

SCREEN BABY SCREEN: PERSPECTIVES ON NEWBORN SCREENING

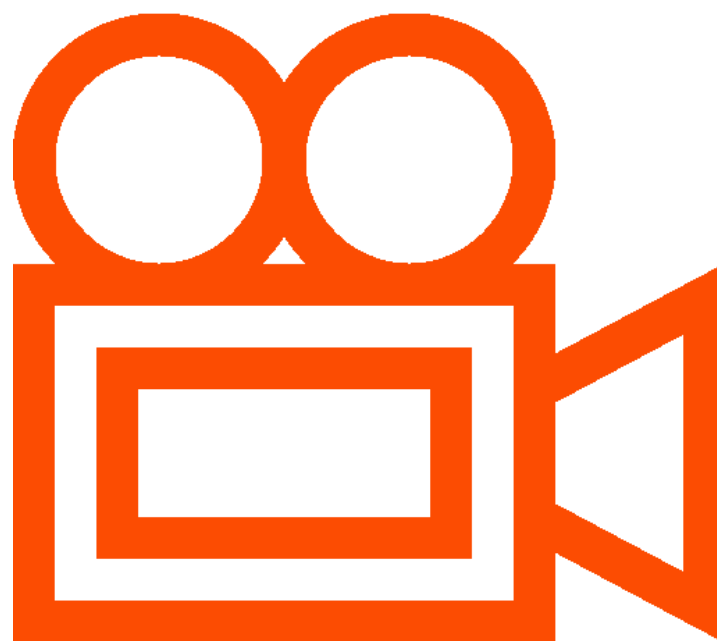
For sound, stream audio through your speakers.

If you are having trouble accessing sound, please send a message using the chat box on the left hand side of the screen.

Alone we are rare. Together we are strong.®



This webinar is being recorded.



Question and Answer Session

Submit your questions using the chat function.
It can be found at the **left hand side** of the window.



Newborn Screening Awareness Month

September is Newborn Screening Awareness Month!



Learn more at rarediseases.org



rarediseases.org



ENTERING A NEW ERA

VIRTUAL EVENT

October 8-9, 2020

#NORDSUMMIT | nordsummit.org

Rare Diseases and Orphan
Products Breakthrough Summit®



rarediseases.org

Speakers



Jose Abduner, MD
Medical Director, Pediatric
Metabolic Disorders
Children's Hospital Orange County



Danyelle Sun, MSW
Rare Mom
Wisconsin State Ambassador
NORD Rare Action Network



Rachel Sher, JD, MPH
Vice President, Policy and
Regulatory Affairs
NORD



National Organization for Rare Disorders

Newborn Screening

J. Abdenur, M.D.

Chief, Division of Metabolic Disorders

Director, Metabolic Laboratory



August 27, 2020

Objectives

- To inform rare disease patients, caregivers and the public about newborn screening
- To communicate the importance newborn screening as a public health priority to prevent premature illness and death

Definitions

- Newborn Screening (NBS) in the US is a Public Health program aimed at the early identification of conditions for which early and timely interventions can prevent or reduce associated mortality and morbidity

Adapted from “Newborn Screening Task Force Report”
Pediatrics 2000; 106: 383-427

The “Guthrie” Newborn Screening Card

- Obtained at the birth hospital
- Usually 24-24 h after delivery



Utah Newborn Screening


SEE BACK FOR BLOOD SPOT COVER

SECOND SCREEN:
General Instructions

Collect specimen after 7 days of life.

COLLECTION INSTRUCTIONS

- Legibly print ALL information in spaces provided using block capital letters.
- Collect specimen with heel stick. See newborn screening handbook for detailed instructions.
- Fill all 7 circles.
- Dry 3-4 hours before mailing.



COLLECT SAMPLE FROM SHADED AREA.

Note: Expiration Date Form CANNOT be used after this date.

Mailing Instructions:

- When blood is dry, fold stock card (from back of form) over blood spots. The flap should enclose the blood spots and reveal a biohazard symbol.
- If using the postal service, place form with blood spots covered into envelope.

Mail to: Newborn Screening Laboratory
Utah Department of Health
49 N Medical Dr.
Salt Lake City UT 84113-9903
Phone: (801) 584-8256

Mother's Information*:
This is used to identify the baby and mother, and to contact mother if necessary.

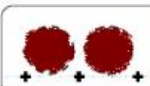
For more information, call, refer to your handbook or visit our website:
http://www.health.utah.gov/newbornscreening/HCP_instructions.htm

Retain this sheet for your records.

XXXXXXX
L.D. Number

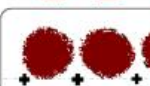
PEEL AWAY THIS PART HERE

2




FOR UDHOH LAB ONLY - DO NOT MARK

XXXXXXX



FOR UDHOH LAB ONLY - DO NOT MARK

XXXXXXX



FOR UDHOH LAB ONLY - DO NOT MARK

XXXXXXX

UTAH DEPARTMENT OF HEALTH
SECOND NEWBORN SCREEN FORM
PRINT ALL CAPS - COMPLETE ENTIRE FORM
FORM EXPIRES DECEMBER 31ST

Patient Record Number: SMITH Baby's full name: JOHN M

Your Hospital: YOUR HOSP Date of birth: 01/01/2007

☒ Adult
 ☐ Infant
 ☐ Transferee
 Write date of transfusion: 3/5/20
 Birthdate (year): 8520

SMITH JANE Mother's last name
 DOE Father's last name
 44 N MEDICAL DR Mother's home address
 SLC UT 84114
 01/02/1986 8015848256 Mother's phone (area code & phone)
 JIM JONES Baby's home address (last name, street address)
 1234 S MEDICAL DR Baby's home phone (area code & number)
 SLC UT 84104 Baby's phone (area code & number)
 801-584-8256 Baby's phone (area code & number)

RECALL SCREEN INFORMATION INSTRUCTED
☐ Unacceptable
 ☒ Positive
 RECALL FOR UDHOH LAB ONLY - DO NOT MARK
 Sample Unacceptable ☐

Fill all 7 circles with blood. The top two blood spots are sent off-site and MUST be filled in addition to the five below.

FOR UDHOH LAB ONLY:
Do not mark or place labels in any of these areas.

Sample Collection Date:
Specimen cannot be processed without this date.

Birthdate:
Several results are based on the baby's age and cannot be processed without the birthdate.

Birth Weight:
Several results are based on birth weight and cannot be processed without it.

Medical Home Information:
Who is called in case of an abnormal screen and where to send results.

Recall Screen Box:
Mark only if instructed by program staff. This is used if another specimen is needed because of specimen being unacceptable or abnormal.

Used by UDHOH lab to mark a specimen unacceptable.

The “Guthrie” Newborn Screening Card

- Sample “vehicle” for collection, transport and storage
- Source of valuable information about the newborn

Infant’s name, birth order

Initial or repeat sample

Race and ethnicity

Birth date, hour

Sample collection date and hour

Feeding type

Transfusion date, hour

Mother’s maiden and surname

Mother’s address and telephone

Hospital ID number

Submitter’s contact information

Sample collector’s initials

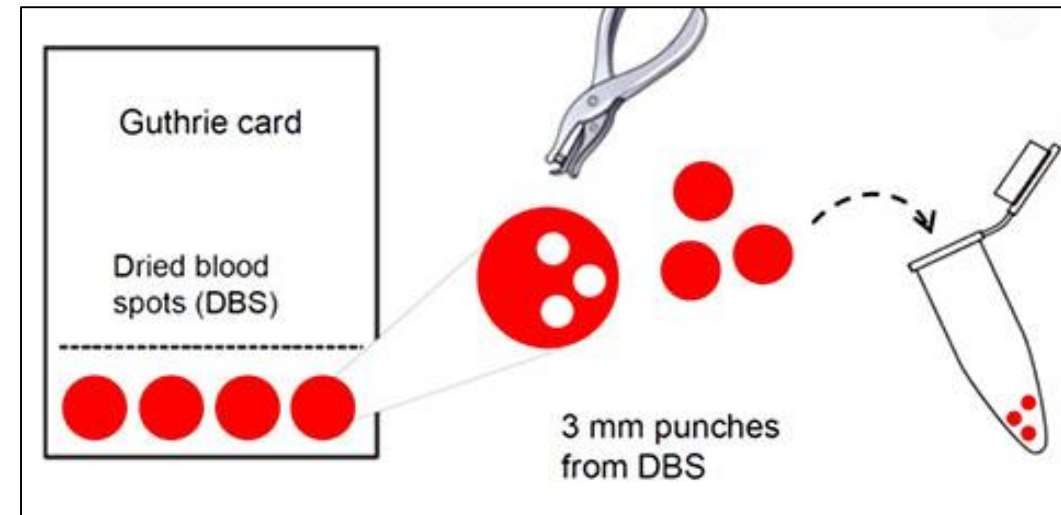
Health care provider’s contact

Hearing screening results (if available)

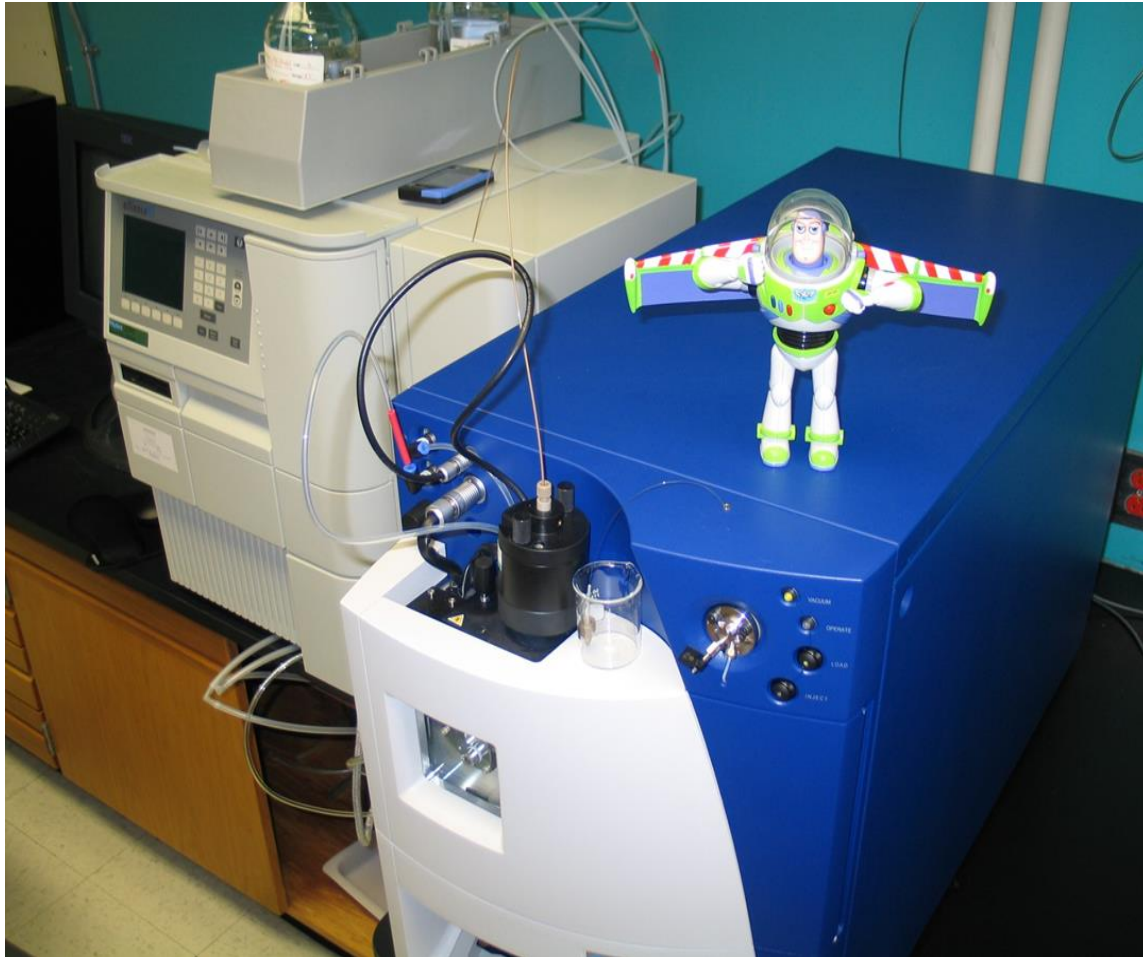
The “Guthrie” Newborn Screening Card

From a 9mm blood spot, small samples are punched and are suitable for testing with a variety of methods

- Fluorometric
- Colorimetric
- Electrophoresis
- Enzymatic analysis
- Molecular testing
- **LC/MS-MS (Tandem MS)**



Tandem Mass Spectrometry



Metabolic Disease Panel
(aminoacids & acylcarnitines)

> 30 Diseases

● One dot of blood

Rapid Preparation & Analysis

High Sensitivity

High Specificity

Which diseases should be included
in a NBS program ?

Criteria for NBS Program: Disease

- “Significant” frequency in the population
- Known clinical course

Significant morbidity / mortality if untreated

Neonatal, early onset manifestations ?

- Available effective treatment

Criteria for NBS Program: Testing

- **Diagnostic methodology**

- Small sample

- Easy collection and transport

- High sensitivity (no false negatives)

- High specificity (few false positives)

- High throughput methodology

- Cost Effective

- Available QC and PT programs

- **Confirmatory tests**

- Easily available

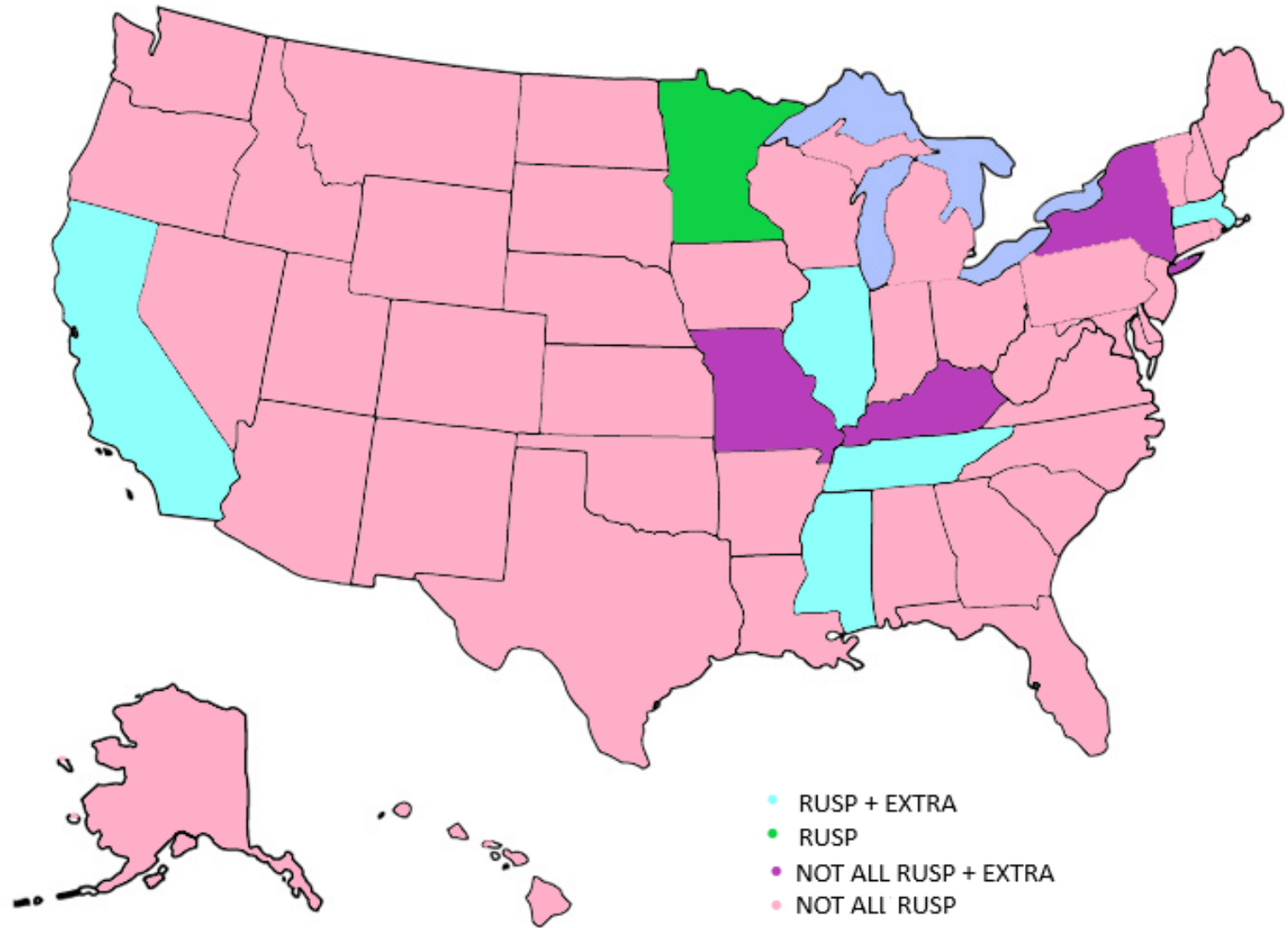
- Rapid results

Recommended Universal NBS Panel (RUSP)

- List of disorders recommended by the **Secretary, Department of Health and Human Services** (HHS) to the states (based on **Advisory Committee** Recommendations)
- Disorders Chosen based on evidence that supports the potential benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments
- Currently: **35 Core** Conditions and **26 Secondary** conditions (disorders that can be detected in the differential diagnosis of a core disorder).
- Newer conditions are still in process of adoption
- States have autonomy, but generally follow HHS recommendations for their NBS
- Some states also screen for additional disorders (not included in RUSP)

<https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html>

Conditions included in State NBS Programs



HHS - Health Resources and Service administration (HRSA)

[Home](#) > [Advisory Committees](#) > [Advisory Committee on Heritable Disorders in Newborns and Children](#) > [Recommended Uniform Screening Panel](#)

Advisory Committees on Heritable Disorders in Newborns and Children

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[Recommended Uniform Screening Panel \(RUSP\)](#)

[Nominate a Condition](#)

[Previously Nominated Conditions](#)

[Newborn Screening Timeliness Goals](#)

Recommended Uniform Screening Panel

The RUSP is a list of disorders that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their state universal newborn screening (NBS) programs.

Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. It is recommended that every newborn be screened for all disorders on the RUSP.

Most states screen for the majority of disorders on the RUSP; newer conditions are still in process of adoption. Some states also screen for additional disorders.


Although states ultimately determine what disorders their NBS program will screen for, the RUSP establishes a standardized list of disorders that have been supported by the Advisory Committee on Heritable Disorders in Newborns and Children and recommended by the Secretary of HHS.

Conditions listed on the RUSP are part of the comprehensive preventive health guidelines supported by HRSA for infants and children under section 2713 of the Public Health Service Act. Non-grandfathered health plans are required to cover screenings included in the HRSA-supported comprehensive guidelines without charging a co-payment, co-insurance, or deductible for plan years beginning on or after the date that is one year from the Secretary's adoption of the condition for screening.

[How to Nominate a Condition](#)

[Previously Nominated Conditions \(Recommended and Not Recommended for the RUSP\)](#)

[Printer-Friendly Recommended Uniform Screening Panel \(PDF - 95 KB\)](#)




[Resources](#)[Your State](#)[Find a Condition](#)[En Español](#)

[About Newborn Screening](#)[Your Baby's Screening](#)[Living With Conditions](#)[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 professionals alike.



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

Newborn Screening – Different Specialties

Metabolic Diseases

Metabolic Disease panel by LC-MS/MS
(> 30 different conditions)

Galactosemia

Biotinidase Deficiency

Adrenoleukodystrophy (ALD)

Pompe Disease

Mucopolysaccharidosis Type 1

Hematology / Immunology

Sickle cell disease

Hemoglobinopathies

Severe Combined Immunodeficiency

Endocrine Disorders

Congenital Hypothyroidism

Congenital Adrenal Hyperplasia

Pulmonary

Cystic Fibrosis

Neurology / Genetics

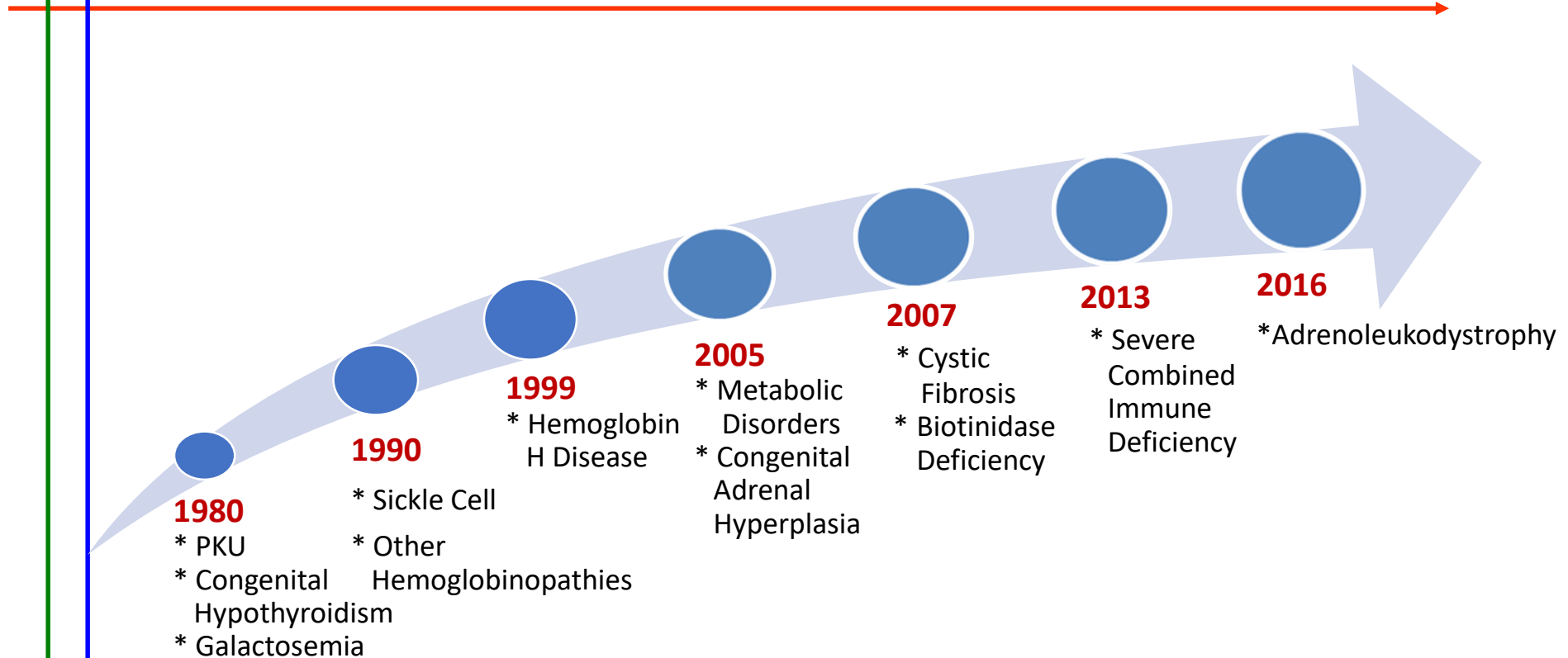
Spinal Muscular Atrophy

Other

Congenital Hearing loss

Critical congenital heart disease

A Brief History of Newborn Screening Expansion In California



Genetic Disease Screening Program

CA – NBS Program 2012-2016

Total Screened > 2,400,000

	# Positive Cases	Rate
Metabolic Disorders	580	1:4236
Galactosemia (GALT)	106	1:23,177
Biotinidase Deficiency (BIOT)	104	1:23,623
Endocrine Disorders	1448	1:1697
Cystic Fibrosis (CF)	257	1:9560
Hemoglobin Disorders (Hb)	1017	1:2416
Severe Combined Immunodeficiencies (SCID)	34	1:55,383
ALL DISORDERS	3546	1:693

Preliminary CDPH data: not for publication or distribution

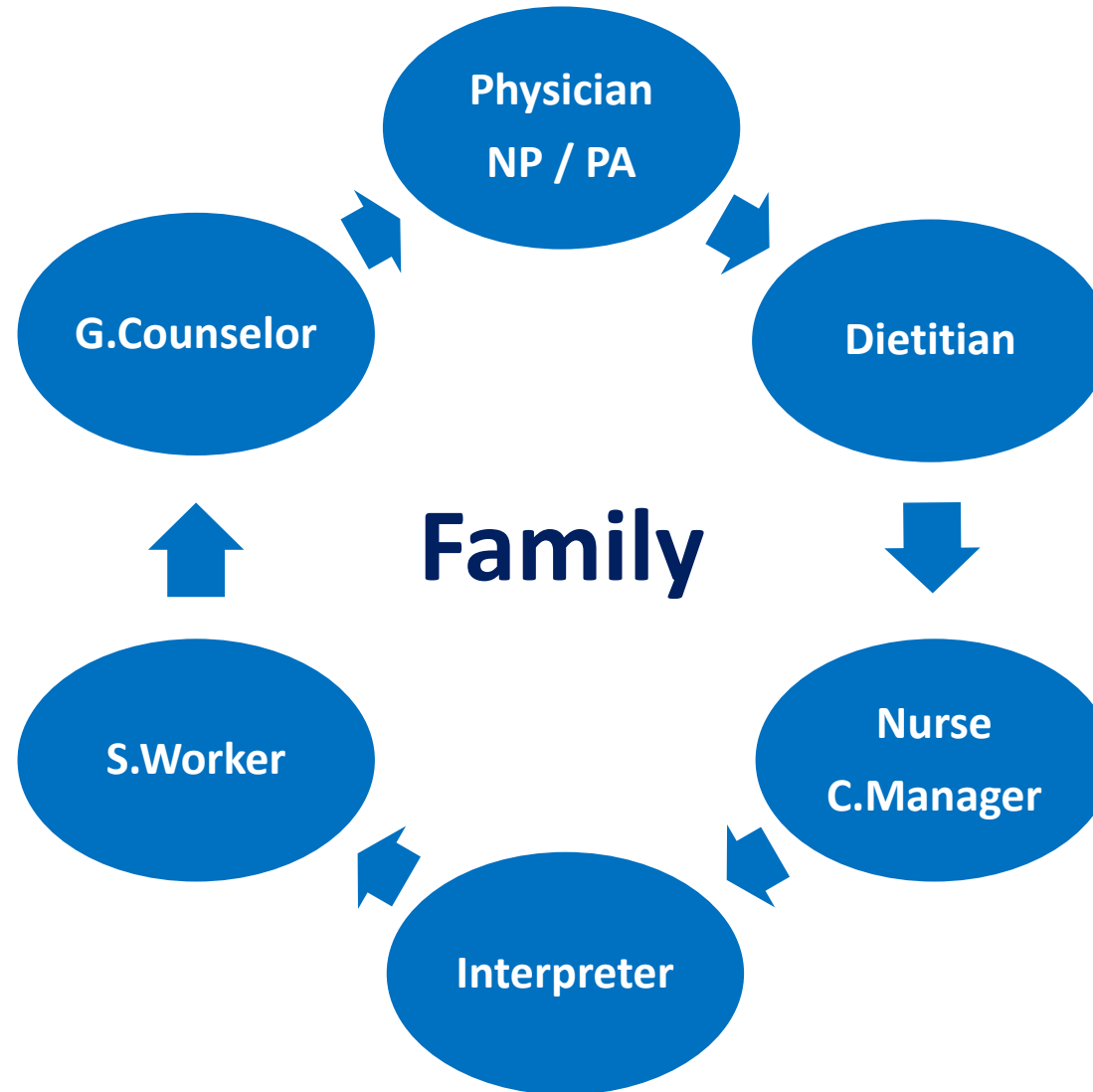
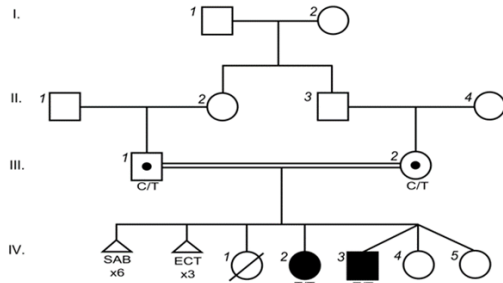
CHOC Children's Metabolic Center



Population
 CA: 39,500,000
 OC: 3,172,000
 RIV: 2,361,000
 SBD: 2,128,000

NB / year:
 500,000
 38,000
 32,000
 31,000

The Metabolic Team



NBS: Success Story

MCAD - Symptomatic



MCAD - NBS Detected

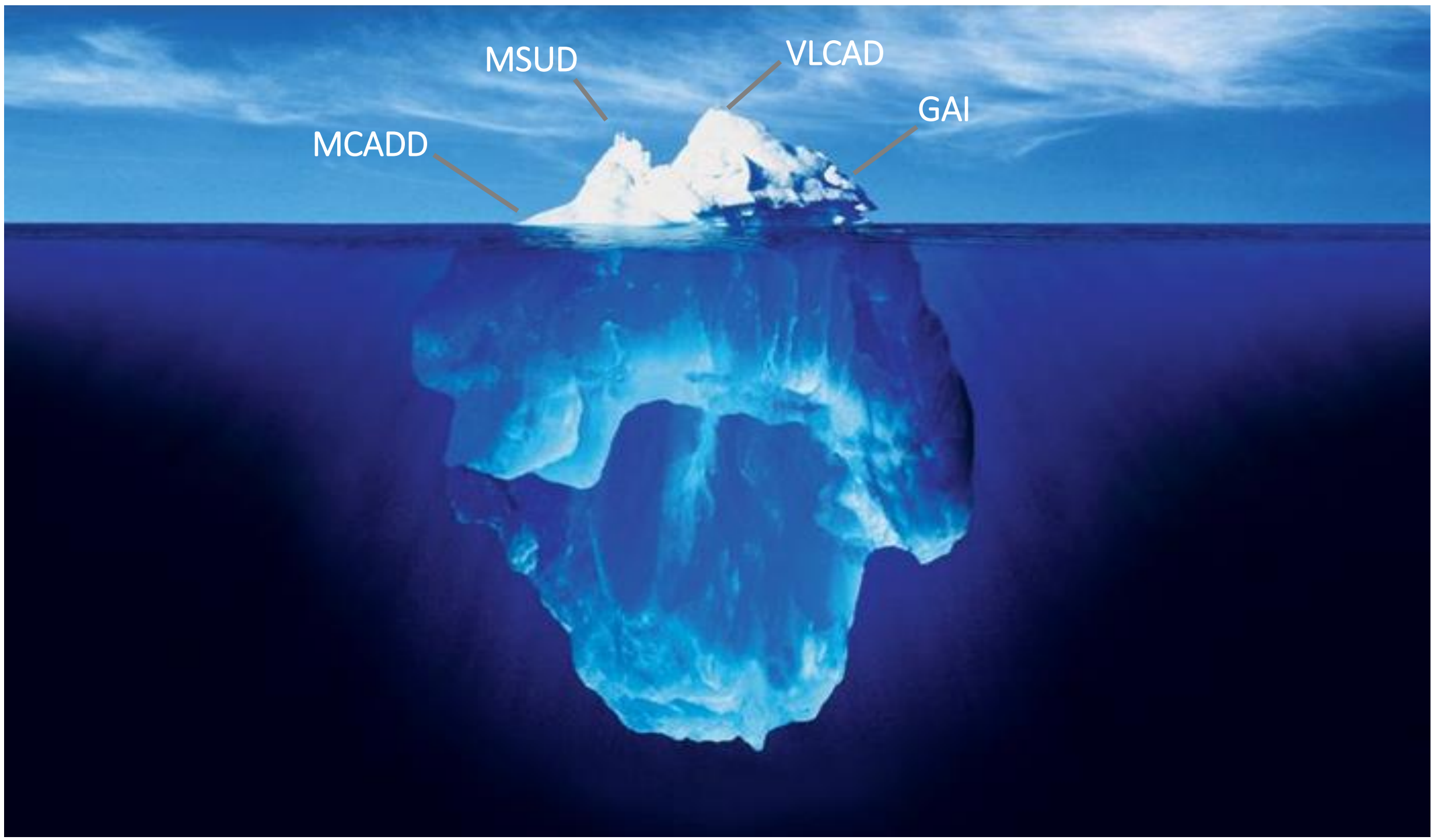


MCADD

MSUD

VLCAD

GAI



MCAD

MSUD

VLCAD

GAI

Detection of
asymptomatic
“patients”

Detection of
carriers

Detection of patients who
may become symptomatic
adolescence / adulthood

Detection of maternal
conditions

Decompensation
prior to NBS results

Too many False
positives

Molecular confirmatory
testing not always covered

Uncertain significance of
molecular results (VUS)

Detection of
Pseudo deficiencies

Insufficient education
HCP and Community

CHOC Metabolic: Clinical

Clinic

MD: J. Abdenur, R. Chang, R. Wang

NP: M. Boyer

RD: M. Sowa, J. Skaar, B. Janda

RN: J. Hagger, C. Daum

NBS Coordinator: R. Sponberg

GC: R. Bressi, K. Schwan

SW: M. Greene

Secretary: L. Esqueda

Metabolic Laboratory:

D. Butoi, CLS

S. Xu, CLS

B. Evans, CLS

C. Aguirre. LT

CHOC Metabolic: Research

Energy - Lab

J. Abdenur, MD

M. Simon, PhD

P. Schwartz, PhD

A. Stover, MS

W. Huang, MS

Foundation of Caring, Lysosomal Lab

R. Wang, MD

J. Huang, PhD

K. Khan, PhD

J. Harb, MS

Thank You !!!

Newborn Screening: Advocate & Parent Perspective

Danyelle Sun, MSW
*Wisconsin State Ambassador
NORD Rare Action Network*



rarediseases.org



*Stacy McCarroll and
daughter, Cystic Fibrosis*

Where I'm Coming From...



- Rare Disease personal experience - Spinal Muscular Atrophy, two children
- Social Work Manager at Cure SMA
- NORD RAN Ambassador since 2018
- Newborn screening advocacy for all rare diseases

Why do I advocate?



- Knowledge is power!
- Gain access to preventative treatment.
- More time to plan.
- The patient and caregiver voice needs to be heard.
- Give meaning to a difficult experience – help others.

Advocacy & Newborn Screening in the Future

- The more diseases or conditions with known causes (RESEARCH)...
- The more treatments available (TRIALS)...
- The more newborn screening panels can be created (NBS)...
- So never stop advocating and learning!

Newborn Screening Program Update

Rachel Sher, JD, MPH
VP, Policy and Regulatory Affairs
NORD



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Vaughn family: Son, Morgan (left), diagnosed with Necrotizing Enterocolitis at four days old

Newborn Screening Saves Lives Act

- Under the law:
 - The CDC manages the Newborn Screening Quality Assurance Program
 - HRSA manages the Advisory Committee on Heritable Disorders which manages the Recommended Uniform Screening Panel
 - NIH Child Health and Human Development conducts research on childhood diseases with funds based on need
- Reauthorization in the works:
 - Program authorization in statute expired in September 2019;
 - H.R. 2507 (Rep. Lucille Roybal-Allard) passed the House; Senate TBD

H. R. 2507

IN THE SENATE OF THE UNITED STATES

JULY 25, 2019

Newborn Screening Advisory Committee



- The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) (Health Resources & Services Administration (HRSA)) meets to determine the diseases on the Recommended Uniform Screening Panel (RUSP)
- The Advisory Committee did not meet for some time but thanks to efforts from NORD, associated advocacy organizations, and Adm. Giroir- ACHDNC is meeting again!
- **CONTINUED FUNDING FOR ALL NBS PROGRAMS IS CRITICAL!**



ACHDNC

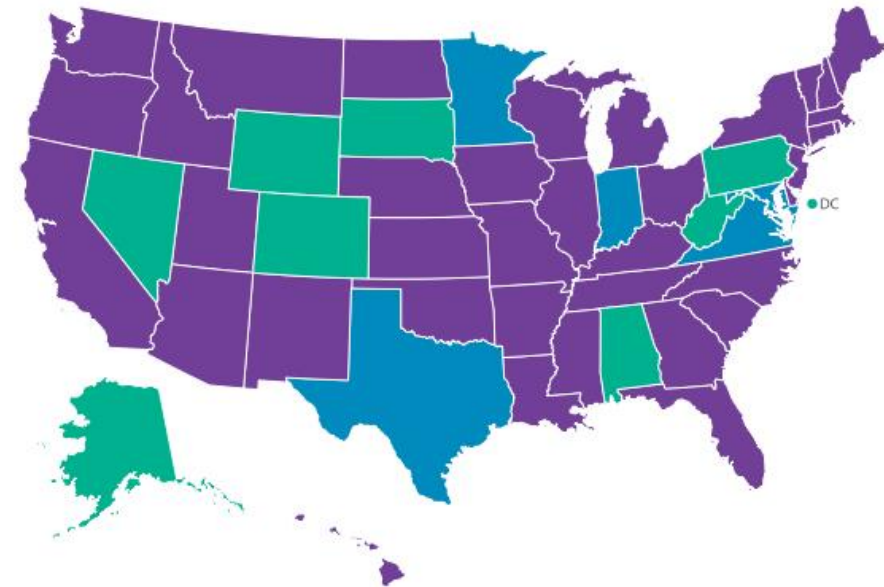
Secretary's Advisory Committee
on Heritable Disorders in
Newborns and Children



rarediseases.org

Newborn Screening at the State Level

- NORD [State of the States Report](#): *NORD supports robust, well-funded newborn screening programs in every state.*
- States are "graded" in 7 areas:
 1. Screening for RUSP core conditions
 2. Adding RUSP core conditions
 3. Funding
 4. Using Dried Blood Spot (DBS)
 5. Follow-up
 6. Quality
 7. Advisory committee



NEWBORN SCREENING OVERALL GRADE SCALE

A B C D F

Find our report here: <https://rareaction.org/resources-for-advocates/nordreport/>

Questions?

Submit your questions in the chat. Email additional questions to education@rarediseases.org



Jose Abduner, MD
Medical Director, Pediatric
Metabolic Disorders
Children's Hospital Orange County



Danyelle Sun, MSW
Rare Mom
Wisconsin State Ambassador
NORD Rare Action Network



Rachel Sher, JD, MPH
Vice President, Policy and
Regulatory Affairs
NORD



Process

Nominate a condition

- Multidisciplinary team: experts, advocacy groups, professional organizations, consumers
- Complete nomination package

Initial Evaluation

- Nomination and prioritization workgroup
- Feed back to Advisory committee
- Vote to continue (external revision) or not.

External Revision Workgroup

- Systematic Evidence based review - updates
- Final report to Advisory Committee

Advisory committee

- Advisory Committee Deliberation
- Decision Matrix
- Vote – Recommendation to Secretary of HHS

Department of HHS

- Recommendation to included a condition as part of State NBS Panels (RUSP)



Thank you.



Alone we are **rare**. Together we are strong.®

rarediseases.org