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

Careema Yusuf, MPH, Manager, NewSTEPs, APHL

Lisa A. (Hom) Wandler, RN Esq. Collaborative Practice Facilitator, Children's National Hospital

Amy Gaviglio, MS, CGC, Public Health Genetics Consultant

September 9, 2021

This presentation is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under grant number #U22MC24078 for $1,500,000. This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

NewSTEPs

A Program of the Association of Public Health Laboratories™

Outline

- ♥ Overview of newborn screening for CCHD
- ♥ Newborn screening implementation
- ♥ Future needs
- ♥ Q&A

NewSTEPs
A Program of the Association of Public Health Laboratories™

NewSTEPs

A Program of the Association of Public Health Laboratories™

Newborn Screening for CCHD

♥ Utilizes pulse oximetry to detect lower oxygen saturations often associated with ductal-dependent Critical Congenital Heart Disease (CCHD)

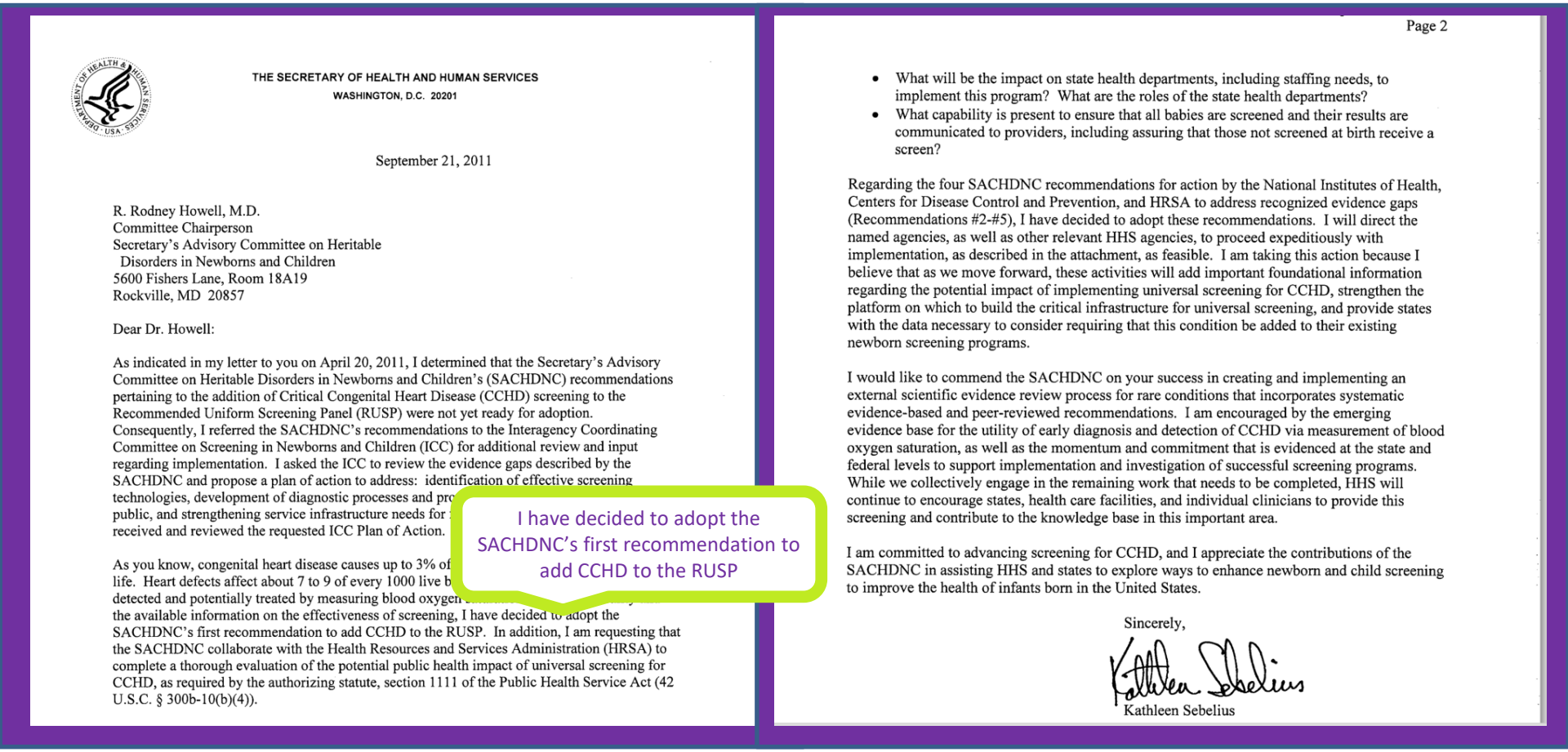
- Critical = surgery or catheter intervention in first year of life

♥ 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

NewSTEPs
A Program of the Association of Public Health Laboratories™

Addition to the RUSP: September 2011



THE SECRETARY OF HEALTH AND HUMAN SERVICES
WASHINGTON, D.C. 20201

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 pro
public, and strengthening service infrastructure needs for
received and reviewed the requested ICC Plan of Action.

As you know, congenital heart disease causes up to 3% of
life. Heart defects affect about 7 to 9 of every 1000 live b
detected and potentially treated by measuring blood oxygen
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-10(b)(4)).

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

- What will be the impact on state health departments, including staffing needs, to implement this program? What are the roles of the state health departments?
- What capability is present to ensure that all babies are screened and their results are communicated to providers, including assuring that those not screened at birth receive a screen?

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.

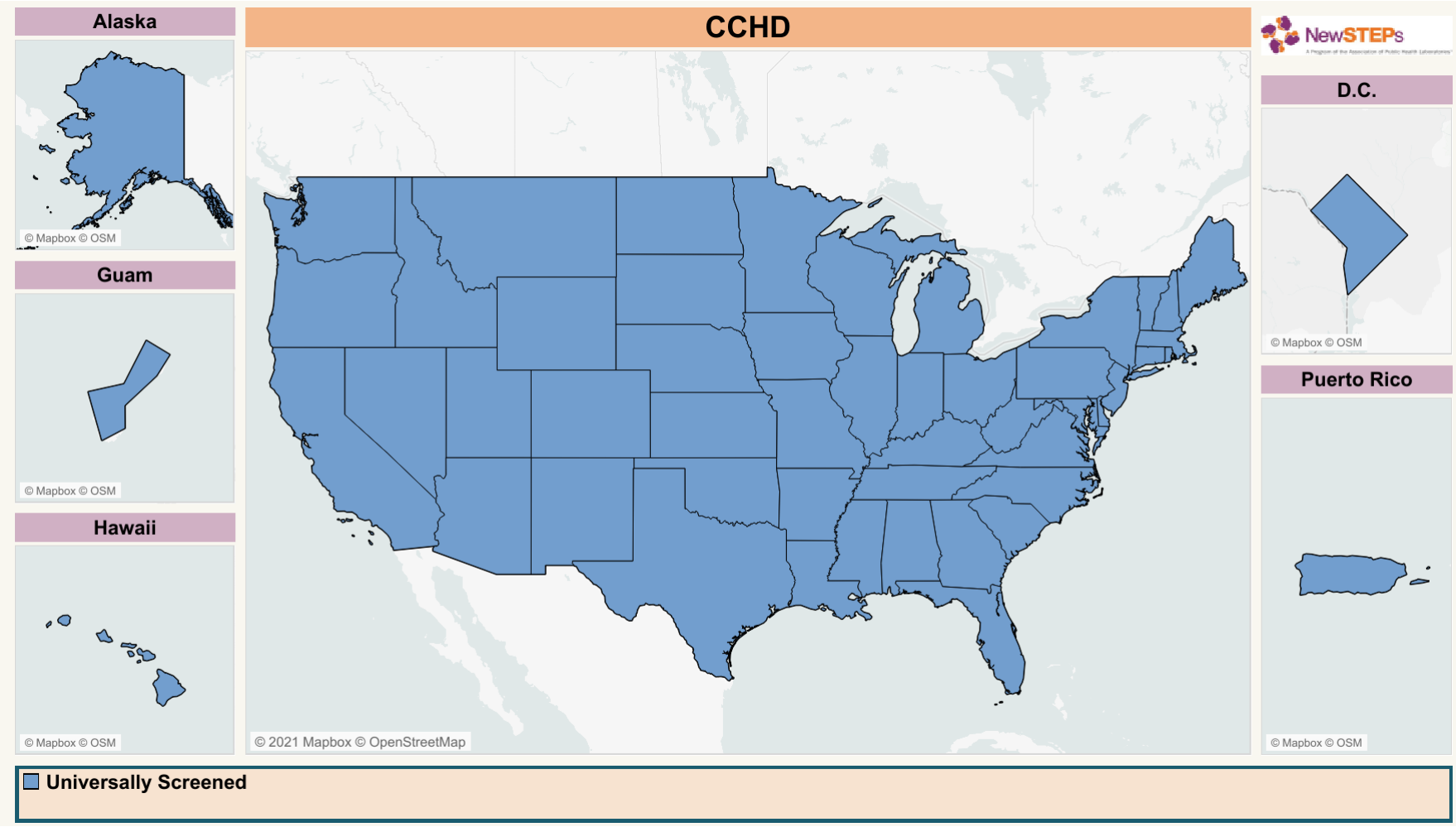
Sincerely,

Kathleen Sebelius

Kathleen Sebelius

NewSTEPs
A Program of the Association of Public Health Laboratories™

Current Status | September 2021



NewSTEPs
A Program of the Association of Public Health Laboratories™

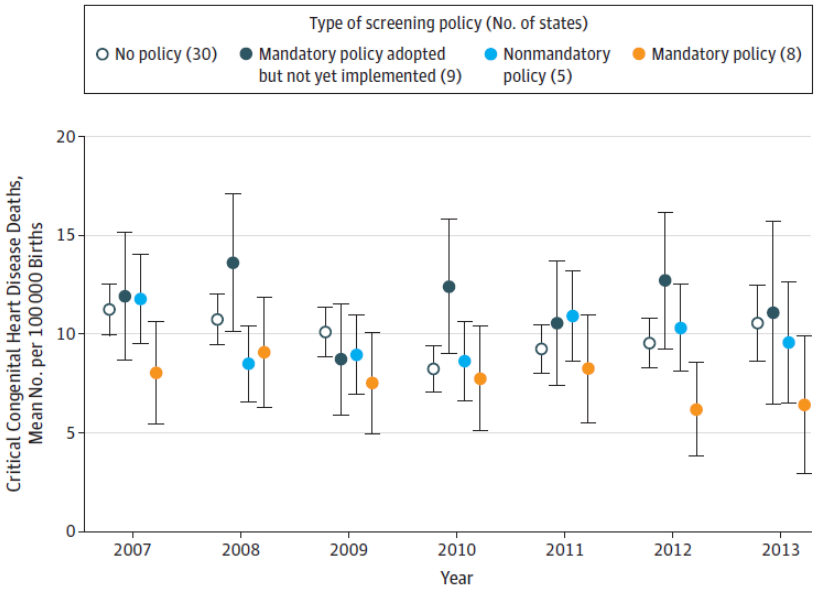
Impact of Mandatory Screening Policy

JAMA | Original Investigation

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. Ailes, 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



JAMA. 2017;318(21):2111-2118. doi:10.1001/jama.2017.17627

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[a]

Age Range of Deaths	Decline in Death Rate, % (95% CI)	
	Critical Congenital Heart Disease Deaths	Other or Unspecified Congenital Heart Disease Deaths
24 h to <6 mo	33.4 (10.6 to 50.3)	21.4 (6.9 to 33.7)
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)	21.7 (8.7 to 32.9)
Implemented July 1, 2012–June 1, 2013	53.6 (36.0 to 66.3)	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)	27.0 (15.1 to 37.3)
Birth to <12 mo	28.4 (8.5 to 44.0)	17.9 (3.0 to 30.6)
24 h to <12 mo	30.5 (12.9 to 44.5)	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)	20.1 (2.3 to 34.7)

NewSTEPs

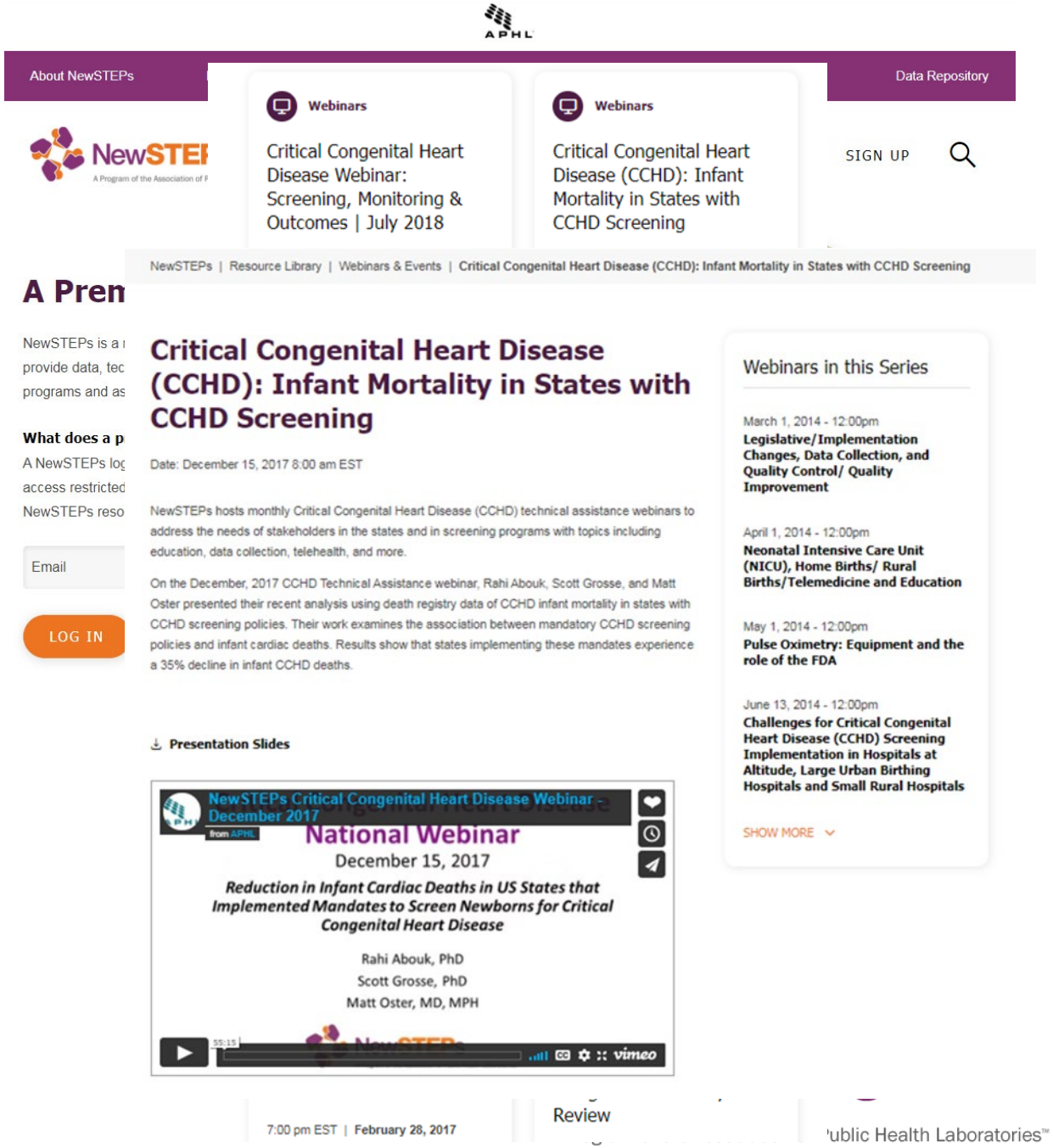
A Program of the Association of Public Health Laboratories™

Unique Challenges and Opportunities CCHD NBS Implementation

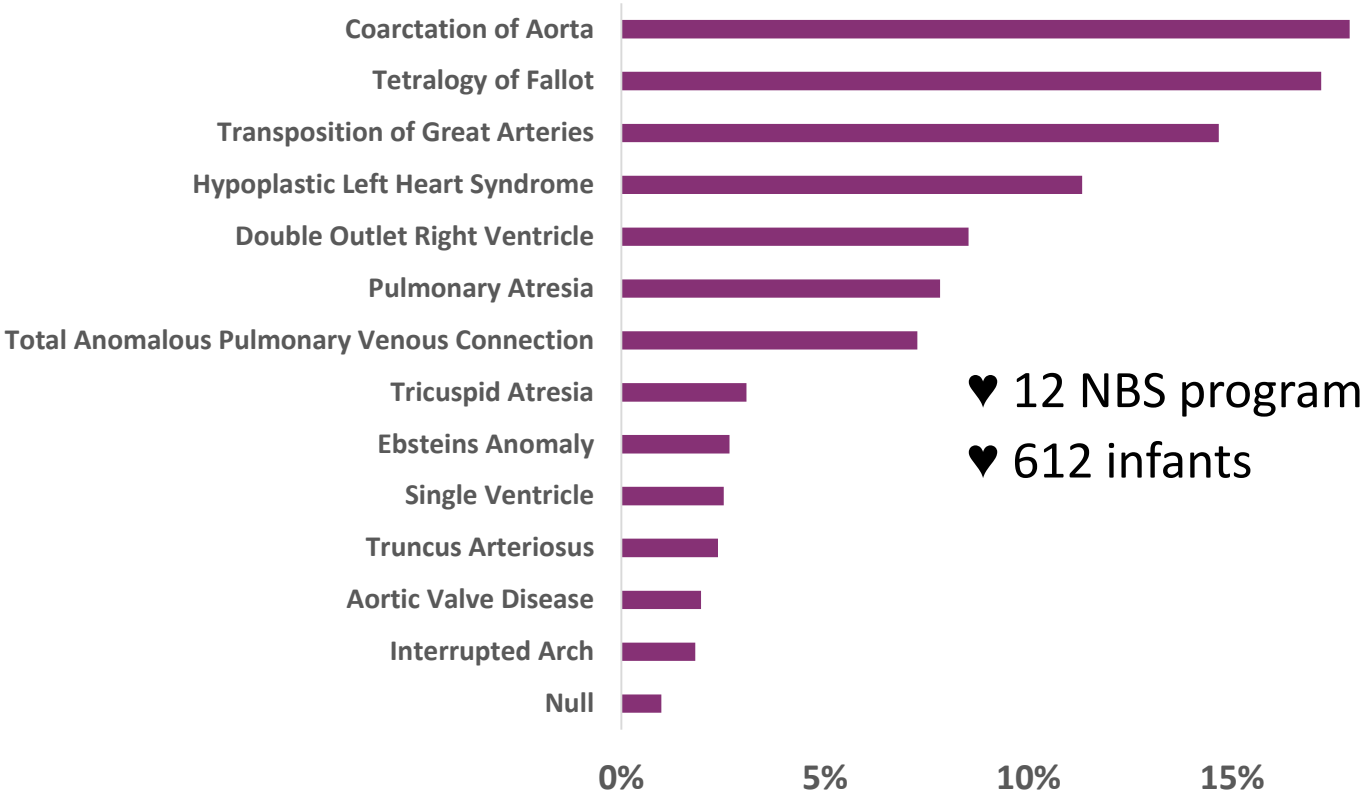
- **Data Collection**
  - State authority to collect data
  - Mechanisms to collect data
  - Hospital time and buy-in to report data
  - Defining minimum data set
  - Funding for surveillance
  - Quality assurance/Quality control
- **Birth Defects Registry**
  - Partner to collect long-term follow-up data
  - Identify false negatives
- **Education**
  - Staff
  - Leadership
  - Clinicians
  - Community/Advocacy

NewSTEPs
A Program of the Association of Public Health Laboratories™

NewSTEPs Technical Assistance CCHD Resources



CCHD Cases Identified via Newborn Screening (2012 – 2021)



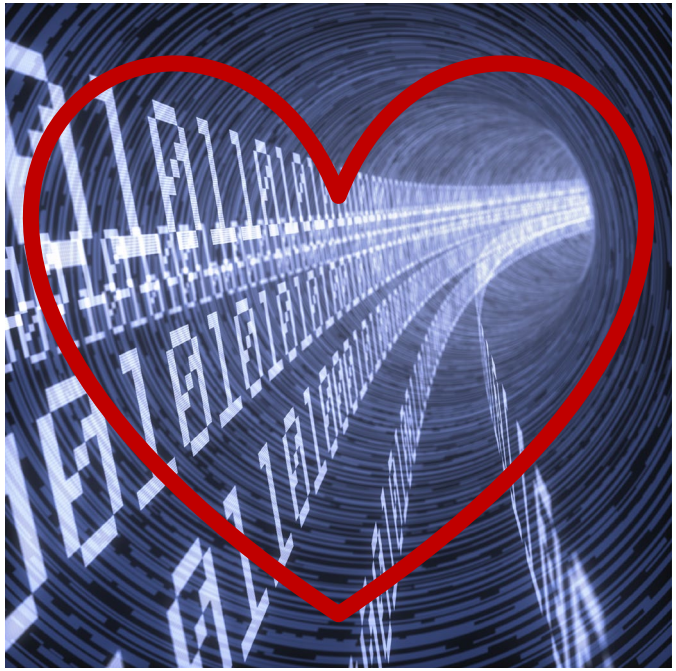
- 12 NBS programs contributing data
- 612 infants

Source: NewSTEPs Data Repository

NewSTEPs
A Program of the Association of Public Health Laboratories™

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



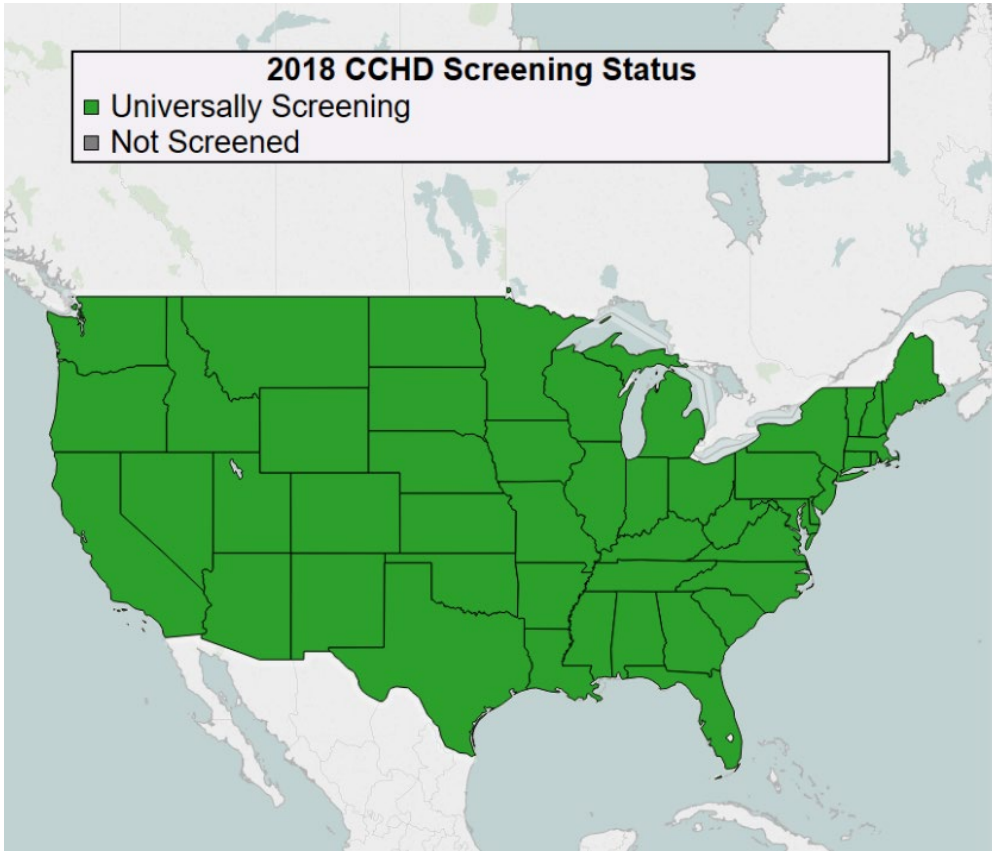
NewSTEPs
A Program of the Association of Public Health Laboratories™

The CCHD Screening Landscape since 2018

A picture may be worth a thousand words…

… but with CCHD screening…

…the devil is in the details.



NewSTEPs
A Program of the Association of Public Health Laboratories™

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

NewSTEPs

A Program of the Association of Public Health Laboratories™

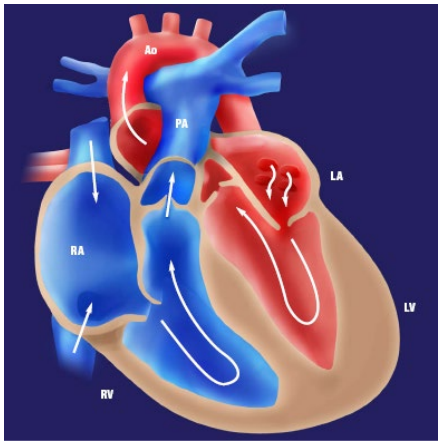
CCHD Pulse Oximetry Screening Is…

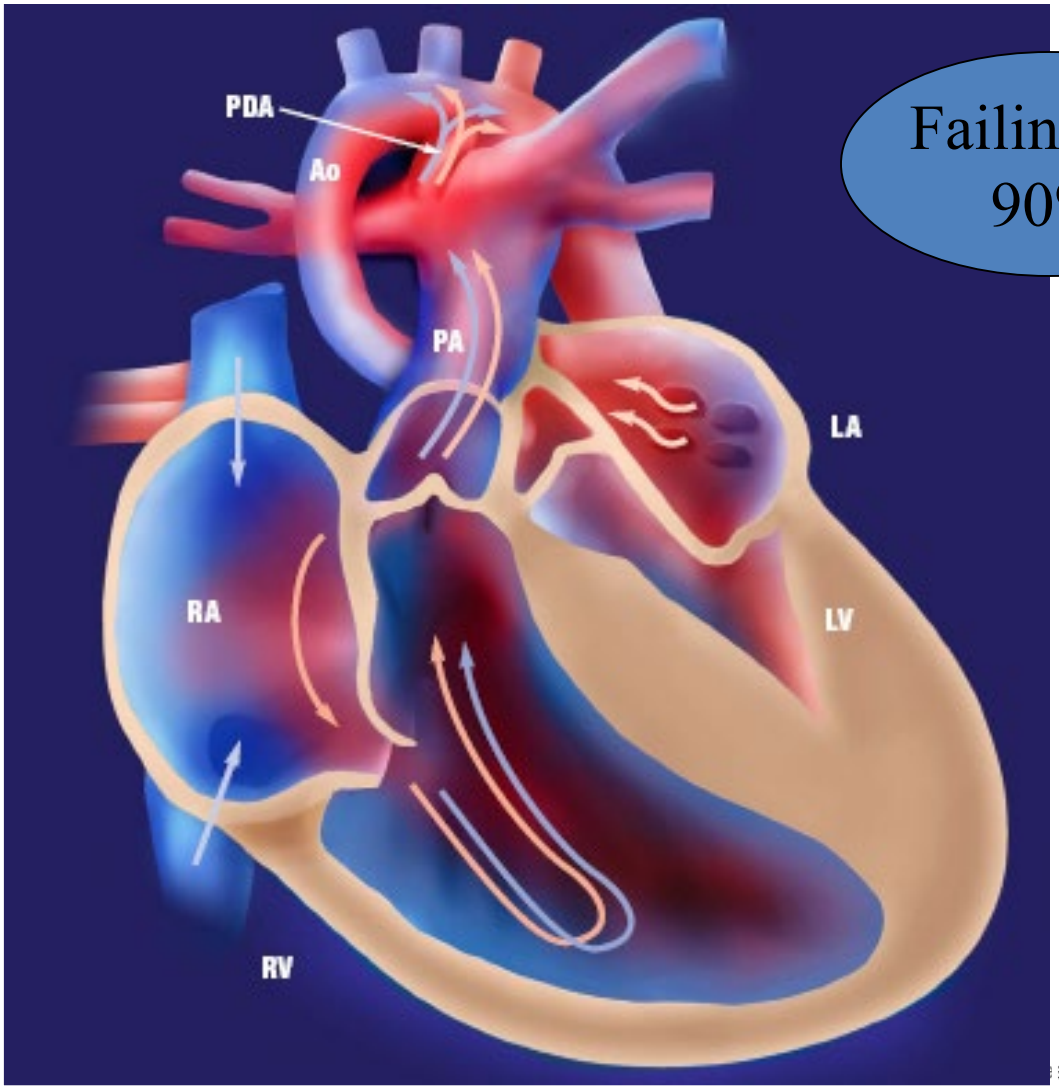
- **Unique to all other NBS conditions**
  - Pulse Oximetry Screening is the third line of defense
    - And the first two lines are getting better (though unlikely to ever be 100%)
  - Other Public Health Programs are involved (e.g., Birth Defects Registries)
    - In most states, identified cases of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

NewSTEPs

A Program of the Association of Public Health Laboratories™

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

NewSTEPs
A Program of the Association of Public Health Laboratories™

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

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™

Additional 5 lesions:

- IAA
- CoA
- Ebstein's
- DORV
- SV

AS/PS?

CDC Centers for Disease Control and Prevention
CDC 24/7: Saving Lives, Protecting People™

NewSTEPs
A Program of the Association of Public Health Laboratories™

Core and Secondary Conditions Detected by CCHD Screening

TABLE 1 Conditions Detected Via Screening for CCHD With the Use of Pulse Oximetry

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

AAP 2016 Expert Panel

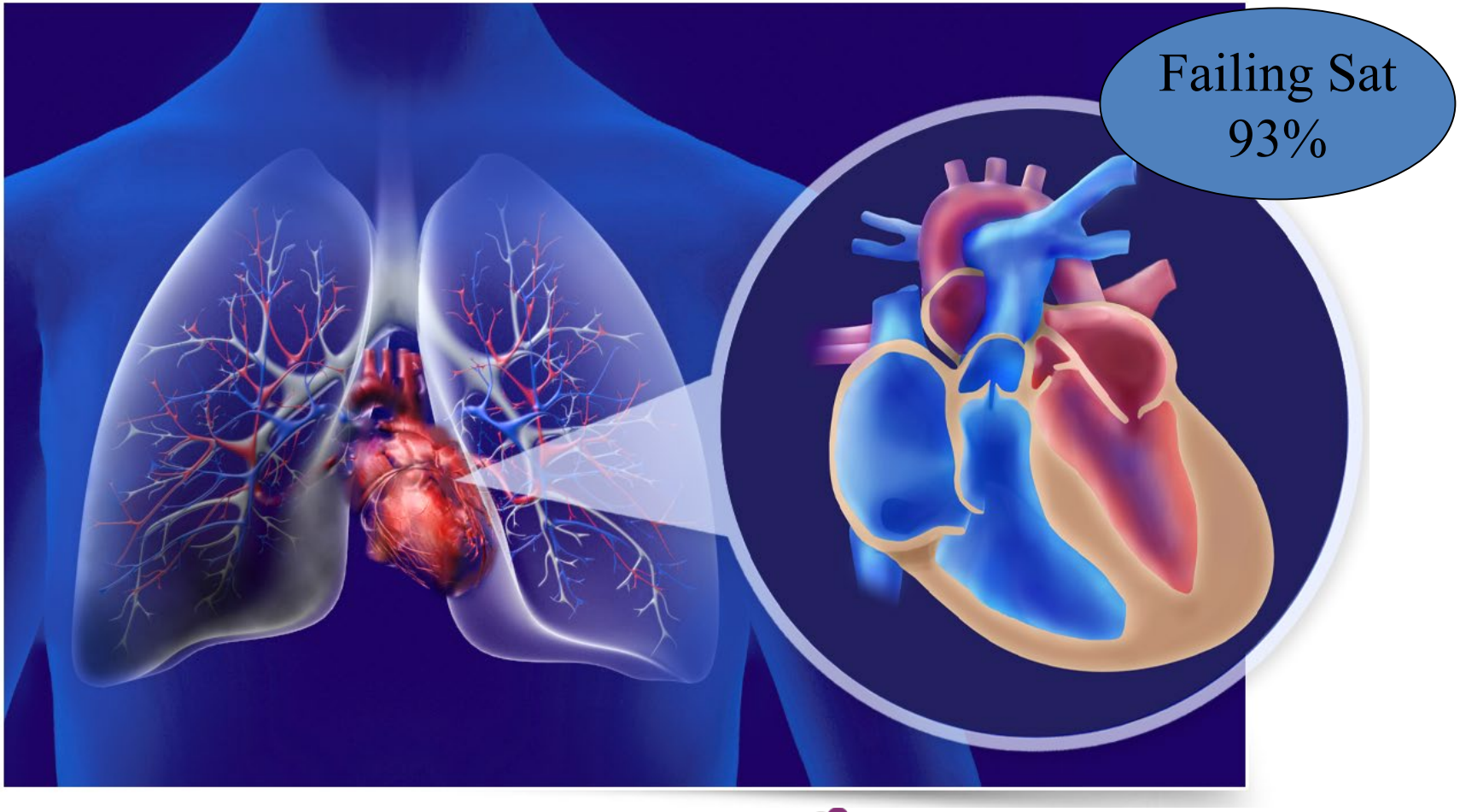
Lessons Learned From Newborn Screening for Critical Congenital Heart Defects

Matthew E. Oster, MD, MPH,[a,b] Susan W. Aucott, MD,[c] Jill Glidewell, APRN, MSN, MPH,[a] Jesse Hackell, MD,[d] Lazaros Kochilas, MD, MSCR,[b] Gerard R. Martin, MD,[e] Julia Phillippi, PhD, CNM,[f] Nelangi M. Pinto, MD,[g] Annamarie Saarinen, MA,[h] Marci Sontag, PhD,[i] Alex R. Kemper, MD, MPH, MS[j]

Pediatrics 137: 2016

NewSTEPs
A Program of the Association of Public Health Laboratories™

Secondary Targets: Pneumonia & Sepsis



NewSTEPs
A Program of the Association of Public Health Laboratories™

EVERY WOMAN
EVERY CHILD

Commitments to support

EVERY NEWBORN

June 2014

World Health Organization

COMMITTING TO CHILD SURVIVAL: A PROMISE RENEWED

ESTIMATED IMPACT 2015–2030

772,000 **Child lives saved**

6% reduction in deaths due to pneumonia

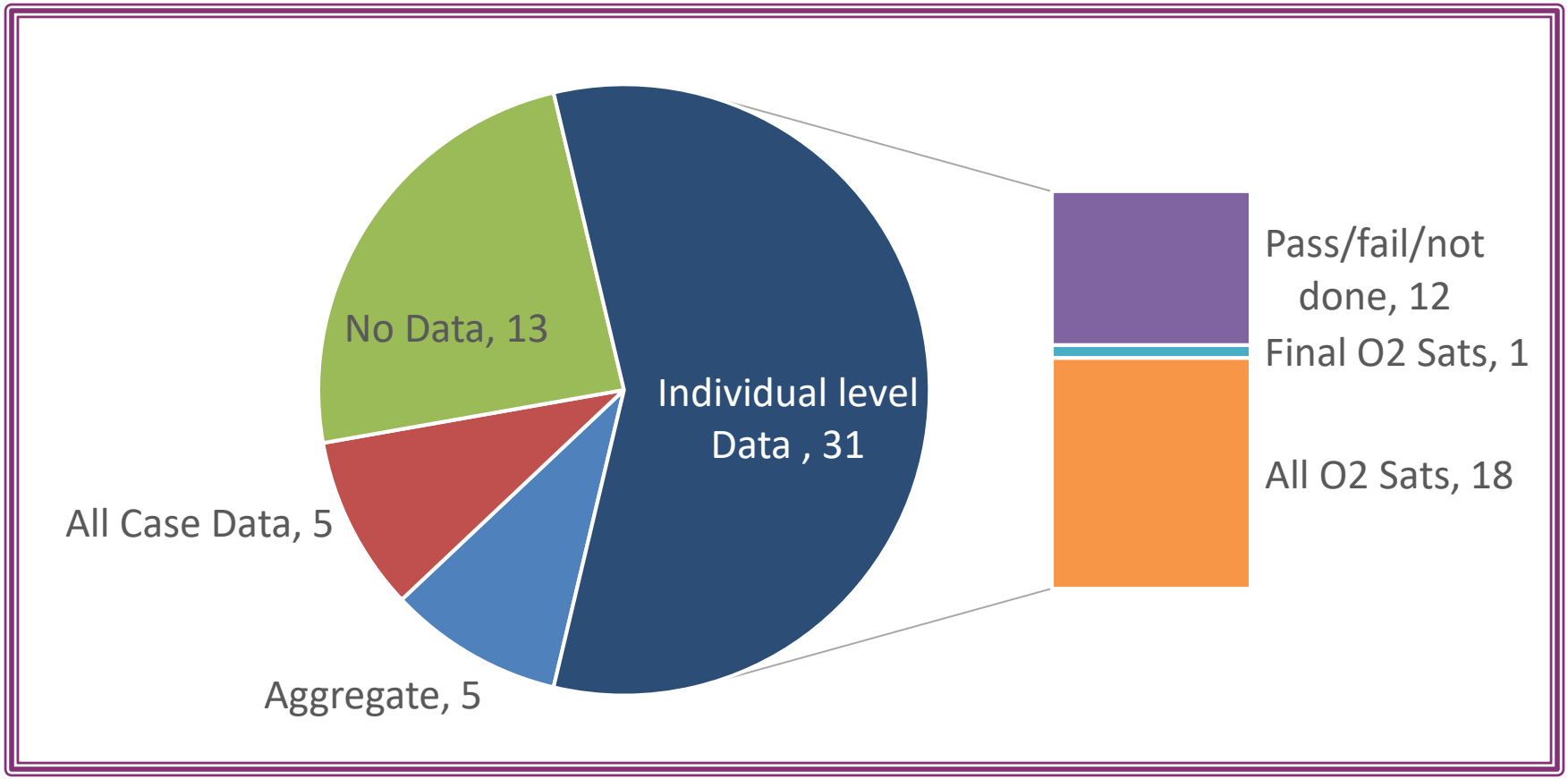
COST	LIVES SAVED SENSITIVITY
$101M	+/- 24,000 for a +/- 5 percentage point change in coverage in the clinic

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.

BORN project
Birth Oximetry Routine for Newborns

What Data are Collected by NBS program?



NewSTEPs
A Program of the Association of Public Health Laboratories™

Aggregate Data

[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.

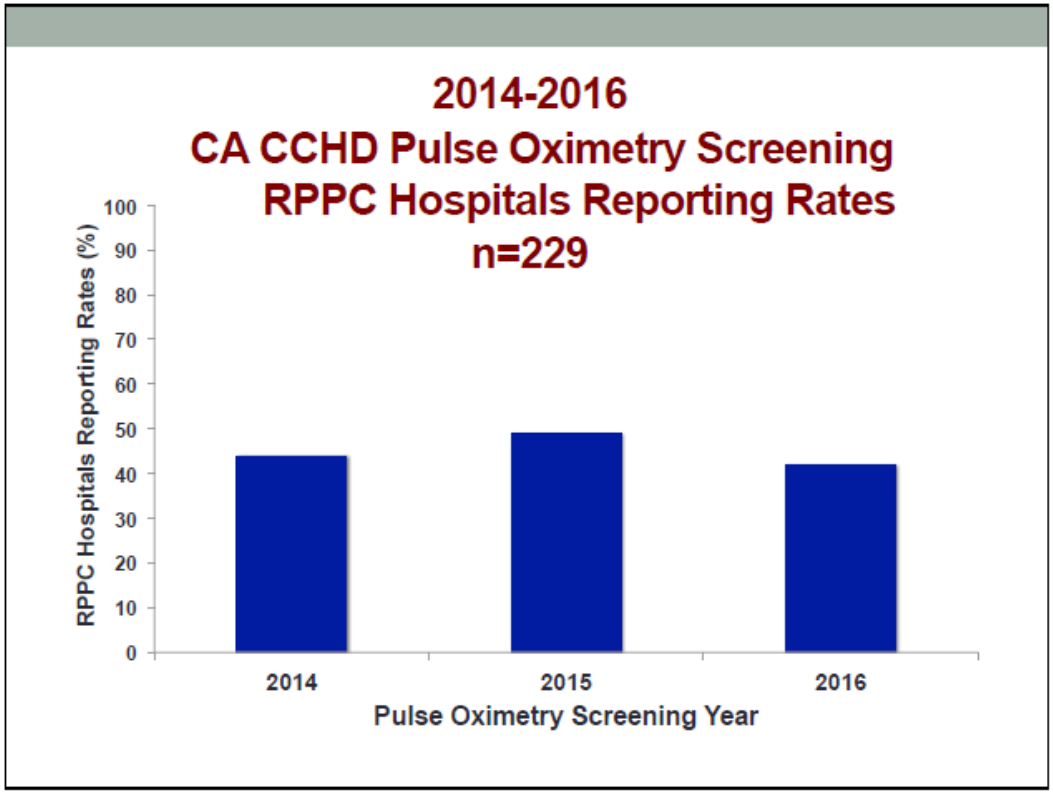
I sit on our child death review committee, so I know when it does not go well. It would be nice to know about the times when it does…

From State "X", Division of Public Health, email response sent August, 2018

NewSTEPs
A Program of the Association of Public Health Laboratories™

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.

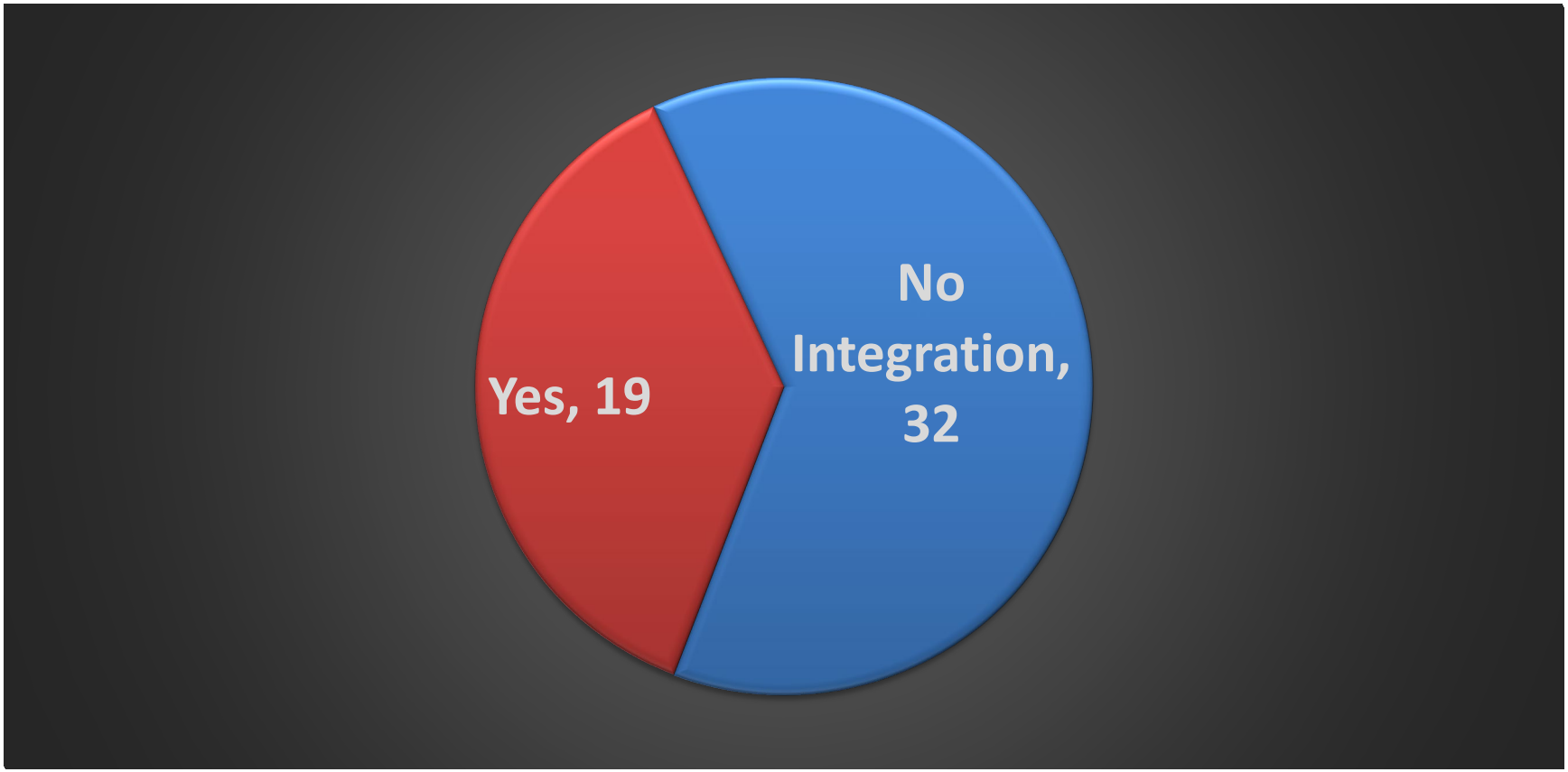
NewSTEPs
A Program of the Association of Public Health Laboratories™

What is counted as a screening find?

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

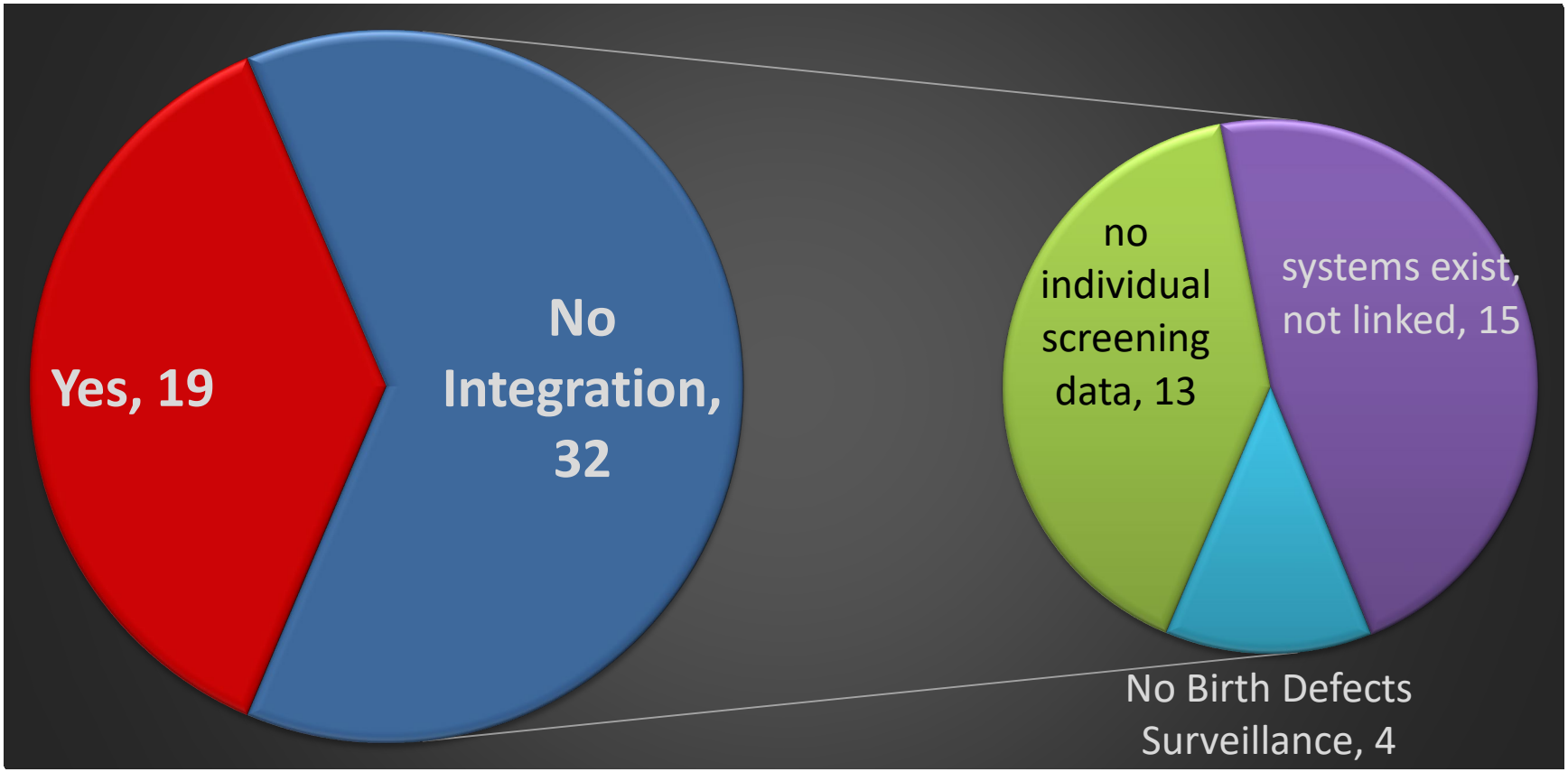
- Are prenatally detected infants excluded?
- Are infants with a prior echocardiogram excluded?
- Clinical assessment leads to early pulse oximetry screen?

Integrated Data System – CCHD and Birth Defects Surveillance



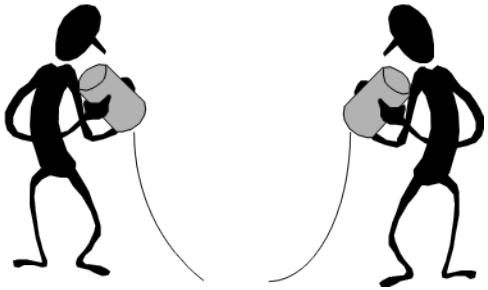
NewSTEPs
A Program of the Association of Public Health Laboratories™

Reasons for *No Integration* Vary



NewSTEPs
A Program of the Association of Public Health Laboratories™

How does bi-directional communication happen between NBS and Birth Defects?



NewSTEPs

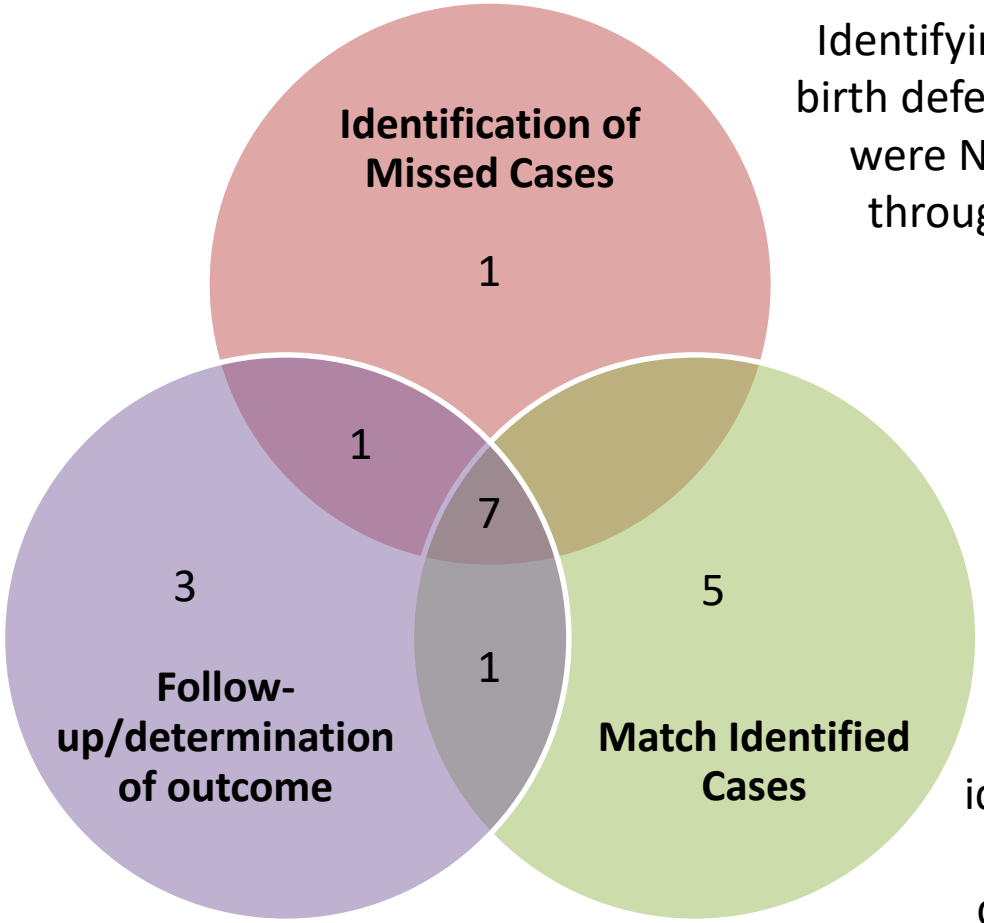
A Program of the Association of Public Health Laboratories™

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**

NewSTEPs
A Program of the Association of Public Health Laboratories™

For those who integrate, how are the data 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

* 1 state unsure of how data were used

NewSTEPs
A Program of the Association of Public Health Laboratories™

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

NewSTEPs
A Program of the Association of Public Health Laboratories™

State variation: population screened

- All newborns
- Special populations/exemptions:
  - NICU
  - births at altitude
  - home births

NewSTEPs
A Program of the Association of Public Health Laboratories™

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

NewSTEPs

A Program of the Association of Public Health Laboratories™

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

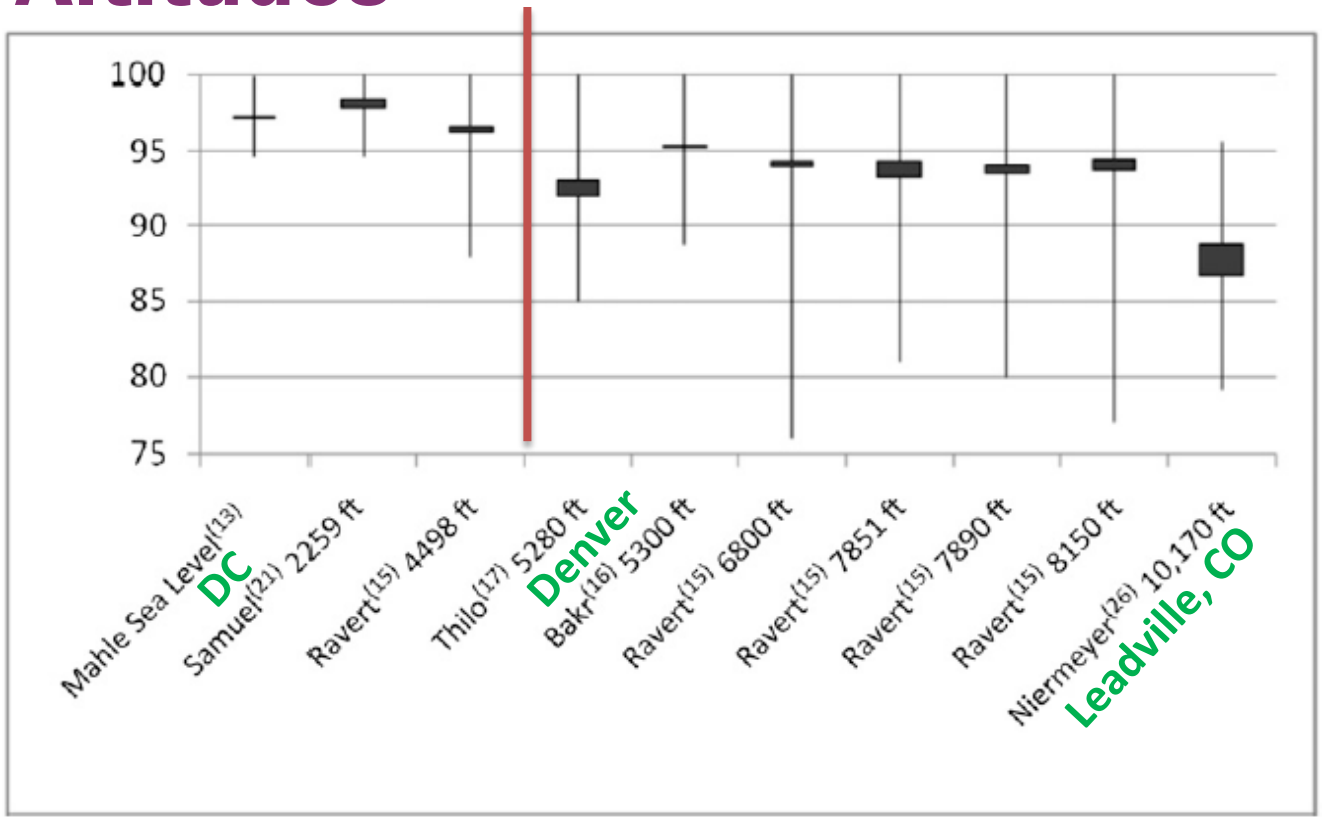


FIGURE 6
All studies used mean saturations ± SD with the exception of Ravert's study[15] who used a mean saturation and saturation range.

Pediatrics 2014;133:e561–e569

NewSTEPs
A Program of the Association of Public Health Laboratories™

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, BS[1], Elizabeth M. Goetz, MD, MPH[2], Jody D. Belling, RN, MSN[2], W. Marijke van Roojen, LM, CPM[3], Gretchen Spicer, LM, CPM[4], and John S. Hokanson, MD[2]

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

NewSTEPs

A Program of the Association of Public Health Laboratories™

“So many protocols, which one is best?”

Algorithm	Extremity Screened	POS for Pass (%)	Difference between arm/leg for Pass (%)	Rescreens (n)	Screen Age (hours)
AAP[10]	RH, Foot	95 in either	≤3	2	>24
New Jersey[18]	RH, Foot	95 in both	≤3	2	>24
Tennessee[24]	Foot (AAP if test Fail)	97	≤3 on rescreen	2	>24
Granelli[5]	RH, Foot	95 in either	≤3	2	<24
Ewer[8]	RH, Foot	95 in both	≤2	1	6-24
Poland[15]	Foot	95		1	<24
Germany[6]	Foot	95		1	>24

Martin *Pediatrics* 2020

NewSTEPs
A Program of the Association of Public Health Laboratories™

U.S. Algorithms

TABLE 2 Common Algorithms for Newborn Screening for CCHD With the Use of Pulse Oximetry in the United States

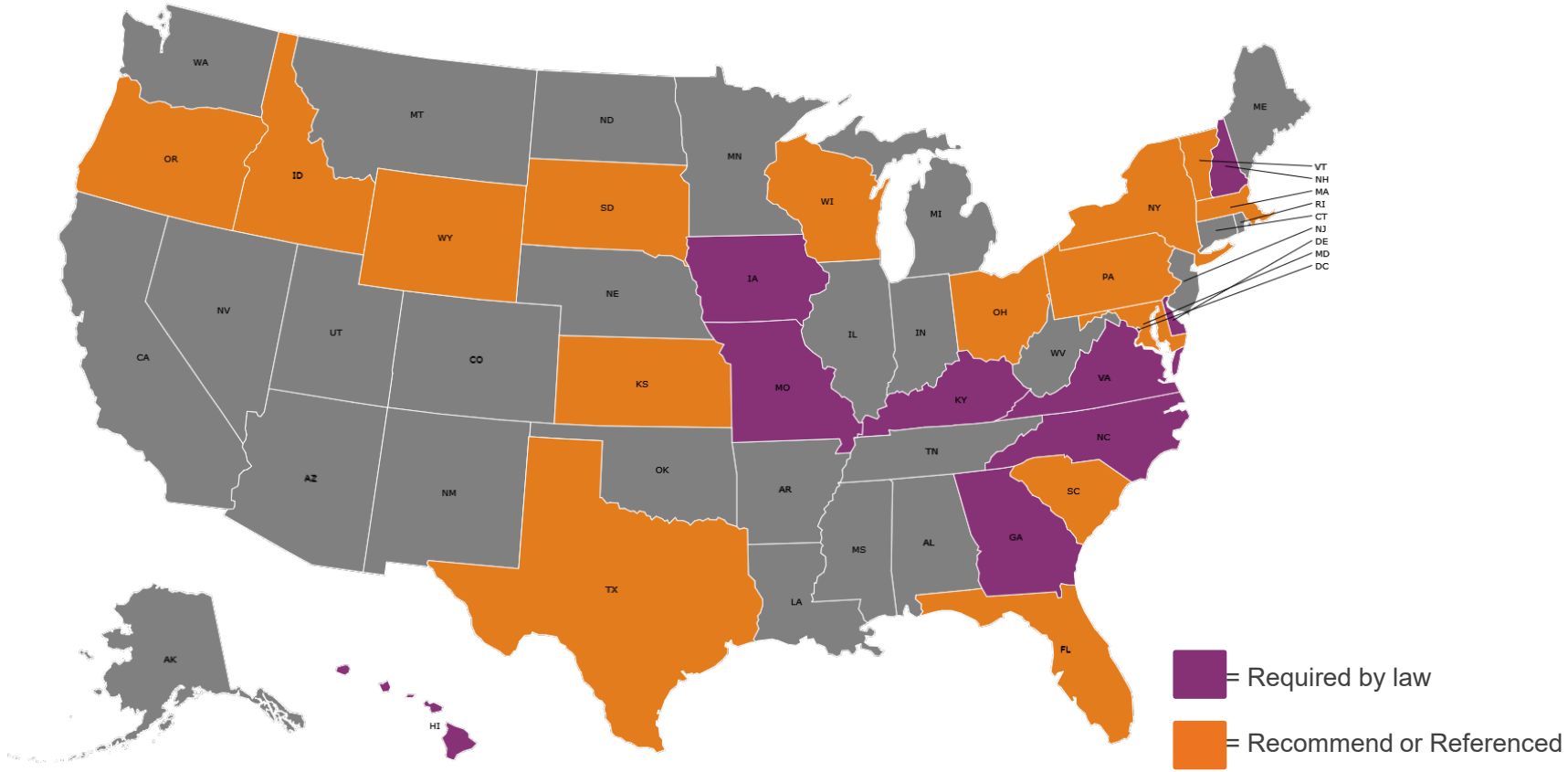
Algorithm Source	Cutoff for Passing With First Measurement	Retest Criteria for Subsequent Measurements	Fail Criteria
AAP	O_2 sat ≥95% (in either RH or F) AND |hand-foot| O_2 sat ≤3%	O_2 sat <95% (in both RH and F) OR |hand-foot| O_2 sat >3%	O_2 sat <90% (either RH or F) OR fail retest criteria × 3
New Jersey	O_2 sat ≥95% (in both RH and F) AND |hand-foot| O_2 sat ≤3%	O_2 sat <95% (in either RH or F) OR |hand-foot| O_2 sat >3%	O_2 sat <90% (either RH or F) OR fail retest criteria × 3
Tennessee	O_2 sat ≥97% (F)	O_2 sat <95% (in both RH and F) OR |hand-foot| O_2 sat >3%	O_2 sat <90% (either RH or F) OR fail retest criteria × 3

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

(Oster Pediatrics 2016)

NewSTEPs
A Program of the Association of Public Health Laboratories™

AAP Protocol U.S. Implementation



Pediatrics, Letter to Editor

https://pediatrics.aappublications.org/content/146/1/e20191650/tab-e-letters

NewSTEPs

A Program of the Association of Public Health Laboratories™

Updated Strategies for Pulse Oximetry Screening for Critical Congenital Heart Disease

Gerard R. Martin, MD,[a] Andrew K. Ewer, MD,[c] Amy Gaviglio, MS, LCGC,[a] Lisa A. Hom, RN, Esq,[a] Annamarie Saarinen, MA,[f] Marci Sontag, PhD,[g] Kristin M. Burns, MD,[a,b,c] Alex R. Kemper, MD, MPH, MS,[h] Matthew E. Oster, MD, MPH[i]

TABLE 1 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

To cite: Martin GR, Ewer AK, Gaviglio A, et al. Updated Strategies for Pulse Oximetry Screening for Critical Congenital Heart Disease. *Pediatrics*. 2020;148(1):e20191650

NewSTEPs

A Program of the Association of Public Health Laboratories™

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

NewSTEPs
A Program of the Association of Public Health Laboratories™

Pulse Oximetry Screening: Future Needs

NewSTEPs

A Program of the Association of Public Health Laboratories™

Three Primary Areas of Need

Data Collection/Integration/Analysis

Follow-Up, Education, and Training

Other Screening/Clinical Considerations

NewSTEPs

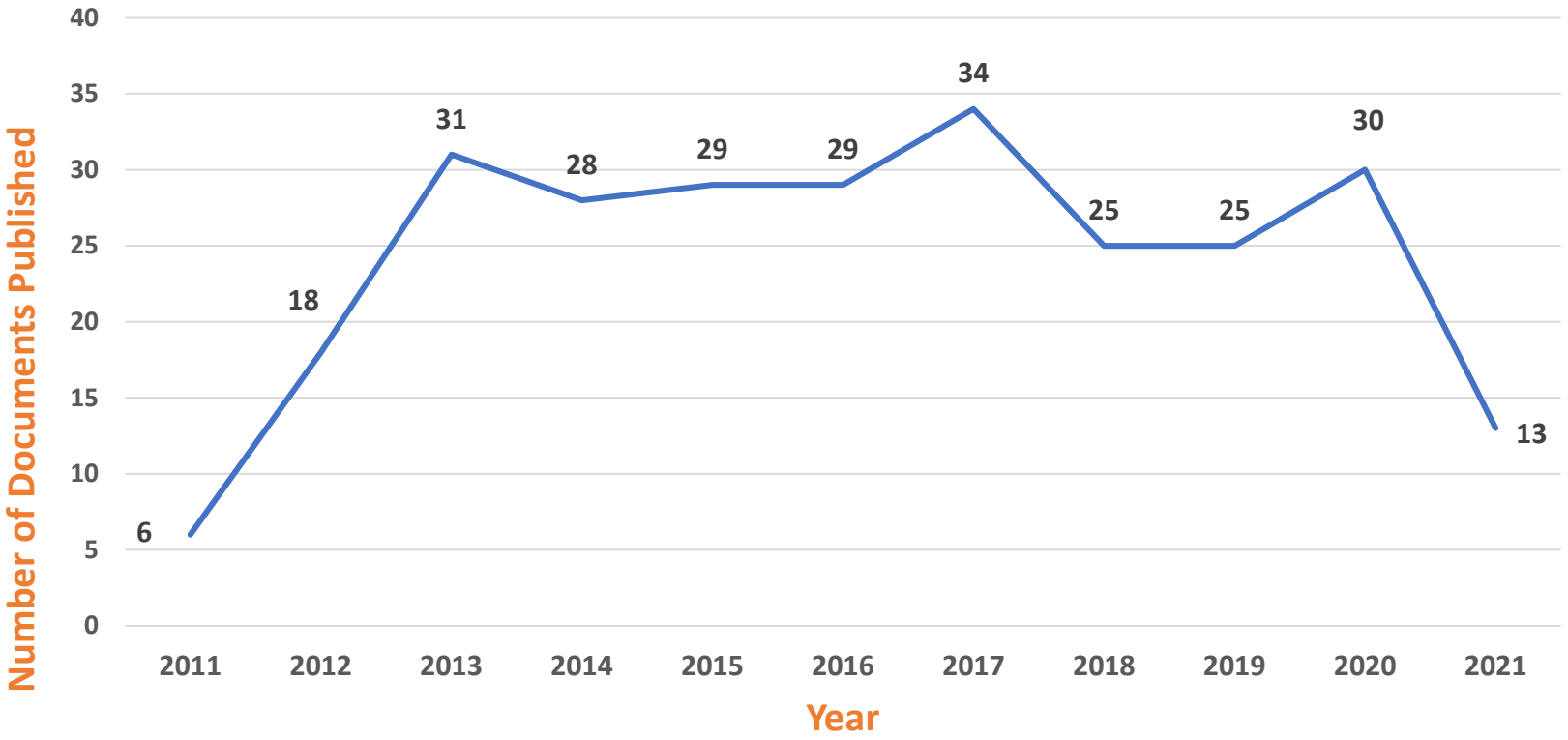
A Program of the Association of Public Health Laboratories™

Data Collection/Integration/Analysis

NewSTEPs
A Program of the Association of Public Health Laboratories™

Where's the Data?

Documents Published 2011-2021; N= 268

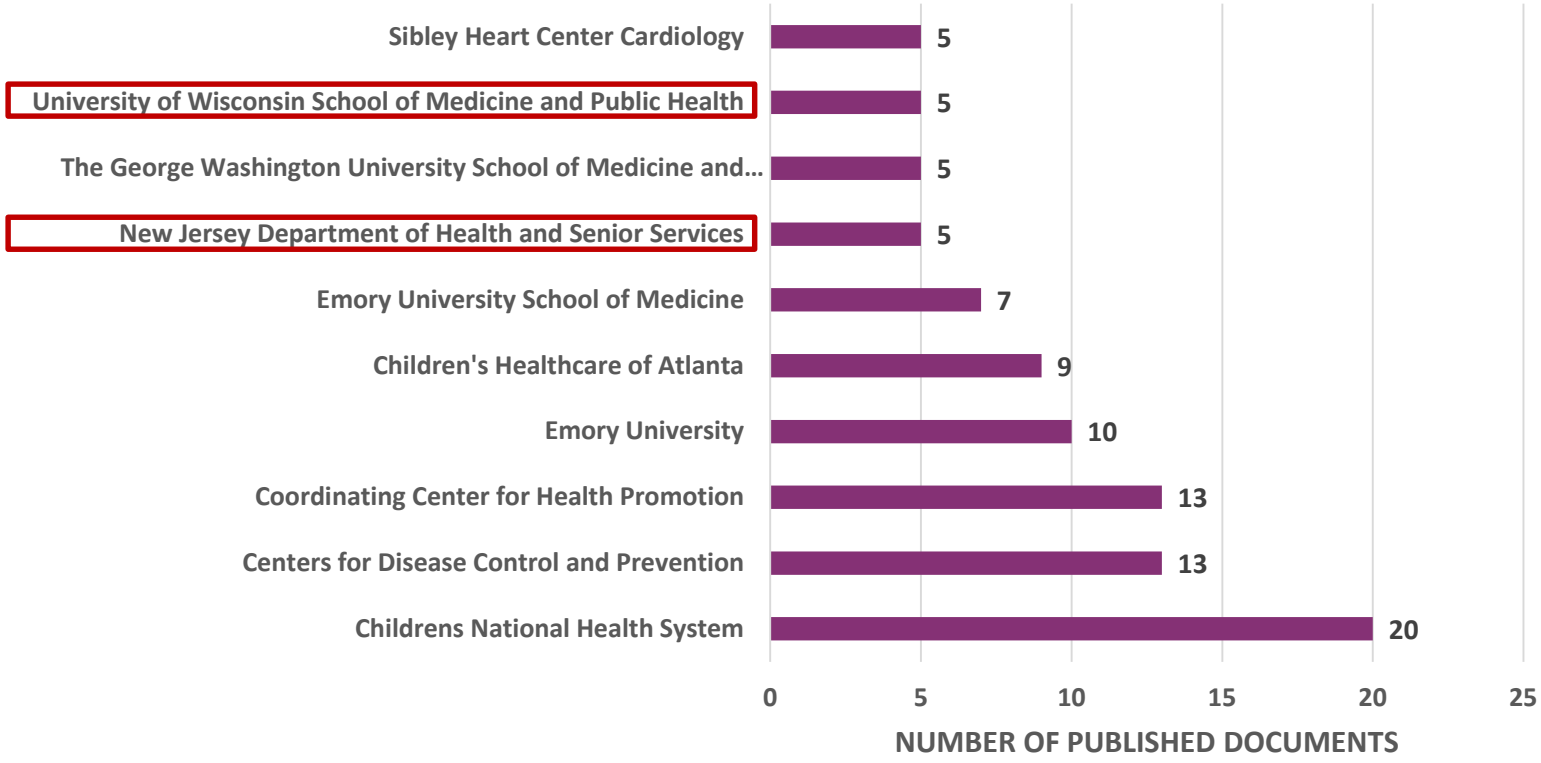


Source: SCOPUS

NewSTEPs
A Program of the Association of Public Health Laboratories™

Where's the Data?

Top 10 Domestic Affiliations 2011-2021; N= 92



Source: SCOPUS

NewSTEPs
A Program of the Association of Public Health Laboratories™

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?

NewSTEPs

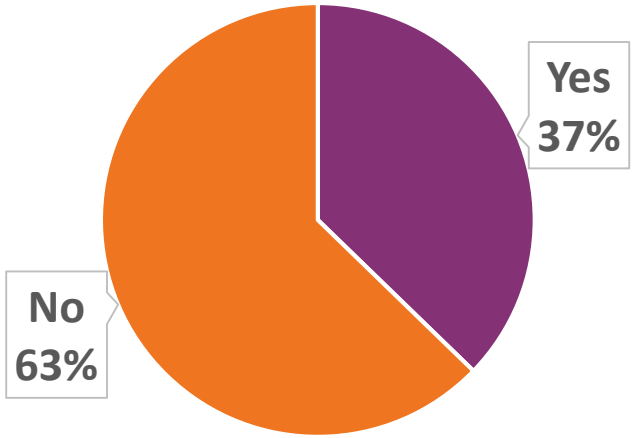
A Program of the Association of Public Health Laboratories™

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



MMWR February 8, 2019 / 68(5);107–111

- **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!!

NewSTEPs

A Program of the Association of Public Health Laboratories™

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

NewSTEPs
A Program of the Association of Public Health Laboratories™

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

NewSTEPs

A Program of the Association of Public Health Laboratories™

So... How Can We Improve Data Collection?

Define and Promote Standard Minimal Data Sets

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

Birth Facility Data	Public Health Program Data
1. Patient-level data: Patient identification data that allows validation that all infants had a valid screen and results Age in hours at time of screening, All oximetry saturations reported (initial screen and any subsequent screens) Final screening result Obstacles encountered during screening process (ie, obstacles with the infant/family, staff, equipment) Diagnostic results 2. Screening program data: Screening protocol being used Type of pulse oximeter used for screening	1. Will vary according to the legislative or executive mandate of each state. 2. Aggregate or individual data may be specified to be provided to and tracked by public health programs 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. 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 5. Ideal for negative screens: Subsequent identification of congenital heart defects (ie, false-negative screens) could be linked within the NBS programs. 6. Summary statistics should be provided by health departments and NBS programs to stakeholders.

NBS, newborn screening program.

Martin, et al (2013) *Pediatrics* (132:1)

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

NewSTEPs

A Program of the Association of Public Health Laboratories™

Develop Public Health Case Definitions

Hypoplastic Left Heart	POX screening should identify all babies with these disorders
Pulmonary Atresia	
Transposition of the Great Arteries	
Single Ventricle	
Truncus arteriosus	
Tricuspid atresia	
Interrupted Aortic Arch with VSD	POX screening may have lower sensitivity due to physiologic variability of oxygen saturations in the newborn
Double-Outlet Right Ventricle	
Aortic Valve Stenosis (with PDA)	POX screening may only detect these cases if the newborn requires intervention in the first 30 days of life
Pulmonary Atresia with Ventricular Septal Defect	
Total anomalous pulmonary venous return	
Tetralogy of Fallot	
Coarctation of the Aorta with PDA	
Ebstein's Anomaly	

Coarctation of the Aorta without a PDA	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.
Aortic Valve Stenosis without a PDA	
Pulmonary Atresia with Ventricular Septal Defect	
Total anomalous pulmonary venous return	
Tetralogy of Fallot	
Ebstein's Anomaly	

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.*

NewSTEPs

A Program of the Association of Public Health Laboratories™

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.

Cardiac Lesion Reported	Public Health Categorization
Cardiac finding (red or yellow); algorithm followed/correct interpretation	Physiological Missed Case
Cardiac finding (green/intervention required within first 30 days of life); algorithm followed/correct interpretation	Physiological Missed Case
Cardiac finding (blue/intervention not required within first 30 days of life); algorithm followed/correct interpretation	Document findings, but is NOT considered a Missed Case
Cardiac finding (any color); algorithm NOT followed/incorrect interpretation	Non-Valid Screen due to Error

NewSTEPs

A Program of the Association of Public Health Laboratories™

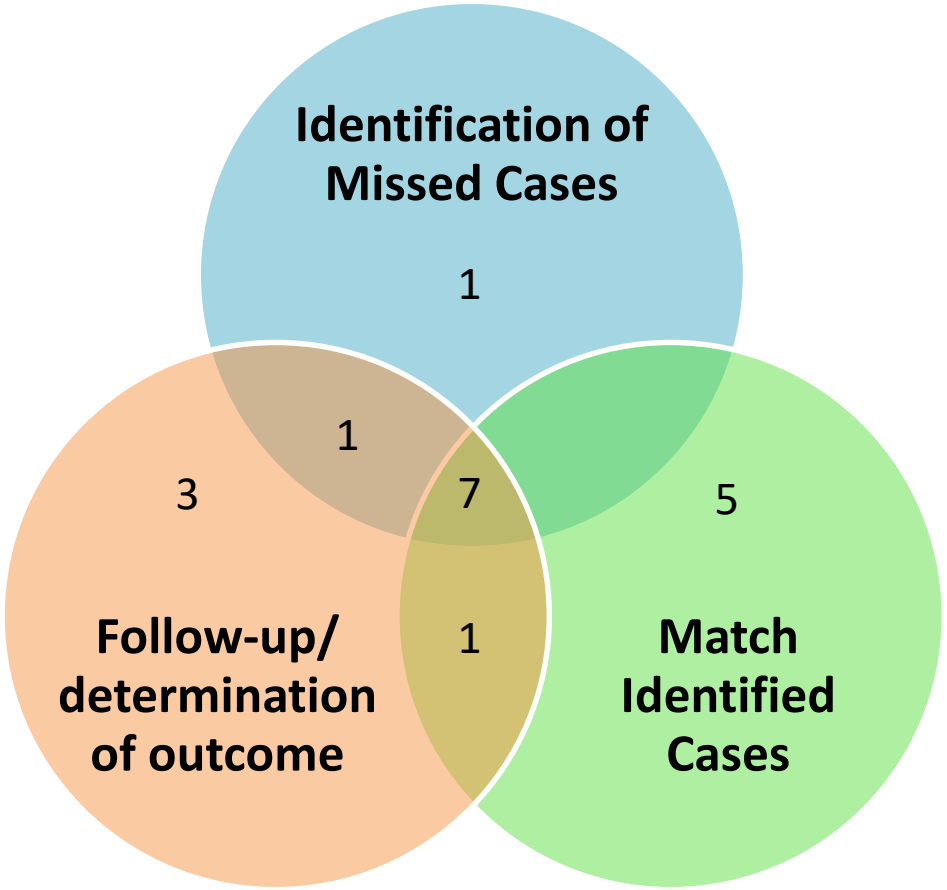
Define What Questions We Should Be Able to Answer

Metric	Question
Screen Rate	What percentage of eligible newborns are getting screened?
Failure Rate	What percentage of newborns fail their pulse oximetry screen?
Detections	What is being detected? Primary and Secondary
Missed Cases	What is not being detected? Why?
Detection Modality	What percentage of cases are detected prenatally, clinically, and via screening?
Other??	

NewSTEPs

A Program of the Association of Public Health Laboratories™

Create Data Sharing and Linkages

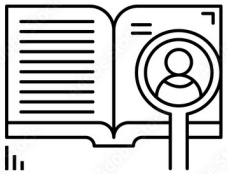


CCHD Screening and Birth Defects Programs MUST work together

NewSTEPs
A Program of the Association of Public Health Laboratories™

Summary of Needs

Standardized Data Recommendations



Improved Case Understanding and Definitions

Enhanced Intra-agency Data Linkages

NewSTEPs

A Program of the Association of Public Health Laboratories™

Follow-Up, Education, and Training

NewSTEPs

A Program of the Association of Public Health Laboratories™

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?**

NewSTEPs
A Program of the Association of Public Health Laboratories™

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?

NewSTEPs
A Program of the Association of Public Health Laboratories™

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?

NewSTEPs
A Program of the Association of Public Health Laboratories™

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?

NewSTEPs
A Program of the Association of Public Health Laboratories™

Family Education and Needs

- Are we meeting the needs of families identified with CCHDs? Do we know what the needs are?

TEPS

ciation of Public Health Laboratories™

Other Screening/Clinical Considerations

NewSTEPs
A Program of the Association of Public Health Laboratories™

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]

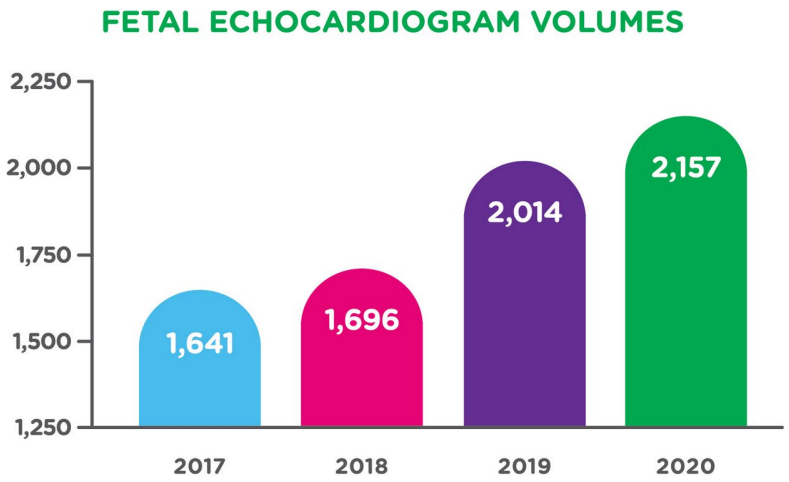
NewSTEPs

A Program of the Association of Public Health Laboratories™

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?



Children's Healthcare of Atlanta

NewSTEPs
A Program of the Association of Public Health Laboratories™

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

Contact Us

www.newsteps.org

Careema Yusuf
Careema.Yusuf@aphl.org

Lisa Hom
lhom@cnmc.org

Amy Gaviglio
amy.gaviglio@outlook.com

NewSTEPs
A Program of the Association of Public Health Laboratories™