Newborn Screening for Critical Congenital Heart Disease: Current Implementation Status and Future

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September 9, 2021

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Outline

❤ Overview of newborn screening for CCHD
❤ Newborn screening implementation
❤ Future needs
❤ Q&A
Newborn Screening for CCHD

♥ Utilizes pulse oximetry to detect lower oxygen saturations often associated with ductal-dependent Critical Congenital Heart Disease (CCHD)
  – Critical = surgery or catheter intervention in first year of life
♥ The screen detects HYPOXEMIA
  – Associated with non-critical CHD
  – Associated with Pulmonary Conditions
    • Pneumonia
    • Persistent Pulmonary Hypertension
  – Associated with Bacterial Infections
    • Sepsis
  – Associated with CCHD
Addition to the RUSP: September 2011

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 18A19
Rockville, MD 20857

Dear Dr. Howell:

As indicated in my letter to you on April 20, 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 procedures, public and strengthening service infrastructure needs for implementation, and reviewed the requested ICC Plan of Action.

As you know, congenital heart disease causes up to 3% of all infant deaths in the first year of life. Heart defects affect about 7 to 9 of every 1000 live births, and many can be detected and potentially treated by measuring blood oxygen saturation. The SACHDNC reviewed the available information on the effectiveness of screening, and 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-1(h)(4)).

I have decided to adopt the SACHDNC’s first recommendation to add CCHD to the RUSP.

Regarding the four SACHDNC recommendations for action by the National Institutes of Health, Centers for Disease Control and Prevention, and HRSA to address recognized evidence gaps (Recommendations #2-#5), I have decided to adopt these recommendations. I will direct the named agencies, as well as other relevant HHS agencies, to proceed expeditiously with implementation, as described in the attachment, as feasible. I am taking this action because I believe that as we move forward, these activities will add important foundational information regarding the potential impact of implementing universal screening for CCHD, strengthen the platform on which to build the critical infrastructure for universal screening, and provide states with the data necessary to consider requiring that this condition be added to their existing newborn screening programs.

I would like to commend the SACHDNC on your success in creating and implementing an external scientific evidence review process for rare conditions that incorporates systematic evidence-based and peer-reviewed recommendations. I am encouraged by the emerging evidence base for the utility of early diagnosis and detection of CCHD via measurement of blood oxygen saturation, as well as the momentum and commitment that is evidenced at the state and federal levels to support implementation and investigation of successful screening programs.

While we collectively engage in the remaining work that needs to be completed, HHS will continue to encourage states, health care facilities, and individual clinicians to provide this screening and contribute to the knowledge base in this important area.

I am committed to advancing screening for CCHD, and I appreciate the contributions of the SACHDNC in assisting HHS and states to explore ways to enhance newborn and child screening to improve the health of infants born in the United States.

Sincerely,

Kathleen Sebelius

Kathleen Sebelius
Current Status | September 2021

[CCHD Map Showing Universally Screened Areas]

[NewSTEPs Logo]

[NewSTEPs: A Program of the Association of Public Health Laboratories]
Impact of Mandatory Screening Policy

Association of US State Implementation of Newborn Screening Policies for Critical Congenital Heart Disease With Early Infant Cardiac Deaths

Rahi Abouk, PhD; Scott D. Grosse, PhD; Elizabeth C. Alles, PhD, MPH; Matthew E. Oster, MD, MPH

Figure. Mean Critical Congenital Heart Disease Early Infant Death Rates by Year, 2007-2013, for States With No Screening Policy, States With Mandatory Screening Policy Not Yet Implemented and Implemented by June 1, 2013, and States With Only Nonmandatory Screening Policies as of June 1, 2013

NewSTEPs
A Program of the Association of Public Health Laboratories
# Impact of Mandatory Screening Policy

## Table 4. Adjusted Percentage Declines in Rates of Deaths Due to Critical Congenital Heart Disease and Other Congenital Heart Disease Associated With State Mandatory Screening Policies, 2011-2013

<table>
<thead>
<tr>
<th>Age Range of Deaths</th>
<th>Decline in Death Rate, % (95% CI)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Critical Congenital Heart Disease Deaths</td>
</tr>
<tr>
<td>24 h to &lt;6 mo</td>
<td>33.4 (10.6 to 50.3)</td>
</tr>
</tbody>
</table>

### Sensitivity analyses of timing of mandate (age at death 24 h to <6 mo)

- Implemented Aug 1, 2011–June 30, 2012: 19.7 (3.1 to 37.1) vs 21.7 (8.7 to 32.9)
- Implemented July 1, 2012–June 1, 2013: 53.6 (36.0 to 66.3) vs 21.0 (0.3 to 37.4)

### Sensitivity analyses of timing of deaths (screening implemented Aug 1, 2011–June 1, 2013)

- Birth to <6 mo: 30.7 (9.3 to 47.1) vs 27.0 (15.1 to 37.3)
- Birth to <12 mo: 28.4 (8.5 to 44.0) vs 17.9 (3.0 to 30.6)
- 24 h to <12 mo: 30.5 (12.9 to 44.5) vs 11.2 (−4.8 to 24.9)
- 24 h to <6 mo, restricted to infants born at >32 wk: 29.5 (5.0 to 50.1) vs 20.1 (2.3 to 34.7)
Unique Challenges and Opportunities
CCHD NBS Implementation

• Data Collection
  – State authority to collect data
  – Mechanisms to collect data
  – Hospital time and buy-in to report data
  – Defining minimum data set
  – Funding for surveillance
  – Quality assurance/Quality control

• Birth Defects Registry
  – Partner to collect long-term follow-up data
  – Identify false negatives

• Education
  – Staff
  – Leadership
  – Clinicians
  – Community/Advocacy
CCHD Cases Identified via Newborn Screening (2012 – 2021)

- Coarctation of Aorta
- Tetralogy of Fallot
- Transposition of Great Arteries
- Hypoplastic Left Heart Syndrome
- Double Outlet Right Ventricle
- Pulmonary Atresia
- Total Anomalous Pulmonary Venous Connection
- Tricuspid Atresia
- Ebsteins Anomaly
- Single Ventricle
- Truncus Arteriosus
- Aortic Valve Disease
- Interrupted Arch
- Null

- 12 NBS programs contributing data
- 612 infants

Source: NewSTEPs Data Repository
CCHD Data Response Team

- Identify gaps in CCHD data collection, analysis, and reporting
- Continue CCHD screening education for state programs and stakeholders
- Collaborate with stakeholders on quality improvement efforts in CCHD screening and data collection
The CCHD Screening Landscape since 2018

A picture may be worth a thousand words...

... but with CCHD screening...

...the devil is in the details.
CCHD Pulse Oximetry Screening Is...

- One of the least uniform of the conditions on the RUSP
  - States utilize various:
    - Primary/Secondary targets
    - Authority to collect data, data collection and analysis
    - Integration methods with Birth Defects Programs
    - Exemptions/Population screened
    - Algorithms
CCHD Pulse Oximetry Screening Is...

• **Unique to all other NBS conditions**
  - Pulse Oximetry Screening is the third line of defense
    • And the first two lines are getting better (though unlikely to ever be 100%)
  
  - Other Public Health Programs are involved (e.g., Birth Defects Registries)
    • In most states, identified cases of primary CCHD targets are being reported

• **Impact of the screen itself varies by individual and location**
  - Dependent upon prenatal and clinical care availability and accessibility
Definition of Critical CHD

• Group of “serious” heart defects
• Typically have low oxygen levels in the newborn
• Conditions that require intervention
  – Soon after birth
  – In the first year of life
• May or may not be ductal dependent
Cyanosis or mixing of oxygenated blood

Failing Sat 90%
Pulse Oximetry as a Screening Method

♥Pulse oximetry measures oxygen saturation of hemoglobin in arterial blood
♥Non-invasive and painless test
♥Overall sensitivity ~76%, specificity 99.9%, false positive rate 0.06%

(Plana Cochrane Database of Systematic Reviews 2018)
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
CDC Expanded List of CCHD

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

Additional 5 lesions:
- IAA
- CoA
- Ebstein’s
- DORV
- SV

AS/PS?

Centers for Disease Control and Prevention
CDC 24/7: Saving Lives. Protecting People™
## Core and Secondary Conditions Detected by CCHD Screening

<table>
<thead>
<tr>
<th>Core conditions (CCHD)</th>
<th>Secondary conditions (non-CCHD)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Coarctation of the aorta</td>
<td>Hemoglobinopathy</td>
</tr>
<tr>
<td>Double-outlet right ventricle</td>
<td>Hypothermia</td>
</tr>
<tr>
<td>Ebstein’s anomaly</td>
<td>Infection, including sepsis</td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome</td>
<td>Lung disease (congenital or acquired)</td>
</tr>
<tr>
<td>Interrupted aortic arch</td>
<td>Noncritical congenital heart defect</td>
</tr>
<tr>
<td>Pulmonary atresia</td>
<td>Persistent pulmonary hypertension</td>
</tr>
<tr>
<td>Single ventricle (not otherwise specified)</td>
<td>Other hypoxemic condition not otherwise specified</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td></td>
</tr>
<tr>
<td>Total anomalous pulmonary venous return</td>
<td></td>
</tr>
<tr>
<td>D-transposition of the great arteries</td>
<td></td>
</tr>
<tr>
<td>Tricuspid atresia</td>
<td></td>
</tr>
<tr>
<td>Truncus arteriosus</td>
<td></td>
</tr>
<tr>
<td>Other critical cyanotic lesions not otherwise specified</td>
<td></td>
</tr>
</tbody>
</table>

**Lessons Learned From Newborn Screening for Critical Congenital Heart Defects**

Matthew E. Deter, MD, MPH, a,b Susan W. Ascroft, MD, a,b Jill Glidewell, APRN, MSN, MPH, a,b Jesse Hackell, MD, a,b Lazaros Kochlias, MD, MSCR, a,b Gerard R. Martin, MD, a,b Julia Phillippi, PhD, CNM, a,b Nelangi M. Pinto, MD, a,b Annamarie Saarinen, MA, a,b Marci Sontag, PhD, a,b Alex R. Kemper, MD, MPH, MS, a,b

Pediatrics 137: 2016

AAP 2016 Expert Panel
Secondary Targets: Pneumonia & Sepsis

Failing Sat 93%
Commitments to support

EVERY
NEWBORN

July 2014

EVERY
WOMAN
EVERY
CHILD

ESTIMATED IMPACT 2015–2030

772,000 Child lives saved

6% reduction in deaths due to pneumonia

<table>
<thead>
<tr>
<th>COST</th>
<th>LIVES SAVED SENSITIVITY</th>
</tr>
</thead>
<tbody>
<tr>
<td>$101M</td>
<td>+/- 24,000 for a +/- 5 percentage point change in coverage in the clinic</td>
</tr>
</tbody>
</table>

Scenario modeled: Expand access to pulse oximeters in clinics and hospitals to more accurately identify children with hypoxic pneumonia and increase percentage of children diagnosed and treated.

Innovation assumptions: Modeled an average peak coverage of 0%, 72%, and 81% in home, clinic, and hospital settings, respectively. Assumes availability of pulse oximeters increases the accuracy of diagnosing hypoxic pneumonia by 15 percentage points to 85% and increases the fraction of children under age five with pneumonia screened for infection by 9 percentage points to an average of 50% across countries in scope. Impact could increase if bundled with other diagnostic tools.
What Data are Collected by NBS program?

- Individual level Data: 31
- All O2 Sats: 18
- Pass/fail/not done: 12
- Final O2 Sats: 1
- No Data: 13
- All Case Data: 5
- Aggregate: 5
Our state does mandate CCHD screening, but the legislature chose to pass it without attaching any funding or mechanism for identifying cases. We get twice yearly reports from facilities with the number of infants born/refused/missed/abnormal screens, but no data on what happens to those abnormal screens.

I sit on our child death review committee, so I know when it does not go well. It would be nice to know about the times when it does...

From State “X”, Division of Public Health, email response sent August, 2018
Individual Data

- CA Department of Health Services (DHCS) receives reports for only about 60% of state births - unfunded mandate

Graph courtesy of Donna Goff, work partially supported by Dr. Goff’s California Community Service Grant, March of Dimes.
What is counted as a screening find?

Also varies, some states are required to screen “all infants”...

• Are prenatally detected infants excluded?
• Are infants with a prior echocardiogram excluded?
• Clinical assessment leads to early pulse oximetry screen?
Integrated Data System – CCHD and Birth Defects Surveillance

Yes, 19

No Integration, 32
Reasons for No Integration Vary

- Yes, 19
- No Integration, 32

Subcategories:
- No individual screening data, 13
- Systems exist, not linked, 15
- No Birth Defects Surveillance, 4
How does bi-directional communication happen between NBS and Birth Defects?
For those who integrate, how are the Data Used?

• Infants determined to have CCHD by the birth defects surveillance program are linked to CCHD screening results to **identify any false negatives**.

• Infants with CCHD identified through CCHD screening are linked to the birth defects surveillance program in order to **match identified cases**

• Infants with failed CCHD screens are linked to the birth defects surveillance program to **aid in follow-up of the failed screen and determination of outcome**
For those who integrate, how are the data used?

- **Identification of Missed Cases**: 1
- **Match Identified Cases**: 5
- **Follow-up/determination of outcome**: 7

* 1 state unsure of how data were used

- Identifying infants from birth defects registry who were NOT identified through screening
- Long-term follow-up of infants with CCHD identified through newborn screening
- Ensure that cases identified by NBS are collected in birth defects surveillance

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Birth defects surveillance

- Two state reports allowed comparison to birth defects registries
  - **State A**: 22 identified by NBS/230 total reported with CCHD
  - **State B**: 8 identified by NBS/561 total reported with CCHD
State variation: population screened

- All newborns
- Special populations/exemptions:
  - NICU
  - births at altitude
  - home births
Screening in the Neonatal Intensive Care Units

• Pre and post ductal oxygen saturations are similar to saturations in late preterm and term infants. Can be safely implemented into NICUs. Iyengar et al. Pediatric Cardiol 2014

• Roughly 1/3 receive echo, many on oxygen, continuous pulse oximetry standard

• Initial U.S. public health outcomes indicate lower yield (NJ, MI)

Photo from www.wspa.com
Exemptions for screening at High or Moderate Altitudes

- 1.1% failure rate at moderate altitude (5557 feet or 1694 meters)
- 0.2% failure rate at sea level

- Delayed transition
- Limited pulmonary vasodilation
- Atrial level shunting right-to-left
- V/Q mismatch

FIGURE 6
All studies used mean saturations ± SD with the exception of Ravert's study \(^{15}\) who used a mean saturation and saturation range.

Pediatrics 2014;133:e561–e569
What do Wisconsin and the Netherlands have in common?

Pulse Oximetry Screening for Critical Congenital Heart Disease in Planned Out-of-Hospital Births

Jennifer J. Lhost, BS¹, Elizabeth M. Goetz, MD, MPH², Jody D. Belling, RN, MSN², W. Marijke van Roojen, LM, CPM³, Gretchen Spicer, LM, CPM¹, and John S. Hokanson, MD²

Adapted protocol for pulse oximetry screening for congenital heart defects in a country with homebirths

Ilona C. Narayen • Nico A. Blom • Marjolein S. Verhart • Marrit Smit • Fennie Posthumus • Annique J. M. van den Broek • Hester Havers • Monique C. Haak • Arjan B. te Pas

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"So many protocols, which one is best?"

<table>
<thead>
<tr>
<th>Algorithm</th>
<th>Extremity Screened</th>
<th>POS for Pass (%)</th>
<th>Difference between arm/leg for Pass (%)</th>
<th>Rescreens (n)</th>
<th>Screen Age (hours)</th>
</tr>
</thead>
<tbody>
<tr>
<td>AAP¹⁰</td>
<td>RH, Foot</td>
<td>95 in either</td>
<td>&lt;3</td>
<td>2</td>
<td>&gt;24</td>
</tr>
<tr>
<td>New Jersey¹⁸</td>
<td>RH, Foot</td>
<td>95 in both</td>
<td>&lt;3</td>
<td>2</td>
<td>&gt;24</td>
</tr>
<tr>
<td>Tennessee²⁴</td>
<td>Foot (AAP if test Fail)</td>
<td>97</td>
<td>≤3 on rescreen</td>
<td>2</td>
<td>&gt;24</td>
</tr>
<tr>
<td>Granelli⁵</td>
<td>RH, Foot</td>
<td>95 in either</td>
<td>&lt;3</td>
<td>2</td>
<td>&lt;24</td>
</tr>
<tr>
<td>Ewer⁸</td>
<td>RH, Foot</td>
<td>95 in both</td>
<td>≤2</td>
<td>1</td>
<td>6-24</td>
</tr>
<tr>
<td>Poland¹⁵</td>
<td>Foot</td>
<td>95</td>
<td></td>
<td>1</td>
<td>&lt;24</td>
</tr>
<tr>
<td>Germany⁶</td>
<td>Foot</td>
<td>95</td>
<td></td>
<td>1</td>
<td>&gt;24</td>
</tr>
</tbody>
</table>

Martin *Pediatrics* 2020
## U.S. Algorithms

<table>
<thead>
<tr>
<th>Algorithm Source</th>
<th>Cutoff for Passing With First Measurement</th>
<th>Retest Criteria for Subsequent Measurements</th>
<th>Fail Criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>AAP</td>
<td>$O_2 \text{ sat} \geq 95%$ (in either RH or F) AND [hand-foot]</td>
<td>$O_2 \text{ sat} &lt; 95%$ (in both RH and F) OR [hand-foot]</td>
<td>$O_2 \text{ sat} &lt; 90%$ (either RH or F) OR fail retest criteria $\times 3$</td>
</tr>
<tr>
<td></td>
<td>$O_2 \text{ sat} \leq 3%$</td>
<td>$O_2 \text{ sat} &gt; 3%$</td>
<td></td>
</tr>
<tr>
<td>New Jersey</td>
<td>$O_2 \text{ sat} \geq 95%$ (in both RH and F) AND [hand-foot]</td>
<td>$O_2 \text{ sat} &lt; 95%$ (in both RH and F) OR [hand-foot]</td>
<td>$O_2 \text{ sat} &lt; 90%$ (either RH or F) OR fail retest criteria $\times 3$</td>
</tr>
<tr>
<td></td>
<td>$O_2 \text{ sat} \leq 3%$</td>
<td>$O_2 \text{ sat} &gt; 3%$</td>
<td></td>
</tr>
<tr>
<td>Tennessee</td>
<td>$O_2 \text{ sat} \geq 97%$ (F)</td>
<td>$O_2 \text{ sat} &lt; 95%$ (in both RH and F) OR [hand-foot]</td>
<td>$O_2 \text{ sat} &lt; 90%$ (either RH or F) OR fail retest criteria $\times 3$</td>
</tr>
<tr>
<td></td>
<td></td>
<td>$O_2 \text{ sat} &gt; 3%$</td>
<td></td>
</tr>
</tbody>
</table>

F, either foot; $O_2$, oxygen; RH, right hand; sat, saturation.

*(Oster Pediatrics 2016)*
AAP Protocol U.S. Implementation

Pediatrics, Letter to Editor
https://pediatrics.aappublications.org/content/146/1/e20191650/tab-e-letters

NewSTEPs
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Updated Strategies for Pulse Oximetry Screening for Critical Congenital Heart Disease

Gerard R. Martin, MD, Andrew K. Ewer, MD, Amy Gaviglio, MS, LCGB, Lisa A. Hom, RN, Esq, Annamarie Saarinen, MA, Marci Sostag, PhD, Kristin M. Burns, MD, Alexa R. Kemper, MD, MPH, MS, Matthew E. Oster, MD, MPH

TABLE I Workgroup Attendees

Clinicians
Pediatricians
Pediatric cardiologists
Neonatologists
Nurses

Representatives from
American College of Cardiology Foundation
AHA
American College of Medical Genetics and Genomics
American Board of Pediatrics
International Society for Neonatal Screening
March of Dimes
Association of Maternal and Child Health Programs
National Association of Neonatal Nurse Practitioners
NewSTEPs (Association of Public Health Laboratories)
Centers for Disease Control and Prevention
US Food and Drug Administration
US HHS
National Institutes of Health
National Library of Medicine
State public health officials
CCHD parent advocates


September 2018

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Success in Screening but challenging to qualify

- CCHD screening is offered in all states
- Fewer U.S. deaths due to CCHD since becoming mandatory
- Outcomes of the screen are still hard to quantify due to differences in how state programs define targets, eligible population, collect data, and vary in algorithm implemented
Pulse Oximetry Screening: Future Needs
Three Primary Areas of Need

Data Collection/Integration/Analysis

Follow-Up, Education, and Training

Other Screening/Clinical Considerations
Data Collection/Integration/Analysis
Where’s the Data?

Documents Published 2011-2021; N= 268

Source: SCOPUS
Where’s the Data?

Top 10 Domestic Affiliations 2011-2021; N= 92

- Sibley Heart Center Cardiology: 5
- University of Wisconsin School of Medicine and Public Health: 5
- The George Washington University School of Medicine and...: 5
- New Jersey Department of Health and Senior Services: 5
- Emory University School of Medicine: 7
- Children's Healthcare of Atlanta: 9
- Emory University: 10
- Coordinating Center for Health Promotion: 13
- Centers for Disease Control and Prevention: 13
- Children's National Health System: 20

Source: SCOPUS
Why is there so little data from Public Health Programs?
Confusion: Responsibilities

- What is the purpose of CCHD Screening Data Collection from a Public Health Perspective?
  - Overarching surveillance
    - Determining incidence of various defects
  - Backend Quality Improvement/Quality Assurance
    - Assessing screening and follow-up performance later in time
  - Real-time Quality Improvement/Quality Assurance
    - Ensuring all eligible babies are screened and followed correctly
  - Program/Algorithm improvement
    - What are we missing? How can we screen better?
Confusion: Roles

• Who should oversee data collection/analysis?
  • NBS programs?
  • Individual birth facilities?
  • Birth defects registries?
  • Cardiology Centers?
  • Combination?
  • Other?
Lack of Intra-Agency Data Linkages

CCHD Screening Programs linked to Birth Defect Registries

- Vital Records/Birth Certificates
  - Aids in determining denominator, unscreened infants
- Neonatal/Infant/Childhood Death Certificates
  - Aids in determining true count of CCHD cases
- Birth Defects Registries
  - Aids in determining detected and missed cases

Register for the APHL and NAPHSIS Newborn Screening and Vital Records Webinar on 9/14!!
Recognition & Funding: Or Lack Thereof

• No annual funding structure
  – E.g., No ongoing grants as is seen in EHDI

• Blood spot NBS programs funded by fees
  – Which may or may not have been increased to add CCHD

• CCHD often done as an “aside”
  – No routine CCHD screening-focused meeting
Critical Congenital Heart Defects (2021-2026)

- Component of Birth Defects Surveillance NOFO
- **Goal:** Understand timing and mode of CCHD detection
- **Activities:**
  - Conduct surveillance on additional CCHD cases
  - Ascertain timing and method of CCHD detection
  - Ascertain individual-level CCHD screening results and timing of confirmatory echocardiogram

- **Funded 8 health departments:**
  - AZ, MI, MN, NJ, NC, SC, TN, UT
So... How Can We Improve Data Collection?
Define and Promote Standard Minimal Data Sets

- Need to relook at minimal data set recommendations
- Need to improve dissemination and promotion

**TABLE 2 Minimum Data Recommendations and Considerations for Data Exchange for Reporting of CCHD Screening Results**

<table>
<thead>
<tr>
<th>Birth Facility Data</th>
<th>Public Health Program Data</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Patient level data: Patient identification data that allows validation that all infants had a valid screen and results</td>
<td>1. Will vary according to the legislative or executive mandate of each state.</td>
</tr>
<tr>
<td>Age in hours at time of screening. All oximetry saturations reported (initial screen and any subsequent screens)</td>
<td>2. Aggregate or individual data may be specified to be provided to and tracked by public health programs.</td>
</tr>
<tr>
<td>Final screening result</td>
<td>3. Birthing facilities required to report to public health programs should provide data sufficient to determine whether all eligible infants were screened and, in the case of positive screens, information about the evaluation performed.</td>
</tr>
<tr>
<td>Obstacles encountered during screening process (i.e., obstacles with the infant/family, staff, equipment)</td>
<td>4. Ideal for positive screens: Final diagnosis should be tracked as well as interventions that follow. Should include whether infants required transport for evaluation and treatment or had evaluation at the birthing facility and what treatment entailed.</td>
</tr>
<tr>
<td>Diagnostic results</td>
<td>5. Ideal for negative screens: Subsequent identification of congenital heart defects (i.e., false-negative screens) could be linked within the NBS programs.</td>
</tr>
<tr>
<td>2. Screening program data: Screening protocol being used</td>
<td>6. Summary statistics should be provided by health departments and NBS programs to stakeholders.</td>
</tr>
<tr>
<td>Type of pulse oximeter used for screening</td>
<td></td>
</tr>
</tbody>
</table>

NBS, newborn screening program.

### Develop Public Health Case Definitions

<table>
<thead>
<tr>
<th>Hypoplastic Left Heart</th>
<th>POX screening should identify all babies with these disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pulmonary Atresia</td>
<td></td>
</tr>
<tr>
<td>Transposition of the Great Arteries</td>
<td></td>
</tr>
<tr>
<td>Single Ventricle</td>
<td></td>
</tr>
<tr>
<td>Truncus arteriosus</td>
<td></td>
</tr>
<tr>
<td>Tricuspid atresia</td>
<td></td>
</tr>
<tr>
<td>Interrupted Aortic Arch with VSD</td>
<td></td>
</tr>
<tr>
<td>Double-Outlet Right Ventricle</td>
<td></td>
</tr>
<tr>
<td>Aortic Valve Stenosis (with PDA)</td>
<td></td>
</tr>
<tr>
<td>Pulmonary Atresia with Ventricular Septal Defect</td>
<td></td>
</tr>
<tr>
<td>Total anomalous pulmonary venous return</td>
<td></td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td></td>
</tr>
<tr>
<td>Coarctation of the Aorta with PDA</td>
<td></td>
</tr>
<tr>
<td>Ebstein's Anomaly</td>
<td></td>
</tr>
</tbody>
</table>

**POX screening may have lower sensitivity due to physiologic variability of oxygen saturations in the newborn.**

<table>
<thead>
<tr>
<th>Coarctation of the Aorta without a PDA</th>
<th>POX screening is unlikely to detect these conditions due to physiologic reasons. This is particularly the case when intervention is not required until after 1 month of age and before 1 year of age.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aortic Valve Stenosis without a PDA</td>
<td></td>
</tr>
<tr>
<td>Pulmonary Atresia with Ventricular Septal Defect</td>
<td></td>
</tr>
<tr>
<td>Total anomalous pulmonary venous return</td>
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<td></td>
</tr>
</tbody>
</table>

**NOTE:** Surveillance case definitions are not intended to be used by healthcare providers for making a clinical diagnosis or determining how to meet an individual patient's health needs.
Public Health Interpretations: Undetected Cardiac Lesions

Follow-up of any missed case should include investigating the adherence to the algorithm and appropriate interpretation of the results.

<table>
<thead>
<tr>
<th>Cardiac Lesion Reported</th>
<th>Public Health Categorization</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cardiac finding (red or yellow); algorithm followed/correct interpretation</td>
<td>Physiological Missed Case</td>
</tr>
<tr>
<td>Cardiac finding (green/intervention required within first 30 days of life); algorithm followed/correct interpretation</td>
<td>Physiological Missed Case</td>
</tr>
<tr>
<td>Cardiac finding (blue/intervention not required within first 30 days of life); algorithm followed/correct interpretation</td>
<td>Document findings, but is NOT considered a Missed Case</td>
</tr>
<tr>
<td>Cardiac finding (any color); algorithm NOT followed/incorrect interpretation</td>
<td>Non-Valid Screen due to Error</td>
</tr>
</tbody>
</table>
# Define What Questions We Should Be Able to Answer

<table>
<thead>
<tr>
<th>Metric</th>
<th>Question</th>
</tr>
</thead>
<tbody>
<tr>
<td>Screen Rate</td>
<td>What percentage of eligible newborns are getting screened?</td>
</tr>
<tr>
<td>Failure Rate</td>
<td>What percentage of newborns fail their pulse oximetry screen?</td>
</tr>
<tr>
<td>Detections</td>
<td>What is being detected? Primary and Secondary</td>
</tr>
<tr>
<td>Missed Cases</td>
<td>What is not being detected? Why?</td>
</tr>
<tr>
<td>Detection Modality</td>
<td>What percentage of cases are detected prenatally, clinically, and via screening?</td>
</tr>
<tr>
<td>Other??</td>
<td></td>
</tr>
</tbody>
</table>
Create Data Sharing and Linkages

Identification of Missed Cases

Follow-up/determination of outcome

Match Identified Cases

CCHD Screening and Birth Defects Programs MUST work together

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Summary of Needs

1. Standardized Data Recommendations
2. Improved Case Understanding and Definitions
3. Enhanced Intra-agency Data Linkages
Follow-Up, Education, and Training
What Should Follow-Up Look Like?

• Follow-Up Should Not Dictate Process
  – Happens on Back End

• When Does Follow-Up End?
  – At Data Collection?
  – At Diagnostic Outcomes?
  – At Longer Term Follow-Up?

• How can Programs Work Together to Achieve Shared Goals?
Provider Education and Training

• Continued lack of understanding of targets and role of Pulse Oximetry Screening
• Routine reminders of importance of clinical vigilance
  – A Passed screen DOES NOT rule out CCHD
• If a screen is missed or algorithm not followed, then what?
Birth Facility Education and Training

• Misinterpretations of the algorithm still occur

• How do we re-educate with a potentially changing recommendation?

• How do we incentivize better data reporting?
Family Education and Needs

- Do families understand Pulse Oximetry Screening? Limitations? What to Look For?

- Are they being given their Pulse Ox Results?

- Do they understand the role of Birth Defects registries?
Family Education and Needs

• Are we meeting the needs of families identified with CCHDs? Do we know what the needs are?
Other Screening/Clinical Considerations
Use of Other Biomarkers

- Can we integrate other analyses to improve detection? Other screens?

Oxygen Saturation and Perfusion Index-Based Enhanced Critical Congenital Heart Disease Screening

Heather Siefkes 1, Laura Kair 1, Daniel J Tancredi 1, Brian Vasquez 2, Lorena Garcia 2, Christa Bedford-Mu 1, Satyan Lakshminrusimha 1

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Other Detection Modalities

• Prenatal Detection
  – Are prenatal detection rates improving? For everyone? Everywhere?

• Clinical Detection
  – Is there improved clinical vigilance leading to earlier postnatal detection?
Let’s Keep Talking!

• Please join us for other webinars in this series:
  
  – Clinical Perspective
    • Delving into the various defects
    • Understanding modes of detection
    • Fall 2021
  
  – Program Perspective
    • Hear about existing projects and lessons learned
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