td>E</td>
<td>Yes</td>
<td>98/96</td>
<td>94/84</td>
</tr>
</tbody>
</table>
Coarctation

Passing Saturation 96%

Modified from Rudolph, 1974, Fig. 10-5
Coarctation

Right Hand: 95%
Foot: 91%

PI less than 0.7

Modified from Rudolph, 1974, Fig. 10-5, Fig. 10-6c
Tetralogy of Fallot

Right Ventricular Outflow Obstruction

Passing Saturation: 95%

Modified from Rudolph, 1974, Fig. 12-4
Tetralogy of Fallot

Failing Saturation: 50%

Modified from Rudolph, 1974, Fig. 12-4, Fig. 12-3
Total Anomalous Pulmonary Venous Drainage (TAPVD)

Passing Saturation: 95%

Modified from Rudolph, 1974, Fig. 17-7
TAPVD

Failing Saturation: 50%

Modified from Rudolph, 1974, Fig. 17-7, Fig. 17-6
Conclusions:

- **Wide inter-hospital variation** in prenatal and antenatal detection for TGA and CoA
- **Manual linkage is impractical** on a national basis
- Need for **national validated database** or registry to raise the standard of care
- Hospitals need **granularity** to understand deficiencies and institute actions to improve performance
The Toolkit & Heart Smart Videos

**Toolkit Includes:**
- Implementation Recommendations
- Screening Protocol
- Education for Families
- Competencies for Providers
- Advocacy Resources and Stories

Educational Videos translated into these five languages:

- مرحبا
- привет
- ¡hola!
- nǐ hǎo
- bonjour

Children's National Heart Institute
Part of the Children's National Health System
Online Resources

Children’s National Pulse Ox Program
www.childrensnational.org/pulseox/

Center for Disease Control
www.cdc.gov/ncbddd/pediatricgenetics/cchdscreening.Html

Baby’s First Test
http://www.babysfirsttest.com//

Parent Advocacy Groups
http://1in100.org/ ; www.tchin.org

Newborn Coalition
http://newborncoalition.com/

ACMG CCHD ACTion Sheet

NewSTEPs
https://www.newsteps.org/
Thank You

Questions?
Contact Information

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