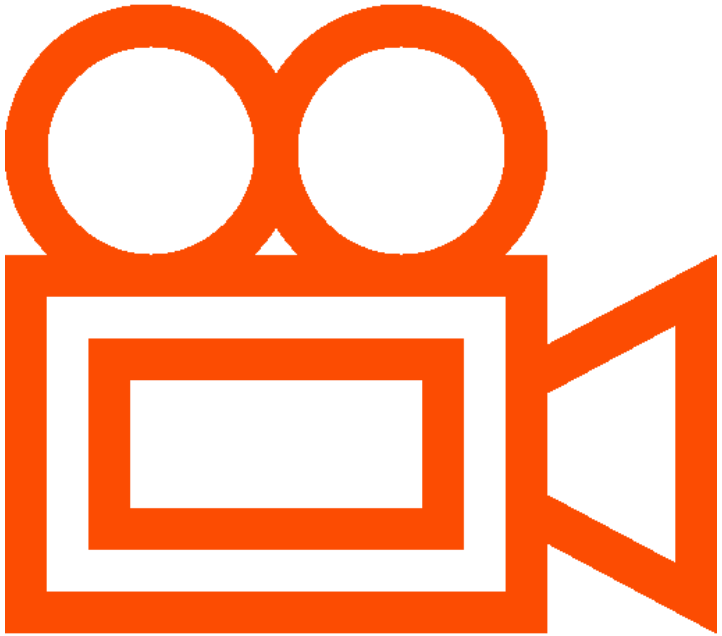
SCREEN BABY SCREEN: PERSPECTIVES ON NEWBORN SCREENING

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.



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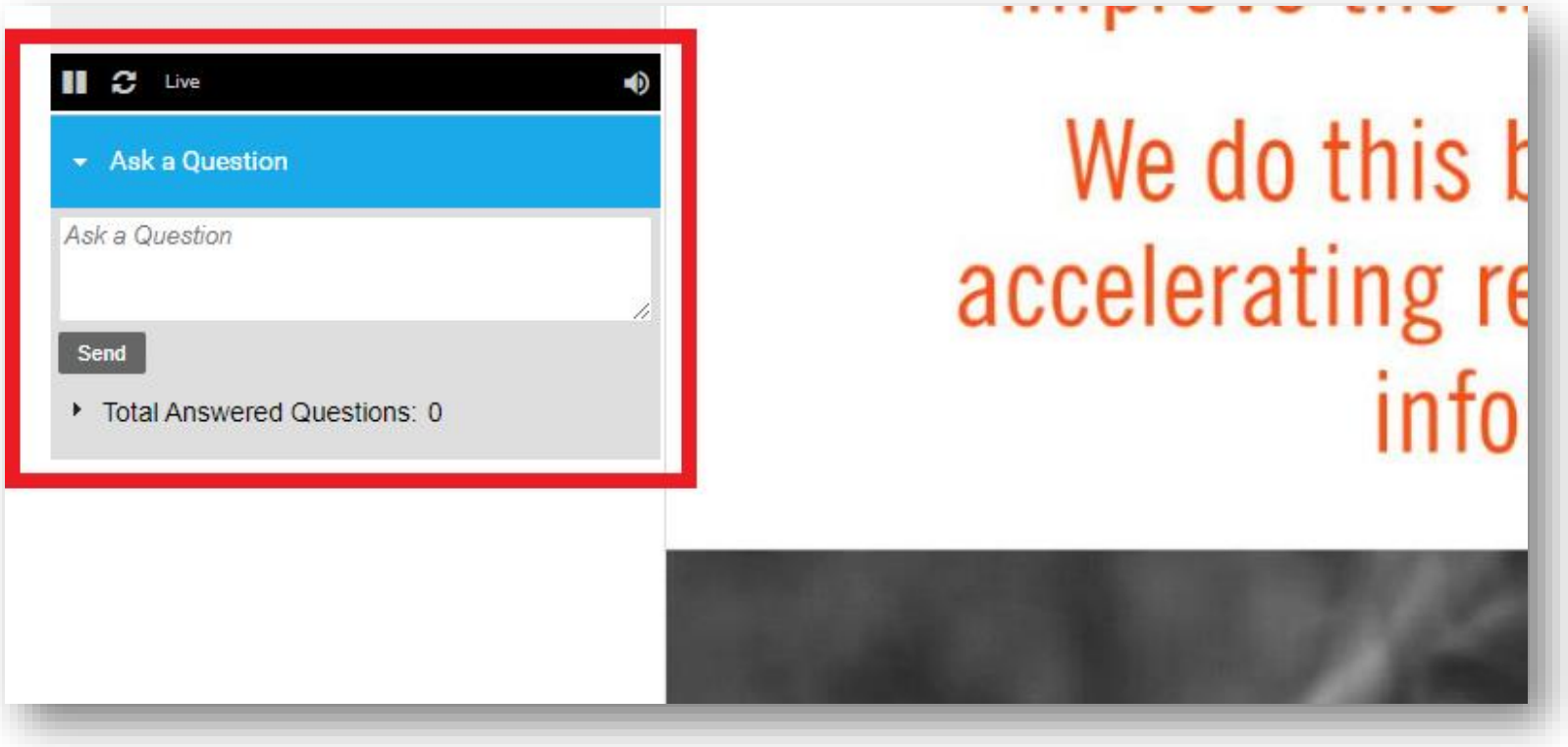
Question and Answer Session

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



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Newborn Screening Awareness Month

September is Newborn Screening Awareness Month!

Learn more at rarediseases.org

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Speakers

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

CHOC Children's.

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

NORD® RARE ACTION NETWORK®

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

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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

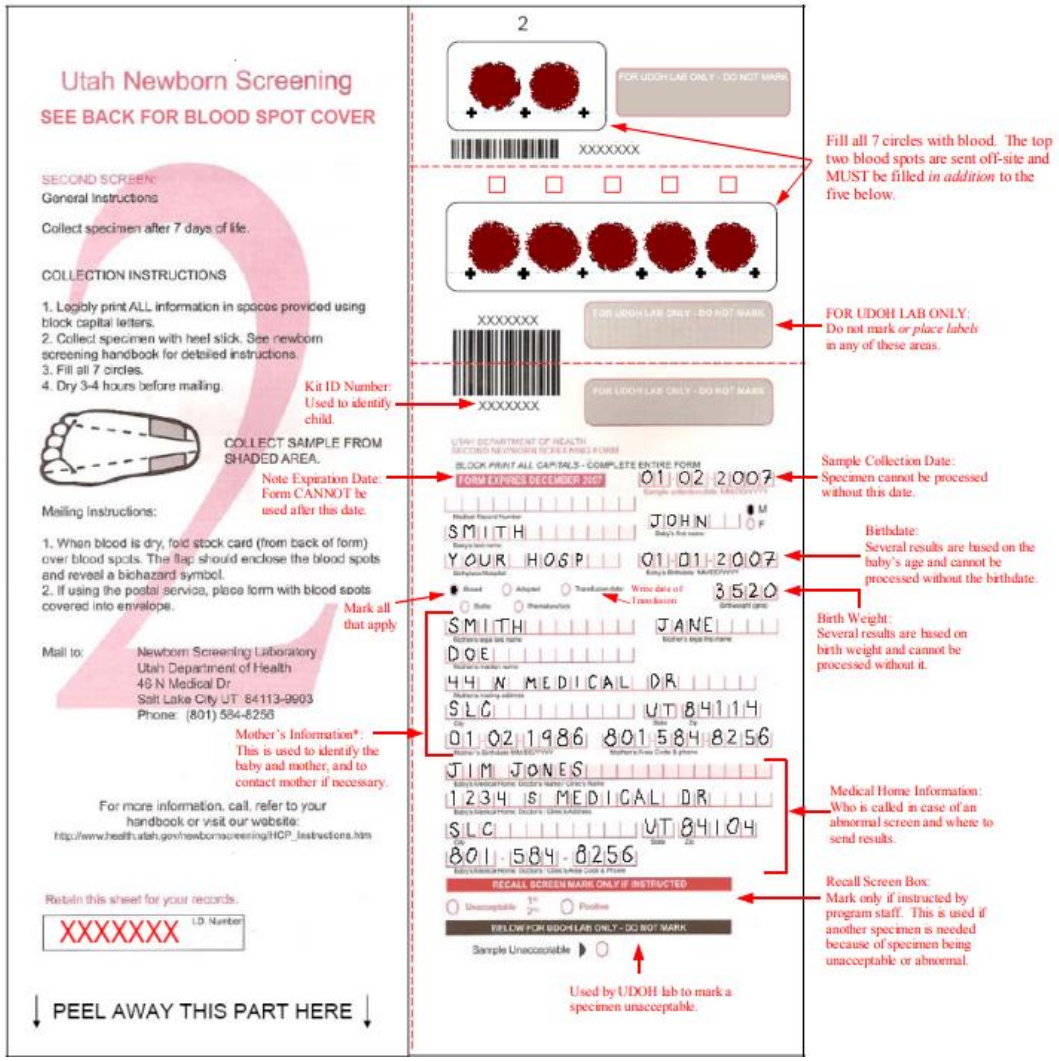
Definitions

- 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



The “Guthrie” Newborn Screening Card

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

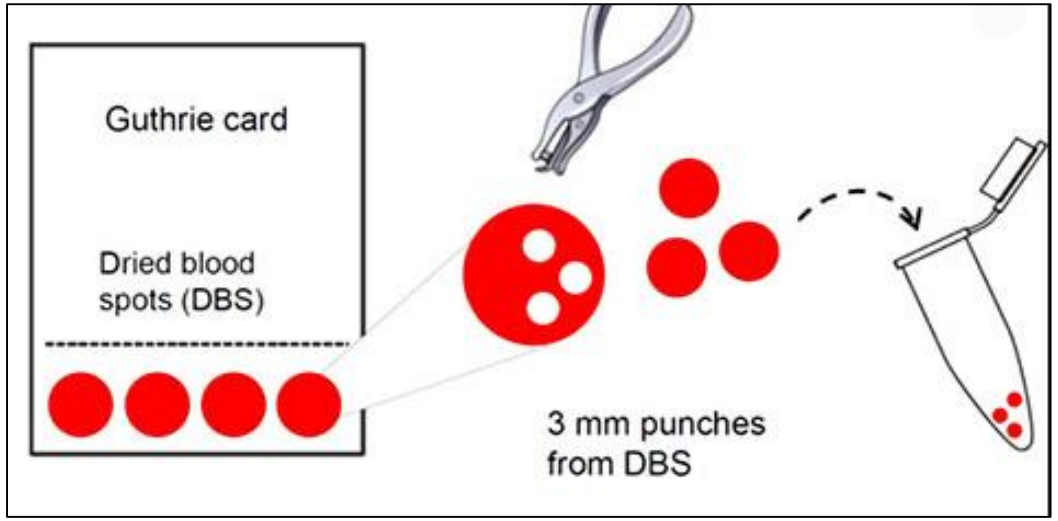
Infant’s name, birth order
Initial or repeat sample
Race and ethnicity
Birth date, hour
Sample collection date and hour
Feeding type
Transfusion date, hour

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)

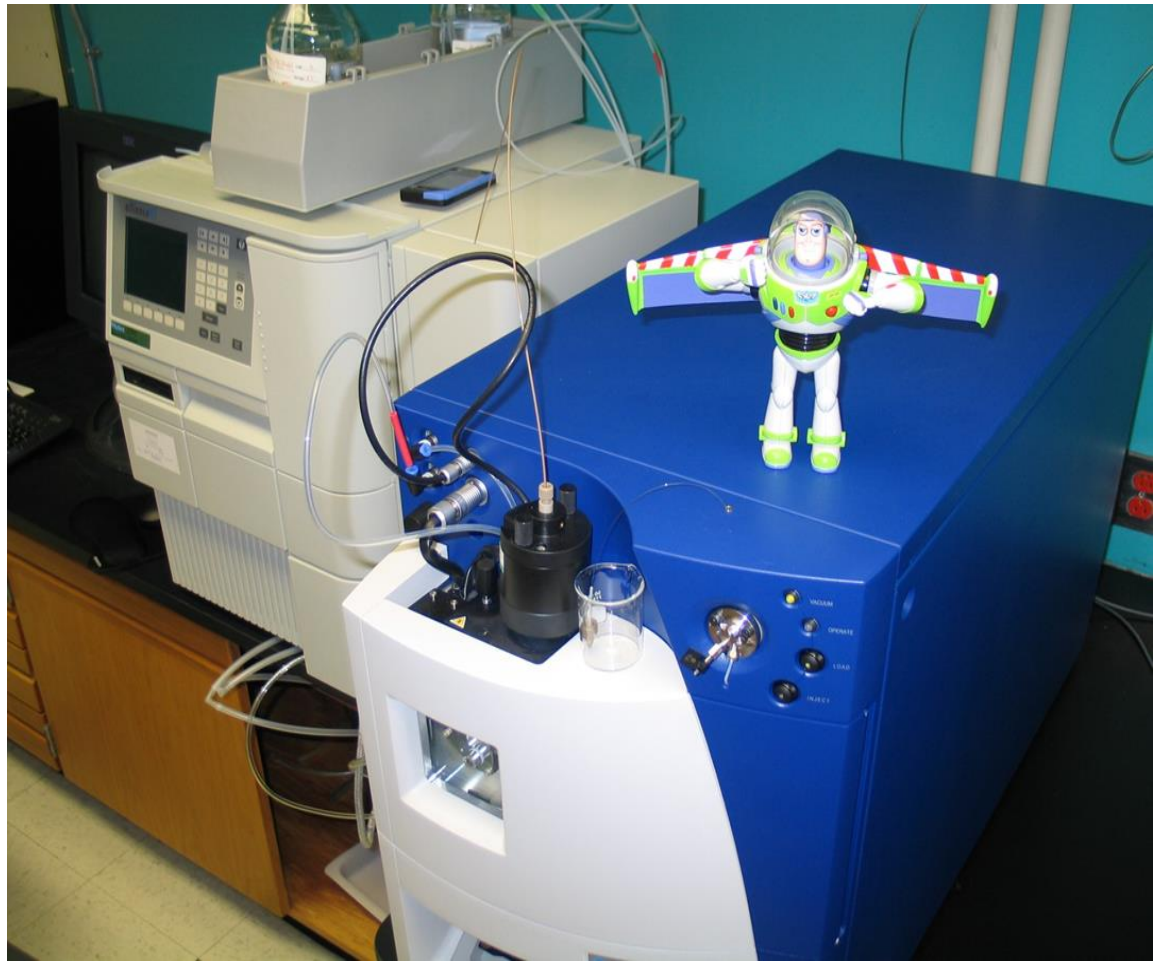
The “Guthrie” Newborn Screening Card

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

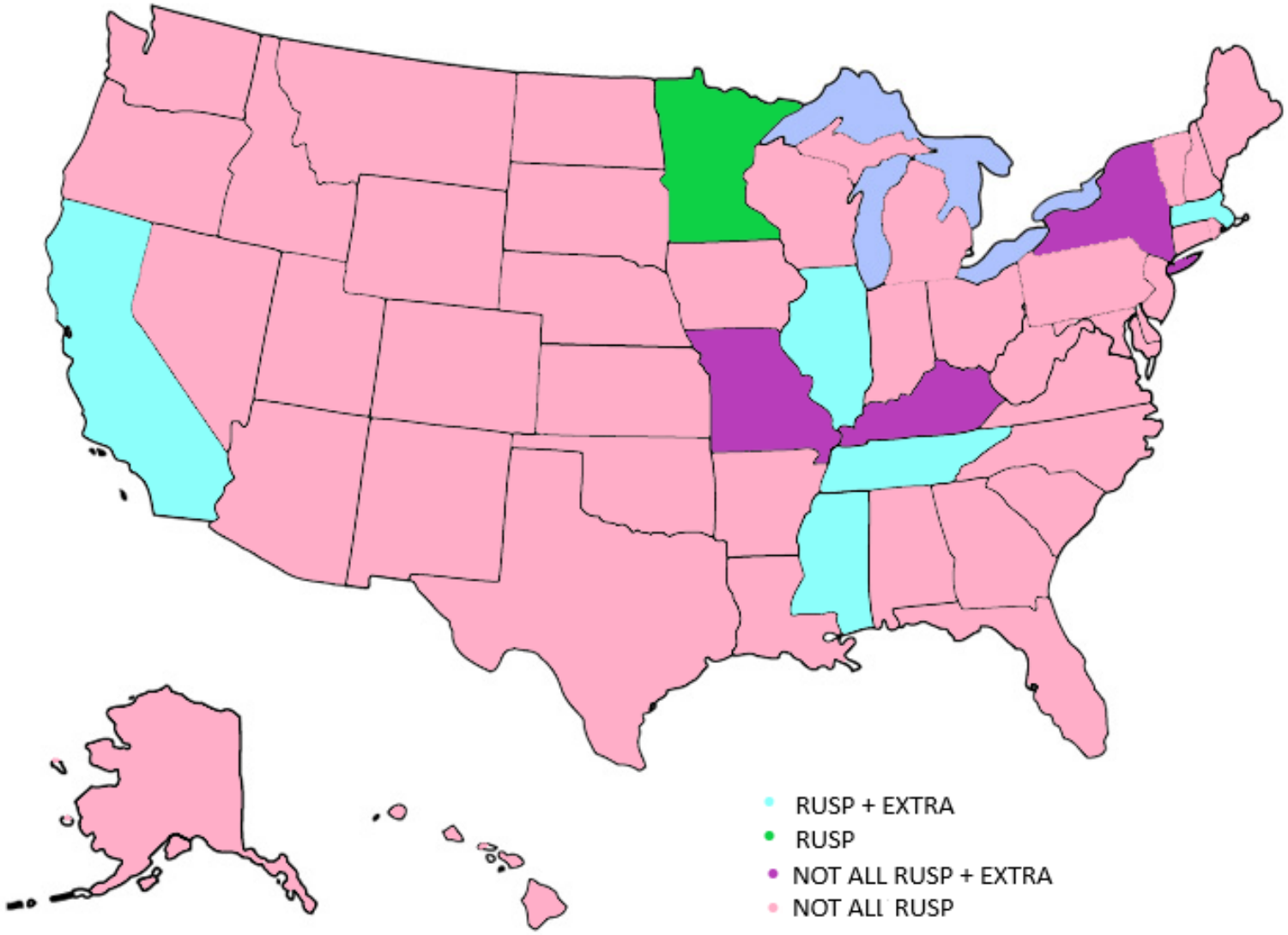
- **Diagnostic methodology**
  - Small sample
  - Easy collection and transport
  - High sensitivity (no false negatives)
  - High specificity (few false positives)
  - High throughput methodology
  - Cost Effective
  - Available QC and PT programs
- **Confirmatory tests**
  - Easily available
  - Rapid 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)

Advisory Committees on Heritable Disorders in Newborns and Children

- Home
- About
- Meetings
- Reports
- Letters
- Resources
- 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.

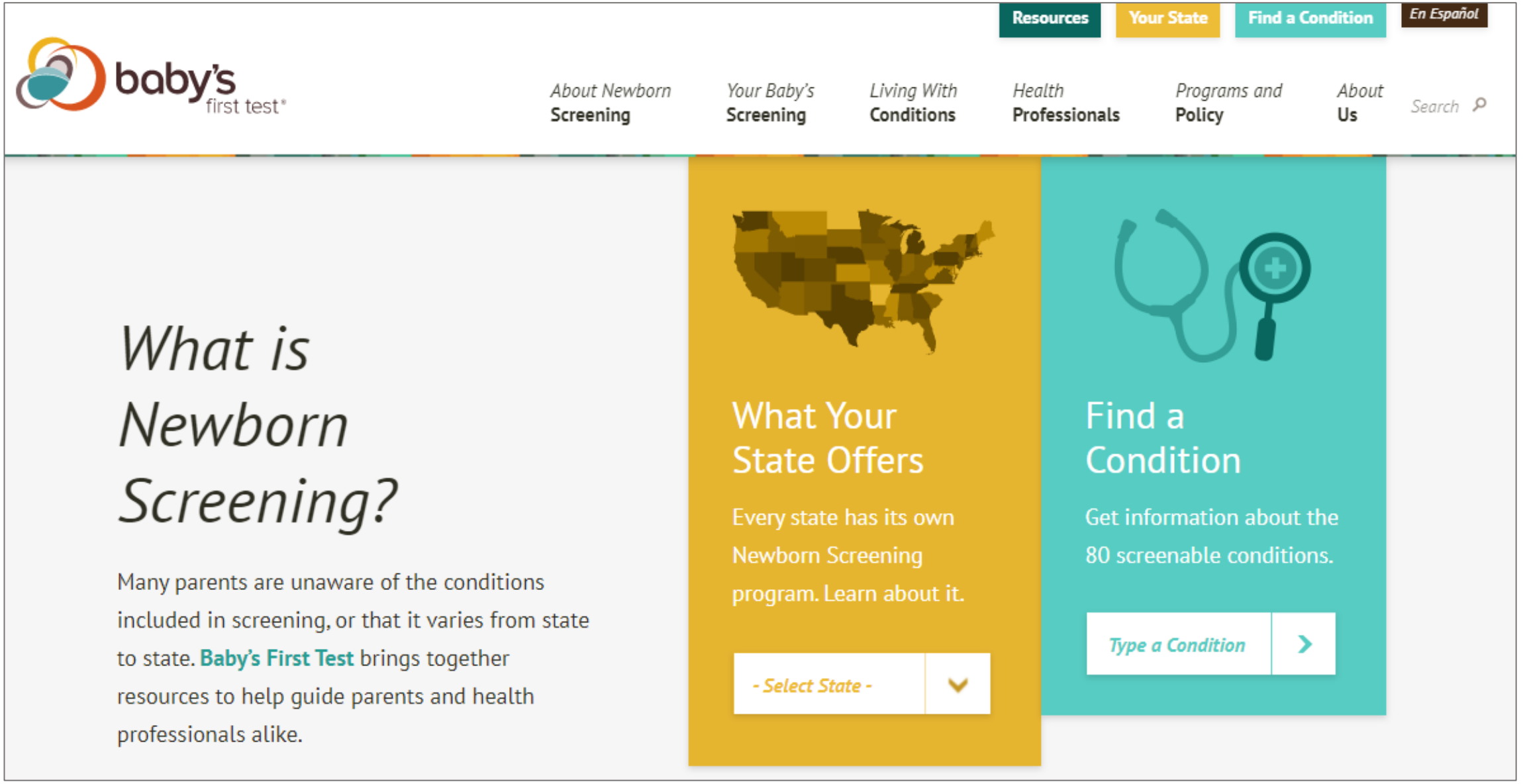
Conditions listed on the RUSP are part of the comprehensive preventive health guidelines supported by HRSA for infants and children under section 2713 of the Public Health Service Act. Non-grandfathered health plans are required to cover screenings included in the HRSA-supported comprehensive guidelines 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)

https://www.babysfirsttest.org/



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 / Immnunology

Sickle cell disease
Hemoglobinopaties
Severe Combined Immunodeficiency

Endocrine Disorders

Congenital Hipothyroidism
Congenital Adrenal Hyperplasia

Pulmonary

Cystic Fibrosis

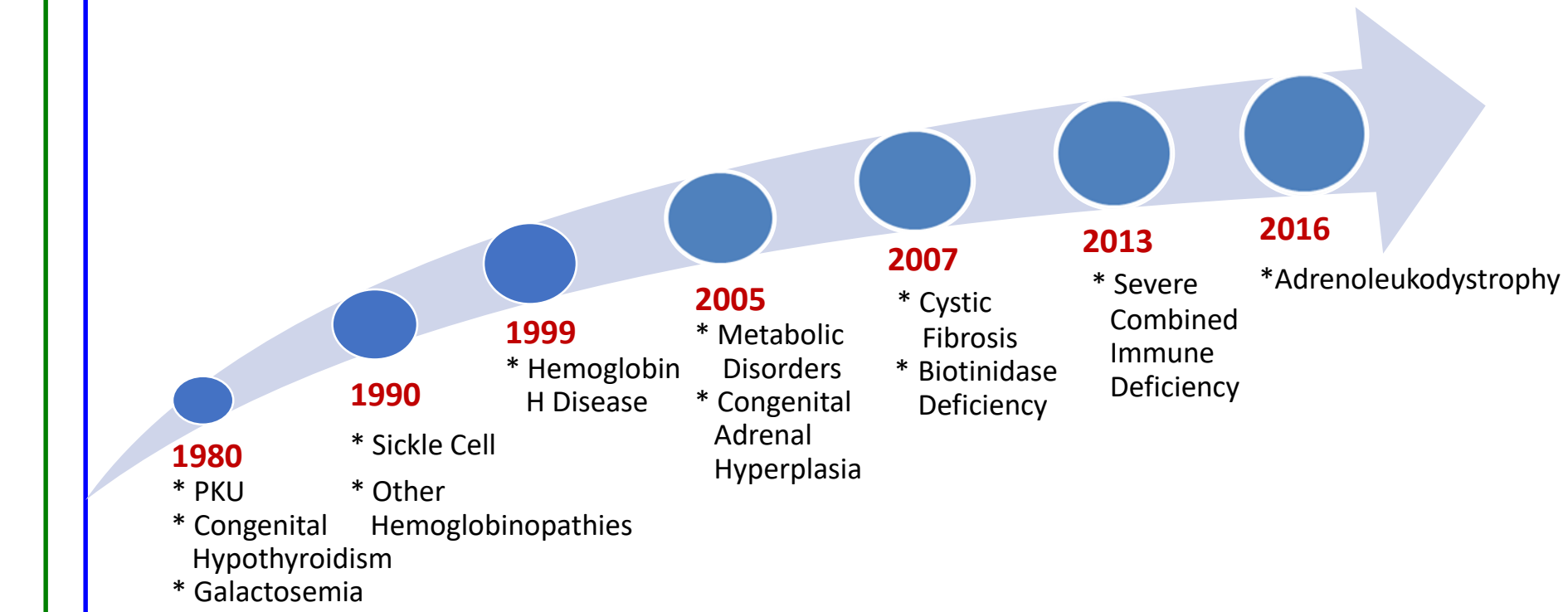
Neurology / Genetics

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

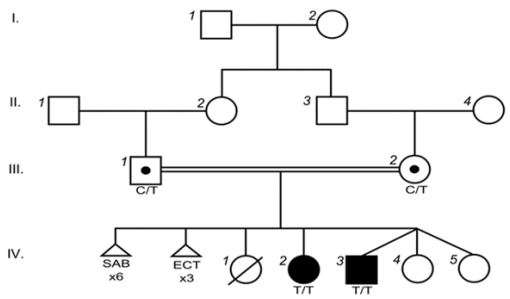
	# Positive Cases	Rate
Metabolic Disorders	580	1:4236
Galactosemia (GALT)	106	1:23,177
Biotinidase Deficiency (BIOT)	104	1:23,623
Endocrine Disorders	1448	1:1697
Cystic Fibrosis (CF)	257	1:9560
Hemoglobin Disorders (Hb)	1017	1:2416
Severe Combined Immunodeficiencies (SCID)	34	1:55,383
ALL DISORDERS	3546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

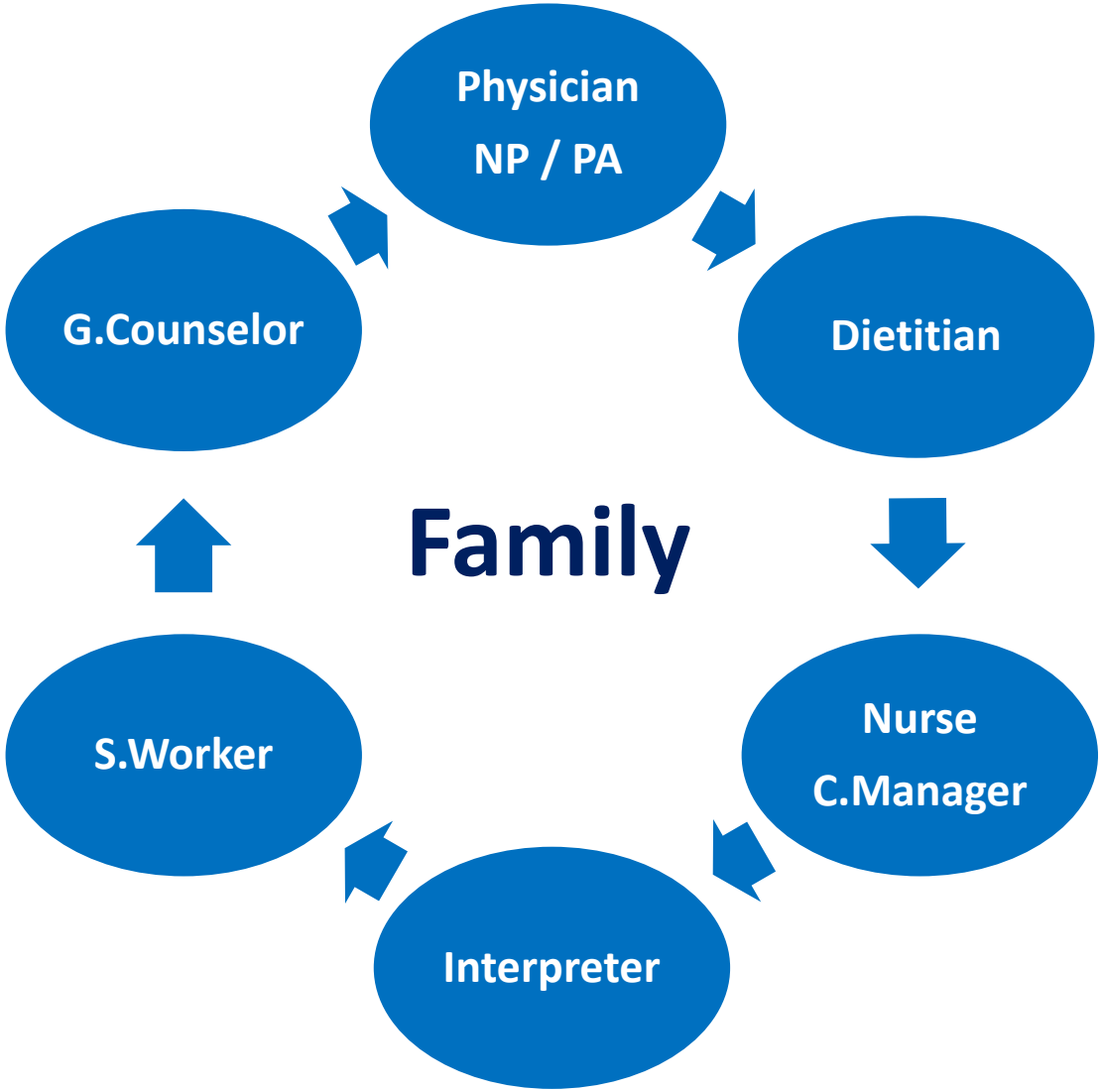
Preliminary CDPH data: not for publication of distribution

CHOC Children's Metabolic Center

Population		NB / year:
CA:	39,500,000	500,000
OC:	3,172,000	38,000
RIV:	2,361,000	32,000
SBD:	2,128,000	31,000

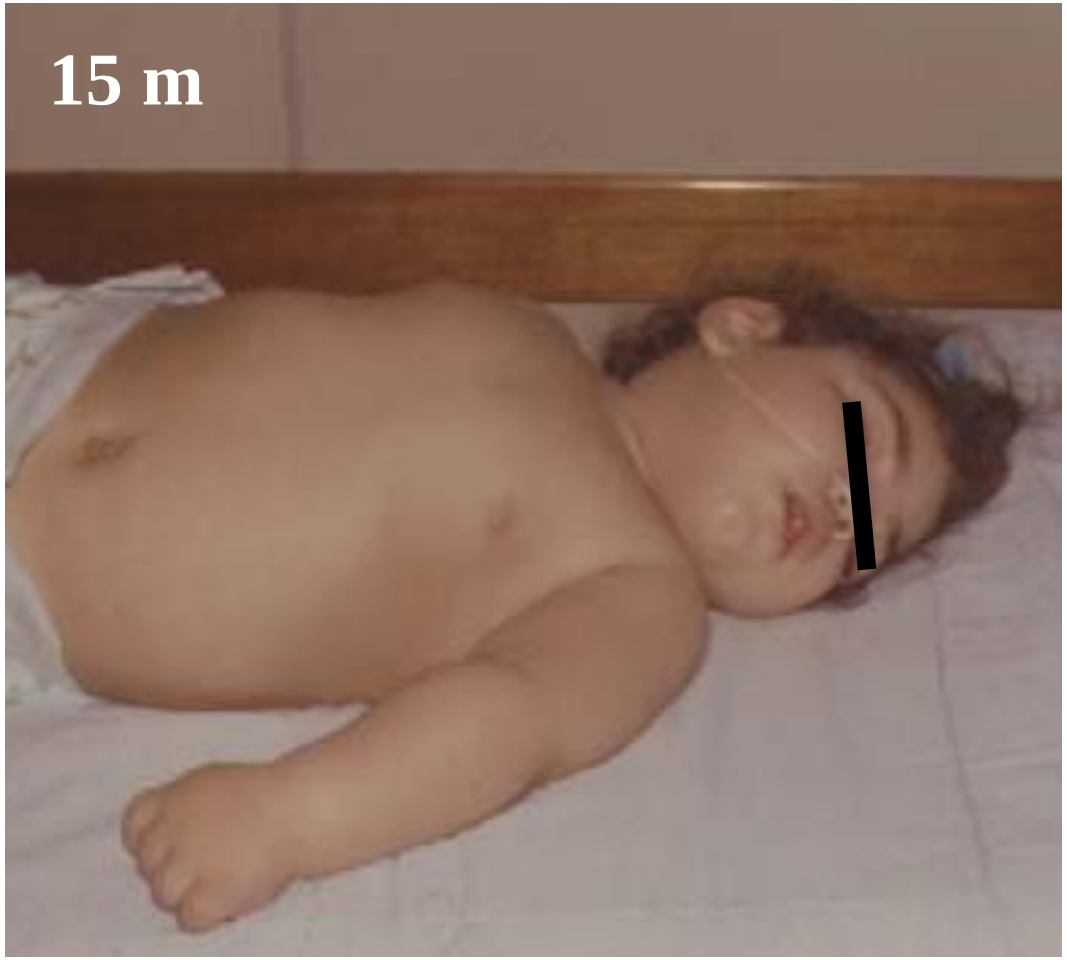
The Metabolic Team



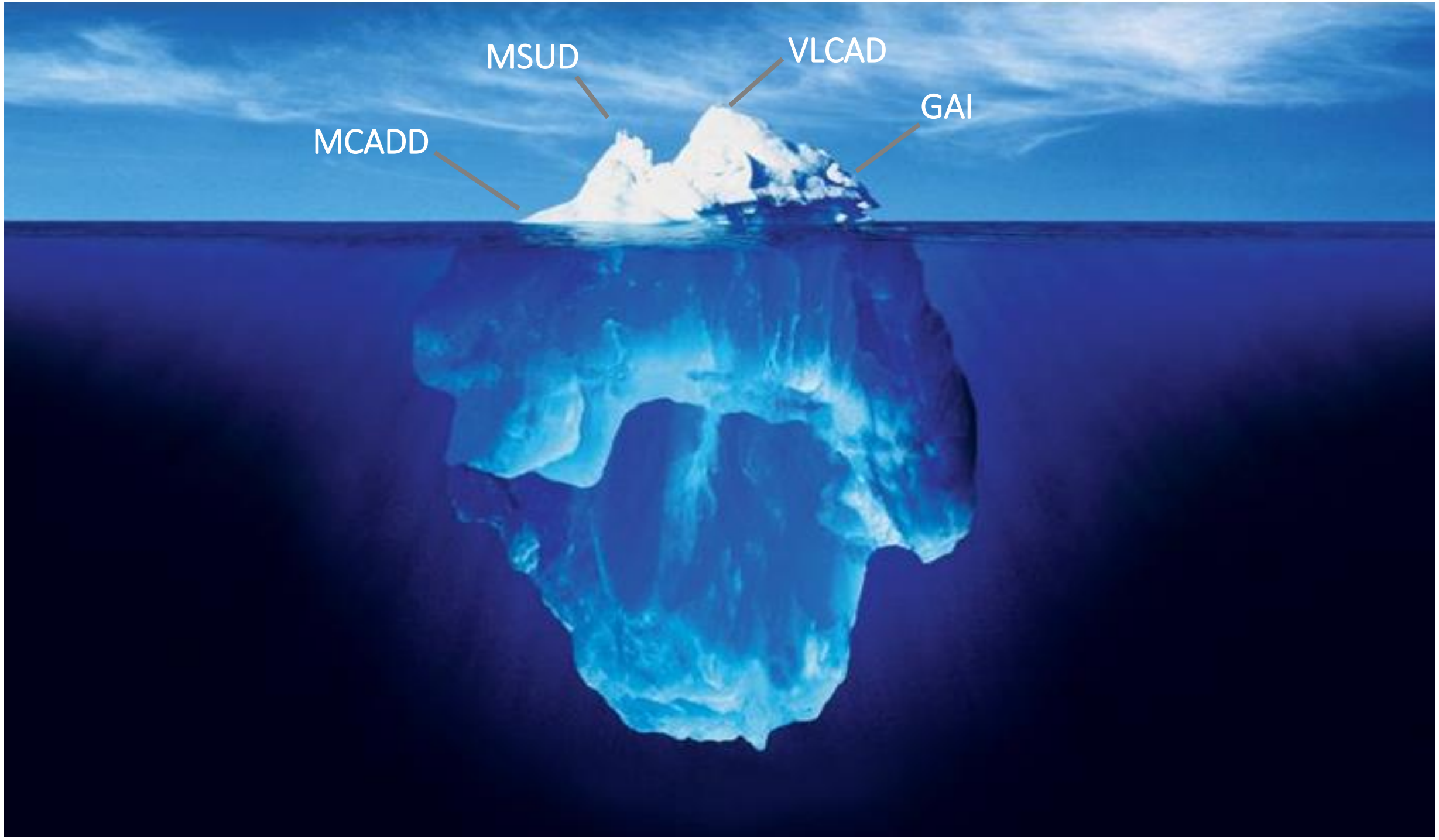


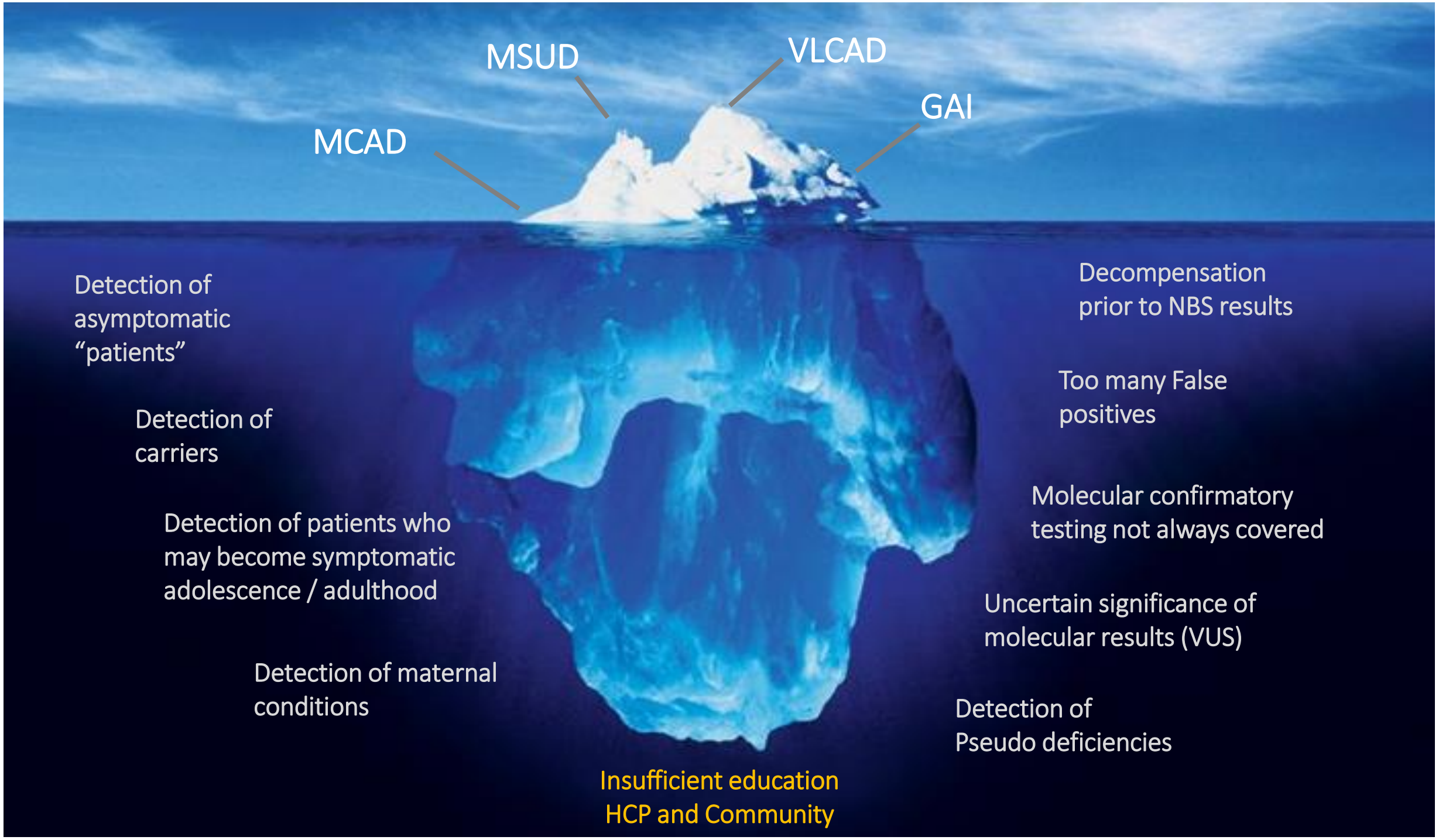
NBS: Success Story

MCAD - Symptomatic



MCAD - NBS Detected


MSUD
VLCAD
GAI
MCADD


MSUD
VLCAD
GAI
MCAD
Detection of asymptomatic "patients"
Detection of carriers
Detection of patients who may become symptomatic adolescence / adulthood
Detection of maternal conditions
Insufficient education HCP and Community
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

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

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Stacy McCaroll 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

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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.

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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!

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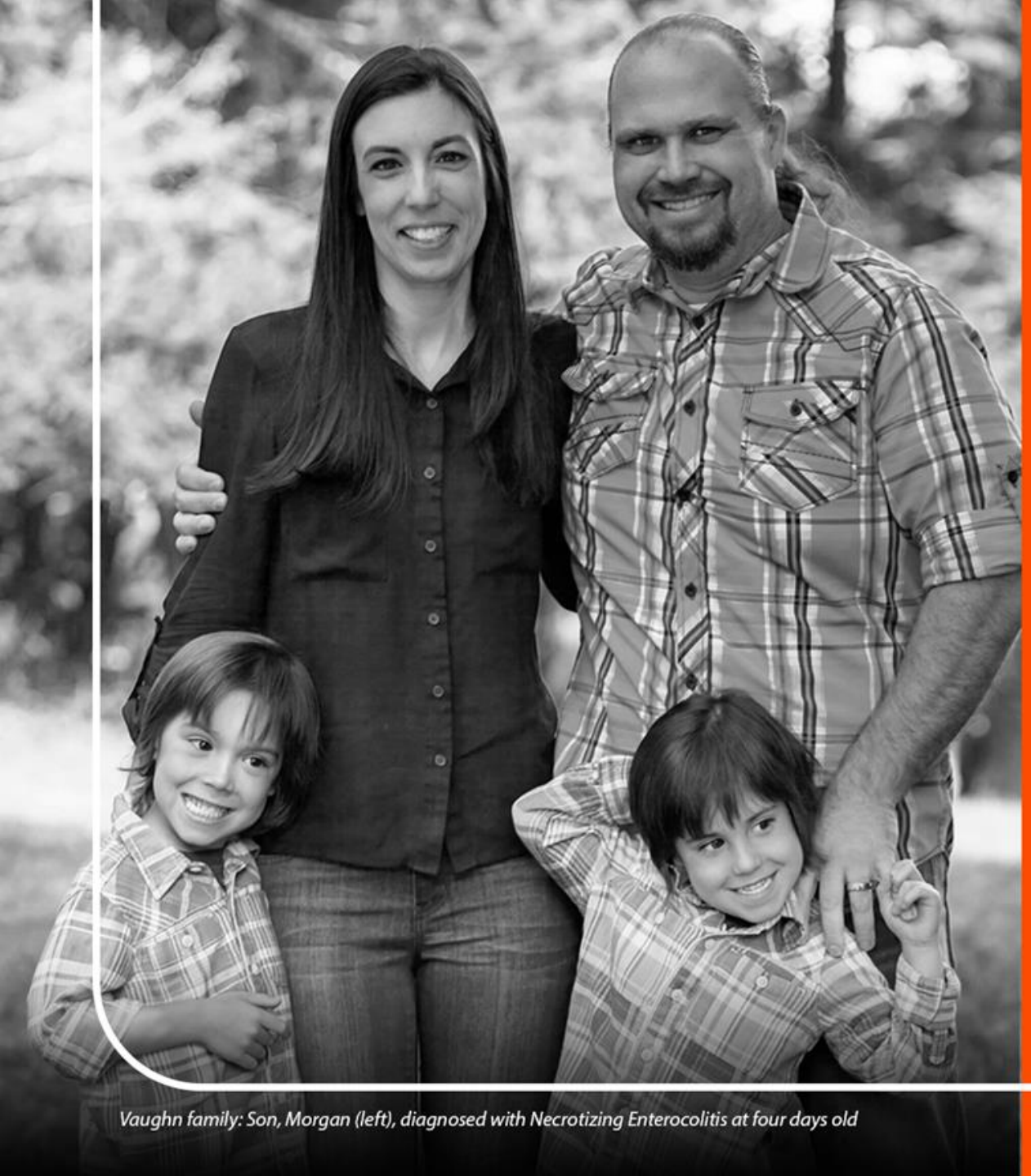

Vaughn family: Son, Morgan (left), diagnosed with Necrotizing Enterocolitis at four days old

Newborn Screening Program Update

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

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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

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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

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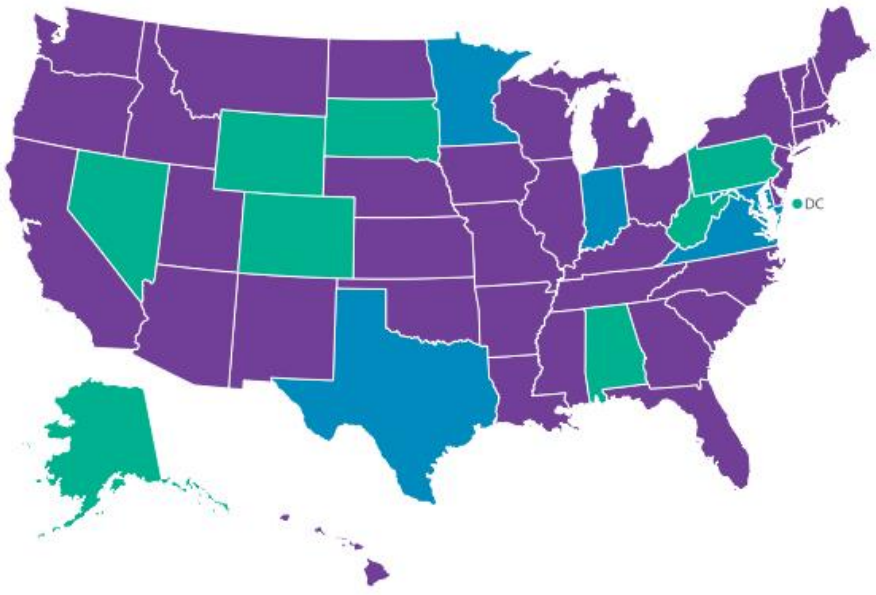
Newborn Screening at the State Level

- 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



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

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Questions?

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

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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)

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