

EMPOWERS.

SUPPORTS.

INFORMS.



# What We Can Learn From Newborn Screening (NBS) and Family Experiences

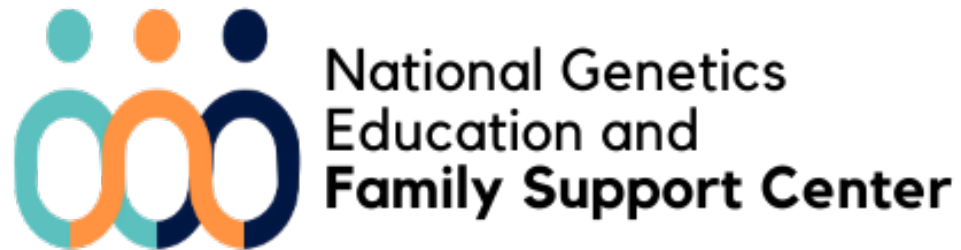




## VISION

The fear and confusion individuals and families face during the prenatal, neonatal, and early childhood years is **replaced by confidence and agency to make the best healthcare decisions for their lives.**

# Our Newborn Screening/Genetics Portfolio



who are building community

and changing the system.

## At the Heart of Newborn Screening: Families Making Change



# WHY DO BABIES NEED NEWBORN SCREENING?

- All babies born in the United States have newborn screening.
- These tests could save a baby's life .
- While most babies are born healthy, some babies are born with serious, but treatable medical conditions.
- Newborn screening helps identify babies who may be at risk of having serious health issues.
- Learning if a baby is at risk for one of these conditions can help prevent more serious medical problems from happening.

Slides Adapted from a module of the Newborn Screening Family Education Program

# WHAT IS NEWBORN SCREENING?

*Newborn screening is a group of screening tests that help the baby's doctor identify and treat these conditions before they make babies sick.*

- While most babies are born healthy, some babies are born with serious, but treatable medical conditions.



**The conditions found through newborn screening can be present in any family, even those without a family history of them.**

- Newborn screening is typically done in the first 24-48 hours after the baby is born.
- Newborn screening includes a blood sample from the baby's heel as well as screening of the baby's hearing and heart to look for different conditions that may cause serious health problems.
- Knowing if the baby is at risk can help make sure he or she gets treatment quickly.



# WHAT TO EXPECT: THREE PARTS TO NEWBORN SCREENING

1

**BLOOD TEST**



2

**PULSE OX**



3

**HEARING TEST**



# WHICH CONDITIONS ARE INCLUDED IN NEWBORN SCREENING?

- **Every state requires newborn screening.** Each state is different and decides which conditions will be included in their screening based on the recommended list provided by the United States Secretary of Health and Human Services (**R**ecommended **U**niformed **S**creening **P**anel – **RUSP**).
- Most states screen for at least 31 of the 35 current core conditions recommended by the United States Secretary of Health and Human Services. Some states screen for over 60 conditions.
- Want to know which conditions are included in your state? You can learn more at [Baby's First Test](#) and the HRSA [Newborn Screening Information Center](#).





# WHAT HAPPENS IF A BABY RECEIVES AN ABNORMAL RESULT?

(Information we provide to parents)

## 1. Don't panic, but act quickly.

It is okay to be scared. But it is important to remember that an abnormal, or out-of-range result, does not mean that a baby has a condition. In fact, some babies with abnormal newborn screens have follow-up test results within the normal range.

## 2. Follow up with the baby's doctor.

Don't delay! If a baby has an abnormal, or out-of-range result, it is very important to follow the baby's doctor or state public health program's instructions quickly and carefully.

## 3. Complete confirmatory testing.

Since newborn screening does not confirm if a baby has a condition, it is very important to complete any additional tests that are recommended. This will help find out if the condition is truly present. Talk to the baby's doctor or state public health program about next steps.





# **NEWBORN SCREENING AT THE STATE LEVEL**

# THE ROLE OF STATES IN NEWBORN SCREENING

- **All states have a law/laws mandating newborn screening**
  - Most states offer options for parents to refuse screening
- Newborn screening programs are organized, governed, and administered by State Public Health Departments within each state's legislative process and regulatory framework
- Fee is often set in statute and is based on cost estimates from programs on financial needs for testing and follow up
- **Decision-makers:**
  - State Legislatures
  - State Health Officers
  - State Boards of Health
  - Advisory Committees



# WHY IS SCREENING A STATE ISSUE?

- Historical
- Various state priorities
- Population, funding, resources, and definition differences

## State and Federal: Working Together

- **State**
  - Create and manage newborn screening program
- **Federal**
  - Support with funding, promoting the development of policies and guidelines, and helping with program improvement and quality assurance



# STATE NEWBORN SCREENING PROGRAMS/ BOARDS OF HEALTH

## Day to Day Operational Concerns:

- **Educating public/families/healthcare professionals**
- Getting specimens (blood spots) to the lab
- Testing methodology
- Reporting abnormal or out-of-range results
- Short term follow-up
- Quality control/assurance
- **Deciding what conditions to screen for**
- **Storage and use of specimens**



# PROCESS AT THE STATE LEVEL- WHY SO DIFFERENT?

*Every state process is different, but these are just a few of the steps and questions that states consider when planning their newborn screening program.*

## Four Main Steps

1

2

3

4

### TRANSPORT

States ensure that blood samples are received in a timely manner and consistently throughout the state.

#### State Considerations:

- How large is the state?
- Use a hospital's courier systems?
- Pay for delivery?

### TESTING

#### State Considerations:

- High sensitivity/ specificity
- High efficiency
- First tier vs second tier testing
- Second screen
- Complete screens

### REPORTING

#### State Considerations:

- Who reports results?
- Who receives the results?
- How are results reported?
- What information is provided?

### FOLLOW UP

#### Short Term Follow Up

- States ensure babies with abnormal or out-of-range screens get needed follow up

#### Long Term Follow Up

- States ensure babies with confirmed diagnoses have access to resources
- States vary in definition



# CONNECTING WITH THOSE IN YOUR STATE



Slides Adapted from a module by the Newborn Screening Family Education Program

1. Connect with local groups:
  - March of Dimes
  - State American Academy of Pediatrics (AAP) chapter
  - Regional Genetic Services Network
  - Support groups/Rare Disease organizations
2. Connect with your state's newborn screening program manager/coordinator
  - Visit [BabysFirstTest.org](http://BabysFirstTest.org) or your state's website for more information
3. Attend your state's advisory committee meeting
  - Check with your state. These meetings are open to the public. Not all states have advisory committees.

***Individuals, families, and legislatures make  
a BIG difference!***



# What is Baby's First Test?



About Newborn Screening

Your Baby's Screening

Living With Conditions

Resources

Your State

Find a Condition

En Español

Health Professionals

Programs and Policy

About Us

Search

## What is Newborn Screening?

Many parents are unaware of the conditions included in screening, or that it varies from state to state. **Baby's First Test** brings together resources to help guide parents and health



### What Your State Offers

Every state has its own Newborn Screening program. Learn about it.

- Select State -



### Find a Condition

Get information about the 80 screenable conditions.

Type a Condition



#2021NBS

## Celebrate Newborn Screening Awareness Month

Share what newborn screening means to you!



### YOUR NEWBORN SCREENING GUIDE TO Newborn Bloodspot Screening

#### What is newborn screening?

While most babies are born healthy, some infants are born with serious but treatable medical conditions. Newborn screening is a public health program that helps healthcare providers identify these conditions before they make a baby sick. Newborn screening usually happens 24 hours after your baby is born and can lead to treatment within the first few days of life.

#### What is bloodspot screening?

Newborn bloodspot screening (sometimes called the "heel poke") is one part of the newborn screening process, in which a small amount of your baby's blood is collected from the heel. The blood is placed on special filter paper and sent to a specialized laboratory for testing.

#### Why is newborn screening important?

Some babies look and act healthy at first, but may have health problems that are causing harm, even before they have symptoms. **These conditions can be present in any family, even those without a family history.**

Alaska's newborn screening program screens for more than 50 different conditions.

#### Did You Know?

Every year, 1 in 12 Alaska babies are diagnosed with a newborn screening condition. When treated early, babies identified through newborn screening can grow up healthy with normal development.

If you have concerns about your baby's health, contact your baby's healthcare provider right away.

For More Information:

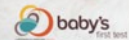
Website:

<http://bit.ly/2InUUNG>



## Community of Newborn Experts Collaborating Together

A collaborative workgroup on congenital cytomegalovirus and newborn screening



@alaska.gov

400

Resources:  
st.org  
rstTest.org



Glance





**What is the public hearing about newborns,  
genetics, and the future?**

HEALTH >

# Privacy concerns prompt states to reexamine storing newborns' heel blood tests

BY MICHELLE ANDREWS

SEPTEMBER 13, 2022 / 5:00 AM / KAISER HEALTH NEWS



Pharma & Life Sciences

# Michigan Parents Mostly Prevail in Newborn Blood Screening Suit

Sept. 14, 2022, 3:26 PM



- Every state and territory has newborn screening requirements
- Ruling turns on parents' right to direct medical care



**Mary Anne Pazanowski**  
Legal Reporter



# Public Health Perspectives on Genetics

Open Access **Commentary**

## Newborn Screening Is on a Collision Course with Public Health Ethics

by  Robert J. Currier  

Department of Pediatrics, University of California, San Francisco, CA 94158, USA

Academic Editors: Lynn Wein Bush and Olaf Bodamer

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Open Access **Article**

## Genomics and Newborn Screening: Perspectives of Public Health Programs

by  Aaron J. Goldenberg <sup>1,\*</sup> ,  Roselle Ponsaran <sup>1</sup>  ,  Amy Gaviglio <sup>2</sup> ,  Dalton Simancek <sup>3</sup>  and  Beth

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Research | [Open Access](#) | [Published: 31 March 2022](#)

## Pain points in parents' interactions with newborn screening systems: a qualitative study

[Mike Conway](#) , [Truc Thuy Vuong](#), [Kim Hart](#), [Andreas Rohrwasser](#) & [Karen Eilbeck](#) 

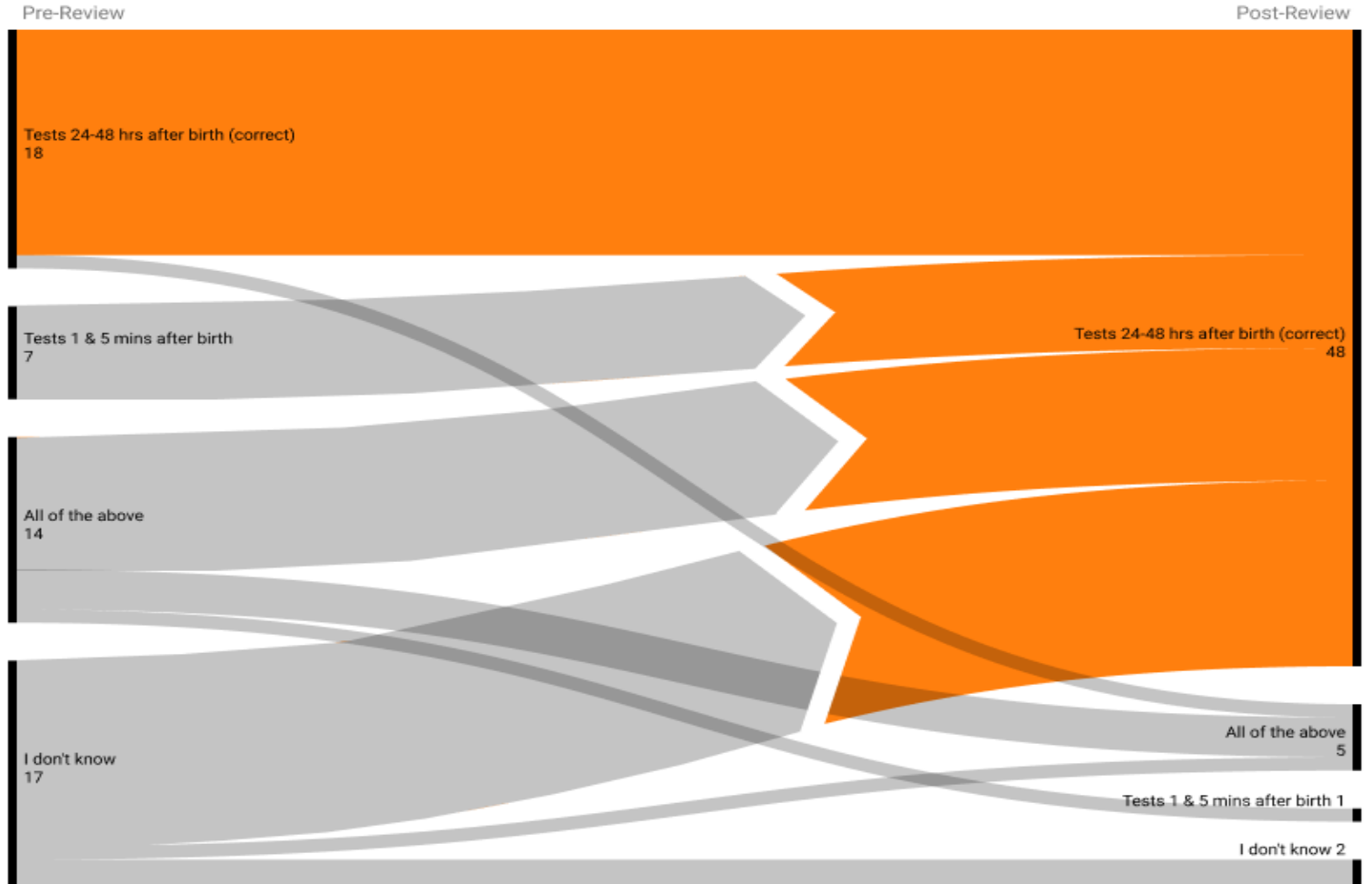
*BMC Pediatrics* **22**, Article number: 167 (2022) | [Cite this article](#)

1024 Accesses | 2 Citations | 9 Altmetric | [Metrics](#)

# Educational Initiatives do work

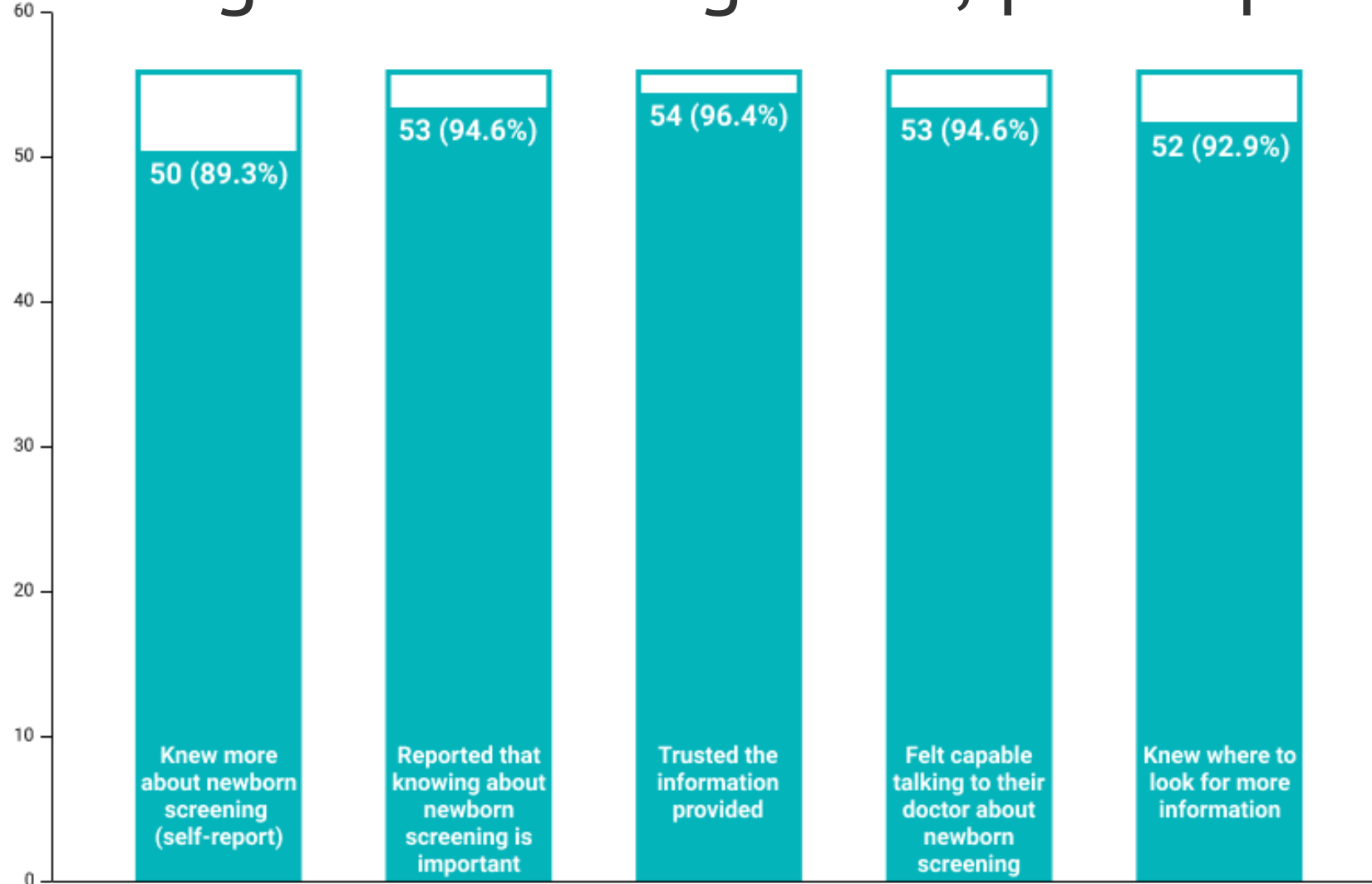
Most participants learned the definition of newborn screening.

What is newborn screening?



*Difference between pretest and posttest is statistically significant at  $p < .001$  using a Wilcoxon signed rank test for nonparametric data.*

# After reading the learning book, participants...



Results reflect participants who agreed or strongly agreed with each statement.



# What is on the radar?

- Recommended Uniform Screening Panel (RUSP) Alignment legislation
- Increase privacy/ownership concerns
- Workforce deficits
- Lack of system-wide investments/upgrades





## *Stay Connected*

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## *Follow Us*

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