

**APHL Newborn Screening
Symposium
2025**



October 5–9, 2025 • Providence, RI

Final Program

Sponsored by the
Association of Public Health Laboratories

Co-Sponsored by the
Rhode Island Department of Health
and the International Society
for Neonatal Screening

www.aphl.org/NBS2025
#APHLNBS

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Agenda at a Glance

(L3) = Convention Center Level 3

(L5) = Convention Center Level 5

Sunday, October 5

2:00 pm – 5:00 pm	Registration Open	West Pre-function (L5)
5:00 pm – 6:30 pm	Exhibit Hall and Posters Open	Exhibit Hall AB (L3)
3:00 pm – 3:30 pm	Welcome Address	Ballroom A (L5)
3:30 pm – 5:00 pm	Newborn Screening Stories	Ballroom A (L5)
5:00 pm – 6:30 pm	Welcome Reception <i>Poster authors P-1 to P-59 available to discuss their posters from 5:00 pm to 6:00 pm</i>	Exhibit Hall AB (L3)
6:30 pm – 8:00 pm	Newborn Screening Laboratory Social Hour The Carol Johnson Follow-Up and Family Social Hour	Ballroom D (L5) Ballroom B (L5)

Monday, October 6

7:30 am – 5:30 pm	Registration Open	West Pre-function (L5)
8:30 am – 4:00 pm	Exhibit Hall and Posters Open	Exhibit Hall AB (L3)
8:00 am – 8:30 am	Innovate! Session: Bio-Rad Laboratories, Inc.	Room 555AB (L5)
8:0 am – 8:45 am	Break in the Exhibit Hall	Exhibit Hall AB (L3)
8:45 am – 10:15 am	Concurrent Sessions	Ballrooms A & BC (L5)
10:15 am – 10:30 am	Break in the Exhibit Hall	Exhibit Hall AB (L3)
10:30 am – 12:30 pm	The Value of the Newborn Screening System	Ballroom A (L5)
12:30 pm – 2:00 pm	Lunch (on your own)	
2:00 pm – 2:30 pm	Sustaining and Advancing Excellence in NBS	Ballroom A (L5)
2:30 pm – 2:45 pm	Break in the Exhibit Hall	Exhibit Hall AB (L3)
2:45 pm – 3:30 pm	Innovate! Session: Waters Corporation	Room 555AB (L5)
3:30 pm – 3:45 pm	Break in the Exhibit Hall	Exhibit Hall AB (L3)
3:45 pm – 5:15 pm	Concurrent Sessions	Ballrooms A & BC (L5)
5:15 pm – 5:30 pm	Break	
5:30 pm – 7:00 pm	Ignite the Night!	Ballroom A (L5)

Tuesday, October 7

7:30 am – 5:00 pm	Registration Open	West Pre-function (L5)
8:30 am – 4:00 pm	Exhibit Hall and Posters Open	Exhibit Hall AB (L3)
8:30 am – 9:00 am	Break in the Exhibit Hall	Exhibit Hall AB (L3)
9:00 am – 10:30 am	Concurrent Sessions	Ballrooms A & BC (L5)
10:30 am – 10:45 am	Break in the Exhibit Hall	Exhibit Hall AB (L3)

US Eastern Daylight Time (EDT)

Sessions in Ballrooms A and BC will be recorded (no livestream).

10:45 am – 11:15 am	Informed Consent for Genomic Sequencing	Ballroom A (L5)
11:15 am – 11:30 am	Break in the Exhibit Hall	Exhibit Hall AB (L3)
11:30 am – 1:00 pm	Concurrent Sessions	Ballrooms A & BC (L5)
1:00 pm – 1:15 pm	Rapid Poster Presentations	Ballroom A (L5)
1:15 pm – 2:30 pm	Lunch in the Exhibit Hall <i>Poster authors P-60 to P-118 available to discuss their posters from 1:15 pm to 2:15 pm</i>	Exhibit Hall AB (L3)
2:30 pm – 3:15 pm	Concurrent Innovate! Sessions Revvity Primary. Health	Room 555AB (L5) Room 556AB (L5)
3:15 pm – 3:30 pm	Break in the Exhibit Hall	Exhibit Hall AB (L3)
3:30 pm – 5:00 pm	Newborn Screening Awards Ceremony	Ballroom A (L5)

Wednesday, October 8

7:30 am – 4:30 pm	Registration Open	West Pre-function (L5)
9:00 am – 4:30 pm	Exhibit Hall and Posters Open	Exhibit Hall AB (L3)

8:00 am – 9:00 am	Concurrent Roundtables	552AB, 555AB, 556AB (L5)
9:00 am – 9:30 am	Break in the Exhibit Hall	Exhibit Hall AB (L3)
9:30 am – 11:00 am	Concurrent Sessions	Ballrooms A & BC (L5)
11:00 am – 11:30 am	Break in the Exhibit Hall	Exhibit Hall AB (L3)
11:30 am – 1:00 pm	Concurrent Sessions	Ballrooms A & BC (L5)
1:00 pm – 2:30 pm	Lunch in the Exhibit Hall	Exhibit Hall AB (L3)
2:30 pm – 4:00 pm	Concurrent Sessions	Ballrooms A & BC (L5)
4:00 pm – 4:30 pm	Raffle Drawing and Break in the Exhibit Hall	Exhibit Hall AB (L3)

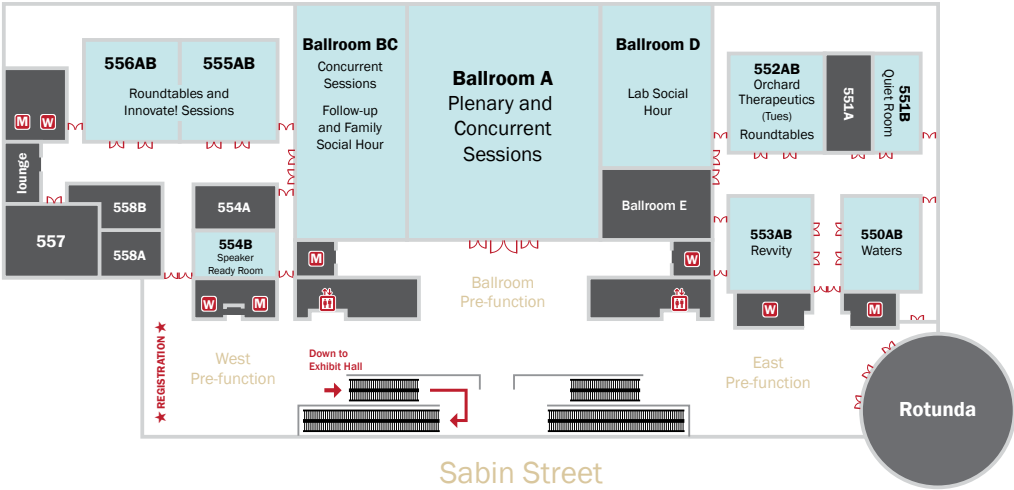
Thursday, October 9

7:30 am – 1:00 pm	Registration Open	West Pre-function (L5)
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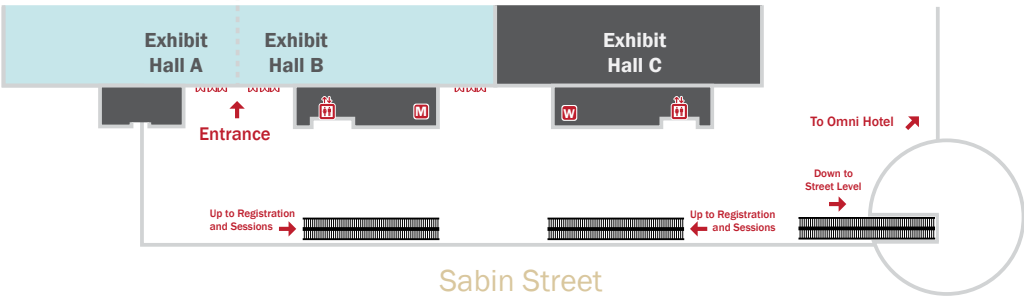
8:00 am – 9:00 am	Concurrent Roundtables	552AB, 555AB, 556AB (L5)
9:00 am – 9:30 am	Break	Ballroom Pre-function (L3)
9:30 am – 11:00 am	Concurrent Sessions	Ballrooms A & BC (L5)
11:00 am – 11:30 am	Break	Ballroom Pre-function (L3)
11:30 am – 1:00 pm	After the Screen: Continuing to Put Families at the Forefront	Ballroom A (L5)
1:00 pm	Closing and Adjournment	Ballroom A (L5)

Convention Center Map

LEVEL 5 • Meeting Rooms



LEVEL 3 • Exhibit Hall



Level 5

- Conference registration
- Plenary sessions
- Concurrent sessions
- Roundtables
- Innovate! sessions
- Quiet room
- Speaker ready room

Level 3

- Exhibit booths
- Posters
- Box lunches, breaks, receptions
- Concurrent sessions
- Roundtables

Welcome to Providence!

On behalf of the Newborn Screening (NBS) Symposium Planning Committee, welcome to the 2025 APHL Newborn Screening Symposium! We are thrilled to have you join us in beautiful Providence, Rhode Island (RI).

The Symposium begins with one of its most valuable sessions—Newborn Screening Stories: Perspectives from Individuals with Lived Experiences. During this session, we'll hear from families about their personal experiences with the NBS system. Once the panel ends, we hope you'll join your fellow attendees for a Welcome Reception in the Exhibit Hall and continue connecting during the evening's social hours.

Throughout the next four days, you will hear sessions on long-term follow-up, quality improvement, engaging with families, adding new disorders to state screening panels, how to improve false-positive rates and more. Make sure you join the Innovate! sessions to hear about new technologies and the roundtables, which will focus on an array of subjects such as higher-tier testing in newborns, workforce resiliency, NBS timeliness and the National Institutes of Health NBS by Whole Genome Sequencing Collaboratory, among other topics.

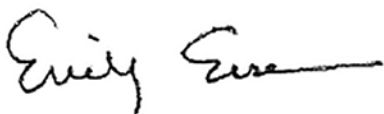
Our keynote session will begin with an official welcome from Dr. Jerome Larkin, the director of the Rhode Island Department of Health. We'll then hear from three speakers, all of whom will share their thoughts on how we can create awareness and appreciation for the NBS system. You'll leave feeling re-energized, with a better understanding of how to manage change. Other not-to-miss events include the Ignite the Night! presentations, a discussion focused on the findings and recommendations from Newborn Screening in the United States: Sustaining and Advancing Excellence and a debate around the benefits and challenges of both consented and un-consented approaches to genomic sequencing in NBS.

Don't forget to stick around for the Rapid Poster Presentations! Following the presentations, head down to the Exhibit Hall where you can continue the conversation with poster presenters and mingle with exhibitors. Attend the Newborn Screening Awards Ceremony on Tuesday, where colleagues will be honored for their amazing achievements and, finally, let loose and relax with your colleagues at our off-site social event on Wednesday evening.

As always, we will continue to put families first by focusing on what happens after the newborn screen in our final session of the conference. If your schedule allows, join us for a bus trip to the New England Newborn Screening Program at the University of Massachusetts Chan Medical School to learn more about how the program provides laboratory screening and clinical follow-up to five New England states.

While you're in Providence, we hope you have a chance to see the city! Come a day early to catch WaterFire, the city's award-winning fire and music installation located in downtown Providence. During the week, stroll the beautiful campuses of Brown University and the RI School of Design (RISD). Foodies won't want to miss all the great dining on Federal Hill, located just a short walk from the Convention Center.

We can't wait to make this a memorable week of connection and learning from each other.



Emily Eisenstein, MPH

Newborn Screening Program Manager, Rhode Island Department of Health
2025 APHL Newborn Screening Symposium Planning Committee Chair

Association of Public Health Laboratories

Vision: A healthier world through quality laboratory systems

Mission: Shape national and global health outcomes by promoting the value and contributions of public health laboratories and continuously improving the public health laboratory system and practice.



The Association of Public Health Laboratories (APHL) is a non-profit 501(c)(3) organization representing governmental laboratories that monitor and detect public health threats, including emerging infectious disease surveillance, detection of metabolic and genetic conditions in newborns, water contamination identification and foodborne outbreak detection. APHL's members are state, local, county and city public health laboratories, state and local environmental health laboratories, state agricultural laboratories, corporations, individual and student members with an interest in public health laboratory issues, and organizations that share common goals with APHL.

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2025 APHL Newborn Screening Symposium Planning Committee

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General Information

Registration Desk Hours

West Pre-function, 5th Level

Sunday, October 5	2:00 pm — 5:00 pm
Monday, October 6	7:30 am — 5:30 pm
Tuesday, October 7	7:30 am — 5:00 pm
Wednesday, October 8	7:30 am — 4:30 pm
Thursday, October 9	7:30 am — 1:00 pm

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Mobility

The Rhode Island Convention Center is an Americans with Disabilities Act (ADA) compliant center. There is a skywalk available from the Omni Providence to the Convention Center. There are entrance doors to the convention center equipped with ADA buttons. Please visit the registration desk with any questions.

Meeting Location and Headquarters Hotel

Conference sessions and events:
Rhode Island Convention Center

Headquarters hotel:
Omni Providence
One Exchange Street, Providence, RI 02903

Secondary hotel:
Hilton Providence
21 Atwells Avenue, Providence, RI 02903

Complimentary Wireless Internet

Network: **APHL2025NBSS**
Password: **2025NBSS**

Lactation Room Available

A Mothering Room is located outside of Exhibit Hall AB. Please visit the registration desk for more information.

Quiet Room Available

Step away from the busy symposium atmosphere and retreat to our Quiet Room. This room will be available throughout the symposium in Room 551B.

Download the Mobile App

Available in your app store! Search for "APHL Conferences" in your app store to download; within the app, you'll find the NBS 2025 event. *You must log in with a **username and password** emailed to each registrant from APHL in the week before the conference.*

Access details on sessions, speakers, posters and exhibitors before the conference and onsite; navigate the convention center; personalize your experience by tagging sessions, exhibitors and creating exportable notes. Receive alerts, reminders and changes in real time. *Session evaluations must be completed in the app in order to receive P.A.C.E.® credit.*



Continuing Education Credits

APHL Audience

This intermediate-level symposium, intended for newborn screening (NBS) and genetics laboratory professionals, follow-up program personnel, genetic counselors, public health nurses, specialists, clinicians, health care practitioners, students, advocacy organizations and individuals with lived experiences will further educate attendees about state, national and international NBS, genetic testing and policy issues important to public health NBS systems.

P.A.C.E.® Continuing Education Credit

APHL is an approved provider of continuing education programs in the clinical laboratory sciences through the American Society of Clinical Laboratory Science (ASCLS) P.A.C.E. program. Attendees may earn up to 18.5 P.A.C.E. credits by attending the entire symposium.

Continuing Medical Education (CME), Continuing Nursing Education (CNE), Continuing Education Contact Hours (CECH) in Health Education (CHES) and Continuing Education Units (CEUs) for Genetic Counselors

CME, CNE, CHES and Genetic Counseling CEUs (up to 18.5 credit hours) will be provided. Please refer to the symposium app for more information.

Exhibit Hall Raffle — Visit with Exhibitors, Win Prizes!

Follow the instructions on the exhibit hall raffle card, found at symposium registration. Turn in your completed card to registration by 2:00 pm, Wednesday, October 8. Winners of the prizes will be announced starting at 4:00 pm on Wednesday in the exhibit hall. You must be present to win. Prizes include:

- **CIL Tote Goodie Bag** compliments of Cambridge Isotope Laboratories, Inc.
- **Apple Watch** compliments of EBF, LLC
- **\$100 Amazon Gift Card** compliments of HCU Network America
- **Gift Basket: Beakers & Brew: Lab-grade Coffee Essentials** compliments of OpenELIS Foundation
- **Sonos Move 2: Bluetooth and WiFi Portable Home Speaker** compliments of Quantabio
- **(2) \$100 Amazon Gift Cards** compliments of STAT Courier Service
- **\$100 Amazon Gift Card** compliments of Waters Corporation
- **\$250 Delta Airlines Voucher** compliments of APHL
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- **2026 NBS Symposium Full Registration** compliments of APHL

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Waters Corporation

Not-to-Miss Events

Newborn Screening Stories: Perspectives from Individuals with Lived Experiences

Ballroom A | Sunday, October 5 | 3:30 pm – 5:00 pm

Hear compelling personal accounts of how newborn screening and rare diseases impact the lives of those in our community and beyond. Follow their journeys from diagnosis to treatment and management and get to know this small but powerful group of individuals in the rare disease community.

Welcome Reception

Exhibit Hall AB | Sunday, October 5 | 5:00 pm – 6:30 pm

Reunite with old friends or meet up with new colleagues. Visit with our industry partners over drinks and hors d'oeuvres and continue the conversation with our panel speakers in The Living Room (front left of the exhibit hall).

Visit with Poster Authors

Exhibit Hall AB

Sunday, October 5 | 5:00 pm – 6:00 pm

Tuesday, October 7 | 1:15 pm – 2:15 pm

Visit the exhibit/poster hall and talk with poster authors who will be standing by their posters.

Visit The Living Room

Exhibit Hall AB

Join us in this cozy and relaxed area located just inside the exhibit hall to chat, connect and share resources with newborn screening advocates and individuals with lived experiences. *Seating kindly reserved for family members.*

Innovate! Sessions

Rooms 555AB and 556AB

Monday, October 6 | 8:00 am – 8:30 am and 2:45 pm – 3:30 pm

Tuesday, October 7 | 2:30 pm – 3:15 pm

Connect with and learn from your industry partners Bio-Rad Laboratories, Inc., Primary.Health, Revvity, Inc., and Waters Corporation.

APHL Newborn Screening Awards Ceremony

Ballroom A | Tuesday, October 7 | 3:30 pm – 5:00 pm

Applaud your colleagues and celebrate their achievements.

Ignite the Night!

Ballroom A | Monday, October 6 | 5:30 pm – 7:00 pm

Grab a bag of popcorn and hear from an array of fast-paced presentations from your newborn screening colleagues!

Agenda of Events

All times are US Eastern
Daylight Time (EDT).

Sunday, October 5

2:00 pm – 5:00 pm **Registration**

West Pre-function

3:00 pm – 3:30 pm **Welcome Address**

Ballroom A

Presenters:

- Emily Eisenstein, MPH, Rhode Island Department of Health
- Scott Becker, MS, Association of Public Health Laboratories
- Scott Shone, PhD, HCLD(ABB), North Carolina Department of Health and Human Services
- Jelili Ojodu, MPH, Association of Public Health Laboratories

3:30 pm – 5:00 pm **Newborn Screening Stories:
Perspectives from Individuals with
Lived Experiences**

Ballroom A

Moderators:

- Jesus Ayala-Figuero, MPH, Rhode Island Department of Health
- Stacey Quesada, MPH, Center for Public Health Innovation

Presenters:

- Giorgi Segama
- Lindsey Coupe
- Faith Porter

Hear compelling personal accounts of how newborn screening and rare diseases impact the lives of those in our community and beyond. Follow their journeys from diagnosis to treatment and management and get to know this small but powerful group of individuals in the rare disease community.

5:00 pm – 6:30 pm **Welcome Reception**

Exhibit Hall AB

Visit the exhibits and posters and continue the conversation with our newborn screening families in “The Living Room,” located near the Exhibit Hall entrance.

5:00 pm – 6:00 pm Poster authors P-1 to P-59 available to discuss
Exhibit Hall AB their posters

Sunday, October 5

6:30 pm – 8:00 pm

Newborn Screening Laboratory Social Hour

Ballroom D

The Carol Johnson Follow-up and Family Social Hour

Ballroom B

Whether you are a seasoned socialite or just looking to meet new people, these mixers promise opportunities to make connections, share unique challenges and achievements and unwind in a fun space. Cash bar and desserts to be provided, and advanced registration is required.

Monday, October 6

7:30 am – 5:30 pm

Registration

West Pre-function

7:30 am – 9:00 am

Coffee and light breakfast

Rooms 555 & 556 Foyer

8:00 am – 8:30 am

Innovate Sessions

Room 555AB

Bio-Rad Laboratories, Inc.

See page 26 for
session details.



8:30 am – 4:00 pm

Exhibit and Poster Hall Open

Exhibit Hall AB

8:30 am – 8:45 am

Break – Visit exhibitors and posters

Exhibit Hall AB

8:45 am – 10:15 am

Concurrent Sessions

Engaging with Families: From Diagnosis to Follow-up

Ballroom A

588-854-25 • 1.5 contact hours for this session

Moderators:

- Jesus Ayala-Figuero, MPH, Rhode Island Department of Health
- Betty Vohr, MD, Women & Infants Hospital of Rhode Island

From Hashtags to Health Decisions: A Qualitative Analysis of Social Media Posts on Newborn Screening

Stacey Quesada, MPH, Center for Public Health Innovation

Monday, October 6

Bridging the Gap: Enhancing Support for Newborns Diagnosed with Sickle Cell Disease through Community-based Partnerships and Digital Innovation

Fran Altmaier, Arizona Department of Health Services

Enhancing Pathways: Streamlining Hearing Loss Referrals to Regional Genetic Centers

Hilary Fryman, RN, Tennessee Department of Health

It's Not the Ride, It's the Resources: Disparities in Newborn Screening Follow-up

Tory Kaye, MPH, Minnesota Department of Health

Evaluating the Early Check Model for Education and Consent in Large-scale Genetic Screening Program: A Mixed-methods Approach

Melissa Raspa, PhD, RTI International

In this session, attendees will identify ways to support families from diagnosis to follow-up. Speakers will delve into reasons why parents may refuse newborn screening (NBS), highlight the impact of community-based interventions on reducing barriers to care, discuss why families may be interested in genetic testing to investigate diagnoses and address disparities between families who complete follow-up and those who do not. Resources for expectant and new families will be highlighted, all with the goal of shaping effective, equity-focused solutions for NBS programs.

After completing this session, the participant will be able to:

- Identify at least one reason as to why families might opt-out of NBS
- Examine ways to create partnership opportunities within the sickle cell disease community
- Summarize guidelines regarding genetic testing and counseling for infants diagnosed with hearing loss
- Identify methods for analyzing newborn screening datasets for disparities
- List the barriers influencing the successful adoption of electronic consent models in population-based genomic NBS

Quality Improvement, Control and Assurance Activities: Part One

Ballroom BC

588-855-25 • 1.5 contact hours for this session

Moderators:

- Elli Laitinen, MS, New York State Department of Health
- Joseph Orsini, PhD, New York State Department of Health

Methionine Makeover: Chopping Cutoffs to Catch Homocystinuria in Colorado's Newborns

Erica Wright, MS, CGC, University of Colorado/Children's Hospital Colorado

Maryland's Adoption of N-acetyltirosine to Reduce False-positive Reports

Suraj Panthi, PhD, Maryland Department of Health

Monday, October 6

One Spot, Two Methods: Multiplexed FIA and LC-MS/MS Analysis of Amino Acid, Organic Acid and Fatty Acid Oxidation Disorders, ADA-SCID, GAMT Deficiency, ALD, Homocysteine and N-acetyltyrosine to Reduce False-positives and Negatives
Adrienne Manning, Connecticut Department of Public Health

NeoBase2 Non-derivatized MSMS kit (NB2): Comparison Between Waters Acquity TQD and SCIEX Triple Quad 4500MD System

Krystyna Weiss-Pawlak, MSE, Michigan Department of Health and Human Services

One and Done: Establishing Confidence in Initial NBS Results

Victoria Floriani, MS, New Jersey Department of Health

In part one of this two-part session, presentations will address ways to enhance the NBS system. Speakers will discuss ways to improve time to diagnosis and reduce false-positives, how to utilize a single blood spot punch, compare the accuracy, sensitivity and precision between two instruments and address ways to decrease the turn-around time of reporting time-critical results. These improvements to the NBS system will ultimately reduce burdens on follow-up teams, decrease stress for providers and families and improve laboratory efficiency.

After completing this session, the participant will be able to:

- Summarize efforts to improve sensitivity for screening for homocystinuria
- Identify ways to reduce false-positive reports in NBS
- Identify internal standards to add to a flow injection analysis tandem mass spectrometry method to improve accuracy
- Compare the instrument performance differences between two systems
- Summarize how discontinuing unnecessary retesting could improve time-to-intervention

10:15 am – 10:30 am Break – Visit exhibitors and posters

Exhibit Hall AB

10:30 am – 12:30 pm

The Value of the Newborn Screening System: How to Create Awareness, Manage Change and Re-energize Our Passion for Public Health

Ballroom A

588-856-25 • 2.0 contact hours for this session

Moderators:

- Patrice K. Held, PhD, FACMG, Oregon State Public Health Laboratory
- Emily Eisenstein, MPH, Rhode Island Department of Health

Presenters:

- Jerome Larkin, MD, Rhode Island Department of Health
- Brian C. Castrucci, DrPH, de Beaumont Foundation

- Kris Liebau, ACC, CMQ/OE, CQE, Action & Ease Leadership Coaching, LLC
- Scott Shone, PhD, HCLD(ABB), North Carolina Department of Health and Human Services

The landscape of public health NBS is continually changing. Whether it is a result of new technologies, environmental and social disruptions or workforce turnover and budgetary concerns, public health professionals must adapt to the many challenges that come their way. This keynote address will focus on how we can build momentum to create awareness and appreciation for NBS. Important questions around how we can be bolder when talking about our work and why we should celebrate the small wins will be addressed. Building a resilient system will take time, but attendees will be given the tools to navigate change and uncertainty and leave feeling re-energized for the work they do every day.

After completing this session, the participant will be able to:

- Identify techniques to build connections within the community
- Examine the current landscape of public health programs
- Plan how to craft the right message when discussing the issues that public health programs are currently facing
- Identify actionable steps to create a resilient public health program

12:30 pm – 2:00 pm

Lunch (on your own)

1:30 pm – 2:00 pm

Dessert

Exhibit Hall AB

2:00 pm – 2:30 pm

Sustaining and Advancing Excellence in Newborn Screening: Implementing the Recommendations of the National Academies Report

Ballroom A

588-857-25 • 0.5 contact hour for this session

Presenters:

- Scott Shone, PhD, HCLD(ABB), North Carolina Department of Health and Human Services
- Beth Tarini, MD, MS, MBA, Children's National Hospital
- Mei Baker, MD, University of Wisconsin School of Medicine and Public Health

This session will highlight findings and recommendations from the report, Newborn Screening in the United States: Sustaining and Advancing Excellence. Grounded in broad input—including families, clinicians, public health officials and researchers, the report calls for increased national coordination to build on the strengths of state and territorial programs and to support more efficient and effective responses to shared challenges. It also calls for actions to advance the excellence of NBS programs, earn and sustain public trust and prepare public health NBS to meet future needs.

Monday, October 6

After completing this session, the participant will be able to:

- Summarize the report's vision into actionable steps that strengthen public health NBS across the United States
- Identify strategies to promote transparency between providers and families
- Advocate for the responsible application of technologies to public health NBS

2:30 pm – 2:45 pm

Exhibit Hall AB

Break – Visit exhibitors and posters

2:45 pm – 3:30 pm

Room 555AB

Innovate Sessions

Waters Corporation

See page 26 for
session details.



3:30 pm – 3:45 pm

Exhibit Hall AB

Break – Visit exhibitors and posters

3:45 pm – 5:15 pm

Concurrent Sessions

Beyond Diagnosis: Strategies for Long-term Follow-up

Ballroom A

588-858-25 • 1.5 contact hours for this session

Moderators:

- Sondra Rosendahl, MS, CGC, Minnesota Department of Health
- Virginia Sack, MS, CGC, New York State Department of Health

Newborn Screening Long-term Follow-up Roadmap: A Pathway for Developing State-based Systems

Marci Sontag, PhD, Center for Public Health Innovation

Time to Pivot? A Threat or an Opportunity to Keep Newborn Screening Long-term Follow-up Alive in New York

Kathy Chou, PhD, New York State Department of Health

From Diagnosis to Lifelong Care: Arizona's Journey in Newborn Screening Long-term Follow-up

Fran Altmaier, Arizona Department of Health Services

Longitudinal Follow-up of Sickle Cell Disease in Minnesota

Lexie Barber, MPH, Minnesota Department of Health

Confounding Genotypes: Clinical and Biochemical Analysis of Individuals with Common GALC Gene Variants and the Impact on Newborn Screening Follow-up for Krabbe Disease

Amy White, MS, CGC, Mayo Clinic

Monday, October 6

Long-term follow-up (LTFU) is an important system created with the intent of providing access to care for families following the short-term follow-up period. However, variability between states around outreach and treatment can cause disparities in who receives care and for how long. In this session, presentations will illuminate how NBS programs can utilize a “roadmap” to successfully implement their LTFU activities, ways to establish an LTFU data repository and the challenges associated with longitudinal follow-up for conditions such as sickle cell disease and Krabbe Disease.

After completing this session, the participant will be able to:

- Design the phases of development for a long-term follow-up program for NBS
- List the collaborators required to build an online long-term follow-up data repository
- Plan how to connect families to resources following an NBS diagnosis
- Identify gaps in care for people living with sickle cell disease
- Identify common gene variants found in infants with abnormal NBS results

Applications for Molecular Technology and Next Generation Sequencing

Ballroom BC

588-859-25 • 1.5 contact hours for this session

Moderators:

- Jerusalem Alleyne, PhD, State Hygienic Laboratory at the University of Iowa
- Christopher N. Greene, PhD, Centers for Disease Control and Prevention

Global Genomic Newborn Screening Initiatives: Diverse Models and Public Health Implications

Nidhi Shah, MD, FACMG, International Consortium on Newborn Sequencing

Newborn Genome Sequencing: Perspectives from a Public Health Laboratory

Denise Kay, PhD, New York State Department of Health

The Spectrum of Variants Detected by Newborn Screening for Multiple Disorders

Anne Comeau, PhD, New England Newborn Screening Program

The Incorporation of Targeted DNA Sequencing in a Multi-omics Screening Strategy into the New South Wales Newborn Screening Programme

Tiffany Wotton, New South Wales Newborn Screening Program

Implications for Cystic Fibrosis (CF) Diagnoses After Newborn Screening in a Heterogeneous Population: A Retrospective Application of Various CFTR Variant Panels or Sequencing to 25 Years of CF Genotypes

Jaime Hale, MS, New England Newborn Screening Program

This session will evaluate the role of genomics in routine NBS. Presentations will focus on comparing programs globally to identify common trends, challenges and implications for sustainable implementation. Speakers will also review how NBS programs manage variants of uncertain significance and utilize targeted DNA sequencing. Ultimately the benefits and

challenges of next-generation sequencing will be addressed, focusing on important factors such as turn-around time, cost and infrastructure.

After completing this session, the participant will be able to:

- Compare genomic NBS initiatives across different regions
- Examine workflows for processing dried blood spots for newborn sequencing studies
- Summarize how variants of uncertain significance can affect ability to predict disease
- State the utility of targeted DNA sequencing in the NBS setting
- Indicate how expanded panels may offer only limited improvements to screening sensitivity

5:15 pm – 5:30 pm

Break

5:30 pm – 7:00 pm

Ignite the Night!

Ballroom A

Grab a bag of popcorn and hear an array of fast-paced presentations from your newborn screening colleagues!

Moderator:

- Breonna Preston, MPH, CHES, California Newborn Screening Program

Calling All Voices: Let's Talk About NBS and Uncertainty

Aaron Goldenberg, PhD, MPH, Case Western Reserve University

Newborn Screening Guidelines: Who Makes Them?

The CLSI Members

Using a Pictograph to Convey NBS Information

Kimberly Noble Piper, RN, CPH, CPHG, Iowa Department of Health and Human Services

What's New with the CDC?

Rachel Lee, PhD, Centers for Disease Control and Prevention,
Carla Cuthbert, PhD, FACMG, Centers for Disease Control and Prevention and
Amy Gaviglio, MS, CGC, Connetics Consulting

Letters in ACTION: Educating PCPs to Accelerate Hemoglobinopathytrait Referrals with Modified NBS ACT Sheets

Tyra Mills, MPH, APHL/CDC Newborn Screening Bioinformatics Fellow, Nebraska
Department of Health and Human Services and
Sarah Ward, MPH, Nebraska Department of Health and Human Services

Alternative Funding Strategies for NBS Programs

Sabra Anckner, RN, MSN, Association of Maternal and Child Health Programs

Monday, October 6

The Long-term Follow-up Expedition: A Practical Guide to Planning a Successful Journey

Yvonne Kellar-Guenther, PhD, Center for Public Health Innovation and
Marci Sontag, PhD, Center for Public Health Innovation and the Expedition Crew

There's a Journal You All Should Know About

Ralph Fingerhut, PhD, FAMH, EuSpLM, Synlab MVZ Weiden

The Public Health Laboratory Fellowship Program: An APHL-CDC Initiative

Courtney Sumbly, MS, Association of Public Health Laboratories and the Newborn
Screening Fellows

The Details are MIRACULOUS

Paloma Juarez, Kansas Department of Health and Environment

Empowering Newborn Sequencing with Streamlined High Molecular Weight Genomic DNA Extraction from DBS and Whole Genome Library Preparation using sparQ DBS-seq Workflow

Subrata Panja, PhD, Quantabio

Born to Screen: How CAPILLARYS 3 DBS Changes the Game

Matthew C. Wagner, PhD, Sebia, Inc.

Automating Routine Tasks in Unsatisfactory Follow-up Workflow with OMNI-LAB

Joe Huynh, PhD, Waters

The Variant Files: Cracking the Case of Rare Hemoglobins

Marco Flamini, Bio-Rad Laboratories, Inc.,
Dusty Rhodes, Bio-Rad Laboratories, Inc. and
Rick Cowan, Bio-Rad Laboratories, Inc.

More than Just Cheerleaders

Dean Suhr, MLD Foundation and Other Newborn Screening Advocates

It's the Little Things

The Revvity Team

Monday, October 6

Tuesday, October 7

7:30 am – 5:00 pm **Registration**

West Pre-function

8:30 am – 9:00 am **Coffee**

Exhibit Hall AB

8:30 am – 4:00 pm **Exhibit and Poster Hall Open**

Exhibit Hall AB

9:00 am – 10:30 am **Concurrent Sessions**

Financial, Legal, Ethical, Policy and Social Implications for Newborn Screening

Ballroom BC

588-860-25 • 1.5 contact hours for this session

Moderators:

- Emily Eisenstein, MPH, Rhode Island Department of Health
- Genevieve Smith Beagen, RN, Rhode Island Department of Health

Evaluating the Cost of Informed Consent in Newborn Screening: Insights from ScreenPlus

Nicole Kelly, MPH, Montefiore Einstein

Assessing the Accuracy of Race and Ethnicity on Newborn Screening Cards

Anna Howard, MPH, Washington State Department of Health

A Qualitative Study on Parental Experiences with Genetic Counseling After a Positive Newborn Screen for Recently Added Conditions on the Recommended Uniform Screening Panel

Macie Hricovec, MS, Case Western Reserve University

Nest Digital Genetics Navigator Facilitates Scale While Meeting Participant Experience and Education Needs

Rebecca Reimers, MD, MPH, Rady's Institute of Genomic Medicine

Evolving ethical and legal standards in the field of NBS have led to NBS programs needing to closely examine the policies and procedures currently in place. This session will help attendees address the need for more scalable, accessible informed consent processes, evaluate the accuracy of race and ethnicity demographic information on NBS cards and examine parental preferences for earlier involvement of genetic counselors. Finally, speakers will discuss why programs should consider multimodal methods of patient engagement, education and consent.

Tuesday, October 7

After completing the session, the participant will be able to:

- Examine the scalability of various informed consent recruitment strategies
- Evaluate the implications of inaccurate NBS race data on health equity analyses
- Define the role of genetic counselors in enhancing NBS programs
- Identify ways to facilitate patient education while minimizing staff time

To Add or Not to Add: Processes and Considerations for Adding New Disorders to State Screening Panels

Ballroom A

588-861-25 • 1.5 contact hours for this session

Moderators:

- Carla Cuthbert, PhD, FACMG, Centers for Disease Control and Prevention
- Denise M. Kay, PhD, New York State Department of Health

Tour the Sausage Factory: A Noteworthy Period of Newborn Screening Policymaking in Washington State

Megan McCrillis, MPH, Washington State Department of Health

Minnesota's Experience of Screening for Duchenne Muscular Dystrophy

Trenna Lapacinski-Ludens, Minnesota Department of Health

Late-infantile Krabbe Disease: Something Worth Screening For!

Dietrich Matern, MD, PhD, Mayo Clinic

The Low Citrulline Conundrum: Reporting Low Citrulline Levels in New York State

Virginia Sack, MS, CGC, New York State Department of Health

Advancing Rare Disease Screening: Development of a Targeted Metabolomics Panel for Biomarker Evaluation

Sara Smith, PhD, Revvity Omics

In this session, presenters will discuss factors to consider when adding new disorders to state screening panels. The first presentation will highlight four recent cases of unique NBS policy in Washington State. Subsequent presenters will examine Minnesota's first six months of screening for Duchenne Muscular Dystrophy (DMD) and ways that programs can implement NBS for Krabbe Disease. Barriers to successfully screening for ornithine transcarbamylase deficiency (OTCD) and a proposal to develop assays that can be employed in NBS for neurodevelopmental disorders will be highlighted.

After completing the session, the participant will be able to:

- Hypothesize alternate pathways that NBS programs can take to enact policy changes
- Reproduce Duchenne Muscular Dystrophy (DMD) screening processes
- Summarize the ethical decisions that need to be made when implementing a new condition
- Identify barriers to successfully screening for ornithine transcarbamylase deficiency (OTCD)
- Argue the importance of pilot studies to identify disorders that should be under consideration for addition to state screening panels

10:30 am – 10:45 am

Break – Visit the exhibitors and posters

Exhibit Hall AB

10:45 am – 11:15 am

Debating the Necessity of Informed Consent for Genomic Sequencing in Newborn Screening

Ballroom A

588-862-25 • 0.5 contact hour for this session

Presenters:

- Aaron Goldenberg, PhD, MPH, Case Western Reserve University
- Amy Gaviglio, MS, CGC, Connetics Consulting

Join us for a debate around the benefits and challenges of both consented and un-consented approaches to genomic sequencing in NBS. Speakers will explore the implications of each approach, including the potential impact on public trust in NBS and the public health system. The practical challenges faced by NBS programs in obtaining meaningful and truly “informed” consent will also be addressed, highlighting the complexities involved in communicating the nuances of genomic data to parents. Through this dialogue, the community can collaboratively address the ethical and practical implications of integrating genomic sequencing into NBS, ultimately guiding policy decisions that reflect both technological advancements and public values.

After completing the session, the participant will be able to:

- Evaluate the ethical foundations of mandatory NBS versus informed consent for genomic sequencing
- Compare the challenges of consented and un-consented approaches to genomic sequencing in NBS
- Summarize the implications of consent for genomic sequencing on the effectiveness of NBS programs

11:15 am – 11:30 am

Break – Visit the exhibitors and posters

Exhibit Hall AB

11:30 am – 1:00 pm

Concurrent Sessions

Experience Counts: Improving Screening Methods for Conditions on the Recommended Uniform Screening Panel (RUSP)

Ballroom A

588-863-25 • 1.5 contact hours for this session

Moderators:

- George Dizikes, PhD, HCLD/CC(ABB), Laboratory Quality Assurance, LLC
- Inderneel Sahai, MD, New England Newborn Screening Program

Tuesday, October 7

California's Experience with Newborn Screening for Guanidinoacetate Methyltransferase Deficiency: Insights into Tier-1 and Tier-2 Testing and the Arginine/Guanidinoacetate Ratio

Kuldeep Dhillon, MS, CCS(CA), MB(ASCP)cm, California Department of Public Health

One Punch

Michael Gelb, PhD, University of Washington

Advancing Newborn Screening for Krabbe Disease Through Accelerated GALC Enzyme Activity Testing

Elya Courtney, MPH, Centers for Disease Control and Prevention

California's Experience with Mucopolysaccharidosis Types I and II Screening: Implementing an Improved Three-tier Strategy

Kuldeep Dhillon, MS, CCS(CA), MB(ASCP)cm, California Department of Public Health

Expanded Homocystinuria Screening in Georgia: A Two-year Pilot Project

Angela Wittenauer, MSN, FNP-RN, Emory University

This session will examine how NBS programs are improving their screening processes for conditions on the Recommended Uniform Screening Panel (RUSP). Presentations will consider strategies for screening for guanidinoacetate methyltransferase (GAMT) deficiency, Krabbe Disease and mucopolysaccharidosis types I and II (MPS-I and MPS-II). Discussions will also focus on how RUSP conditions can be consolidated into a single liquid chromatography-tandem mass spectrometry (LC-MSMS) assay using one dried blood spot punch and how a pilot project helped better detect disorders with elevated homocysteine while also reducing false-positives.

After completing the session, the participant will be able to:

- Illustrate the importance of tiered testing and analyte ratio analysis in early detection of rare metabolic disorders
- Indicate how conditions can be consolidated into a single liquid chromatography-tandem mass spectrometry (LC-MSMS) assay using one dried blood spot punch
- Examine method-based advancements for first-tier lysosomal storage disorder screening
- Define the role of NBS in the early detection of mucopolysaccharidosis types I and II (MPS I & II)
- Summarize the challenges in current methodologies for classic homocystinuria (HCU) screening

Improving Training, Education and Communication

Ballroom BC

588-864-25 • 1.5 contact hours for this session

Moderators:

- Breonna Preston, MPH, CHES, California Newborn Screening Program
- Emily Reeves, BSN, RN, CPM, Florida Department of Health

From Storyboard to Screen: Lessons Learned from Arizona's Parent-focused Newborn Screening Video Series

Fran Altmaier, Arizona Department of Health Services

Tuesday, October 7

Survey of Primary Care Provider Knowledge of Equity Within the Cystic Fibrosis Screening Process

Shelby Heppe, MPH, Michigan Department of Health and Human Services

Developing a Standardized Training Protocol: Training Specialist Perspectives

Marina Choi, MPH, Washington State Department of Health

Participatory Design of a Chatbot for Sickle Cell Trait Newborn Screening Results

Courtney Gauchel, RN, MS, Association of Public Health Laboratories

In this session, presenters will discuss ways to strengthen community engagement with the NBS process. Attendees will leave with practical tips for effectively integrating multimedia storytelling into outreach to parents, identifying ways to support the educational needs of primary care providers, implementing a training position to improve follow-up and using a scripted chatbot to offer timely, accessible education and emotional reassurances to parents.

After completing the session, the participant will be able to:

- Illustrate the end-to-end process of developing parent-centered multimedia content for NBS education
- Identify inequities in screening protocols for cystic fibrosis (CF)
- Evaluate the potential benefit of developing a standardized training protocol for NBS program staff
- Summarize the participatory design process used to develop a digital health intervention resource for NBS follow-up

1:00 pm – 1:15 pm

Ballroom A

Rapid Poster Presentations

See page 25 for session details.

1:15 pm – 2:30 pm

Exhibit Hall AB

Lunch provided while you visit the exhibitors and posters

1:15 pm – 2:15 pm

Exhibit Hall AB

Poster authors P-60 to P-118 available to discuss their posters

2:30 pm – 3:15 pm

Room 555AB

Room 556AB

**Innovate Sessions
Revvity
Primary.Health**

See page 27 for session details.

3:15 pm – 3:30 pm

Break – Visit exhibitors and posters

3:30 pm – 5:00 pm

Ballroom A

2025 APHL Newborn Screening Awards Ceremony

Applaud your colleagues and celebrate their achievements. Moderated by Scott Becker, MS, Association of Public Health Laboratories and Scott Shone, PhD, HCLD(ABB), North Carolina Department of Health and Human Services.

Tuesday, October 7

Rapid Poster Presentations

Tuesday, October 7, 1:00 pm – 1:15 pm

This session will include short presentations of the following select posters. The full posters may be seen in the exhibit hall.

P-42: Implementing Next Generation Sequencing as Part of Newborn Screening in State Public Health Laboratories: Status and Barriers

Presenter: Holly Snyder, MS, LGC, Illumina, hsnyder@illumina.com

P-45: Enhancing Kansas Newborn Screening by Launching a Medical Directorship

Presenter: Kourtney Bettinger, MD, MPH, FAAP, Kansas Department of Health and Environment, kourtney.bettinger@ks.gov

P-18: Engaging Midwives in Washington State

Presenter: Lauren Barringer, PT, DPT, MPH, Center for Public Health Innovation, Lauren.Barringer@cphinnovation.org

P-86: Empowering Newborn Sequencing with Streamlined High Molecular Weight Genomic DNA Extraction from Dried Blood Spot and Whole Genome Library Preparation Using sparQ DBS-seq Workflow

Presenter: Subrata Panja, PhD, Quantabio, Subrata.Panja@quantabio.com

P-108: An Interactive, Searchable Database of LC-FAOD Gene Variants, Genotypes and Phenotypes

Presenter: Vanessa Rangel Miller, MS, MBA, CGC, Ultragenyx Pharmaceutical, Inc., vrangelmiller@ultragenyx.com

P-39: Establishing a Newborn Screening Center of Excellence to Advance Testing Technologies and Enhance Data Infrastructure in the Southeastern US

Presenter: Marie-Claire Rowlinson, PhD, D(ABMM), Florida Department of Health, marie-claire.rowlinson@flhealth.gov

P-44: Sowing the Seeds of Newborn Screening: The Iowa NBS Lab's STEM Outreach Initiative

Presenter: Jerusalem Alleyne, PhD, State Hygienic Laboratory as the University of Iowa, jerusalem-alleyne@uiowa.edu

P-1: digitalMLPA as a Potential Confirmatory Test for Congenital Adrenal Hyperplasia, Glucose-6-Phosphate Dehydrogenase Deficiency and Hemoglobinopathies in Newborn Screening

Presenter: Terence Diane Fabella, MS, PhD candidate, Amsterdam University Medical College, t.d.fabella@amsterdamumc.nl

P-4: Impacts of Genome Supplemented Newborn Screening

Presenter: Gianna D'Apolito, MS, Association of Public Health Laboratories, gdapolit@asu.edu

P-20: Creating a Shared Decision-making Toolkit for Midwives to Assist with Newborn Screening Discussions

Presenter: Yvonne Kellar-Guenther, PhD, Center for Public Health Innovation, Yvonne. Kellar-Guenther@CPHInnovation.org

P-75: Shortening Time to Diagnosis and Treatment: Adjusting Follow-up Action Schedules for Aminoacidopathy Borderline Results

Presenter: Elli Laitinen, MS, New York State Department of Health, elli.laitinen@health.ny.gov

P-19: Building a Newborn Screening Long-term Follow-up Program? There's a Guide for That

Presenter: Yvonne Kellar-Guenther, PhD, Center for Public Health Innovation, Yvonne. Kellar-Guenther@CPHInnovation.org

Innovate! Sessions

Monday, October 6

8:00 am – 8:30 am

Mastering Newborn Hemoglobinopathy Screens: Clear, Confident Interpretation for Better Outcomes

Bio-Rad Laboratories, Inc. • Room 555AB

Diamond Level Sustaining Member

Presenter:

Marco Flamini, Bio-Rad Laboratories, Inc.

Accurate interpretation of NBS results for hemoglobinopathies is critical as an aid for diagnosis and timely intervention. This session equips laboratories with the confidence and clinical insight needed to evaluate chromatograms effectively. Through case-based discussions from Bio-Rad's Library of Variants and expert-led guidance, attendees will learn to distinguish between different patterns, evaluate clinically meaningful findings, plan further actions for genotype confirmation, and communicate results with clarity to clinicians and care teams. Whether you are new to hemoglobinopathy screening or looking to refine your approach, this session will strengthen your diagnostic confidence and decision making.

2:45 pm – 3:30 pm

Transforming Multiplex Workflows: Innovation in LC-MS/MS QC and Data Review

Waters Corporation • Room 555AB

Gold Level Sustaining Member

Presenter:

Zackary (Ari) M. Herbst, Enfanos and Heather A. Brown, PhD, Waters

Join us for an in-depth look at the latest innovations driving efficiency, accuracy and flexibility in multiplex LC-MS/MS workflows. This session brings together expert speakers to highlight practical solutions for today's most complex analytical challenges. We will explore Enfanos MegaQC DBS for Multiplex Methods, a powerful QC solution designed to streamline validation and improve confidence in multi-analyte assays. We will also present a range of LC-MS/MS applications showcasing how Waters technologies are helping laboratories optimize performance.

Tuesday, October 7

2:30 pm – 3:15 pm

Seven in One — Advancing Lysosomal Storage Disorder Detection with a Single Injection, Including Mucopolysaccharidosis Type II

Revvity • Room 555AB

Diamond Level Sustaining Member

Presenter:

Hanna Polari, Revvity

Come learn about our newest, most advanced assay for screening of lysosomal storage disorders (LSDs): The NeoLSD™ 7plex LC-MSMS. During this presentation, you will learn how this kit brings efficiency to the forefront by simultaneously analyzing seven LSDs, including MPS II (Hunter syndrome)—a meaningful addition to all laboratories for early detection and timely intervention. This powerful assay combines trusted accuracy with our intuitive, streamlined workflow, helping laboratories get more out of every run. For research use only. Not for use in diagnostic procedures. Product under development.

Disrupting the Status Quo: Building the Next Generation of Surveillance and Case Management Systems

Primary.Health • Room 556AB

Gold Level Sustaining Member

Presenter:

Jack Hysell, Primary.Health and Amy Gaviglio, MS, CGC, Connetics Consulting

NBS is a cornerstone of public health, yet the technology supporting it has often lagged behind modern healthcare systems. This session will introduce how Primary.Health, in partnership with Amy Gaviglio of Connetics Consulting, is advancing new solutions through the development of a modern NBS surveillance system and a case management platform. We will explore how interoperability, electronic test ordering and results (ETOR) and real-time data sharing can strengthen case follow-up, improve coordination among programs and support families impacted by rare conditions. Attendees will not only learn about these emerging technologies, but also have the opportunity to provide feedback, share challenges and shape the future direction of this work. This interactive format is designed to spark discussion, exchange ideas and highlight the collective expertise of the NBS community. Come prepared to engage, learn and collaborate — and yes, there will be swag!

Wednesday, October 8

7:30 am – 4:30 pm **Registration**

West Pre-function

7:30 am – 9:30 am **Coffee**

Ballroom Pre-function

8:00 am – 9:00 am **Concurrent Roundtables**

Education and Outreach Success Stories: Innovative Strategies for Engaging Key Audiences in Newborn Screening

Room 555AB

588-865-25 • 1.0 contact hour for this session

Presenter: Marianna Raia, MS, CGC, Expecting Health

This roundtable will highlight a series of creative and adaptable NBS outreach initiatives implemented nationwide. These initiatives include engaging tools such as NBS-themed band-aids and bite-sized educational podcasts. Through small break-out discussion, participants will be introduced to 2–3 proven approaches for each collaborator group and gain insights into what made these efforts successful. Whether you are just beginning your education and outreach journey or looking to refresh and expand your current strategies, this roundtable offers an opportunity to gain practical ideas, build connections and feel inspired about the possibilities ahead.

After completing the session, the participant will be able to:

- Identify education strategies used by NBS programs to engage key collaborator groups
- Evaluate how successful outreach initiatives can be adapted to meet diverse program needs
- Design a collaborative network with peers to exchange NBS outreach success stories

Higher-tier Testing in Newborn Screening: Barriers, Facilitators and What's Next

Room 556AB

588-866-25 • 1.0 contact hour for this session

Presenters:

- Shawn Moloney, MPH, MLS(ASCP)cm, Michigan Department of Health and Human Services
- George Dizikes, PhD, HCLD/CC(ABB), Laboratory Quality Assurance, LLC
- Amy Gaviglio, MS, CGC, Connetics Consulting
- Kshea Hale, MPH, Association of Public Health Laboratories

Wednesday, October 8

As the NBS system expands to include new and more complex disorders, NBS programs must strive to provide comprehensive testing for these disorders while minimizing both false-positive and false-negative results. For many disorders, higher-tier testing is utilized to improve screening performance and is an approach already employed at some level by every NBS program in the US. Despite this reality, there are still obstacles programs must overcome to expand their higher-tier testing offerings, whether performed in-house or outsourced. APHL's New Disorders Subcommittee instituted a Higher-Tier Testing Workgroup to better understand the needs, barriers and obstacles to implementing higher-tier testing across NBS programs. Work on this topic has included administering a survey and performing an environmental scan of available higher-tier testing options across all currently screened disorders. This roundtable discussion will review the results from the survey and facilitate discussions around higher-tier testing with the goal of helping all NBS programs perform the highest quality and most impactful testing.

After completing the session, the participant will be able to:

- Identify the main higher-tier testing facilitators for NBS programs
- Summarize common themes in experiences regarding higher-tier testing
- Formulate strategies for overcoming challenges to higher-tier testing

Maintaining Workforce Resiliency in the Face of Change

Room 552AB

588-867-25 • 1.0 contact hour for this session

Presenter: Susan Tanksley, PhD, Texas Department of State Health Services

Workforce resiliency has never been more critical—or more challenging—as NBS programs navigate funding cuts, return-to-office mandates and persistent staffing shortages. This roundtable session invites participants to engage in an open, solutions-focused discussion about sustaining program operations, retention and morale during times of change. Mental health and wellness will also be a key theme, and participants will be invited to share programs, resources and initiatives that have effectively contributed to workforce resilience in their programs. Through peer-to-peer exchange, attendees will identify shared challenges, creative solutions and practices that can strengthen their program's capacity to navigate uncertainty. Participants will leave with a toolkit of new strategies to foster a resilient, supported and engaged workforce.

After completing the session, the participant will be able to:

- Examine practical strategies to enhance workforce retention
- Identify new approaches to navigating organizational change amid resource constraints
- Construct peer connections among professionals addressing similar workforce challenges

9:00 am – 4:30 pm

Exhibit and Poster Hall Open

Exhibit Hall AB

9:00 am – 9:30 am

Break – Visit the exhibitors and posters

Exhibit Hall AB

Wednesday, October 8

9:30 am – 11:00 am Concurrent Sessions

It's All About the Babies: How to Build Resiliency, Reduce Barriers and Improve the System

Ballroom A

588-868-25 • 1.5 contact hours for this session

Moderators:

- Patrice K. Held, PhD, FACMG, Oregon State Public Health Laboratory
- Aaron Goldenberg, PhD, MPH, Case Western Reserve University

Welcoming Kansas Babies Initiative

Drew Duncan, MA, Kansas Department of Health and Environment

Rethinking Timeliness: A Proposal for Updated Timeliness Categories

Amy Gaviglio, MS, CGC, Connetics Consulting

Scaling Mountains: A Journey of Advocacy, Resiliency and Quality Improvement of the Colorado Newborn Screening Program

Erica Wright, MS, CGC, University of Colorado/Children's Hospital Colorado

Reducing Barriers for Timely Recollection Utilizing Partnerships with Traveling Midwives

Drew Duncan, MA, Kansas Department of Health and Environment

South Carolina Improves Continuity of Operations Planning for Newborn Screening Program and Partner States

Beth Bair, MS, PhD, South Carolina Department of Public Health

This session will reinforce the importance of NBS and explore ways that NBS programs can continue to advance their missions. Presentations will highlight how traditional public health platforms can be used in creative ways as well as how to establish new initiatives to meet existing needs. As NBS programs continue to evolve, they must consider ways to establish timeliness goals, enhance quality improvement needs, establish community-based support for families and develop a Continuity of Operations Plan (COOP).

After completing the session, the participant will be able to:

- Indicate how NBS platforms can be leveraged to enhance postpartum outreach
- Compare the newly proposed timeliness categories
- Identify ways to engage stakeholders for legislative efforts
- Summarize how to facilitate timely recollection for a newborn with a suspected time-critical condition in the midwife community
- Design a Continuity of Operations Plan (COOP) or strategy to manage an emergency or disruptive event

Advances in Newborn Screening from Around the World

Ballroom BC

588-869-25 • 1.5 contact hours for the session

Moderators:

- Inderneel Sahai, MD, New England Newborn Screening Program
- Joseph Orsini, PhD, New York State Department of Health

NS2400: First Complete Solution for Automation of Immunoassays, Enzyme Assays and LC-MS/MS in Newborn Screening

Ralph Fingerhut, PhD, FAMH, EuSpLM, Synlab MVZ Weiden

MetabPlus: Enzymatic Microarrays for Enhanced Detection of Inherited Metabolic Disorders

Raquel Yahyaoui, MD, PhD, Malaga Regional University Hospital

Combined Biochemical and Genetic Analysis Enhances Diagnosis and Prenatal Assessment of Multiple Acyl-CoA Dehydrogenase Deficiency

Ting Chen, Xinhua Hospital, Shanghai Jiao Tong University School of Medicine

Japan's Nationwide EQCP Pilot for Severe Combined Immunodeficiency and Spinal Muscular Atrophy: Ensuring Accuracy and Promoting Standardization of qPCR-Based Newborn Screening

Nobuyuki Ishige, Tokyo Health Services Association

In this session, attendees will learn about NBS practices from programs around the world. Presentations will examine the possibility of complete automation in the NBS laboratory, discuss the experimental results of a study aimed at overcoming limitations to current NBS methods and highlight the critical role of combined biochemical and genetic screening in early diagnosis of multiple acyl-CoA dehydrogenase deficiency (MADD). Lastly, the development of a quality control program to ensure accuracy and consistency of severe combined immunodeficiency (SCID) and spinal muscular atrophy (SMA) will be discussed.

After completing the session, the participant will be able to:

- Advocate for the possibility of complete automation in the NBS laboratory
- Summarize the potential of reactomics-based enzyme microarrays to enhance the specificity of newborn screening for inherited metabolic disorders
- Identify the critical role of combined biochemical and genetic screening in early diagnosis of multiple acyl-CoA dehydrogenase deficiency (MADD)
- Design a quality control program for NBS conditions

11:00 am – 11:30 am Break – Visit the exhibitors and posters

Exhibit Hall AB

Wednesday, October 8

11:30 am – 1:00 pm Concurrent Sessions

Newborn Screening Panels and Performance

Ballroom A

588-870-25 • 1.5 contact hours for this session

Moderator:

- Guisou Zarbalian, MS, MPH, Association of Public Health Laboratories

Proposed RUSP Changes for Consistency and Transparency

Susan Tanksley, PhD, Texas Department of State Health Services

Evaluating False-positives and Positive Predictive Value in Newborn Screening:

A 3-part Discussion

Sarah McKasson, MPH, Association of Public Health Laboratories,

Ashley Comer, Association of Public Health Laboratories and

Amy Gaviglio, MS, CGC, Connetics Consulting

This session opens with a presentation from the Counting Conditions Workgroup, formed to address differences among states in counting primary and secondary conditions and in listing conditions on NBS panels. The speakers will share the workgroup's proposed condition naming conventions and a modified Recommended Uniform Screening Panel (RUSP). Next, NewSTEPS will present findings from a national assessment of outcomes among screen-positive newborns, including true positives, false-positives and other clinically relevant conditions. This multi-part analysis examines NBS program performance through the lens of positive predictive value (PPV), highlighting factors that drive variability and opportunities for improvement.

After completing the session, the participant will be able to:

- Summarize proposed naming conventions for conditions on the Recommended Uniform Screening Panel (RUSP)
- Indicate the role of positive predictive value (PPV) to evaluate newborn screening performance
- Examine false-positive trends in state and territorial NBS programs
- Identify strategies to reduce false-positives

Are We There Yet: Tools for Newborn Screening Health Information Technology

Ballroom BC

588-871-25 • 1.5 contact hours for this session

Moderators:

- Brendan Reilly, Texas Department of State Health Services
- Hugh Peebles, MLS, Tennessee Department of Health

Detor: A New Path for Newborn Screening Electronic Test Orders and Results

Rachel Shepherd, Association of Public Health Laboratories

Minnesota’s Roadmap for Electronic Test Orders and Results Testing

Heather Brand, Minnesota Department of Health

A Decade of Working with Newborn Screening Submitters Across Virginia to Improve Quality and Timeliness Through the Implementation and Expansion of Electronic Data Exchange

Emily Hopkins, MS, Virginia Division of Consolidated Laboratory Services and Willie Andrews, BSMT(ASCP), River Views Consulting, LLC

Utilizing Rhode Island’s Unique Integrated Child Health Information System to Improve Newborn Screening Follow-up

Emily Eisenstein, MPH, Rhode Island Department of Health

A Pilot Study to Implement FHIR Queries for Newborn Screening Clinical Follow-up Data

Gregory Bonn, MT(ASCP), Colorado State Public Health and Environment Laboratory

The landscape of NBS health information technology (HIT) is ever evolving, leaving NBS programs wondering, “Are we there yet?” In this session, presentations will explore data exchange solutions aimed at solving the challenges faced by public health laboratories. Speakers will illustrate how to map out ways to effectively send and receive accurate order and result messages, evaluate the processes and lessons learned from an electronic messaging solution meant to support all types of NBS submitters and reveal how programs can create more fluid and standardized follow-up care by investing in unique health information systems.

After completing the session, the participant will be able to:

- Indicate the benefits of a national standardized solution for electronic data exchange
- Plan for an NBS program’s electronic test orders and results (ETOR) needs
- Summarize the challenges within an organization for messaging implementation
- Differentiate the quality improvements of electronic messaging from manual submission
- Identify the benefits of an integrated child health information system
- Formulate the steps needed to gain buy-in to implement interoperability methods in NBS

1:00 pm – 2:30 pm
Exhibit Hall AB

Lunch provided while you visit the exhibitors and posters

Wednesday, October 8

2:30 pm – 4:00 pm Concurrent Sessions

The Newest Conditions: Newborn Screening for Metachromatic Leukodystrophy (MLD) and Multiplexing Strategies

Ballroom A

588-872-25 • 1.5 contact hours for this session

Moderators:

- George Dizikes, PhD, HCLD/CC(ABB), Laboratory Quality Assurance, LLC
- Jing Xiao, PhD, New York State Department of Health

The MLD Alliance: Facilitating Advancement of a Candidate Condition to Screen-ready Status

Charlotte Chanson, PGCert, MS, MSPH, Orchard Therapeutics

Newborn Screening for Metachromatic Leukodystrophy: Second-tier ARSA Activity Test

Dietrich Matern, MD, PhD, Mayo Clinic

Newborn Screening for Metachromatic Leukodystrophy and Cerebrotendinous Xanthomatosis in New York State

Joseph Orsini, PhD, New York State Department of Health

New Disorders and New Treatments: Considerations for Therapeutic Access for Newborn Screening Programs

Amy Gaviglio, MS, CGC, Connetics Consulting

Aaron Goldenberg, PhD, MPH, Case Western Reserve University

Optimizing Newborn Screening Efficiency Through Multiplex Biomarker Detection: Evaluation of a Triplex Assay for X-ALD, MLD and GAMT Deficiency

Khaja Basheeruddin, PhD, Illinois Department of Public Health

The session will delve into the newest conditions that NBS programs are exploring by highlighting existing resources, real-world experiences and implementation models. Speakers will address ways to reduce or eliminate false-positive results and explore lessons learned from a pilot screening for metachromatic leukodystrophy (MLD). Potential considerations around access to treatment and how to optimize the use of limited dried blood spots for three neurodegenerative disorders will be discussed.

After completing the session, the participant will be able to:

- Summarize strategies to develop actionable guidance for MLD NBS implementation
- Compare in-house second and third-tier testing
- Advocate the use of secondary markers to reduce first-tier false-positive rates
- Identify barriers to treatment for patients identified through NBS
- Indicate the importance of multiplexed biomarker detection

Quality Improvement, Control and Assurance Activities: Part Two

Ballroom BC

588-873-25 • 1.5 contact hours for this session

Moderators:

- Chanika Phornphutkul, MD, Rhode Island Hospital/Brown University Health
- Kathy Chou, PhD, New York State Department of Health

Resource Centers, Collaboratives and Grants, Oh My! Tools for Laboratories to Improve Cystic Fibrosis Newborn Screening

Marissa Taylor, MPH, Cystic Fibrosis Foundation

Comprehensive LC-MS/MS Method for Newborn Screening: Resolving Interferences, Reducing Second-tier Reflexes and Quantifying Over 70 Biomarkers

C. Austin Pickens, PhD, Centers for Disease Control and Prevention

Qualifying the Benefits of Active Hospital Outreach with Dynamic Quality Improvement

Bryan Wendel, Florida Department of Health

The Cystic Fibrosis ConSEQUENCE: Michigan's Experience Transitioning to Sequencing-based CF Screening, Implications of NGS and Lessons Learned

Erika Jackson, Michigan Department of Health and Human Services

Using CRISPR/Cas9 Genome Editing to Create Custom Cell Lines for Newborn Screening Quality Assurance Materials

David Cobb, PhD, Centers for Disease Control and Prevention

In part two of this two-part session, attendees will continue to learn ways to improve the NBS system. The first presentation will delve into how NBS guidelines can be time- and resource-intensive for state laboratories and ways to alleviate this stress. Subsequent presentations will focus on ways to define new conditions using a first-tier LC-MS/MS method, how to create accountability for birthing facilities and ways to think outside the box to accurately screen for conditions. The final presentation will discuss how CDC is developing new quality assurance materials for the NBS community using genome editing technology.

After completing the session, the participant will be able to:

- Identify resources to help improve sensitivity for CF NBS programs
- Compare the benefits of using first-tier LC-MS/MS over flow injection analysis
- Identify strategies to implement accountability changes in administrative rule
- Evaluate the benefits of NBS next generation sequencing (NGS) implementation
- Summarize the new quality assurance materials developed by CDC

4:00 pm – 4:30 pm

Raffle drawing

Exhibit Hall AB

6:00 pm – 10:00 pm

Off-site Social Event

Sponsored by Revvity

Wednesday, October 8

7:30 am – 1:00 pm **Registration**

West Pre-function

7:30 am – 9:30 am **Coffee**

Ballroom Pre-function

8:00 am – 9:00 am **Concurrent Roundtables**

Bridging Gaps in Newborn Screening Timeliness for Time-critical Disorders

Room 555AB

588-874-25 • 1.0 contact hour for this session

Presenter: Amanda Jenkins, MPH, MLS(ASCP), Association of Public Health Laboratories

The NewSTEPS Quality Indicators (QI) are performance metrics used to evaluate the NBS system. Longitudinal comparisons of these QI metrics within a program and comparisons to aggregate data across programs highlight opportunities for system-wide quality improvement initiatives. This roundtable will explore findings from NewSTEPS' analysis of turnaround times for time-critical disorders in both one- and two-screen programs. The goal is to identify key barriers and actionable strategies to improve reporting timeliness. Participants will collectively identify practical solutions and opportunities for quality improvements that can be tailored to their unique program.

After completing the session, the participant will be able to:

- Summarize the reasons behind the differences in reporting metrics between one- and two-screen programs
- Identify specific testing and reporting practices in one- and two-screen programs that contribute to variations in timeliness metrics
- Examine program practices to find quality improvement opportunities to enhance timely reporting

NBSxWGS Collaboratory: Development of a Virtual Gene Panel

Room 556AB

588-875-25 • 1.0 contact hour for this session

Presenters:

- Melissa Parisi, MD, PhD, Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health
- Ingrid A. Holm, MD, MPH, Boston Children's Hospital

The goal of this informal roundtable discussion will be to hear from newborn screening (NBS) program representatives and staff, such as those involved in NBS follow-up and care of screen-positive newborns, regarding the development of the virtual gene panel. The National Institutes of Health (NIH) Newborn Screening by Whole Genome Sequencing Collaboratory (NBSxWGS) will discuss potential criteria to use when selecting the conditions and genes, as well as gather NBS community feedback on which genes to include. The NIH team can use the feedback obtained from the roundtable to help inform the work and ensure the perspectives of the NBS community are incorporated.

After completing the session, the participant will be able to:

- Summarize the goals of NBSxWGS Collaboratory project
- Examine the feasibility of using sequencing in a state and territorial NBS program
- Identify criteria to use when selecting the conditions and genes for a virtual gene panel

Design That Listens: Building Better Reports with User-centered Design

Room 552AB

588-876-25 • 1.0 contact hour for this session

Presenter: Michele Gornick, PhD, Children's National Hospital

Effective NBS communication depends on role-specific, visually clear and action-oriented guidance using standardized terminology and explicit delineation of follow-up responsibilities. Given that the NBS results report is the primary communication tool for conveying findings to pediatric providers, it is critical that the report prioritizes their needs. We invite all NBS stakeholders to engage in this roundtable to learn about and apply principles of user-centered design to critically review their own NBS reports. In this roundtable, we will discuss feedback from pediatric providers regarding the perceived usability of current NBS results reports, identify redundancies and explore ways to reformat NBS reports using principles of user-centered design. The goal of this roundtable is to initiate a critical discussion about pediatric providers' experiences with NBS results reports, reflect on ways to reformat the reports to align with real user needs, enhance the process of communicating findings to providers and ultimately improve care for patients and families.

After completing the session, the participant will be able to:

- Summarize pediatric providers' experiences with NBS results reports
- Identify ways to reformat the NBS results report to align with real user needs
- Formulate ways to enhance the process of communicating findings to providers

9:00 am – 9:30 am

Break

Ballroom Pre-function

Thursday, October 9

9:30 am – 11:00 am Concurrent Sessions

Newborn Screening for Congenital Cytomegalovirus (cCMV)

Ballroom BC

588-877-25 • 1.5 contact hours for this session

Moderators:

- Norma Tavakoli, PhD, New York State Department of Health
- Carrie Wolf, MBS, Minnesota Department of Health

Universal or Hearing-targeted? That Is the Congenital Cytomegalovirus Screening Question

Sondra Rosendahl, MS, CGC, Minnesota Department of Health

Opting Out of Congenital Cytomegalovirus Screening in New York State

Virginia Sack, MS, CGC, New York State Department of Health

Congenital Cytomegalovirus Culture of Collaboration to Streamline Regulatory Workflows

Rachel McCaffery, RN, Wolfson Children's Hospital

This session will focus on three state NBS programs and their experiences with screening for congenital cytomegalovirus (cCMV). First, Minnesota will discuss the benefits and limitations of universal cCMV screening and highlight signs and symptoms in newborns that would be missed using a targeted approach. Next, New York will examine how information garnered from an opt-out survey for a cCMV pilot study will help inform research on parents' attitudes towards NBS. Our final presentation will illustrate a collaborative approach between the Florida NBS program and Wolfson's Children's Hospital to solve issues around compliance with testing and reporting requirements for cCMV.

After completing the session, the participant will be able to:

- Compare universal and hearing-targeted screening for cCMV
- Summarize an opt-out procedure for families who do not want their child's results reported
- Formulate workflows to improve the rapid identification of at-risk populations to provide timely diagnosis

Challenges to Newborn Screening and Building a Resilient System

Ballroom A

588-878-25 • 1.5 contact hours for this session

Moderators:

- Michele Caggana, ScD, FACMG, New York State Department of Health, retired
- Sulay Rivera, PhD, Puerto Rico Newborn Screening Program

Utilizing Machine Learning for Improved Determination of Cystic Fibrosis Status in Newborn Screening Samples

Alankar Kamppowale, MPH, The University of Iowa

From Computer to Clinic: Calibrating Variant Impact Predictors Yields a ClinGen Clinical Recommendation Up to Strong Evidence in Genome Variation Interpretation
Steven Brenner, PhD, University of California, Berkley

Statistical Challenges in Newborn Screening Parental Stress Index Time Series Data
Thevaa Chandereng, PhD, Children's National Hospital

Newborn Screening in North Carolina in the Aftermath of Hurricane Helene and the Program's Ongoing Continuity of Operations Planning
Kimberly Blake, MPH, North Carolina State Laboratory of Public Health

NBS programs must be ready to face the many challenges that will come their way. Whether these challenges are brought about by internal program disruptions or external factors, public health professionals must learn to react, adapt and grow. In this session, speakers will discuss a project which utilized machine learning to improve the prediction of a newborn's cystic fibrosis (CF) status, examine how to more reliably and effectively classify genetic variants for diagnosis and screening, investigate the long-term impact of false-positive NBS results on parental well-being and highlight ways to work collaboratively with partner organizations during unexpected natural disasters.

After completing the session, the participant will be able to:

- Indicate how machine learning techniques can be utilized to improve cutoff algorithms
- Classify genetic variants for diagnosis and screening
- Identify the potential psychosocial burdens experienced by families receiving false-positive results
- Summarize the logistical procedures to consider when developing a state-specific Continuity of Operations Plan (COOP)

11:00 am – 11:30 am Break

Ballroom Pre-function

11:30 am – 1:00 pm

After the Screen: Continuing to Put Families at the Forefront

Ballroom A

588-879-25 • 1.5 contact hours for this session

Moderators:

- Marianna Raia, MS, CGC, Expecting Health
- Lesa Brackbill, MA, Leukodystrophy Newborn Screening Action Network

Developing a Framework to Assess Family Outcomes After Newborn Screening
Don Bailey, PhD, RTI International

Parent and Provider Evaluation of Long-term Follow-up in North Dakota
Yvonne Kellar-Guenther, PhD, Center for Public Health Innovation

Thursday, October 9

Recruitment Strategies and Demographics from a Multi-site Longitudinal Study of False-positive Newborn Screening Results

Kelly Christensen, Children’s National Hospital

Parents’ Perspectives on the Newborn Screening Experience: Comparing False-positive and Normal Result Experiences

Anne Atkins, MPH, Children’s National Hospital

A Multi-state Population-based Study of Parental Stress After a False-positive Newborn Screening Result

Beth Tarini, MD, MS, MBA, Children’s National Hospital

Our final session will underscore the importance of family outcomes after NBS. Speakers will report on steps taken to develop an instrument to access family outcomes, discuss ways to improve long-term follow-up care, emphasize the importance of recognizing the psychological impact of receiving false-positive results and offer suggestions for how to better communicate results to parents, all with the goal of creating an NBS system that puts families at the forefront today and every day.

After completing the session, the participant will be able to:

- List eight family outcomes after NBS
- Examine a care coordination approach for NBS long-term follow-up
- Identify strategies that impact recruitment for large-scale parent participation in NBS research
- Contrast differences in the experiences between parents who self-report false-positive and normal NBS results
- Summarize the findings from a longitudinal study on parent stress after receipt of a false-positive NBS result

1:00 pm

Ballroom A

Closing and Symposium Adjourns

1:30 pm – 5:30 pm

Ballroom A

**Tour of the New England NBS Program
at the University of Massachusetts
Chan Medical School**

Join us for a bus trip to the New England Newborn Screening Program at the University of Massachusetts Chan Medical School to learn more about how the program provides laboratory screening and clinical follow-up to five New England states.

Advanced registration is required. Please check the registration desk starting on Tuesday for possible space availability.

Thursday, October 9



Shaping the future of newborn screening... one baby at a time.

Newborn screening is evolving rapidly, and together, we're helping labs like yours deliver faster, more reliable answers for families. With solutions that screen for over 50 congenital disorders, Revvity enables providers and patients to make faster, trusted treatment decisions.

Visit us at **booth 500** during the APHL Newborn Screening Symposium to explore what's new:

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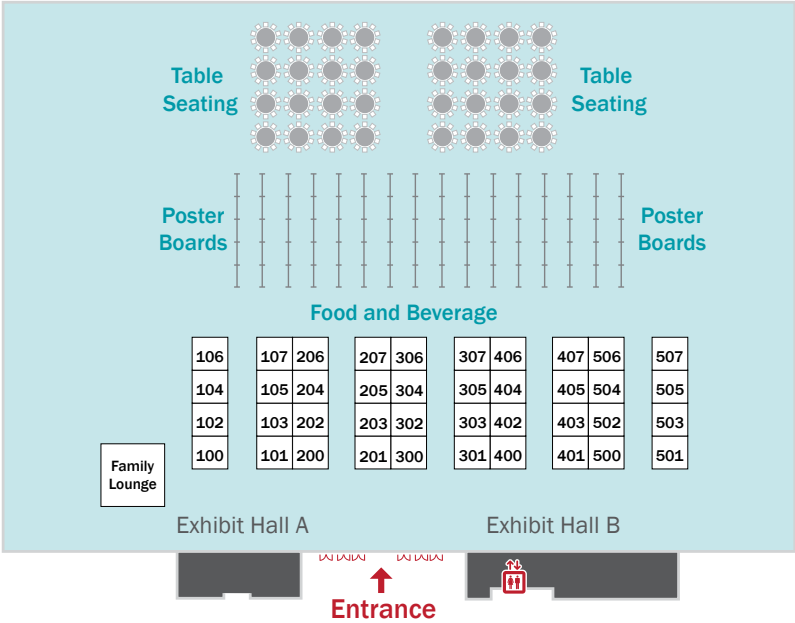
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Exhibit Hall Floor Plan



AGENA BIOSCIENCE

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www.agenabio.com



Booth 107

Agena Bioscience enables laboratories worldwide to deliver affordable targeted genomic analysis with patented high-multiplexing capabilities. Our advanced genotyping platforms deliver timely, accurate and targeted results, to improve decision making and laboratory economics.

Association of Public Health Laboratories Career Pathways

Booth 104

www.aphl.org/Career-Pathways

APHL Career Pathways program representatives will be available to discuss career opportunities in public health laboratory science. Stop by our booth to explore fellowship, internship and leadership programs. Discover how you can make an impact in the field by becoming a Public Health Laboratory Ambassador, Fellow, Intern or Mentor.

Astoria-Pacific

Booth 507

www.astoria-pacific.com

Astoria-Pacific is a small business that is fully dedicated to providing our clients with the best support and applications as possible. We are honored that Newborn Screening Programs rely on us to keep their newborns safe. We offer Laboratory Automation, Diagnostic Reagent Kits, and Reference Materials to neonatal laboratories worldwide.

Asuragen, a Bio-Techne Brand

Booth 207

www.asuragen.com

Asuragen provides best-in-class molecular diagnostics, including proprietary chemistry and software, designed for use with widely available instruments to ease adoption and quickly expand lab test menus. Our diagnostic tests and companion diagnostic partnerships address current and emerging clinical needs in reproductive health, cancer diagnosis and monitoring, and inherited disease. Asuragen also offers a suite of unique, target-specific molecular controls that have been used in IVD assays for more than 20 years.

Bio-Rad Laboratories, Inc.

Booth 301

Diamond Level Sustaining Member

www.bio-rad.com

Bio-Rad is a global leader in developing, manufacturing, and marketing a broad range of innovative products for the life science research and clinical diagnostic markets. With a focus on quality and customer service for 70 years, our products advance the discovery process and improve healthcare.

BSD Robotics

Booth 501/503

www.bsdrobotics.com

BSD Robotics has over 30 years' experience providing innovative biosample punch instruments for life science applications. The BSD brand represents a dedication and specialization in the design, manufacture, and lifetime support of sample preparation instruments. Supporting life science laboratories worldwide, our highly experienced service team provide assurance for maximum instrument performance and workflow efficiency. BSD Products are proudly recognized worldwide as the gold standard in Forensic databasing laboratories and as the preferred alternative in newborn screening.

Cambridge Isotope Laboratories

Booth 401

Bronze Circle Sponsor

www.isotope.com



Since 1981, Cambridge Isotope Laboratories, Inc. (CIL) has been known for manufacturing quality analytical standards. CIL offers stable isotope-labeled standards, including amino acids, carnitine/acylcarnitines, organic acids, steroids, and many others, for use in lab-developed tests employing tandem mass spectrometry-based systems. CIL has obtained the CE mark and manufactures standards that are compliant with ISO 13485.

Centers for Disease Control and Prevention

Booth 105

www.cdc.gov/newbornscreening

CDC is the nation's leading science-based, data-driven, service organization that protects the public's health. For more than 70 years, we've put science into action to help children stay healthy so they can grow and learn; to help families, businesses, and communities fight disease and stay strong; and to protect the public's health.

Devyser

Booth 304

www.devyser.com

We are pioneers in kits and solutions for advanced DNA testing. Our goal is to eliminate tedious protocols and streamline laboratory workflows with simple, fast, and easy-to-use solutions.

DiaSorin Molecular

Booth 400

Silver Level Sustaining Member

www.molecular.diasorin.com

DiaSorin Molecular is a global provider of sample-to-answer molecular products for encephalitis, respiratory, and neonatal health. Our Simplexa® Congenital CMV Direct kit is the only FDA cleared PCR assay for the detection of cCMV in both saliva swabs and urine. This solution provides faster diagnosis to facilitate better patient outcomes.

EBF LLC

Booth 402

Bronze Circle Sponsor

www.ebf-inc.com



Eastern Business Forms, Inc. has been a leading provider of newborn screening collection devices for more than 50 years. Our goal is to exceed customer requirements by leveraging our extensive manufacturing experience and providing superior customer service. EBF is the exclusive provider of Whatman 903 filter paper in the United States



**Meet our team Booth
305/307**

Enfanos, LLC

Booth 204

www.enfanos.com

Enfanos (formerly GelbChem) is passionate about providing the most scientifically sound, up-to-date, and cost-effective LC-MS/MS methodologies for Newborn Screening and diagnostics. We back our products with comprehensive, personalized customer service from our team of experts in enzymology, mass spectrometry, analytical biochemistry, and synthetic chemistry.

EveryLife Foundation for Rare Diseases

Booth 203

everylifefoundation.org

The EveryLife Foundation for Rare Diseases is a 501(c)(3) nonprofit, nonpartisan organization dedicated to empowering the rare disease patient community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments and cures.

Expecting Health/Baby's First Test

Booth 100

www.expectinghealth.org

Born from Genetic Alliance, a nonprofit organization rooted in nearly 40 years of community programs and representing national voices and family-centered experiences, Expecting Health shares science-based and policy-informed information that reflects the lived experiences of individuals and their families. We do this through the power of relationships; convening the top experts; working with key leaders in health; and engaging with families and communities at the center of the conversation.

HCU Network America

Booth 306

www.hcunetworkamerica.org

HCU Network America strives to inform and provide resources for patients and families, create connections, influence state and federal policy, and support the advancement of diagnosis and treatment for HCU and related disorders.

Labsystems Diagnostics Oy

Booth 206

www.labsystemsdx.com

Labsystems Diagnostics develops, manufactures and markets high quality screening and diagnostics solutions for Newborn screening. Labsystems Diagnostics provides a one stop solution with Fluorescence Enzymatic assays, Molecular Real Time PCR, Massspectrometry and NGS assays.

LaCAR US, Inc.

Silver Level Sustaining Member
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Booths 305/307

www.lacar-mdx.com/en

LaCAR MDx Technologies is an innovative company specializing in rapid, reliable and automated in vitro genetic diagnostic (IVD) solutions, focusing on Newborn Screening and Pharmacogenetics. Founded in 2013, LaCAR is committed to improving global public health through cutting-edge technologies tailored to the specific needs of each laboratory.

Mirum Pharma

Booth 404

mirumpharma.com

Mirum proudly serves children, adults, and families with rare disease, working closely with these communities to intimately understand the challenges they face day to day. It's with this deep understanding, empathy, and urgency that we seek to develop remarkable therapies for rare diseases that have limited or no approved treatment options. Because ultimately, our goal is to transform the lives of these individuals so that they have the opportunity to live life to the fullest.



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by genetic and other severe diseases
by unlocking the transformative potential
of HSC gene therapy**

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Novartis

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www.novartis.com



Booth 303

Novartis is an innovative medicines company. Every day, we work to reimagine medicine to improve and extend people's lives so that patients, healthcare professionals and societies are empowered in the face of serious disease. Our medicines reach nearly 300 million people worldwide.

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OpenELIS Foundation

Silver Level Sustaining Member

www.openelis.org

Booth 101

The OpenELIS Foundation is a 501(c)(3) nonprofit corporation whose mission is to provide open source software and services to public health laboratories that will meet the demands of today's responsive public health laboratory. Designed to be an enterprise solution that scales to the needs of any PHL.

Orchard Therapeutics

Silver Circle Sponsor

www.orchard-tx.com



Booth 102

Orchard was founded in 2015—but our roots run deeper, going back to some of the first research and clinical development involving hematopoietic stem cell, or HSC, gene therapy. Our team has played a central role in the evolution of this technology from a promising idea to a potentially life-transforming reality. Today, we carry forward our history of visionary science to imagine new possibilities for people and families affected by genetic and other severe diseases.

Primary.Health

Gold Level Sustaining Member

Bronze Circle Sponsor

www.primary.health



Booth 106

Primary.Health partners with public health agencies and labs to modernize testing, streamline data exchange, and expand access to care. From NBS to infectious disease, our flexible platform and nationwide provider network support scalable, equitable solutions that strengthen public health infrastructure and unlock new possibilities for early detection and intervention.

Quantabio

Booth 317

Silver Level Sustaining Member

www.quantabio.com

Quantabio is a leading provider of advanced DNA and RNA amplification reagents for the most demanding molecular testing applications in applied, translational, and life science research. Our team leverages decades of experience in developing pioneering amplification technologies to deliver cutting-edge products to researchers focused on critical cloning, PCR, qPCR, and Next-Generation Sequencing (NGS) applications.

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Revvity

*Diamond Level Sustaining Member
Gold Circle Sponsor*



Booths 500/502/
504/506

www.revvity.com

Revvity provides health science solutions, technologies, expertise, and services that deliver complete workflows from discovery to development, and diagnosis to cure.

Revvity is pushing the limits of what's possible in healthcare, with specialized focus areas in translational multi-omics technologies, biomarker identification, imaging, prediction, screening, detection and diagnosis, informatics, and more.

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RTI International

Booth 205

www.rti.org

RTI International supports critical initiatives to improve international public health outcomes for all. With a multidisciplinary team of experts and established relationships across the public health research community, our team is poised to create a world where people thrive in health and wellness. Learn more about RTI's expertise in designing, implementing, and evaluating scientifically based programs that can inform public health policy and promote healthy behaviors.

Sebia, Inc.

Booths 405/407

Silver Level Sustaining

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www.sebia-usa.com

Sebia is a global specialty IVD company focused on protein analysis. The new CAPILLARYS 3 DBS* instrument utilizes Sebia's proprietary and best-in-class capillary electrophoresis separation technology to maximize resolution for newborn Hemoglobin (Hb) disorder screening. By offering NBS labs a fully automated, walkaway platform, Sebia seeks to improve NBS laboratory workflows, freeing up precious time for labs to focus on value-added tasks. *CAPILLARYS 3 DBS not currently available for sale in the USA.

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STAT Courier Service, Inc.

Booth 201

www.stat-courier.com

STAT Courier Service Inc. connects birthing centers and State Public Health Laboratories statewide with logistical solutions to drive better healthcare outcomes for all newborn citizens, through reliable medical courier service delivery for newborn screening tests in addition to clinical and environmental specimens and medical supplies.

Sumitomo Pharma America

Booth 103

www.us.sumitomo-pharma.com

We drive innovative treatments, science, and technology to address patient needs in the critical areas of oncology, women's health, urology, rare disease, CNS, and cell & gene therapies.

Thermo Fisher Scientific

Booth 302

Diamond Level Sustaining Member

www.thermofisher.com/us/en/home.html

Thermo Fisher Scientific is the world leader in serving science. Our mission is to enable our customers to make the world healthier, cleaner and safer. We support reproductive health researchers with innovative technologies throughout the reproductive lifecycle – such as genetic analysis for newborn screening and sequencing.

Ultragenyx

Booth 300

www.ultragenyx.com

Ultragenyx was founded to advance innovative medicines for rare and ultrarare diseases that have never been treated before. We are delivering transformative therapies across multiple indications, and we have one of the most robust and diverse clinical pipelines in rare disease.

Waters Corporation

Gold Level Sustaining Member

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www.waters.com



Booths 200/202

Waters mission is to optimally enable global Newborn Screening laboratories to deliver 1st Tier and 2nd Tier screening. With >20 years of experience supporting Newborn Screening we understand the wide variety of solutions required and provide a range of tried and tested mass spectrometry, informatics, training and support to meet multiple user needs daily.

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Posters

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*P-1

digitalMLPA as a Potential Confirmatory Test for Congenital Adrenal Hyperplasia, Glucose-6-Phosphate Dehydrogenase Deficiency and Hemoglobinopathies in Newborn Screening

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Presenter: Terence Diane Fabella, MS, PhD candidate, Amsterdam University Medical College, t.d.fabella@amsterdamumc.nl

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Addressing Low Levels of Newborn Screening Awareness Pre- and Post-prenatal Education

E. Yang, K. Fullerton, F. Altmaier, T. Huynh, C. Clabaugh, K. Fitzpatrick; Arizona Department of Health Services

Presenter: Fran Altmaier, Arizona Department of Health Services, fran.altmaier@azdhs.gov

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Can Education Alone Address Newborn Screening Unsatisfactory Samples/Rise in Unsatisfactory Samples Despite Consistent Education? What Else Is Needed

E. Yang, K. Fullerton, F. Altmaier, T. Huynh, K. Fitzpatrick, C. Clabaugh; Arizona Department of Health Services

Presenter: Fran Altmaier, Arizona Department of Health Services, fran.altmaier@azdhs.gov

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Impacts of Genome Supplemented Newborn Screening

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Presenter: Gianna D'Apolito, MS, Association of Public Health Laboratories, gdapolit@asu.edu

P-5

A “Detor” for Test Orders and Results

R. Shepherd, H. McDavid, D. Shirazi, E. Sudduth; Association of Public Health Laboratories

Presenter: Rachel Shepherd, Association of Public Health Laboratories, rachel.shepherd@aphl.org

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The Newborn Screening Fellowship Program

C. Sumbly, Association of Public Health Laboratories

Presenter: Courtney Sumbly, MS, Association of Public Health Laboratories, courtney.sumbly@aphl.org

P-7

Quantitative Dried Blood Spot Verification Tool for Sample Quality Assessment

A. Stewart, BSD Robotics

Presenter: Andrew Stewart, MPhil, BSD Robotics, andrew.s@bsdrobotics.com

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California’s 20-year Experience with Newborn Screening for Congenital Adrenal Hyperplasia: Insights into Tier-1 and Tier-2 Testing

K. Dhillon, P. Rich-Crawford, R. Miranda, L. Nguyen, R. Sharma; California Department of Public Health

Presenter: Kuldeep Dhillon, MS, CCS(CA), MB(ASCP)cm, California Department of Public Health, kuldeep.dhillon@cdph.ca.gov

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Enhancing Quality in Clinical Testing Laboratory Through Automated Temperature Monitoring

Y. Bai, K. Fujimura, R. Zahedi, K. Ramanathan, S. Wu, S. Sharma, R. Rawat, G. Kuntamallappanavar, P. Rich-Crawford, L. Nguyen, R. Sharma; California Department of Public Health

Presenter: Kuldeep Dhillon, MS, CCS(CA), MB(ASCP)cm, California Department of Public Health, kuldeep.dhillon@cdph.ca.gov

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Newborn Screening for Hemoglobinopathies in California: The Past 20 Years (2004-2024)

K. Fujimura, P. Roworth, R. Rawat, R. Sharma; California Department of Public Health

Presenter: Kuldeep Dhillon, MS, CCS(CA), MB(ASCP)cm, California Department of Public Health, kuldeep.dhillon@cdph.ca.gov

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Validation of a Multiplexed LC-MS/MS Assay for Pompe, MPS-I, and MPS-II for Newborn Screening of Lysosomal Storage Disorders

K. Ramanathan, P. Roworth, J. Vongsa, K. Dhillon, C. Ortega, J. Velasquez, S. Bailey, A. Mercado, J. Johnson, J. Aduviso, J. Qiang, L. Nguyen, R. Zahedi, R. Sharma; California Department of Public Health

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P-12

Newborn Screening for Severe Combined Immunodeficiency (SCID) in California: Ten Years of Experience

R. Rawat, G. Kuntamallappanavar, A. Lovely, D. Simon, L. Zhang, P. Roworth, S. Sharma, R. Sharma; California Department of Public Health

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California Newborn Screening for Pompe Disease Using FIA and LC-MS/MS Assays: The Past Six Years

P. Roworth, K. Ramanathan, J. Vongsa, K. Dhillon, C. Ortega, J. Velasquez, S. Bailey, A. Mercado, J. Johnson, J. Aduviso, J. Zhou, L. Nguyn, R. Zahedi, R. Sharma; California Department of Public Health

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A Synopsis of Five Years of Spinal Muscular Atrophy Screening in California

S. Sharma, L. Shih, L. Wu, L. Tom, C. Wu, J. Matteson, B. Sohal, S. Bailey, C. Perez, R. Sharma; California Department of Public Health

Presenter: Kuldeep Dhillon, MS, CCS(CA), MB(ASCP)cm, California Department of Public Health, kuldeep.dhillon@cdph.ca.gov

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A High Throughput Workflow for Newborn Screening of Spinal Muscular Atrophy in California

L. Shih, L. Wu, L. Tom, C. Wu, S. Sharma, R. Rawat, J. Matteson, B. Sohal, J. Velasquez, B. Espinosa, D. Simon, S. Bailey, C. Perez, R. Sharma; California Department of Public Health

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Strengthening Laboratory Compliance and Efficiency Through Robust Document Control

S. Wu, Y. Bai, R. Sharma; California Department of Public Health

Presenter: Kuldeep Dhillon, MS, CCS(CA), MB(ASCP)cm, California Department of Public Health, kuldeep.dhillon@cdph.ca.gov

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Analysis of Lead and Other Elements Using 10 and 50 µL Dried Microsample Collection Devices

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Presenter: Donald Chace, PhD, MSFS, FADMD, Capitainer, Inc., donald.chace@capitainer.com

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Engaging Midwives in Washington State

L. Barringer¹, A. Watkins², G. Gerboth³, Y. Kellar-Guenther¹, S. Quesada¹; ¹Center for Public Health Innovation, ²Washington State Department of Health, ³Midwives College of Utah

Presenter: Lauren Barringer, PT, DPT, MPH, Center for Public Health Innovation, Lauren.Barringer@cphinnovation.org

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Building a Newborn Screening Long-term Follow-up Program? There's a Guide for That

M. Sontag¹, A. Steyermark², Y. Kellar-Guenther¹, L. Barringer¹; ¹Center for Public Health Innovation, ²ACS Public Health Consulting

Presenter: Yvonne Kellar-Guenther, PhD, Center for Public Health Innovation, Yvonne.Kellar-Guenther@CPHInnovation.org

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Creating a Shared Decision Making Toolkit for Midwives to Assist with Newborn Screening Discussions

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Presenter: Yvonne Kellar-Guenther, PhD, Center for Public Health Innovation, Yvonne.Kellar-Guenther@CPHInnovation.org

P-21

Analyzing CFTR Panel Performance by Race: Using Registry Data to Support State-level Review

R. West¹, M. McGarry², K. Raraigh³, M. Sontag¹; ¹Center for Public Health Innovation, ²Seattle Children's Research Institute, ³John Hopkins University

Presenter: Marci Sontag, PhD, Center for Public Health and Innovation, marci.sontag@cphinnovation.org

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Development of Dried Blood Spot Based Lysosomal Storage Disorders Linearity Materials

E. Courtney, S. Isenberg, T. Lim, C. Pickens, R. Lee, C. Cuthbert, K. Petritis; Centers for Disease Control and Prevention

Presenter: Elya Courtney, MPH, Centers for Disease Control and Prevention, pli3@cdc.gov

P-23

Effect of Storage Conditions on the Stability of Immunoreactive Trypsinogen in Dried Blood Spots Quality Assurance Materials

E. Gonzalez Reyes¹, L. Li¹, T. Trosclair², C. Brown¹, A. Johnson³, J. Bernstein¹, R. Lee¹, J. Mei¹; ¹Centers for Disease Control and Prevention, ²Neptune and Company, Inc., ³Cherokee Nation Operation Solutions

Presenter: Ernesto Gonzalez Reyes, PhD, Centers for Disease Control and Prevention, tku0@cdc.gov

P-24

Results of CDC's Pilot Combined SMA-TREC QC Programs

C. Greene, S. Cordovado, I. Onyechi, C. Cuthbert, R. Lee; ¹Centers for Disease Control and Prevention

Presenter: Christopher Greene, PhD, Centers for Disease Control and Prevention, crg0@cdc.gov

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Evaluation of Short-term Stability of Lab-created Dried Blood Spot Quality Assurance Materials for Newborn Screening for Congenital Cytomegalovirus

G. Kim, D. Cobb, R. Hage, C. Greene, S. Cordovado, R. Lee, C. Cuthbert; Centers for Disease Control and Prevention

Presenter: Rosemary Hage, PhD, Centers for Disease Control and Prevention, tol5@cdc.gov

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Quality Assurance Specimens for Variant Detection Using Next Generation Sequencing and Sanger Sequencing Methods

M. Hendix, R. Hage, S. Cordovado, R. Lee, C. Cuthbert, C. Greene; Centers for Disease Control and Prevention

Presenter: Miyono Hendrix, MS, Centers for Disease Control and Prevention, zuj7@cdc.gov

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A Pilot Program for Psychosine Quality Assurance Dried Blood Spot Materials for Krabbe Screening

S. Isenberg, S. Sah, C. Freeman, E. Courtney, T. Lim, C. Pickens, R. Lee, C. Cuthbert, K. Petritis; Centers for Disease Control and Prevention

Presenter: Samantha Isenberg, PhD, Centers for Disease Control and Prevention, yhx1@cdc.gov

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Evaluation of D-ASA and Blood Heat Treatment for Development of Dried Blood Spot Quality Assurance Materials for Argininosuccinic Aciduria

T. Lim, D. Peppers, S. Isenberg, C. Pickens, E. Courtney, R. Lee, C. Cuthbert, K. Petritis; Centers for Disease Control and Prevention

Presenter: Timothy Lim, PhD, Centers for Disease Control and Prevention, tlim@cdc.gov

P-29

Two Year Stability of TREC and SMN1 Laboratory Created Dried Blood Spot Materials for Use with the SMN1/TREC/RPP30 Triplex qPCR Assay

I. Onyechi, C. Greene, R. Lee, C. Cuthbert; Centers for Disease Control and Prevention

Presenter: Ivy Onyechi, MS, Centers for Disease Control and Prevention, kru1@cdc.gov

P-30

Preparation and Storage of Pilot Dried Blood Spot Quality Assurance Materials for Duchenne Muscular Dystrophy (DMD) Using Human-tissue-derived Creatine Kinase-MM

G. Pena¹, E. McCown¹, A. Johnson², S. Zobel¹, I. Barham¹, F. Dola¹, J. Bernstein¹, R. Lee¹, S. Nikolova¹, J. Mei¹; ¹Centers for Disease Control and Prevention, ²Cherokee Nation Operation Solutions

Presenter: Elizabeth McCown, Centers for Disease Control and Prevention, erm5@cdc.gov

P-31

Comparison of the Capillary Electrophoresis System to Detect Hemoglobin Proteins in Proficiency Testing Material with Data Collected from NSQAP Participating Laboratories (2022–2025)

G. Pena, S. Zobel, C. Brown, A. Alvarado, J. Bernstein, S. Nikolova, R. Lee, J. Mei; Centers for Disease Control and Prevention

Presenter: Christofer Brown, Centers for Disease Control and Prevention, qhk3@cdc.gov

P-32

Development of Homocystinuria, ADA-SCID, Argininosuccinic Aciduria, CPT-I and CACT/CPT-II Dried Blood Spot Based Proficiency Testing Materials

D. Peppers, S. Isenberg, T. Lim, C. Pickens, R. Lee, C. Cuthbert, K. Petritis; Centers for Disease Control and Prevention

Presenter: Daquille Peppers, MS, Centers for Disease Control and Prevention, sy77@cdc.gov

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Performance of Pilot Quality Assurance Materials for Creatine Kinase MM in Dried Blood Spots

J. Mei, G. Pena, E. McCown, A. Johnson, S. Zobel, R. Lee, M. Nikolova, Centers for Disease Control and Prevention

Presenter: Mimi Nikolova, PhD, Centers for Disease Control and Prevention, sjn4@cdc.gov

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Expanding Cystic Fibrosis DNA Screening for Colorado Newborn Screening: Adventures of Implementing Next Generation Sequencing in a Mid-size Newborn Screening Program

K. Hanson¹, K. Croak², K. Fater², C. Kirkland², G. Bonn², ¹Association of Public Health Laboratories, ²Colorado Department of Public Health and Environment

Presenters: Kendra Croak, MS, Colorado Department of Public Health and Environment, Kendra.Jones@state.co.us and Krysten Hanson, APHL Bioinformatics Fellow, Colorado Department of Public Health and Environment, Kyrsten.hall@state.co.us

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The Impact of Prenatal Education Packets: A Retrospective Observational Analysis Examining Colorado Newborn Screening Refusal Rates

A. Howk, K. Inkhamfong, J. Lucero, G. Bonn; Colorado Department of Public Health and Environment

Presenter: Abigail Howk, MPH, Colorado Department of Public Health and Environment, abigail.howk@state.co.us

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WITHDRAWN

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Implementing Hearing-targeted Congenital Cytomegalovirus Testing for Infants in Georgia: The First Six Months

A. Wittenauer¹, T. Kavanaugh², M. Morris², B. Culpepper², H. Juca¹; ¹Emory University, ²Georgia Department of Public Health

Presenter: Angela Wittenauer, MSN, FNP-C, RN, Emory University, alwitte@emory.edu

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Synthesis of Quantitative Standards for the Endogenous Glycosaminoglycan Non-reducing End Method for Mucopolysaccharidosis-I, -II, and -IIIA

Z. Herbst^{1, 2}, S. Lim¹, S. Gholap², H. Khaledi¹, M. Gelb²; ¹Enfanos, LLC, ²University of Washington

Presenter: Sanghyun Lim, Enfanos, LLC, sanghyun@enfanos.com

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Establishing a Newborn Screening Center of Excellence to Advance Testing Technologies and Enhance Data Infrastructure in the Southeastern U.S.

M. Rowlinson¹, S. Crowe¹, P. Ryland¹, I. Sunitha¹, D. Stern¹, E. Reeves¹, T. Garret², C. Souders II²; ¹Florida Department of Health, ²University of Florida

Presenter: Marie-Claire Rowlinson, PhD, D(ABMM), Florida Department of Health, marie-claire.rowlinson@flhealth.gov

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WITHDRAWN

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Overview of 30 Years of Newborn Screening in Lebanon: Impact of the Recent Economic Crisis and War

H. Mansour¹, I. Khneisser²; ¹Saint Georges University of Beirut², HVD Vertriebs Lifesciences gmbh

Presenter: Issam Khneisser, MS, MBA, HVD Vertriebs Lifesciences gmbh, issam.khneisser@usj.edu.lb

***P-42**

Implementing Next Generation Sequencing as Part of Newborn Screening in State Public Health Laboratories: Status and Barriers

H. Snyder¹, K. Sund², C. Seymour³, M. Haiber⁴; ¹Illumina, ²Independent, ³Rutgers, the State University of New Jersey, ⁴GeneDx

Presenter: Holly Snyder, MS, LGC, Illumina, hsnyder@illumina.com

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Performance Evaluation of a Multiplex Kit for Simultaneous Detection of Phenylalanine, 17OHP, IRT, and TSH for Newborn Screening Programs

R. Almeida, M. Sampaio; Interscientific Corporation

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Sowing the Seeds of Newborn Screening: The Iowa NBS Lab's STEM Outreach Initiative

J. Alleyne, L. Jaramillo, M. Pentella; State Hygienic Laboratory at the University of Iowa

Presenter: Jerusalem Alleyne, PhD, State Hygienic Laboratory at the University of Iowa, jerusalem-alleyne@uiowa.edu

***P-45**

Enhancing Kansas Newborn Screening by Launching a Medical Directorship

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Presenter: Kourtney Bettinger, MD, MPH, FAAP, Kansas Department of Health and Environment, kourtney.bettinger@ks.gov

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Comparing Top Ten CFTR Variants in Kansas Between State of Kansas Newborn Screening CFTR 2nd Tier Luminex 39 Variant Panel from 2016 to 2020 and the Wisconsin State Laboratory of Hygiene CFTR 2nd Tier MiSeq Expanded Panel from 2020 to 2024

S. Brownlee, Kansas Department of Health and Environment

Presenter: Sally Brownlee, Kansas Department of Health and Environment, sally.brownlee@ks.gov

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A New Public Health Laboratory - Design, Construction and Moving

P. Harrison, Kansas Department of Health and Environment

Presenter: Paul Harrison, Kansas Department of Health and Environment, paul.harrison@ks.gov

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Increase in TREC False Positive Rate Due to Environmental Factors 2020 to 2023 - The State of Kansas Experience

M. Mills, Kansas Department of Health and Environment

Presenter: Michelle Mills, MS, Kansas Department of Health and Environment, Michelle.J.Mills@ks.gov

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The Impact of the State of Kansas Department of Health and Environmental Laboratories New Building in Comparison to the Old Building on T-cell Receptor Excision Circles (TREC) Out of Range/Abnormal Rate

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Presenter: Faith Smith, Kansas Department of Health and Environment, Faith.Smith@ks.gov

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Characterizing Irritability in Krabbe Disease: Implications for Early Diagnosis and Treatment Decisions

S. Pike-Langenhof¹, P. Engel², S. Jackson², P. Orchard³, N. Bascou⁴; ¹KrabbeConnect, ²Engage Health, ³University of Minnesota, ⁴John Hopkins University

Presenter: Stacy Pike-Langenhof, KrabbeConnect, stacy.pike@krabbeconnect.org

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Development of a High-throughput Fluorescence-based Screening Assay for Krabbe Disease Using Dried Blood Spot Samples

J. Chen, S. Vodehnal, K. Alpadi, LabSystems Diagnostics

Presenter: Kannan Alpadi, LabSystems Diagnostics, kannan.alpadi@labsystemsdx.com

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Development of a High-throughput Fluorescence-based Screening Assay for Hunter Syndrome (MPS II) Using Dried Blood Spot Samples

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Presenter: Kannan Alpadi, LabSystems Diagnostics, kannan.alpadi@labsystemsdx.com

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Tandem Mass Spectrometry for Routine Screening of Sickle Cell Disease

M. Greffe¹, J. Washburn², H. Hiertz¹, C. Barbe¹, C. Sepult¹, L. Detemmerman¹; ¹LaCAR MDx, ²LaCAR US

Presenter: Jon Washburn, LaCAR US, jon.washburn@lacar-mdx.com

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Performance Evaluation of the NEONATAL SCID&SMA Screening qPCR Assay in Newborn Dried Blood Spots

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Presenter: Jon Washburn, LaCAR US, jon.washburn@lacar-mdx.com

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Automated Newborn Screening for GALT and Biotinidase on Dried Blood Spots

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Presenter: Jon Washburn, LaCAR US, jon.washburn@lacar-mdx.com

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SMA/SCID Control Panel: A Novel Panel of Biologically-relevant, Allele-based Dried Blood Spot Mimic Controls Accurately Monitoring SMA/SCID Assay Performance and SMN1/SMN2 Copy Number Variation

L. Cooper, J. Gordon, J. Hayden, A. Karaczyn, L. Krott, L. Lenart, D. Magoon, S. Pelsue, A. Reilly, J. Ross, N. Sivaji, T. Spenlinhauer; Maine Molecular Quality Controls

Presenter: Aldona Karaczyn, Maine Molecular Quality Controls, akaraczyn@mmqci.com

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Expanded Cystic Fibrosis Screening through Next Generation Sequencing Improves Detection and Diagnosis for Maryland Infants

E. Moradel, H. Dholakia, K. Brown, M. Dorley, T. Wavra; Maryland Department of Health

Presenter: Edgar Moradel, MD, Maryland Department of Health, edgar.moradel@maryland.gov

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A Three-hour Incubation for Krabbe Disease: Results from Maryland Newborn Screening

P. Paudyal, S. Gayen Betal, K. Smith, A. Willoughby-Spriggs, M. Dorley, K. Brown; Maryland Department of Health

Presenter: *Prakash Paudyal, Maryland Department of Health, prakash.paudyal@maryland.gov*

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Migration from Digital Microfluidics and Fluorometry to Liquid Chromatography Tandem Mass Spectrometry to Screen for Five Lysosomal Disorders: The Maryland Newborn Screening Experience

P. Paudyal, K. Smith, S. Gayen Betal, A. Willoughby-Spriggs, M. Dorley, K. Brown; Maryland Department of Health

Presenter: *Prakash Paudyal, Maryland Department of Health, prakash.paudyal@maryland.gov*

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Evaluation of CLIR Post-analytical Tools for Cystic Fibrosis Screening Performance Improvement

P. Hall¹, E. Barr², K. McKie³, R. Linemann², A. Wittenauer²; ¹Mayo Clinic, ²Emory University, ³Augusta University

Presenter: *Patricia Hall, PhD, Mayo Clinic, hall.patricia@mayo.edu*

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Breaking Down Barriers: How a Legislative Change is Improving Newborn Screening Follow-up

M. Read, L. Thompson, N. Champaigne, J. Griffin; Medical University of South Carolina

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Validating a Fluorometric Enzymatic Assay for Mucopolysaccharidosis Type II in Michigan

A. Strange, S. Moloney, E. Wotring; Michigan Department of Health and Human Services

Presenter: *Lohgan Schlaack, Michigan Department of Health and Human Services, schlaackl@michigan.gov*

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Development of a Multiplex LC-MS/MS Method for Newborn Screening of MLD, X-ALD and MPS II

J. Huang, C. Lord, A. Hietala; Minnesota Department of Health

Presenter: Amy Hietala, MS, Minnesota Department of Health, amy.hietala@state.mn.us

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Process of Matching Infants with Critical Congenital Heart Defects in Short-term and Long-term Follow-up: Methodology and Outcomes

S. Lammert, H. Pint, A. Pavan, A. Strong, K. Yang; Minnesota Department of Health

Presenter: Sara Lammert, PhD, MPH, Minnesota Department of Health, sara.lammert@state.mn.us

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The Parent Experience: Methodology for Engaging with Families to Learn about Their Newborn Screening Follow-up Experience

B. Walde¹, G. Liverseed¹, L. Barber¹, J. Soukup¹, J. Hullerman-Umar¹, K. Kass¹, M. Raspa², E. Cheves², S. Andrews²; ¹Minnesota Department of Health, ¹RTI International

Presenter: Bridget Walde, MPH, Minnesota Department of Health, bridget.walde@state.mn.us

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When Data Doesn't Add Up

J. Hale¹, A. Counihan¹, M. Colantuoni¹, G. Sawicki², H. Dorkin², T. Kremer³, L. Yonker⁴, N. Vehse⁵, A. Comeau¹; ¹New England Newborn Screening Program, ²Boston Children's Hospital, ³UMass Memorial, ⁴Massachusetts General Hospital, ⁵Baystate Medical Center

Presenter: Jaime Hale, MS, New England Newborn Screening Program, jaime.hale@umassmed.edu

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Evaluation of Follow-up Turnaround Times Across Healthcare Provider Types

J. Hale, A. Counihan, M. Colantuoni, B. Kumar, I. Sahai, D. Britton, A. Comeau, R. Eaton, D. Kaur; New England Newborn Screening Program

Presenter: Jaime Hale, MS, New England Newborn Screening Program, jaime.hale@umassmed.edu

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Leveraging Classical Mass Spectrometry Assay to Expand Screening for GAMT Deficiency and to Improve Accuracy for Acylcarnitines Detection

D. Kaur, C. Stockhaus, S. Goff, D. Britton, A. Comeau, R. Eaton, I. Sahai; New England Newborn Screening Program

Presenter: Devinder Kaur, PhD, New England Newborn Screening Program, devinder.kaur@umassmed.edu

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Feasibility of Next Generation Sequencing in Newborn Screening Algorithms to Enhance the Diagnostic Potential of Cystic Fibrosis Newborn Screening

S. Barton, J. Kordowska, J. Gerstel-Thompson, J. Hale, A. Counihan, D. Britton, M. Colantuoni, B. Kumar, A. Comeau; New England Newborn Screening Program

Presenter: Binod Kumar, PhD, New England Newborn Screening Program, binod.kumar@umassmed.edu

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Evaluating the Biochemical Profiles, Genotypes and Clinical Phenotypes of Screen Positives for MPS-1 and Pompe Disease, and the Evolution and Improvement of the Newborn Screening Algorithms

I. Sahai, A. Comeau, R. Eaton, C. Villaruz, J. Hale, B. Kumar, D. Kaur, D. Britton, S. Goff; New England Newborn Screening Program

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A New Direction for Newborn Screening - Implications from New Jersey Biomonitoring and Rutgers Health's Lead and Mercury Screening Program

E. Bind¹, O. Fofah², D. Campbell², H. Kaur², Z. Fan¹; ¹New Jersey Department of Health, ²Rutgers Health

Presenter: Sarah Mustaqali, New Jersey Department of Health, sarah.mustaqali@doh.nj.gov

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Analyzing Delays in Newborn Screening Timeliness: A Baseline Assessment Using Data in New York State

H. Ahmed, N. Guilfoyle, M. Caggana, K. Chou; New York State Department of Health

Presenter: Kathy Chou, PhD, New York State Department of Health, kathy.chou@health.ny.gov

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Development of a Newborn Screening Long-term Follow-up Data Repository in New York State

S. Fandl, R. Wilson, M. Mortimer, P. Santos, M. Caggana, K. Chou; New York State Department of Health

Presenter: Kathy Chou, PhD, New York State Department of Health, kathy.chou@health.ny.gov

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Extended Short-term Follow-up for Endocrine Disorders: Modification of a Previously Established Framework

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Presenter: Kathy Chou, PhD, New York State Department of Health, kathy.chou@health.ny.gov

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Shortening Time to Diagnosis and Treatment: Adjusting Follow-up Action Schedules for Aminoacidopathy Borderline Results

E. Laitinen, V. Sack, J. Xiao, D. Kay; New York State Department of Health

Presenter: Elli Laitinen, MS, Biology, New York State Department of Health, elli.laitinen@health.ny.gov

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Fatty Acid Oxidation Disorder or Food Toxicity? A Case Report

J. Patel¹, V. Sack¹, C. Saavedra-Matiz¹, J. Gold², C. Colefield², J. Xiao¹, T. Whispell¹, D. Kay¹, M. Caggana¹; ¹New York State Department of Health, ²Northwell Health

Presenter: Virginia Sack, MS, CGC, New York State Department of Health, virginia.sack@health.ny.gov

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Short-term Follow-up at 13 Weeks: Case Reviews and Outcomes

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Presenter: Virginia Sack, MS, CGC, New York State Department of Health, virginia.sack@health.ny.gov

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50 Years of Newborn Screening for Sickle Cell Disease in New York State: Evolution, Impact and Future Directions

E. Tabe, C. Saavedra-Matiz, J. Yeman, E. Washor, I. Whitlingam, C. Brandon, S. Gagnon, C. Kim, L. Diantonio, K. Chou, R. Wilson, D. Kay; New York State Department of Health

Presenter: Denise Kay, PhD, New York State Department of Health, denise.kay@health.ny.gov

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Newborn Screening for Human Immunodeficiency Virus in New York State

N. Tavakoli, R. McMahon, M. Pearce, C. McManaman, M. Erickson, M. Caggana; New York State Department of Health

Presenter: Norma Tavakoli, PhD, New York State Department of Health, norma.tavakoli@health.ny.gov

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Cytomegalovirus Infection Rate and Concordance/Discordance in Twin Births

C. McManaman, M. Pearce, V. Sack, C. Brandon, C. Saavedra-Matiz, D. Kay, M. Caggana, N. Tavakoli; New York State Department of Health

Presenter: Norma Tavakoli, PhD, New York State Department of Health, norma.tavakoli@health.ny.gov

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Results and Challenges of Newborn Screening for Metachromatic Leukodystrophy in New York State

J. Xiao¹, C. Saavedra-Matiz¹, J. Orsini¹, M. Wasserstein², M. Nichols¹, D. Kay¹; ¹New York State Department of Health, ²Children's Hospital at Montefiore/Montefiore Medical Center/Albert Einstein College of Medicine

Presenter: Jing Xiao, PhD, New York State Department of Health, jing.xiao@health.ny.gov

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North Carolina Phenylketonuria Diet Monitoring Program to Support Long-term Follow-up

E. Keel¹, J. Mills¹, E. Ramsey², C. Hall², D. Pettit¹, S. Shone¹, K. Blake¹; ¹North Carolina State Laboratory of Public Health, ²University of North Carolina Division of Genetics and Metabolism

Presenter: Kimberly Blake, MPH, North Carolina State Laboratory of Public Health, Kimberly.blake@dhhs.nc.gov

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Pre and Post Move Quality Assurance Studies on Fluoroimmunoassay and Hemoglobinopathy Instrumentation in the North Carolina Newborn Screening Laboratory

C. LaPradd, P. Deloatch, D. Pettit, S. Shone, K. Blake; North Carolina State Laboratory of Public Health

Presenter: Cecily LaPradd, MLS(ASCP), North Carolina State Laboratory of Public Health, cecily.lapradd@dhhs.nc.gov

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North Carolina Cut-off Analysis of Amino Acids Affected by Total Parenteral Nutrition Administration in Newborns Utilizing N-Acetyl Tyrosine to Enhance Sensitivity and Specificity

J. Mills, S. Freeman, D. Pettit, S. Shone, K. Blake; North Carolina State Laboratory of Public Health

Presenter: Jamie M. Mills, MLS(ASCP), North Carolina State Laboratory of Public Health

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Supporting Hospital Newborn Screening Performance - Building Relationships Yielding Results

S. Arceneaux, L. Caton, J. Baysinger, M. Birchell; Oklahoma State Department of Health

Presenter: Shari Arceneaux, RN, Oklahoma State Department of Health, shari.arceneaux@health.ok.gov

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Empowering Newborn Sequencing with Streamlined High Molecular Weight Genomic DNA Extraction from Dried Blood Spot and Whole Genome Library Preparation Using sparQ DBS-seq Workflow

S. Panja, M. Aitichou, S. Chung, B. Komorous, D. Schuster; Quantabio

Presenter: Subrata Panja, PhD, Quantabio, Subrata.Panja@quantabio.com

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Enhancing Laboratory Efficiency: Implementation of Revvity Transcribe AI for Automated Data Entry in Newborn Screening

J. Anderle, E. Chin, A. Millerschoen, A. Mikkonen, C. Murphy, J. Decker, K. Sredzienski, M. Ralph, M. Hegde; Revvity

Presenter: Justin Anderle, Revvity, justin.anderle@revvity.com

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Screening Performance of a New Revvity NeoLSD™ 7plex LC-MSMS Kit

H. Appelblom¹, Y. Chen², M. Nguyen², S. Aguado², E. Obispo², P. Ryland², W. Barnhart¹, H. Lindroos¹, T. Hanses², A. Kiviniemi¹, T. Leppänen¹, C. Rautanen¹, J. Suvanto¹;
¹Revvity, ²Florida Department of Health

Presenter: Heidi Appelblom, Revvity, heidi.appelblom@revvity.com

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Multiplexed LC-MS/MS Measurement of MLD, CTX, NP-C and X-ALD Markers from a Single Dried Blood Spot Sample

M. Manninen, A. Tuominen, S. Talha, J. Huhtala, T. Lehtonen, H. Polari, A. Kiviniemi, J. Brozinski; Revvity

Presenter: Jenny-Maria Brozinski, PhD, Revvity, jenny-maria.brozinski@revvity.com

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Measurement of GlcA-tetrol, t-CDCA, t-THCA and Bile Acid B from a Single Dried Blood Spot Sample Using the Revvity QSight™ 225 MD UHPLC-MSMS System

J. Huhtala, T. Lehtonen, H. Polari, V. Simonian, J. Trometer, T. Au Yeung; Revvity

Presenter: Juuso Huhtala, MS, Revvity, juuso.huhtala@revvity.com

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Adding Metachromatic Leukodystrophy to Existing Newborn Screening Panels: A Laboratory Perspective

S. Smith¹, R. Bearden¹, A. Park¹, H. Lindberg¹, J. Rabbitt¹, T. Donti¹, M. Hegde²;
¹Revvity Omics, ²Revvity

Presenter: Sara Smith, MPS, PhD, Revvity Omics, Sara.Smith@revvity.com

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Simultaneous Detection of Creatine, Guanidinoacetate, Lysophosphatidylcholines, C16:0 Sulfatides and C16:1-OH Sulfatides from Newborn Dried Blood Samples

J. Trometer, V. Simonian, T. Au Yeung, S. Rakus, M. Weng, H. Polari, J. Huhtala; Revvity

Presenter: Tsun Au Yeung, Revvity, Tsun.AuYeung@revvity.com

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Less Burden, More Information: Engaging State Newborn Screening Programs

M. Raspa, M. Lynch, B. Wright, C. Horvitz; RTI International

Presenter: Melissa Raspa, PhD, RTI International, mraspa@rti.org

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A Systematic Process to Match Study Data to State Newborn Screening Data in Early Check

E. Cheves, H. Frawley, J. Dibble, A. Forsythe, S. Scott, V. Copeland, C. Matthews, A. Gwaltney, H. Cope, H. Peay, C. Scharfe; RTI International

Presenter: Curt Scharfe, RTI International, cscharfe@rti.org

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Identifying Quality Improvement Activities in North Carolina: Use of a Modified Site Review

M. Raspa¹, B. Wright¹, E. Reynolds¹, C. Hill¹, E. Cheves¹, A. Forsythe¹, K. Blake², S. Shone²; ¹RTI International, ²North Carolina State Laboratory of Public Health

Presenter: Becca Wright, MPH, RD, LDN, RTI International, rwright@rti.org

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Neonatal Screening for Lysosomal Diseases in the Federal District: Two-year Experience of the Public Health Service

G. Carvalho¹, L. Santos¹, V. Araujo², R. Oliveira², J. Santos², M. Viegas², L. Avelino², D. Caldas², A. Fonseca², A. Monteiro², F. Fustinoni², L. Almeida², M. Costa², V. Freitas², N. Noletto², L. Toledo², F. Soares², M. Rosa², K. Gameleira¹, M. Cardoso¹; ¹Secretaria de Estado de Saúde do Distrito Federal, ²Serviço de Referência em Triagem Neonatal do Distrito Federal

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Pioneering Newborn Screening for Spinal Muscular Atrophy, Severe Combined Immunodeficiency, and X-Linked Agammaglobulinemia in Latin America: Experience from the First Two Years in the Public Health Service of the Federal District, Brazil

C. Oliveira^{1,2}, C. Moura², G. Vasconcelos², D. Amaral², D. Silva², A. Vaqueiro², S. Ribeiro², T. Almeida², L. Coutinho², E. Oliveira², G. Dourado², A. Melo¹, P. Gomes², J. Chaves³, M. Rosa², F. Moraes², J. Thomas², K. Gameleira², G. Carvalho², M. Cardoso²; ¹Serviço de Referência em Doenças Raras e Triagem Neonatal do Distrito Federal, ²Secretaria de Estado de Saúde do Distrito Federal, ³Hospital da Criança de Brasília

Presenter: Claudiner Oliveira, Serviço de Referência em Doenças Raras e Triagem Neonatal do Distrito Federal - Secretaria de Estado de Saúde do Distrito Federal, claudinero@yahoo.com.br

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Topical Interference: When Creams Leave Their Mark on Dried Blood Spots

M. Kjær¹, F. Ottosson², L. Melgaard¹, M. Thorbek¹, M. Nyegaard¹, M. Ernst¹;

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Presenter: *Martin Kjær, Statens Serum Institut, MWKJ@ssi.dk*

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Development and Early Implementation of a Long-term Follow-up Program: Pitfalls, Successes and Program Framework

E. Bradley, J. Smith, A. Porter, A. Ingram; Tennessee Department of Health

Presenter: *Emma Bradley, MS, CGC, Tennessee Department of Health, emma.bradley@tn.gov*

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Retrospective Analysis of Immunoreactive Trypsinogen Reflex Patterns to Cystic Fibrosis DNA Testing Applying Consensus Guidelines to Determine Potential Impact on Tennessee Newborn Screening

D. Godfrey, T. Childs, J. McKenzie, H. Peeples, A. Keyton, Y. Li, A. Ingram; Tennessee Department of Health

Presenter: *Deborah Godfrey, Tennessee Department of Health, deborah.godfrey@tn.gov*

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Building the Car While Driving: Implementing New Disorders & Full Instrument Replacement Without Disrupting Testing

A. Morgan, L. Tyler, G. McKee, M. Finlay, S. Lampson, B. Swanagan, J. Mckenzie; Tennessee Department of Health

Presenter: *Azsa Morgan, Tennessee Department of Health, Azsa.Morgan@tn.gov*

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Cutting Through the Noise and Getting Clearer Answers: Improving Tennessee's Steroid Profile Panel's Accuracy

G. McKee¹, A. Morgan¹, J. McKenzie¹, M. Dorley², M. Finlay¹, S. Lampson¹, A. Bowman¹, L. Tyler¹; ¹Tennessee Department of Health, ²Maryland Department of Health

Presenter: *Lauren Tyler, MLS(ASCP)CMP, Tennessee Department of Health, Lauren.Tyler@tn.gov*

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Bridging Tradition and Timeliness: Strengthening Newborn Screening Awareness Among Texas Midwives

K. Connell, C. Prinz; Texas Department of State Health Services

Presenter: Kelli Connell, ASCP, Texas Department of State Health Services.
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Tackling Transport Barriers in the Texas Rio Grande Valley: Improving Timeliness of Newborn Screening Specimens

C. Prinz, K. Connell, E. Villarreal, K. Kneupper; Texas Department of State Health Service

Presenter: Codie Prinz, Texas Department of State Health Service,
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Optimizing Congenital Hypothyroidism Screening in Texas: Implementation of a Hybrid Model

N. Choo, G. Bryars, L. Sanchez, G. Vargas, B. Reilly, S. Tanksley; Texas Department of State Health Services

Presenter: Amy Schlabach, Texas Department of State Health Services,
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Texas Department of State Health Services Changes to Instrumentation and Algorithm for IRT Screening

A. Schlabach, G. Vargas; Texas Department of State Health Services

Presenter: Amy Schlabach, Texas Department of State Health Services,
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Ten Years of Newborn Screening Follow-up in Rural Northern Indiana

J. Werker¹, Z. Ammous¹, B. Lesko²; ¹The Community Health Clinic, ²Indiana Newborn Screening Laboratory

Presenter: Jody Werker, RN, The Community Health Clinic, jwerker@indianachc.org

***P-108**

An Interactive, Searchable Database of LC-FAOD Gene Variants, Genotypes and Phenotypes

V. Rangel Miller¹, O. Japalaghi¹, H. Richbourg¹, M. AlSayed², P. Baker II³, S. Daugherty¹, S. Grünert⁴, A. Khan⁵, H. Kobayashi⁶, L. Korngut⁷, I. Schwartz⁸, J. Vockley⁹; ¹Ultragenyx Pharmaceutical, Inc., ²Al-Faisal University, ³University of Colorado, ⁴Freiburg University Hospital, ⁵M.A.G.I.C. (Metabolics and Genetics in Canada) Clinic Ltd., ⁶Shimane University Hospital, ⁷University of Calgary, ⁸Hospital de Clinicas, ⁹UPMC Children's Hospital of Pittsburgh

Presenter: Vanessa Rangel Miller, MS, MBA, CGC, Ultragenyx Pharmaceutical, Inc., vrangelmiller@ultragenyx.com

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Mucopolysaccharidosis Type VII: A Locus-specific Database of GUSB Gene Variants, Genotypes and Phenotypes

V. Rangel Miller¹, T. Hamazaki², M. Hegde³, R. Giugliani⁴, T. MacKenzie⁵, L. Dilworth¹, M. Kiel⁶, O. Japalaghi¹, B. Lianoglou⁵, J. Merritt II¹, T. Nalin¹, V. Pansare¹, L. Pollard⁷, H. Richbourg¹, T. Sparks⁵, S. Daugherty¹; ¹Ultragenyx Pharmaceutical, Inc., ²Osaka Metropolitan University, ³Revvity, ⁴DASA and Casa dos Raros, ⁵University of California, San Francisco School of Medicine, ⁶Genomenon, ⁷Greenwood Genetic Center

Presenter: Vanessa Rangel Miller, MS, MBA, CGC, Ultragenyx Pharmaceutical, Inc., vrangelmiller@ultragenyx.com

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Expansion of Newborn Screening Short-term Follow-up for Hemoglobin Traits – Leveraging Funding to Reach an Additional 1000+ Families in a Rural Southern State

L. Hays, S. Fendley;¹University of Arkansas for Medical Sciences

Presenter: Laura Hays, PhD, APRN, CPNP-PC, FAHA, University of Arkansas for Medical Sciences, lhays@uams.edu

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Implementation of Electronic Test Orders and Results for Newborn Screening in Utah

A. Farrington, J. Himes, M. Rindler, N. Szabo-Fresnais, J. Shao; Utah Department of Health and Human Services

Presenter: Amy Farrington, MLS(ASCP)CM, Utah Department of Health and Human Services, afarrington@utah.gov

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MPS-I and MPS-II Newborn Screening with One Dried Blood Spot Punch: Expansion of the Utah LC-MS/MS Platform

F. Mesmar¹, H. Golsan¹, N. Szabo-Fresnais¹, H. Khaledi², M. Gelb³; ¹Utah Department of Health and Human Services, ²Enfanos, ³University of Washington

Presenter: Fahmi Mesmar, PhD, Utah Department of Health and Human Services, fmesmar@utah.gov

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Lysosomal Storage Disorders and Adrenoleukodystrophy Newborn Screenings with One Punch Using High Performance Chromatography-tandem Mass Spectrometry: From Method Development to Universal Population Screening in Utah

N. Szabo-Fresnais, Utah Department of Health and Human Services

Presenter: Nicolas Szabo-Fresnais, PhD, Utah Department of Health and Human Services, nszabo@utah.gov

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Six Months of Next Generation Sequencing for Cystic Fibrosis as a Second Tier in Virginia

C. Alcorta, L. Lion, G. Cote, E. Hopkins; Virginia Division of Consolidated Laboratory Services

Presenter: Christian Alcorta, MS, Virginia Division of Consolidated Laboratory Services, christian.alcorta@dgs.virginia.gov

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Opportunities to Improve Short- and Long-term Follow-up Data Collection with FHIR-based Queries

D. Paseltiner¹, A. Howard², J. Thompson², A. Ragsdale², M. Sontag³; ¹Skylight, ²Washington State Department of Health, ³Center for Public Health Innovation

Presenter: Anna Howard, MPH, Washington State Department of Health, Anna.Howard@doh.wa.gov

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WITHDRAWN

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Analytical Performance Evaluation of a Reagent Kit Under Development for the Measurement of Biological Metabolites in Dried Blood Spots, by FIA-MS/MS

P. Rossiter, N. Stafford, R. McBrinn, H. Brown; Waters Corporation

Presenter: Heather Brown, PhD, Waters Corporation, heather_brown1@waters.com

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Comparison of a Reagent Kit Under Development for the Measurement of Biological Metabolites in Dried Blood Spots, by FIA-MS/MS

N. Stafford, P. Rossiter, R. McBrinn, H. Brown; Waters Corporation

Presenter: Heather Brown, PhD, Waters Corporation, heather_brown1@waters.com

Notes

Symposia History

National Neonatal Screening Symposium (1981-1999)

Newborn Screening & Genetic Testing Symposium (2001-2019)

Newborn Screening Symposium (2020 onward)

1981	Austin, TX	2005	Portland, OR
1982	Chicago, IL	2007	Minneapolis, MN
1984	Orlando, FL	2008	San Antonio, TX
1985	Columbus, OH	2010	Orlando, FL
1986	Austin, TX	2011	San Diego, CA
1988	Portland, OR	2013	Atlanta, GA (with ISNS)
1989	New Orleans, LA	2014	Anaheim, CA
1991	Saratoga, NY	2016	St. Louis, MO
1992	Raleigh, NC	2017	New Orleans, LA
1994	Seattle, WA	2019	Chicago, IL
1995	Corpus Christi, TX	2020	Virtual
1996	Boston, MA	2021	Virtual
1998	San Diego, CA	2022	Tacoma, WA
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