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.
This webinar is being recorded.
Submit your questions using the chat function. It can be found at the **left hand side** of the window.
September is Newborn Screening Awareness Month!

Learn more at rarediseases.org
ENTERING A NEW ERA
VIRTUAL EVENT

October 8-9, 2020
#NORDSUMMIT | nordsummit.org

Rare Diseases and Orphan Products Breakthrough Summit®
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

CHOC Children's  
NORD  
RARE ACTION NETWORK®  
NORD® National Organization for Rare Disorders

rarediseases.org
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
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)

Conditions included in State NBS Programs
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)
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.
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

1980
- PKU
- Congenital Hypothyroidism
- Galactosemia

1990
- Sickle Cell
- Other Hemoglobinopathies

1999
- Hemoglobin H Disease

2005
- Metabolic Disorders
- Congenital Adrenal Hyperplasia

2007
- Cystic Fibrosis
- Biotinidase Deficiency

2013
- Severe Combined Immune Deficiency

2016
- Adrenoleukodystrophy

Genetic Disease Screening Program
## CA – NBS Program 2012-2016
Total Screened > 2,400,000

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

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

Family

Physician NP / PA

G.Counselor

Dietitian

S.Worker

Nurse C.Manager

Interpreter
NBS: Success Story

MCAD - Symptomatic

15 m

MCAD - NBS Detected

14 m
Detection of carriers
Detection of asymptomatic “patients”
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
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
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
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!
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/
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.