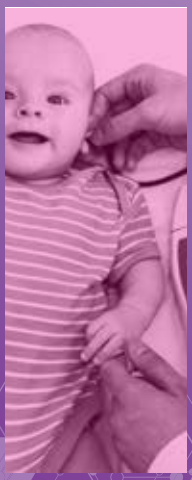
APHL Newborn Screening Symposium 2024

October 20–24, 2024 • Omaha, NE

APHL™


Final Program

Sponsored by the
Association of Public Health Laboratories

Co-Sponsored by the
Nebraska Department of Health and Human Services and the International Society for Neonatal Screening

www.aphl.org/NBS2024

#APHLNBS

Agenda at a Glance

All listed times are Central Daylight Time (CDT).

Sessions in teal will be recorded.

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 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.

APHL is a national leader in:

- Scientific Expertise
- Education and Training
- Health Policy
- Informatics
- Quality Assurance
- Workforce Development
- Laboratory Systems
- Global Laboratory Capacity

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Chief Executive Officer, APHL

Welcome to Omaha!

On behalf of the entire planning committee, welcome to the 2024 Association of Public Health Laboratories (APHL) Newborn Screening (NBS) Symposium in Omaha, Nebraska! We are thrilled to have a diverse group of NBS advocates, healthcare experts and families on a mission to promote healthier beginnings for all. Throughout the week, we hope you dive into sessions, network and explore innovative offerings at the exhibit hall.

Get ready for an exciting lineup of events, starting with the Parent/Patient Panel on Sunday to center our thoughts on the "why" of our work. Following the Parent/Patient Panel, please join us for the Welcome Reception, a fantastic opportunity to network, make new connections and shape the future of NBS through collaborations and friendships.

We are excited about Monday, which will start with the Innovate! sessions and an engaging keynote around the importance of fostering a healthy and dynamic public health workforce. We look forward to your active participation in the day's discussions on system improvements, ethical considerations and challenges around privacy in NBS.

Tuesday will offer a variety of roundtable discussions on topics such as regulatory compliance, advanced challenges in hemoglobinopathy screening and the impact of artificial intelligence (AI) in NBS. The day will also include in-depth sessions on health information technology, the importance of collaboration, lysosomal storage disorders (LSDs) and conditions currently on the recommended uniform screening panel

(RUSP), as well as a Rapid Posters session. We will honor our peers and colleagues during the APHL Newborn Screening Awards Ceremony and end the day with fast and fun presentations during the Ignite the Night session.

Take advantage of additional concurrent roundtable discussions covering family engagement, thyroid cutoffs and strategies for handling refusal rates on Wednesday morning followed by sessions on strengthening our NBS systems through follow-up and education. Make sure to visit all of the vendor booths by Wednesday morning before the afternoon raffle for your chance to win exciting prizes. For Wednesday afternoon, prepare for sessions on next generation sequencing (NGS) and quality assurance activities, followed by the always popular off-site social event in the evening.

We will conclude the symposium on Thursday with talks on refining testing procedures and exploring new screening frontiers before we announce the location of the 2025 APHL Newborn Screening Symposium!

During your stay, we hope you take the chance to explore Omaha. Known for its vibrant arts scene, you can visit the Joslyn Art Museum or catch a live performance at the Holland Performing Arts Center. For history enthusiasts, the Durham Museum offers fascinating exhibits in a stunning Art Deco building. The Henry Doorly Zoo and Aquarium, one of the best in the world, is perfect for a family outing. Stroll through the Old Market, where you'll find unique shops, charming cafes and some of Omaha's best dining options.

Let's make it a memorable week in Omaha filled with inspiration, knowledge sharing and enjoyable surprises, both within the symposium and throughout this vibrant city.

Jillian Chance DNP, APRN-NP, FNP-BC

Program Manager, Newborn Screening & Genetics

Nebraska Department of Health and Human Services

Chair, 2024 APHL Newborn Screening Symposium Planning Committee

APHL 2024 Newborn Screening Symposium Planning Committee

Chair, Jillian Chance, DNP, APRN-NP, FNP-BC, Nebraska Department of Health and Human Services

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Amy R.U.L. Calhoun, MD, Stead Family Department of Pediatrics, University of Iowa Health Care

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Peter Schielen, PhD, International Society for Neonatal Screening

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General Information

Meeting Location and Headquarters Hotel

Conference sessions and events: Omaha CHI Health Center Convention Center

Headquarters hotel: Hilton Omaha
1001 Cass St, Omaha, NE 68102

Secondary hotels: Hilton Garden Inn Omaha Downtown and
Omaha Marriott Downtown at the Capitol District

Registration Hours

Sunday, October 20	1:00 pm – 6:30 pm
Monday, October 21	7:30 am – 6:30 pm
Tuesday, October 22	7:30 am – 6:30 pm
Wednesday, October 23	7:30 am – 6:30 pm
Thursday, October 24	7:30 am – 12:00 pm

Lactation Rooms Available

A Mom Pod is available on the exhibit hall level. An extra room is located just outside of the South Ballroom Foyer. 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 212.

Consent to Use Photographic Images

Registration and attendance at or participation in APHL conferences and other activities constitutes an agreement by the registrant to APHL's use and distribution (both now and in the future) of the registrant's or attendee's image or voice, without compensation, in photographs, video and audio recordings of such events and activities.

Complimentary Wireless Internet in the Meeting Space

There is complimentary wireless internet in the public areas.

Network: APHL_NBSS2024

Password: APHL2024

Enhance Your Experience with the Mobile App

Available in your app store in October! Search for "APHL Conferences" in your app store to download.

Access details on sessions, posters and exhibitors before the symposium 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.

Tribal Support

The 2024 APHL Newborn Screening Symposium will take place in Omaha, NE, which occupies the ancestral homelands of the Umoⁿhoⁿ (Omaha), Otoe-Missouria, and Očhéthi Šakówiŋ (Sioux) tribes. APHL expresses its support to these tribes in their struggle for federal recognition and restoration of their ancestral homelands.

Inclusivity and Accessibility

As part of our ongoing commitment to inclusivity and respect for diversity, we want to ensure that all attendees feel valued and accommodated, especially regarding religious observances and holidays. We are proud that our team and members represent a variety of faiths and beliefs, and we are committed to making every effort to honor dietary restrictions, accessibility needs and other accommodations during our scheduled meetings and events.

If you have specific requirements or preferences related to religious observances, dietary restrictions, accessibility or any other accommodations, please let us know in advance so that we can make the necessary arrangements. Your comfort and well-being are important to us, and we want to create an environment where everyone can fully participate and feel included.

Well-Being

In order to ensure the health and well-being of everyone participating in this meeting, we kindly remind you to prioritize your health and that of others when deciding whether to attend in-person meetings, especially if you are feeling unwell.

If you are experiencing any symptoms of illness or have been in contact with someone who is sick, we strongly encourage you to reschedule your participation in the meeting. Your health and safety are our top priorities, and we appreciate your cooperation in maintaining a healthy work environment for all.

Please don't hesitate to reach out if you have any concerns or questions regarding this matter. Let's continue to support each other and prioritize our collective well-being.

Mobility

The Omaha CHI Health Center Convention Center is an Americans with Disabilities Act (ADA) compliant center.

There is a skywalk available from the Hilton Omaha to the Convention Center as well as elevators available from the Hilton Omaha to the Convention Center level. All entrance doors to the building also have ADA buttons. Please visit the registration desk with any questions.

Food Restrictions and Allergies

We ensure that APHL events offer vegetarian, vegan, gluten-free and dairy-free food options. Buffets will have menu labels clearly indicating ingredients and/or accommodations. Please include your food restrictions and specific allergies when registering.

Continuing Education Credits

P.A.C.E.® Continuing Education Credits

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 21.5 P.A.C.E. credits by attending the entire symposium. Please refer to your tote bag insert for more information.

Certified Public Health Recertification Credits

APHL is an approved provider of Certified in Public Health (CPH) recertification credits through the National Board of Public Health Examiners (NBPHE). Attendees have the opportunity to earn up to 22 hours of credit by attending the entire symposium. APHL will not issue certificates of CPH credits earned. The attendee is responsible for keeping track of the hours earned.

Continuing Medical Education/Continuing Nursing Education/Certified Health Education Specialist/ Genetic Counselor Credits

CME, CNE, CHES and Genetic Counseling CEUs (up to 21.5 credit hours) will be provided. Please refer to the tote bag insert for more information.

Not-to-Miss Events

All listed times are Central Daylight Time (CDT).

Parent/Patient Panel

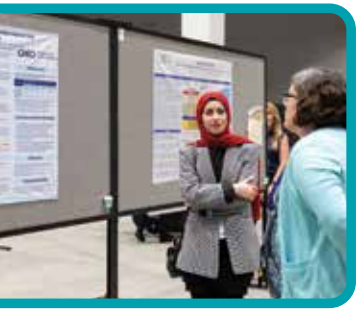
Sunday, October 20 | 4:30 pm – 6:00 pm

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

Sunday, October 20 | 6:00 pm – 7:30 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 newborn screening family advocates in 'The Living Room.'



Visit with Poster Authors

Sunday, October 20 | 6:00 pm – 7:00 pm

Tuesday, October 22 | 12:30 pm – 1:30 pm

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

Visit with Newborn Screening Families and Advocates in 'The Living Room'

Visit 'The Living Room' with Newborn Screening Families and Advocates

Join us in this cozy and relaxed area located just inside the exhibit hall to chat, connect and share with newborn screening families and advocates.

Innovate! Sessions

Monday, October 21 | 7:00 am – 7:30 am and 7:45 am – 8:15 am

Connect with your industry partners and learn of new technologies and services.

APHL Newborn Screening Awards Ceremony

Tuesday, October 22 | 1:30 pm – 3:30 pm

Applaud your colleagues and celebrate their achievements.

Ignite the Night!

Tuesday, October 22 | 5:30 pm – 6:30 pm

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

MLD Newborn Screening Enables Access to Gene Therapy — Documentary Screening and Discussion

Thursday, October 24 | 7:00 am – 8:00 am

This session will include a screening of the documentary "Sequencing Hope," followed by an interactive discussion of the collaborative implementation of MLD newborn screening.

Convention Center Floor Plans

Meeting Room Level (2nd floor)

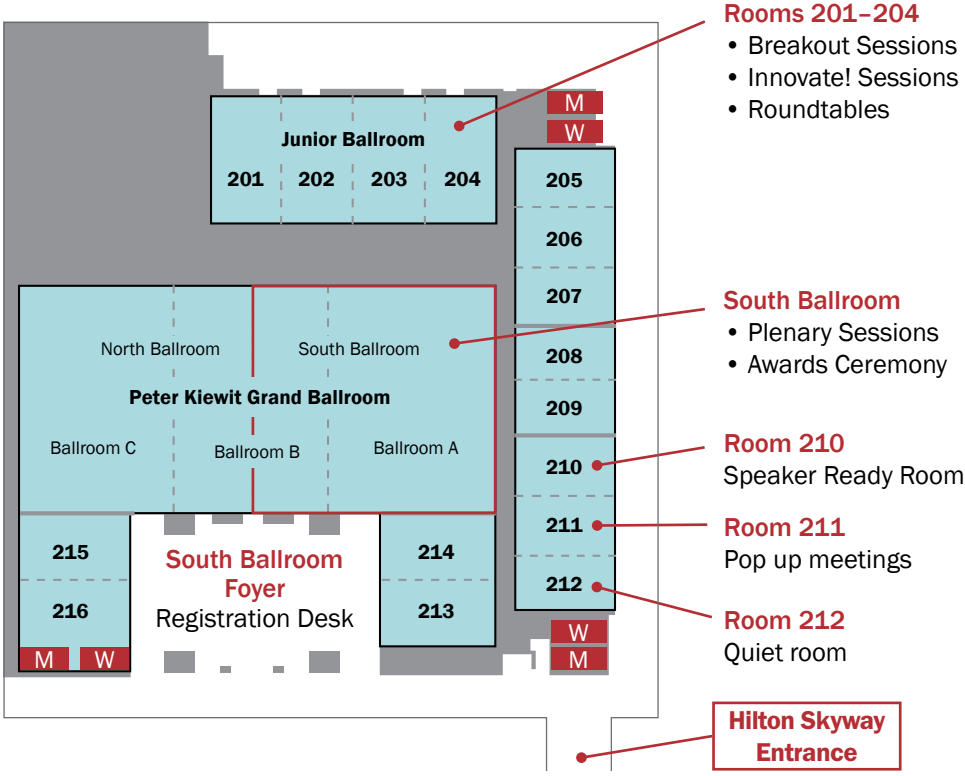
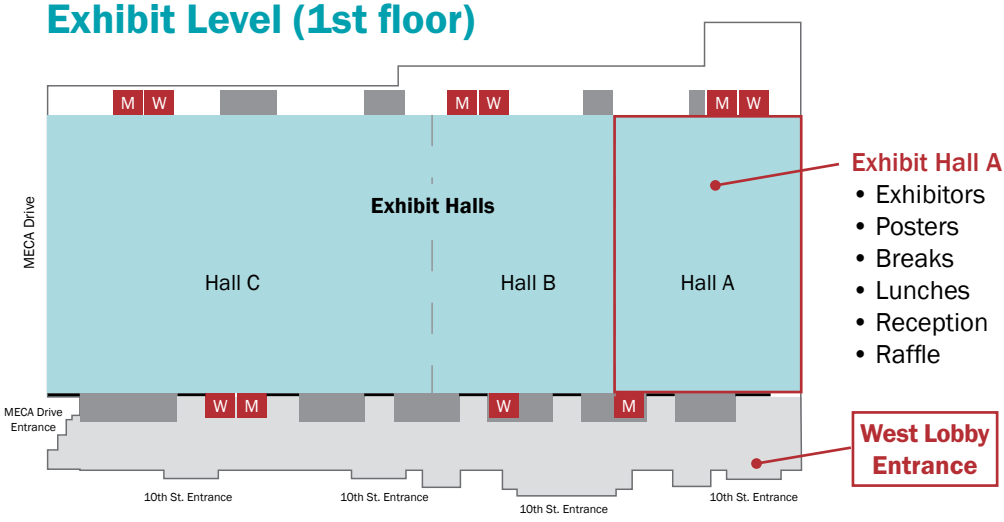


Exhibit Level (1st floor)



Agenda of Events

All times are Central Daylight Time (CDT).

Sunday, October 20

1:00 pm – 6:30 pm Registration

South Ballroom Foyer

4:00 pm – 4:30 pm Welcome

South Ballroom

Speakers:

- Scott J. Becker, MS, Association of Public Health Laboratories
- Jelili Ojodu, MPH, Association of Public Health Laboratories
- Jillian Chance, DNP, APRN-NP, FNP-BC, Nebraska Department of Health and Human Services

4:30 pm – 6:00 pm Parent/Patient Panel

South Ballroom

Moderators:

- Alyssa Keller, MS, CGC, University of Nebraska Medical Center Munroe-Meyer Institute
- Robert Rauner, United Leukodystrophy Foundation

Speakers:

- Jamie Slaughter
- Jennifer Wallin
- Kala McWain
- Brittany Parke

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.

6:00 pm – 7:30 pm Welcome Reception

Exhibit Hall

Visit the exhibits and posters and continue the conversation with our newborn screening family advocates in 'The Living Room,' located near the exhibit hall entrance.

6:00 pm – 7:00 pm

Exhibit Hall

Poster authors P-1 to P-65 available to discuss their posters

7:00 pm – 8:30 pm

Newborn Screening Laboratory Social Hour Room 208

Newborn Screening Follow-up & Family Social Hour Room 209

Whether you're 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!

Monday, October 21

7:30 am – 6:30 pm — Registration

South Ballroom Foyer

6:30 am – 8:00 am — Coffee/Light Breakfast

Room 201-204 Foyer

7:00 am – 8:15 am — Concurrent Innovate Sessions

Thermo Fisher Scientific	Room 201
Quantabio	Room 202
Agilent Technologies	Room 203
Revvity, Inc.	Room 201
Sebia	Room 202
Waters Corporation	Room 203

See page 16 for session details.

8:15 am – 8:30 am — Break

South Ballroom Foyer

8:30 am – 10:00 am — Newborn Screening System Issues, Updates and Initiatives

South Ballroom

588-846-24 • 1.5 contact hours for this session

Moderators:

- Holly Winslow, BS, Minnesota Department of Health
- M. Christine Dorley, PhD, Maryland Department of Health

International Survey on Practice of Newborn Screening for Phenylketonuria: Preliminary Report

Domen Trampuz, University Medical Center Ljubljana, Slovenia

Weathering The Storm: The Iowa NBS Lab's Unexpected Replacement of its CFTR Screening Test

Jerusalem Alleyne, PhD, Iowa State Hygienic Laboratory

Updates to the National Newborn Screening Contingency Plan (CONPLAN)
Scott Shone, PhD, HCLD(ABB), North Carolina State Laboratory of Public Health

An Initiative for Tokyo's Newborn Screening: A Comparative Approach to Enhancing Child Health Outcomes
Kimihiko Oishi, MD, The Jikei University School of Medicine

This session will explore best practices that newborn screening (NBS) programs in the United States and globally can adopt in order to improve outcomes for newborns with rare diseases. Speakers will focus on how to ensure all babies receive screening in a timely manner during periods of turmoil, provide updates to the National Newborn Screening Contingency Plan (CONPLAN) and identify how countries with limited resources can learn from more established NBS programs to enhance screening quality and health outcomes everywhere.

10:00 am – 4:00 pm Exhibit and Poster Hall Open

Exhibit Hall

10:00 am – 10:30 am Break – Visit the exhibitors and posters

Exhibit Hall

10:30 am – 12:00 pm Keynote Session

South Ballroom

588-847-24 • 1.5 contact hours for this session

Fostering the Workforce of Tomorrow: How We Can Work Together to Build Trust, Inclusion and Morale in Public Health

Moderators:

- Michele Caggana, ScD, FACMG, New York State Department of Health
- Emily Eisenstein, MPH, Rhode Island Department of Health

Advancing Public Health Together: Nebraska's Commitment to Workforce Development
Timothy A. Tesmer, MD, Nebraska Department of Health and Human Services

Bridging the Gap: Engaging the Next Generation of Public Health Professionals
Omari Richins, MPH, The Public Health Millennial, LLC

The Importance of Health Equity in Building a Dedicated Public Health Workforce
Johanne E. Morne, New York State Department of Health

Our keynote session will focus on strategies for recruiting younger generations into public health while also retaining the current public health workforce in order to strengthen the newborn screening (NBS) system as a whole. Speakers will address ways to promote interest in public health careers and build trust, interconnectedness, inclusion and morale to ensure a strong and committed workforce of tomorrow.

Innovate! Sessions

Monday, October 21

7:00 am – 7:30 am

Changing the Course of Inherited Disorders — Utilizing the Quantitative Strength of LC-MS to Advance Newborn Screening Biomarker Measurement with the Thermo Scientific Stellar Mass Spectrometer

Thermo Fisher Scientific • Room 201

Speaker:

- Jingshu Guo, PhD, Thermo Fisher

The advancement of newborn screening biomarker measurement for enhanced detection accuracy necessitates confident quantification of more analytes with increased sensitivity, specificity and throughput. Thermo Scientific™ Stellar™ mass spectrometer synergistically combines the robust quantitative performance of triple-quadrupole technology with the sensitive, hyper-fast full-scan MSn acquisition of dual-pressure linear ion trap technology. These unprecedented analytical capabilities empower laboratories to expand their newborn screening system capacity while upholding unwavering confidence and speed in detecting inherited genetic disorder biomarkers.

Rapid Extraction of High-quality Genomic DNA from Dried Blood Spots and Library Preparation Using SparQ DBS Library Prep Kit for Whole Genome Sequencing

Quantabio • Room 202

Speaker:

- Subrata Panja, PhD, Quantabio

Quantabio has developed a simple and efficient method for isolation of high purity, high molecular weight (>50 kb) genomic DNA from dried blood spots (DBS) in 40 minutes. High yields of gDNA from as few as 3, 3 mm DBS punches generated sufficient gDNA for preparation of high-quality PCR-free libraries using the sparQ DBS Library Prep Kit in just 90 minutes. Minimal use of each DBS sample enabled conservation of the remaining DBS or utilization for other analyses. Sequencing results showed minimal bias, low duplication rate and robust variant detection. Overall, the sparQ DBS Library Prep Kit offers a rapid, cost-effective and reliable solution for sequencing newborn samples.

Automate Screening and Confirmation of NBS Workflows — Convert GCMS Organic Acids to LCMS

Agilent Technologies • Room 203

Speaker:

- Julie Cichelli, PhD, Agilent

Automate screening and confirmation of newborn screening (NBS) workflows overnight with the Agilent Intelligent Reflux using LCMS QQQ. Following the screening for NBS metabolite Biomarkers, automatically switch to a confirmatory method on presumptive positives - screening and confirmation completed overnight using the intelligent reflexive injection logic. Adapt GCMS organic acids to LCMS QTOF > 60 organic acids. No derivatization required. Selectivity improved with HRAM. Dramatically reduce analysis time vs GCMS. Early maintenance feedback, clock time scheduled tuning.

7:30 am – 7:45 am

Break • Room 201-204 Foyer

7:45 am – 8:15 am

Informatic Solution to Support NGS in the Newborn Screening Laboratory

Revvity, Inc. • Room 201

Speaker:
- Zeq Ma, MS, PhD, Revvity

This presentation will review Revvity's end-to-end software applications. These applications can be used by laboratories to perform and track next generation sequencing (NGS) experimental data as well as bioinformatics pipelines to support both primary and secondary genomic data analysis. Additionally the software is available for genomic variant analysis, interpretation, classification and reporting.

For research use only. Not for use in diagnostic procedures.

Transforming NBS for Hb Disorders: Introducing the CAPILLARYS 3 DBS

Sebia • Room 202

Speaker:
- Katherine Berg, Sebia, Inc.

Discover the CAPILLARYS 3 DBS, an automated, high-throughput system tailored for newborn hemoglobin disorder screening. This session will delve into Capillary Electrophoresis technology, optimized for newborn screening (NBS), offering clear, high-resolution detection of Hb S, C, D, E and Bart's. Discover how this innovation enhances lab workflows and delivers high quality results. Join us to learn about the system's features and benefits, followed by an interactive Q&A.

Enabling Innovation by Progressing Together

Waters Corporation • Room 203

Speakers:
- Curt Paulette, Waters Corporation
- Ari Herbst, GelbChem

Newborn screening (NBS) service delivery and the continuing evolution of needs is a journey that leans into the skills and strengths of colleagues as well as company partners. Learn how waters_connect for IVD QUAN review software can save up to 50% of your second-tier screening review and from our colleagues at GelbChem, how modern solutions for NBS labs must deliver flexibility, technical support and premium quality at a budget-friendly price.

12:00 pm – 2:00 pm **Lunch (on your own)**

1:15 pm – 2:00 pm **Dessert**
Exhibit Hall

2:00 pm – 3:30 pm **Process Improvements in Newborn Screening**
South Ballroom

588-848-24 • 1.5 contact hours for this session

Moderators:

- Jillian Chance, DNP, APRN-NP, FNP-BC, Nebraska Department of Health and Human Services
- Michael Cellucci, MD, Nemours Children's Hospital, Delaware

Current Patterns of Beta Thalassemia Newborn Screening and Reporting in the United States
Yao Fu, MPH, Vertex Pharmaceuticals Incorporated

Unite Newborn Screening: An Early Hearing Detection and Intervention and Dried Blood Spot Learning Community
Alyson Ward, PhD, MS, IA CHES, National Center for Hearing Assessment and Management

Validating a Custom Next-generation Sequencing Assay for Cystic Fibrosis Newborn Screening
Christian Alcorta, MS, Virginia Division of Consolidated Laboratory Services and Aki Harada, PhD, Virginia Division of Consolidated Laboratory Services

Strengthening Newborn Screening Systems Quality and Partnerships by Implementing a Hospital Communication Portal
Kathy Chou, PhD, New York State Department of Health

Clearing the Air: Strategies for Improving Communication with Providers on Cystic Fibrosis in Tennessee
Angela Keyton, RN, Tennessee Department of Health

This session will highlight strategies for improving processes within newborn screening (NBS) programs. Discussions will focus on the importance of standardized reporting in NBS, the vital role that collaboration plays to ensure unified and cohesive messaging to families and ways to ensure timely diagnosis to initiate appropriate interventions and improve patient outcomes.

3:30 pm – 4:00 pm **Break – Visit the exhibits and posters**
Exhibit Hall

4:00 pm – 5:30 pm
South Ballroom

Access, Ethics and Equity in Newborn Screening

588-849-24 • 1.5 contact hours for this session

Moderators:

- Carla Cuthbert, PhD, FACMG, Centers for Disease Control and Prevention
- George Dizikes, PhD, HCLD/CC(ABB), Illinois Department of Public Health

Ensuring Access to Screening
Alyssa Rapp, BS, Oregon State Public Health Laboratory

Expressions of (Mis)trust: Engaging Underrepresented Communities About the Storage and Use of Newborn Screening Bloodspots
Aaron Goldenberg, PhD, MPH, Case Western Reserve University

Tackling Institutional Review Board, Regulatory, Recruitment and Study Design Challenges to Reach Families for Newborn Screening Research
Anne Atkins, MPH, Children's National Hospital

Parental Perspectives on the Utilization of Residual Dried Blood Spots: Insights from ScreenPlus
Katrina Paleologos, MPH, Albert Einstein College of Medicine/Children's Hospital at Montefiore

This session will focus on the intersection between access, ethics and equity in newborn screening (NBS) and how programs can improve how they communicate with their community. Presentations include: a look into why certain populations opt out of NBS and ways to address any barriers to screening that families may face; how NBS programs can establish and preserve trust and open communication among parents and the public, especially in light of concerns around storage and usage of residual bloodspots and data; and how to incorporate parent perspectives into policy in order to mitigate the risk of ongoing legal and social injustices.

5:30 pm – 6:00 pm
South Ballroom Foyer

Break

6:00 pm – 7:30 pm
South Ballroom

National Institutes of Health Townhall/Focus Group

NBSXWGS: An NIH Pilot Project to Assess the Feasibility of Adding a WGS Component into the US NBS Program

Moderators:

- Melissa A. Parisi, M.D., Ph.D., Intellectual & Developmental Disabilities Branch, Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health
- Dominique C. Pichard, M.D., M.S., Division of Rare Diseases Research Innovation, National Center for Advancing Translational Sciences, National Institutes of Health

- Philip J. (P.J.) Brooks, Ph.D., Division of Rare Diseases Research Innovation, National Center for Advancing Translational Sciences, National Institutes of Health

Please join our partners from NIH for a townhall meeting/focus group discussion on accessing the feasibility of adding a whole genome sequencing (WGS) component into the US newborn screening (NBS) program. Moderators will gather feedback, including challenges and opportunities, about a NBS pilot research project that would evaluate WGS for a panel of treatable genetic conditions at the statewide laboratory level in newborns.

Tuesday, October 22

7:30 am – 6:30 pm **Registration**

South Ballroom Foyer

6:30 am – 9:00 am **Coffee**

Room 201-204 Foyer

7:00 am – 8:00 am **Concurrent Roundtables**

Model Language for State Regulations Regarding NBS Specimen and Data Retention

Room 201

588-850-24 • 1 contact hour for this session

Kimberly Noble Piper, RN, BS, CPH, CPHG, Iowa Department of Health and Human Services and Aaron Goldenberg, PhD, MPH, Case Western Reserve University

The APHL Newborn Screening (NBS) Ethical, Legal, Social and Policy Issues (ELSPI) Subcommittee reviewed existing state statutes regarding law enforcement access to NBS resources in developing language to help states clearly restrict law enforcement access and use of these residual samples and associated data in such investigations. This roundtable will present several possible policy options, and their rationale, which attendees can modify to suit their state's needs. Attendees will leave with model language ready for incorporation into state regulation, policy or statute after appropriate review and approval by their policy makers.

Tough Calls in Hemoglobinopathy Screening: Stump the Experts with Your Difficult Cases and Learn from Others

Room 202

588-851-24 • 1 contact hour for this session

Patrick Hopkins, BS, Missouri State Public Health Laboratory

While most newborn screening (NBS) for hemoglobinopathies (HGBs) is straight forward, subtleties in interpreting protein data can be difficult and confusing, requiring experience and a deep understanding of the caveats of the platforms used. High workforce turnover and loss of institutional knowledge can lead to problems in accurately determining some HGB screening results. This roundtable will make use of the experts in the audience to share and

work through several demonstrative cases. Attendees should bring their thinking caps and be prepared for a fun and interactive roundtable that will allow everyone to learn from best practices and encourage ongoing discussions among states.

Assessing the Need for Artificial Intelligence in Newborn Screening
Room 203

588-852-24 • 1 contact hour for this session

Marianna Raia, MS, CGC Expecting Health and Brendan Reilly, BS, Texas Department of State Health Services

As medical technologies continue to advance, the integration of automation and artificial intelligence (AI) has become increasingly prevalent, offering promising avenues for enhancing healthcare practices. The newborn screening (NBS) landscape is also evolving and stands to benefit significantly from AI applications. This roundtable aims to explore the opportunity and feasibility of integrating AI into NBS processes and will discuss its applications in key areas including variant interpretation, biochemical cutoff and algorithm determination, results communication, consent processes and standardization across the system.

8:00 am – 8:30 am Break

Room 201-204 Foyer

8:30 am – 10:00 am Newborn Screening Health Information Technology

South Ballroom

588-853-24 • 1.5 contact hours for this session

Moderators:

- Sarah Ward, BS, Nebraska Department of Health and Human Services
- Sarah McKasson, MPH, Association of Public Health Laboratories

Informatics Gap Analysis Aids NBS System Updates
Mary Kate Yost-Daljev, PhD, J Michael Consulting

Leveraging Existing Infrastructures to Solve the Follow-up Data Puzzle: The Case for eCR
Ashley Nash, DrPH, MPH, PMP, Centers for Disease Control and Prevention

Enhancing Public Health Outcomes Using a Fast Healthcare Interoperability Resources (FHIR) Query Client: Applications in Newborn Screening
Marcelle Goggins, Skylight and Daniel Paseltiner, Skylight

Implementing Electronic Test Orders and Results for Newborn Screening Using an Intermediary
Rebecca McNall, PhD, Centers for Disease Control and Prevention

This session will delve into platforms that newborn screening (NBS) programs can use to effectively collect, interpret and exchange information. Presentations will focus on ways the Laboratory Information Management System (LIMS) can ensure long-term informatics strategy for NBS programs, provide information on how to implement Electronic Case Reporting (eCR)

infrastructure in order to reduce the manual burden of exchanging case report information, how to confront challenges associated with collecting results for critical congenital heart disease (CCHD) and Early Hearing Detection and Intervention (EHDI), and ways to ensure a faster and more complete exchange of information using Electronic Test Orders and Results (ETOR) in order to benefit all NBS partners.

10:00 am – 4:00 pm Exhibit and Poster Hall Open

Exhibit Hall

10:00 am – 10:30 am Break – Visit the exhibits and posters

Exhibit Hall

10:30 am – 12:00 pm Concurrent Sessions

Collaboration Through Training, Education and Communication

Room 204

588-854-24 • 1.5 contact hours for this session

Moderators:

- Marianna Raia, MS, CGC, Expecting Health
- Liz Carter, Patient Advocate, Rare & Metabolic Diseases Communications Manager, HCU Network America

A Collaborative Approach to Newborn Screening Education in Community Birth Settings
Kristen Thompson, MPH, Michigan Department of Health and Human Services

Creating a Positive Snowball Effect: Targeted Education to Reduce High Initial Fail Rates for Hearing Screenings at Hospitals
Hilary Fryman, BSN, RN, Tennessee Department of Health

Implementing a Formal Training Program for Hospitals and Midwives: A 3-year Review
Hilary Fryman, BSN, RN, Tennessee Department of Health

Newborn Screening ACT Sheets for Cystic Fibrosis: Current Use, Revisions and Next Steps
Marci Sontag, PhD, Center for Public Health Innovation

This session will focus on ways to enhance best practices within newborn screening (NBS) programs to ensure prompt care for families. Attendees will learn how to improve education for midwives through collaboration among NBS programs, how issues with hearing and critical congenital heart disease (CCHD) screenings can be improved through training, and the importance of continual updates to guidelines and resources in order to meet the current needs of NBS programs and primary care providers (PCPs).

Exploring Lysosomal Storage Disorders (LSDs)

South Ballroom

588-855-24 • 1.5 contact hours for this session

Moderators:

- George Dizikes, PhD, HCLD/CC(ABB), Illinois Department of Public Health
- Alyssa Keller, MS, CGC, University of Nebraska Medical Center Munroe-Meyer Institute

Newborn Screening Pilot Study for Mucopolysaccharidosis Type II in North Carolina
Katerina Kucera, PhD, RTI International

Optimized Extraction of Endogenous Biomarkers for MPS I and MPS II Prior to LC-MS/MS Analysis: Implications for Newborn Screening and Follow-up
Craig Seymour, PhD, Mayo Clinic

Adaptation of a Cold-induced Aqueous Acetonitrile Phase Separation Extraction Method for Multiplexed (LC)-MS/MS Analysis of Iduronate-2-Suphatase (MPS-II) with Alpha-L-iduronidase (MPS-I) and Acid alpha-glucosidase (Pompe)
Adrienne Manning, BS, Connecticut Department of Health

Performance Analysis of a Second Tier Test Designed to Reduce False Positive Screens for Pompe Disease
Patricia Hall, PhD, Mayo Clinic

Five Years of Newborn Screening for Pompe, Mucopolysaccharidosis Type I, Gaucher and Fabry
Patrice Held, PhD, Oregon State Public Health Laboratory

This session will explore how newborn screening (NBS) programs adapt their screening processes as more disorders are added to the Recommended Uniform Screening Panel (RUSP). Presentations will focus on the results from a pilot study for mucopolysaccharidosis type II (MPS II), ways to best optimize the method of detection for MPS I and MPS II from dried blood spots (DBS), a review of modifications that need to be made to current methods when conditions are added to the RUSP, how reducing false positive results for Pompe can help preserve financial and clinical resources for NBS programs, and a long-term investigation into testing methods for four lysosomal storage disorders (LSDs) to provide considerations for long-term follow-up.

12:00 pm – 12:30 pm
South Ballroom

Rapid Poster Presentations

See page 24 for session details.

12:30 pm – 1:30 pm
Exhibit Hall

Lunch – Visit exhibits and posters

12:30 pm – 1:30 pm
Exhibit Hall

Poster authors P-66 to P-129 available to discuss their posters

Rapid Poster Presentations

Tuesday, October 22, 12:00 pm – 12:30 pm

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

P-34: From Heel Prick to Recommendation Statements: CF Foundation Newborn Screening Consensus Recommendations
Marissa Taylor, MPH, Cystic Fibrosis Foundation and Marci Sontag, PhD, Center for Public Health Innovation

P-57: Genetic Providers' Views on Trauma-informed Care in Genetics Clinics
Jessica Scott Schwoerer, MD, Medical College of Wisconsin

P-101: PEDI CAMPAIGN — A Newborn Screening Working Group US-wide Pediatricians Education Initiative on Newborn Screening for Lysosomal Storage Diseases
Elizabeth Vengoechea, MS, CGC, Sanofi

P-58: Improving Communication with a Positive Newborn Screen
Jessica Scott Schwoerer, MD, Medical College of Wisconsin

P-117: Parallel Biochemical and Genetic Testing Informs a Timely and Accurate Diagnosis of MPS VII: Findings from 5 Years of Sponsored Testing Programs
Nicole Miller, PhD, Ultragenyx Pharmaceutical, Inc.

P-96: Improved IEF workflow by using Migele Gel Analyzer
Lynn Lira, BS, MT(ASCP), Revvity Omics

P-41: AI Cloud-based End-to-End Technology for Accurate, Fast & Affordable Newborn Screening of Genetic Disorders / Rare Diseases Towards Precision Treatments
Chris Kyriakidis, PhD, MSc, MBA, gMendel

P-24: Comparison of Method Performance in Hemoglobinopathy Phenotype Identification
Sherri Zobel, MT, ASCP, Centers for Disease Control and Prevention

P-120: Newborn Sequencing: Approaches Taken by Programs Around the Globe
Nicole Miller, PhD, Ultragenyx Pharmaceutical, Inc.

1:30 pm – 3:00 pm
South Ballroom

2024 APHL Newborn Screening Awards Ceremony

Applaud your colleagues and celebrate their achievements. Moderated by Scott J. Becker, MS, Association of Public Health Laboratories and Timothy Southern, PhD, D(ABMM), South Dakota Public Health Laboratory.

3:00 pm – 3:30 pm
Exhibit Hall

Break – Visit the exhibits and posters

3:30 pm – 5:00 pm
South Ballroom

Recommended Uniform Screening Panel (RUSP) Conditions

588-856-24 • 1.5 contact hours for this session

Moderators:

- Kostas Petritis, PhD, Centers for Disease Control and Prevention
- Jess Jorgensen, BSN, Children's Nebraska

Recommendations from the ACHDNC Condition Counting Workgroup: A Step Towards National Harmonization
Susan Tanksley, PhD, Texas Department of State Health Services

Consistency or Inconsistency: A 5-year Review of Congenital Hypothyroid Referrals in Delaware
Michael Cellucci, MD, Nemours Children's Hospital, Delaware

Elucidation of Several ACADVL Pathogenic Variants in Texas Through NBS Cross-functional Collaboration
Brooke Allen, PhD, Texas Department of State Health Services

CDC's Laboratory Activities to Support Newborn Screening for Spinal Muscular Atrophy
Christopher Greene, PhD, Centers for Disease Control and Prevention

Reducing the Burden of Confirmatory Testing on Specialists and Providers: New England Newborn Screening Program Experience
Inderneel Sahai, MD, New England Newborn Screening Program

This session will explore conditions currently on the Recommended Uniform Screening Panel (RUSP). Discussion will include recommendations for how states can count primary and secondary conditions, the findings of a 5-year review of congenital hypothyroidism (CH) referrals and ensuing action items, how the Texas newborn screening (NBS) program collaborated with various groups to assist with their screening processes, and ways the Centers for Disease Control and Prevention (CDC) supports the implementation of spinal muscular atrophy (SMA) screening. The session will conclude with a look into how programs can ensure their NBS system is not being overwhelmed as the number of disorders added to the RUSP continues to expand.

5:00 pm – 5:30 pm Break

South Ballroom Foyer

5:30 pm – 6:30 pm Ignite the Night!

South Ballroom

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

Effortless Data Automation with Revvity Transcribe AI
Justin Anderle, Revvity, Inc.

Simplifying Your LCMS Operation and Reducing Cost of Ownership
Julie Cichelli, PhD, Agilent Technologies

A Circular Journey for HbP Screening in the Netherlands: From HPLC to Capillary Electrophoresis and Back Again
Rose Maase, Dutch National Institute for Public Health and the Environment (RIVM) and Auke Otten, M.Sc, Dutch National Institute for Public Health and the Environment (RIVM)

Multiplexing Is Your Ticket to Minimum Hassle Expansion
Michael Gelb, PhD, GelbChem LLC

The Journey of Long-term Follow-up: Partnerships to Measure Success
Jennifer Baysinger, MSN, RN, Oklahoma State Department of Health,
Jennifer Hauser, MPH, RN, PHN, Minnesota Department of Health,
Yvonne Kellar-Guenther, PhD, Center for Public Health Innovation and
Marci Sontag, PhD, Center for Public Heath Innovation

Hospital In-Service Blitz: The New Jersey Newborn Screening Eras State Tour
Mary Carayannopolous, PhD, New Jersey Department of Health and
Victoria Floriani, MS, New Jersey Department of Health

Bridging Worlds: My Journey and Japan's Innovations in Newborn Care
Kimihiko Oishi, MD, The Jikei University School of Medicine

Erin and the Terrible, Horrible, No Good, Very Bad Day
Michelle Mills, MS, Kansas Department of Health and Environment

Participatory Chatbot Design to Deliver Carrier Status Hemoglobinopathy Newborn Screen Results to Parents
Courtney M. Gauchel, RN-BSN, MS in Progress, APHL Fellow, 2nd Year NTMS Graduate Student at the University of Utah in Biomedical Informatics

Building the Next Generation of Laboratory Scientists Through the Internship and Fellowship Programs
Mariane S. Wolfe, MS, MLS(ASCP)^{CM}, Association of Public Health Laboratories

Wednesday, October 23

7:30 am – 6:30 pm **Registration**
South Ballroom Foyer

6:30 am – 9:00 am **Coffee**
Room 201-204 Foyer

7:00 am – 8:00 am **Concurrent Roundtables**

Age Adjusted Cutoffs in Newborn Screening: The T4 TSH Tango
Room 201

588-857-24 • 1 contact hour for this session

Victoria Floriani, MS, New Jersey Department of Health and Mary Carayannopoulos, PhD, New Jersey Department of Health

Newborn screening (NBS) is meant to detect rare conditions in asymptomatic infants, allowing for early treatment and improved long-term outcomes. While NBS is universally acknowledged as a major public health triumph, screening can cause harm by medicalizing families who receive a false positive result or over-treating children with an ambiguous or mild phenotype. Using congenital hypothyroidism (CH) as the index disorder, this roundtable will provide a forum for state programs to discuss challenges with their current CH screening algorithm. Participants will complete a survey, split into groups, work with other states to outline challenges, and suggest possible solutions to improve screening outcomes for CH, with emphasis on the impact of incorporating age-adjusted cut-offs.

Refusing to Let It Go: A Long Overdue Conversation About Rising Refusal Rates
Room 202

588-858-24 • 1 contact hour for this session

Tory Kaye, MPH, Minnesota Department of Health

In Minnesota (MN), birthing providers have a duty to perform newborn screening (NBS), though state statute allows families to opt-out of screening with written refusal for any reason. The MN newborn screening program utilizes vital record data to identify infants who are unscreened and program staff follow all unscreened infants to determine if a family chose to opt-out of screening. The goal of this roundtable is to reduce the number of families opting out of newborn screening through collaboration with other state programs. Attendees will compare strategies for reducing the rate of refusal across programs and discuss what other states have done to understand why families opt out of screening.

Leveraging Family Experience to Create an Action Plan for Engagement
Room 203

588-859-24 • 1 contact hour for this session

Marianna Raia, MS, CGC, Expecting Health

Family engagement plays a pivotal role in ensuring the success and efficacy of newborn screening (NBS) programs. However, to fully harness its benefits, it is essential to involve families actively in the entire system. This roundtable will explore the strategies needed to enhance family engagement in NBS initiatives. Attendees will hear directly from a panel of family representatives who are participating members of the newly formed APHL Newborn Screening Technical assistance and Evaluation Program (NewSTEPS) workgroups and/or subcommittee. After, attendees will focus on creating an "action plan" for engaging families and will discuss the barriers and challenges that hinder effective family engagement in NBS.

8:00 am – 8:30 am Break

South Ballroom Foyer

8:30 am – 10:00 am Phases of Newborn Screening Long-term Follow-up

South Ballroom

588-860-24 • 1.5 contact hours for this session

Moderators:

- Lisa Shook-Chiles, DHPE, MA, MCHES, Cincinnati Children's Hospital Medical Center
- Jennifer Hauser, MPH, RN, PHN, Minnesota Department of Health

A Model for Long-term Follow-up: Integrating Epic Registries, Dashboards and Workflows
Katherine Raboin, RN, BS, The Connecticut Newborn Screening Network

Comprehensive Long-term Follow-up Program for Newborn Screening in Colorado and Wyoming
Erica Wright, MS, CGC, Children's Hospital Colorado/University of Colorado

Engaging Families in Long-term Follow-up Care: Perspectives from the Comprehensive Long-term Follow-up Program in Colorado and Wyoming
Stacey Quesada, MPH, Center for Public Health Innovation

Five Years of Longitudinal Follow-up of Children with Critical Congenital Heart Disease in Minnesota
Sara Lammert, PhD, MPH, Minnesota Department of Health

Critical Congenital Heart Disease Screening in Michigan: A 10-year Review
Kristen Thompson, MPH, Michigan Department of Health and Human Services

This session will analyze the positive effects of newborn screening (NBS) long-term follow-up (LTFU). Discussions will focus on how integrating systems of care within NBS can help address identified gaps to improve long-term health outcomes for patients, how to communicate

with families to identify and achieve their long-term goals for their child and what tools NBS programs can utilize to ensure continuous quality improvement throughout the LTFU system's lifespan. The session will conclude with two programs each discussing the long-term outcomes of screening and follow-up for critical congenital heart disease (CCHD).

10:00 am – 4:00 pm Exhibit and Poster Hall Open

Exhibit Hall

10:00 am – 10:30 am Break – Visit the exhibits and posters

Exhibit Hall

10:30 am – 12:00 pm Strengthening the Newborn Screening System Through Education and Follow-up

South Ballroom

588-861-24 • 1.5 contact hours for this session

Moderators:

- Kathy Inkhamfong, MS-HSA, Colorado Department of Public Health and Environment
- Susan Mays, BA Sociology, parent of a child with MSUD, NBS Advocate, Expecting Health Ambassador

Next Generation Sequencing for Cystic Fibrosis: The Florida Newborn Screening Program Experience
Emily Reeves, BSN, RN CPN, Florida Department of Health and Kristin Barnette, RN, Florida Department of Health

Universal Newborn Screening for Congenital Cytomegalovirus: What Information Do Families Want After Diagnosis?
Gina Liverseed, APRN, DNP, Minnesota Department of Health

Strengthening the Newborn Screening System: Expansion of Follow-up and Education Initiatives at NewSTEPs
Noah Sinangil, Association of Public Health Laboratories

Evaluating the Biochemical Profiles, Clinical Phenotypes and Genotypes of X-ALD Detected by Newborn Screening and Feasibility of Adjusting Algorithms to Identify Only Clinically Significant X-ALD
Inderneel Sahai, MD, New England Newborn Screening Program

Caregiver Reports of Developmental Services Following a Newborn Screening Diagnosis
Elizabeth Reynolds, PhD, RTI International

This session will focus on ways newborn screening (NBS) programs can strengthen their NBS system to ensure quality screening for all. Presentations will address how next generation sequencing (NGS) can be used to aid in screening for Cystic Fibrosis (CF), the important role that NBS programs play in ensuring up-to-date, relevant information reaches parents after a

diagnosis, how the creation of subcommittees and workgroups has organized partners across the NBS system to successfully identify areas of need for improved workflows, and a look into the New England NBS program's experience with screening for X-linked adrenoleukodystrophy (X-ALD). The session will conclude with a discussion about the importance of early intervention (EI) and how EI and NBS coordinators collaborate to serve children and their families.

12:00 pm – 1:30 pm Lunch – Visit the exhibits and posters

Exhibit Hall

1:30 pm – 3:00 pm Advancements in Molecular Technology and Next Generation Sequencing

South Ballroom

588-862-24 • 1.5 contact hours for this session

Moderators:

- Michele Caggana, ScD, FACMG, New York State Department of Health
- Rachel Lee, PhD, Centers for Disease Control and Prevention

Next Generation Sequencing for Newborn Screening: It's Really Complicated!
John Leavitt, PhD, HCLD(ABB), Texas Department of State Health Services

CDC's Vision to Support the Expansion of Molecular Testing in Newborn Screening
Suzanne Cordovado, PhD, Centers for Disease Control and Prevention

Using Genome Sequencing to Screen Newborns in North Carolina: Successes and Challenges in the First Six Months
Katerina Kucera, PhD, RTI International

Road to Expansion: Michigan's Experience with Next Generation Sequencing on the illumina TruSight™ Cystic Fibrosis 139-Variant Assay
Erika Jackson, BS, AAS, Michigan Department of Health and Human Services

Leveraging Large Population Databases and Machine Learning Models to Scale Newborn Genome Sequencing
Liana Protopsaltis, MS, CGC, Rady Children's Institute for Genomic Medicine

This session will focus on how advancements to screening technologies have improved health outcomes for newborns. Presentations will detail the hurdles and opportunities encountered when implementing next generation sequencing (NGS), a look into how the Centers for Disease Control and Prevention (CDC) is creating tools and resources to support molecular testing, how Early Check, a voluntary NBS research program, has successfully implemented voluntary state-wide supplemental screening with genome sequencing, and how one program's experience with NBS for cystic fibrosis (CF) can help inform screening for other disorders using NGS technology.

3:00 pm – 4:00 pm Break and raffle drawing – Visit exhibits and posters

Exhibit Hall

4:00 pm – 5:30 pm
South Ballroom

Quality Improvement, Control and Assurance Activities

588-863-24 • 1.5 contact hours for this session

Moderators:

- Jill Simonetti, BS, Minnesota Department of Health
- Patrice Held, PhD, Oregon State Public Health Laboratory

You Gotta Have a Plan: System-wide and Systematic New Disorder Implementation
Holly Winslow, BS, Minnesota Department of Health

North Carolina Population Study for N-Acetyl Tyrosine (NAT) Multiplexed into NeoBase2TM Non-derivatized MSMS Kit as an Indicator of Total Parenteral Nutrition (TPN) Administration in Newborns
Jamie Mills, MLS(ASCP), North Carolina State Laboratory of Public Health

Expanding CDC Quality Assurance Materials for Lysosomal Storage Disorders: QC, PT and Linearity Programs
Elya Courtney, MPH, Centers for Disease Control and Prevention

Preparation for a Combined Molecular QC Program for TREC and SMA
Christopher Greene, PhD, Centers for Disease Control and Prevention

Partnerships in Newborn Screening: A Promising Pathway for Emerging Newborn Screening Programs
John Anetor, PhD, FIBMS, FACN, FRSC, FADLM, University of Ibadan, Nigeria

This session will evaluate how newborn screening (NBS) programs can continually improve their systems as more disorders are added to the screening panel. Presentations will describe the structure, organization, processes and activities necessary when preparing to implement screening for new disorders; one program's experience with how best to incorporate n-acetyl tyrosine (NAT) into their NBS system with the goal of reducing the burden on the NBS program, NBS system partners, and families; a look into how the Centers for Disease Control and Prevention (CDC) is expanding their quality assurance materials to include newly added conditions to the Recommended Uniform Screening Panel (RUSP); and how programs can partner with each other to further the goals of NBS everywhere.

5:30 pm – 6:00 pm
South Ballroom Foyer

Break

6:00 pm – 10:00 pm

Off-site Social – Sponsored by Revvity, Inc.

Thursday, October 24

7:30 am – 12:00 pm Registration

South Ballroom Foyer

7:00 am – 8:00 am MLD Newborn Screening Enables Access to Gene Therapy — Documentary Screening and Discussion

South Ballroom

Sequencing Hope follows the intimate and often gut-wrenching journey of an Alabama family grappling with the unimaginable risks and potential rewards of new frontiers in medicine as their child becomes the first patient in the United States to receive a then-experimental gene therapy treatment for metachromatic leukodystrophy (MLD). It gives insight into the medical processes patients and families endure to access this therapy, and contrasts outcomes with newborn screening (NBS) to those without the early detection of NBS.

Sequencing Hope premiered at the 2024 Minneapolis St. Paul International Film Festival as an Official Selection and is directed by Maribeth Romslo and Lindsey Seavert.

After over two decades of work, the FDA approved MLD gene therapy in March 2024, and five months later, MLD Foundation submitted the MLD RUSP Nomination to the HHS ACHDNC, where it was accepted and referred to expert review. Funding for this viewing is provided by MLD Foundation.

This screening will be co-moderated by Dean Suhr, President, and Teryn Suhr RN (retired), Executive Director, MLD Foundation and include an interactive discussion of the collaborative implementation of MLD Newborn Screening.

6:30 am – 9:00 am Coffee

South Ballroom Foyer

8:30 am – 10:00 am Improving Testing Methods and Follow-up in Newborn Screening

South Ballroom

588-864-24 • 1.5 contact hours for this session

Moderators:

- M. Christine Dorley, PhD, Maryland Department of Health
- Alisha Keehn, MPA, Health Resources and Services Administration

Increased Sensitivity for First-tier Homocystinuria Newborn Screening: Overcoming Limitations, New Biomarker Additions and Pilot Screening Results
Austin Pickens, PhD, Centers for Disease Control and Prevention

New Treatments, New Challenges: The Impact of Highly Effective Modulator Therapy on Cystic Fibrosis Newborn Screening Results in New York State
Denise Kay, PhD, New York State Department of Health

Streamlining the Onboarding Process for New Conditions in North Carolina Through Standardized Project Plans, Engaging with External Stakeholders and Early Staff Involvement
Jamie Mills, MPS(ASCP), North Carolina State Laboratory of Public Health

Effective and Efficient Newborn Screening for X-linked Adrenoleukodystrophy: One Extraction with Two Injections
Mei Baker, MD, Wisconsin State Laboratory of Hygiene

Second Tier Testing for a Time Critical Lysosomal Storage Disorder: Psychosine and Time to a Definitive Diagnosis of Krabbe Disease
Patricia Hall, PhD, Mayo Clinic

This session will focus on how newborn screening (NBS) programs can improve their processes in order to better detect and treat newborns diagnosed with a variety of disorders. Discussions will center around how the Centers for Disease Control and Prevention (CDC) is improving screening methods for several homocystinuria (HCU) diseases, a look into how the New York State NBS program is adapting their diagnosis and treatment methods as the number of birthing parents with cystic fibrosis (CF) and on CFTR modulators increases, and how the North Carolina NBS program maintained program agility and readiness while adding new disorders over a 3-year timeline. Attendees will also examine ways to enhance NBS for X-linked adrenoleukodystrophy (X-ALD) performance with the goal of retaining screening effectiveness and improving screening efficiency as well as examine second-tier testing for Krabbe Disease (KD) and the importance of timeliness when referring an infant with early infantile KD.

10:00 am – 10:30 am Break

South Ballroom Foyer

10:30 am – 12:00 pm New Conditions on State and National Screening Panels

South Ballroom

588-865-24 • 1.5 contact hours for this session

Moderators:

- Amy R.U.L Calhoun, MD, Stead Family Department of Pediatrics, University of Iowa Health Care
- Alison Breitbarth, Grant's Giants Pompe Nonprofit

One Year of Newborn Screening Follow-up for Congenital Cytomegalovirus in Minnesota and Lessons Learned
Sondra Rosendahl, MS, LCGC, Minnesota Department of Health

A Pilot Study in New York State to Screen Newborns for Congenital Cytomegalovirus
Norma Tavakoli, PhD, New York State Department of Health

Expanding LSD Screening to Include Additional RUSP Conditions for New England States Using an Efficient Multiplex LC-MS/MS Assay
Devinder Kaur, PhD, New England Newborn Screening Program

From Matrix to Reference Ranges, Innovation in the Diagnostic Field of Lysosomal Storage Disorders
Amber Van Baelen, PhD, MD, Antwerp University Hospital

Newborn Screening for Duchenne Muscular Dystrophy in Ohio
Rashmi Gaur, PhD, Ohio Department of Health Laboratory and
Sharon Linard, MS, MEd, CGC, Ohio Department of Health Laboratory

Our closing session will focus on how four newborn screening (NBS) programs have adapted to account for new conditions added to their NBS panels. The Minnesota NBS program will discuss follow-up processes and outcomes for universal congenital cytomegalovirus (cCMV) screening, the New York State NBS program will highlight the first pilot study to include an opt-out method for screening newborns for cCMV, the New England NBS program will review their plans for expanding lysosomal storyage disorder (LSD) screening, the Antwerp University Hospital will discuss how a newly accredited simultaneous detection method for LSDs enables treatment in an earlier stage of disease, and finally the Ohio NBS program will highlight the successes, lessons learned and pain points of being the first program in the nation to provide population-based NBS for Duchenne Muscular Dystrophy (DMD).

12:00 pm Closing and Symposium Adjourns

South Ballroom

Speaker:

- Emily Eisenstein, MPH, Rhode Island Department of Health

Exhibitors and Sponsors

Thank you to our exhibitors and sponsors for their support of the symposium! Be sure to visit them in the exhibit hall. Details and contact information can be found on pages 39–53.

Gold Circle

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Expecting Health/Baby's First Test

GelbChem LLC

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ImmunoIVD AB

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LabVantage Solutions, Inc.

Exhibit Hall Schedule

October 20–23, 2024

Sunday, October 20

5:30 pm – 7:30 pm	Hall Open
6:00 pm – 7:30 pm	Welcome Reception

Monday, October 21

10:00 am – 4:00 pm	Hall Open
10:00 am – 10:30 am	Break
1:15 pm – 2:00 pm	Dessert
3:30 pm – 4:00 pm	Break

Tuesday, October 22

10:00 am – 10:30 am	Break
12:30 pm – 1:30 pm	Box Lunch
3:00 pm – 3:30 pm	Break

Wednesday, October 23

10:30 am – 11:00 am	Break
12:00 pm – 1:30 pm	Box Lunch
3:00 pm – 4:00 pm	Break/Raffle Drawing

Mayo Clinic Laboratories

Natus Medical Incorporated

OpenELIS Foundation

Quantabio

Ruvos

SNP Biotechnology R&D Ltd.

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Exhibitors by Booth Number

100 Cambridge Isotope Laboratories

101 OpenELIS Foundation

102 Expecting Health/Baby's First Test

103 Novartis Pharmaceuticals

104 LabVantage Solutions, Inc.

105 Ruvos

109 Natus Medical Incorporated

110 DiaSorin Molecular/Luminex, a DiaSorin Company

111 Thermo Fisher Scientific

112 Ultragenyx

114 ImmunoIVD AB

200 STAT Courier Service Inc.

201 Centers for Disease Control and Prevention

202/204 Sebia

203 EBF, Inc.

205 EveryLife Foundation for Rare Diseases

206 GelbChem LLC

207 Bio-Rad Laboratories

208/210 Waters Corporation

209/211 Revvity, Inc.

300 Astoria-Pacific

301 BSD Robotics

302 APHL Career Pathways

303 SNP Biotechnology R&D Ltd.

304 Labsystems Diagnostics Oy

305 Agilent Technologies

307 Asuragen, a Bio-Techne brand

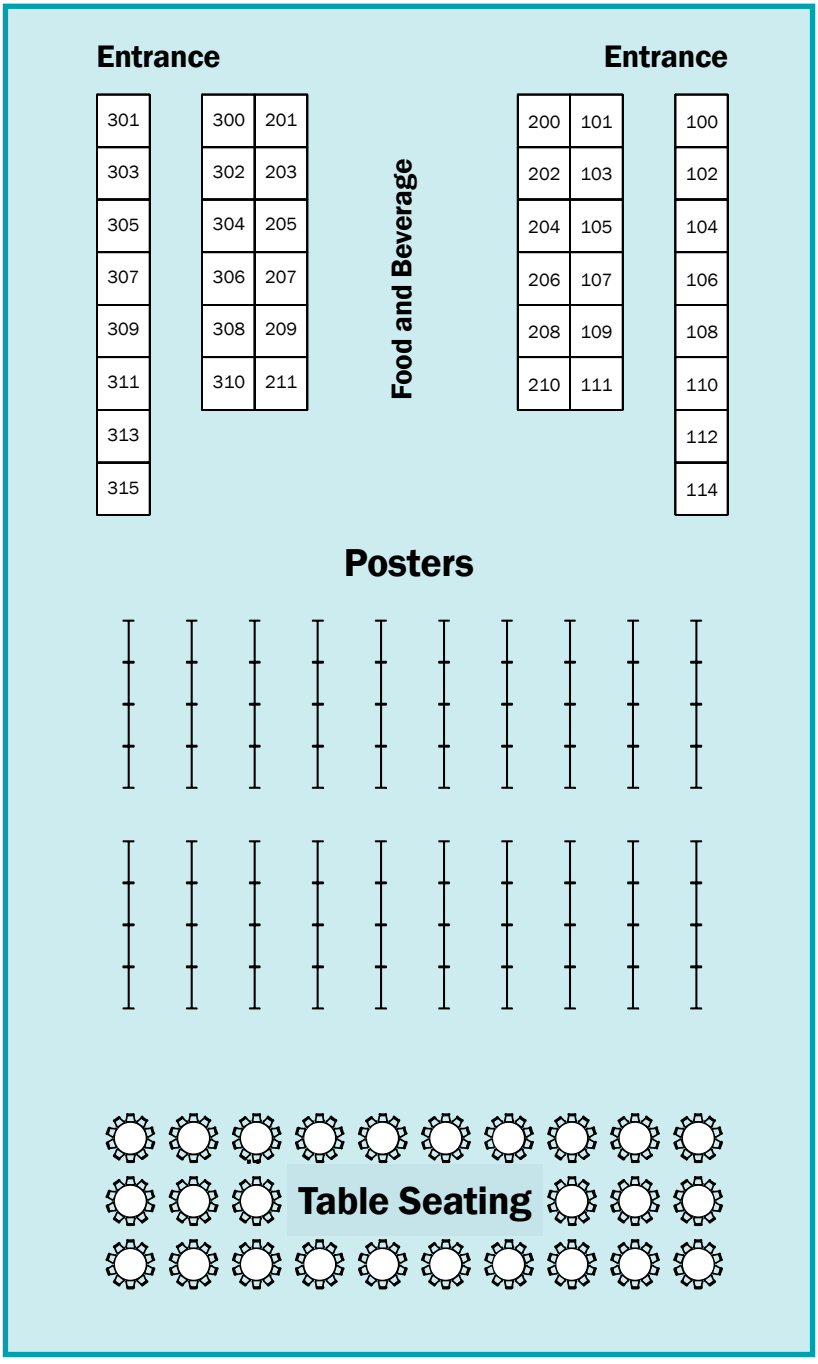
308 Quantabio

310 HCU Network America

311 Mayo Clinic Laboratories

313/315 Baebies

Exhibit Hall Floor Plan



Visit the Exhibitors! Win Prizes!

Exhibit Hall Connection Contest

Visit with the vendors and win prizes! Take the exhibit raffle card from your tote bag and visit each exhibit booth. Ask one of the booth personnel to sign their initials in the box corresponding to their booth number. When ALL of the boxes are filled, turn the card in at the registration desk. Cards must be turned in at the registration desk no later than 2:00 pm on Wednesday, October 23. Be sure to include your name on the card. Winners of the prizes will be announced starting at 3:00 pm on Wednesday in the exhibit hall. You must be present to win.

Prizes

- Two $500 ticket vouchers on Delta Airlines
- $100 gift card (compliments of Astoria-Pacific)
- Complimentary registration to the 2025 Newborn Screening Symposium
- 2 CIL vests (compliments of Cambridge Isotope Laboratories)
- One $250 ticket voucher on Delta Airlines
- Apple Watch (compliments of EBF, Inc.)
- $150 Amazon gift card (compliments of HCU Network America)
- $150 Amazon gift card (compliments of Labsystems Diagnostics Oy)
- $200 gift card (compliments of Natus Medical Incorporated)
- $200 Amazon gift card (compliments of STAT Courier Services, Inc.)
- $150 gift card (compliments of Waters Corporation)

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APHL's Career Pathways are represented by four programs: Internships, Fellowships, Leadership and Academic Partnerships. These APHL and CDC initiatives support the development of the future public health laboratory workforce from early college careers all the way through to programs that support laboratory professionals becoming laboratory leaders. Learn more about opportunities to develop your leadership skills, connect with community partners to recruit the future workforce and how you can inspire interns or fellows through mentorship.

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510.741.1000 • **www.bio-rad.com**

Bio-Rad Laboratories develops, manufactures, sells, and supports a large portfolio of products for newborn screening and diagnostics. Bio-Rad has been a global leader in thalassemia and sickle cell screening for over 30 years, providing "Gold Standard" automated solutions for newborn programs worldwide. We also offer a suite of market-leading qPCR and digital PCR technologies to support molecular testing.

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11 Aldinga St., Brendale, QLD 4500, Australia
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Booth 201

1600 Clifton Rd., Atlanta, GA 30329
770.488.7571 • **www.cdc.gov/newbornscreening/**

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11331 Valley View St., Cypress, CA 90630
562.240.6500 • **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, Inc.

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Booth 205

1012 14th St., Suite 500, Washington, DC 20001
202.697.7273 • **www.everylifefoundation.com**

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 102

26400 Woodfield Rd., #189, Damascus, MD 20872
www.expectinghealth.org

Expecting Health provides families with the most current tools and information - from planning a pregnancy and early infant care to empowering family leadership and engagement. Baby's First Test, an Expecting Health program, provides newborn screening information and resources to more than 800,000 people annually. Our work in newborn screening is to convene stakeholders and experts, connect people to relevant and action-oriented information, and center the experiences of families as we build towards a more inclusive and responsive newborn screening system.

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Booth 206

4000 Mason Rd., Suite 300, Seattle, WA 98195
206.717.3515 • **www.gelbchem.com**

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HCU Network America

Booth 310

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630.546.6452 • **www.hcunetworkamerica.org**

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- Support research that improves diagnosis and treatment including a cure for the disease
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NOTE: SPOT-it™ is currently not available for sales in the USA.

Labsystems Diagnostics Oy

Booth 304

Tiilitie 3, Vantaa 01720, Finland
358.2015.57530 • **www.labsystemsdx.com**

Labsystems Diagnostics Oy, Finland has been developing, manufacturing, and distributing innovative products for newborn screening for over 3 decades. Labsystems cover the entire range of methods for newborn screening and offer complete solutions including automation and software options. Labsystems distribute products to over 70 countries worldwide and all processes and products are CE IVD certified. Labsystems' goal for the future is to develop new innovative products for the detection of inborn errors of metabolism.

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Booth 104

Platinum Member

265 Davidson Ave., Suite 220, Somerset, NJ 08873
843.489.2220 • **www.labvantage.com**

LabVantage offers a true Next Generation Public Health Newborn Screening LIMS. In a single LIMS instance, LVS LIMS can serve State Public Health lab departments and Newborn Screening without compromising functionality. The platform includes an embedded electronic lab notebook, lab execution system to assure compliance in a sample life cycle, scientific data management system, ETOR portal, and comprehensive business intelligence & predictive analytics. LVS provides a unique graphic designer to precisely accommodate your workflows.

Mayo Clinic Laboratories

Booth 311

3050 Superior Drive NW, Rochester, MN 55905
507-273-4253 • **www.mayocliniclabs.com**

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3150 Pleasant View Rd., Middleton, WI 53562
608.829.8500 • **www.natus.com**

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Booth 103

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573.301.8222 • **www.openelis.org**

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978.869.1505 • **www.quantabio.com**

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Posters

*Rapid poster presentations

P-1

A Preliminary Exploratory Comparison of the Consent and Decline Rates Between 10 Languages for ScreenPlus: a Pilot Newborn Screening Study

N. Kelly[1], M. Wasserstein[2], K. Paleologos[2], M. Clarke[2], [1]Children's Hospital at Montefiore, [2]Albert Einstetin College of Medicine

Presenter: *Megan Clarke, MS, Albert Einstein College of Medicine, megan.clarke@einsteinmed.edu*

P-2

ScreenPlus: Why do parents decline participation for expanded newborn screening?

N. Kelly[1], K. Paleologos[2], M. Clarke[2], M. Wasserstein[2] [1]Children's Hospital at Montefiore, [2]Albert Einstein College of Medicine

Presenter: *Nicole Kelly, MPH, Albert Einstein College of Medicine, nikell@montefiore.org*

P-3

Enhancing Newborn Screening Education: A Comparative Analysis of Sample Quality Before and After Educational Intervention in Arizona Hospitals

K. Fullerton[1], M. Contursi[1], C. Clabaugh[1], F. Altmaier[1], T. Huynh[1], [1]Arizona Department of Health Services

Presenter: *Trung Huynh, PhD, MB(ASCP), Arizona Department of Health Services, trung.huynh@azdhs.gov*

P-4

Arizona's Approach to X-ALD and GAMT Screening

O. Bilante[1], A. Hauser[1], S. Hylsope[1], C. Clabaugh[1], M. Contursi[1], C. Hill[2], T. Huynh[1], [1]Arizona Department of Health Services, [2]Revvity

Presenter: *Trung Huynh, PhD, MB(ASCP), Arizona Department of Health Services, trung.huynh@azdhs.gov*

P-5

Program-Specific Aims, Collective Impact: NewSTEPs Quality Improvement Projects Collaborative Success

A. Comer[1], S. McKasson[1], E. Jones[1], S. Singh[1], [1]Association of Public Health Laboratories

Presenter: *Ashley Comer, Association of Public Health Laboratories, ashley.comer@aphl.org*

P-6

NCAA sickle cell trait result requests in the California Newborn Screening Program

S. Carter[1], J. Matteson[1], H. Haran[1], T. Bishop[1], S. Asfaha[1], G. Nash[1], C. Wu[1], S. Sciortino[1], [1]California Department of Public Health

Presenter: *Sarah Carter, PhD, California Department of Public Health, Sarah.Carter@cdph.ca.gov*

P-7

An evolving treatment landscape for Spinal Muscular Atrophy (SMA), 2020-2023

J. Matteson[1], T. Bishop[1], G. Nash[1], C. Wu[1], L. Shih[1], L. Wu[1], L. Tom[1], S. Diaz[1], S. Sharma[1], S. Sciortino[1], [1]California Department of Public Health

Presenter: *Sarah Carter, PhD, California Department of Public Health, Sarah.Carter@cdph.ca.gov*

P-8

Short term stability study of guanidinoacetate, creatine and creatinine in dried blood spot samples for use with the Guanidinoacetate Methyltransferase enzyme deficiency assay

K. Dhillon[1], J. Aduviso[1], J. Zhou[1], P. Roworth[1], B. Espinosa[1], P. Neogi[1], R. Sharma[1], [1]California Department of Public Health

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

P-9

Short term stability study of acid α-glucosidase, α-iduronidase and iduronate-2-sulfatase in dried blood spot samples for use with the Pompe, Mucopolysaccharidosis type I and II disorders

K. Dhillon[1], J. Aduviso[1], J. Zhou[1], K. Ramanathan[1], J. Vongsa[1], P. Roworth[1], B. Espinosa[1], P. Neogi[1], R. Sharma[1], [1]California Department of Public Health

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

P-10

Short term stability study of Amino Acids, Acylcarnitine, and Lysophosphatidylcholines in dried blood spot samples for use with the mass spectrometry assay for newborn metabolic disorders

J. Aduviso[1], K. Dhillon[1], J. Zhou[1], K. Ramanathan[1], P. Roworth[1], B. Espinosa[1], P. Neogi[1], R. Sharma[1], [1]California Department of Public Health

Presenter: *Rajesh Sharma, PhD, FACMG, California Department of Public Health, rajesh.sharma@cdph.ca.gov*

P-11

Short-term Stability Study of Immunoreactive Trypsinogen in Dried Blood Spot Samples

J. Aduviso[1], K. Dhillon[1], R. Kaur[1], M. Jain[1], S. Singh[1], J. Zhou[1], P. Rich[1], R. Sharma[1], [1]California Department of Public Health

Presenter: *Rajesh Sharma, PhD, FACMG, California Department of Public Health, rajesh.sharma@cdph.ca.gov*

P-12

Short-term stability study of Hemoglobinopathies in dried blood spot samples for use with the high-performance liquid chromatography system assay

J. Aduviso[1], R. Kaur[1], M. Jain[1], S. Singh[1], C. Dominguez[1], J. Zhou[1], Y. Hou[1], P. Rich[1], K. Dhillon[1], R. Sharma[1], [1]California Department of Public Health

Presenter: *Rajesh Sharma, PhD, FACMG, California Department of Public Health, rajesh.sharma@cdph.ca.gov*

P-13

Short-term Stability Study of Galactose-1-Phophate Uridyltransferase and Biotinidase Activity in Dried Blood Spot Samples

J. Aduviso[1], K. Dhillon[1], R. Kaur[1], M. Jain[1], S. Singh[1], J. Zhou[1], P. Rich[1], R. Sharma[1], [1]California Department of Public Health

Presenter: *Rajesh Sharma, PhD, FACMG, California Department of Public Health, rajesh.sharma@cdph.ca.gov*

P-14

Impact of storage temperature and duration on the stability of Dried Blood Spot (DBS) cards for Spinal Muscular Atrophy (SMA) screening

S. Sharma[1], J. Aduviso[1], L. Wu[1], K. Dhillon[1], L. Shih[1], J. Zhou[1], L. Tom[1], P. Roworth[1], S. Diaz[1], R. Sharma[1], [1]California Department of Public Health

Presenter: *Rajesh Sharma, PhD, FACMG, California Department of Public Health, rajesh.sharma@cdph.ca.gov*

P-15

Looking Back at 4 Years of Spinal Muscular Atrophy (SMA) Screening in California

S. Sharma[1], L. Shih[1], L. Wu[1], L. Tom[1], S. Diaz[1], R. Sharma[1], [1]California Department of Public Health

Presenter: *Rajesh Sharma, PhD, FACMG, California Department of Public Health, rajesh.sharma@cdph.ca.gov*

P-16

Development of Psychosine Quality Assurance Dried Blood Spot Materials for Krabbe Screening

S. Isenberg[1], S. Sah[1], E. Courtney[1], T. Lim[1], C. Pickens[1], R. Lee[1], C. Cuthbert[1], K. Petritis[1], [1]Centers for Disease Control and Prevention

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

P-17

Customization of a Liquid Handling System to Produce Dried Blood Spots for Quality Assurance

C. Brown[1], T. Trosclair[2], J. Bernstein[1], A. Johnson[3], I. Barham[1], J. Mei[1], [1]Centers for Disease Control and Prevention, [2]Neptune and Company, Inc., [3]Cherokee Nation Operation Solutions

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

P-18

Short Term and Freeze/Thaw stability studies of Recombinant Human TSH in dried blood spots

E. Gonzalez Reyes[1], C. Brown[1], L. Li[1], T. Trosclair[2], G. Peña[1], S. Simmons[3], J. Mei[1], [1]Centers for Disease Control and Prevention, [2]Neptune and Company, Inc., [3]Cherokee Nation Operation Solutions

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

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Withdrawn

P-20

Short Term Stability Study of Cystic Fibrosis Patient and Family Dried Blood Spot Quality Assurance Samples for CFTR Pathogenic Variant Testing

M. Hendrix[1], K. Duneman[1], A. Colvin[1], S. Nikolova[1], R. Lee[1], C. Cuthbert[1], S. Cordovado[1], [1]Centers for Disease Control and Prevention

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

P-21

Developing and Evaluating Dried Blood Spot Materials to Support Congenital Cytomegalovirus Screening in Newborns

J. Kim[1], D. Cobb[1], F. Lee[1], R. Hage[1], S. Cordovado[1], R. Lee[1], C. Cuthbert[1], [1]Centers for Disease Control and Prevention

Presenter: *Grace Kim, MS, Centers for Disease Control and Prevention, ueb5@cdc.gov*

P-22

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

I. Onyechi[1], A. Cervalli[1], A. Moseley[1], C. Greene[1], F. Lee[1], S. Cordovado[1], R. Lee[1], C. Cuthbert[1], [1]Centers for Disease Control and Prevention

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

P-23

Effects of Hydrazine on Newborn Screening Analytes and Alternatives for Succinylacetone Detection in Dried Blood Spots

D. Peppers[1], C. Pickens[1], R. Lee[1], C. Cuthbert[1], K. Petritis[1], [1]Centers for Disease Control and Prevention

Presenter: *Daquille Peppers, MS, Centers for Disease Control and Prevention, syy7@cdc.gov*

*P-24

Comparison of Method Performance in Hemoglobinopathy Phenotype Identification

S. Zobel[1], G. Peña[1], J. Mei[1], J. Bernstein[1], [1]Centers for Disease Control and Prevention

Presenter: *Sherri Zobel, MT, ASCP, Centers for Disease Control and Prevention, szobel@cdc.gov*

P-25

Analysis of Laboratory Performance Using Summary Data from CDC's Newborn Screening Quality Assurance Program: Opportunities for Quality Improvement

E. Gonzalez Reyes[1], S. Zobel[1], K. Stewart[1], J. Bernstein[1], R. Lee[1], J. Mei[1], [1]Centers for Disease Control and Prevention

Presenter: *Sherri Zobel, MT, ASCP, Centers for Disease Control and Prevention, zyu1@cdc.gov*

P-26

Enhancing Data-driven Disease Detection in Newborns (ED3N): Designing a model to Improve Newborn Screening Risk Assessment

C. Cuthbert[1], R. Lee[1], A. Gaviglio[2], [1]Centers for Disease Control and Prevention, [2]Connetics Consulting, LLC

Presenter: *Rachel Lee, PhD, Centers for Disease Control and Prevention, nmo1@cdc.gov*

P-27

Analyte Expansion of Nine-Level Linearity Quality Assurance Dried Blood Spot Materials for Newborn Screening

T. Lim[1], S. Isenberg[1], D. Peppers[1], C. Pickens[1], E. Courtney[1], S. Sah[1], R. Lee[1], C. Cuthbert[1], K. Petritis[1], [1]Centers for Disease Control and Prevention

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

P-28

Using CRISPR/Cas9 genome editing for custom production of quality assurance materials for newborn screening molecular assays

D. Cobb[1], J. Kim[1], F. Lee[1], R. Lee[1], R. Hage[1], S. Cordovado[1], [1]Centers for Disease Control and Prevention

Presenter: *David Cobb, PhD, Centers for Disease Control and Prevention, ugh1@cdc.gov*

P-29

Development and validation for second-tier liquid chromatography tandem-mass spectrometry analysis of psychosine

S. Sah[1], C. Pickens[1], S. Isenberg[1], E. Courtney[1], T. Lim[1], R. Lee[1], C. Cuthbert[1], K. Petritis[1], [1]Centers for Disease Control and Prevention

Presenter: *Samyukta Sah, PhD, Centers for Disease Control and Prevention, jyw9@cdc.gov*

P-30

Higher Tier Testing: The Colorado Approach

E. Wright[1], M. Sands[2], P. Baker[1], C. Porter[3], A. Watson-Siriboe[3], G. Bonn[3], [1]Clinical Genetics and Metabolism, Department of Pediatrics, University of Colorado Anschutz Medical Campus, [2]Genetic Counseling Graduate Program, University of Colorado, [3]Colorado Department of Public Health and Environment

Presenter: *Erica Wright, MS, CGC, Clinical Genetics and Metabolism, Department of Pediatrics, University of Colorado Anschutz Medical Campus, erica.wright@childrenscolorado.org*

P-31

A Quality Improvement Project to Test the Clinical Utility of Integrated Genetic Testing for Alpha Thalassemia in Newborn Screening

L. Shook-Chiles[1], D. Haygood[1], C. Quinn[1], [1]Cincinnati Children's Hospital Medical Center

Presenter: *Lisa Shook-Chiles, DHPE, MA, MCHES, Cincinnati Children's Hospital Medical Center, lisa.shook@cchmc.org*

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A Design-Thinking Model for Co-Developing Educational Materials about Sickle Cell Trait

L. Shook-Chiles[1], D. Haygood[1], C. Quinn[1], [1]Cincinnati Children's Hospital Medical Center

Presenter: *Lisa Shook-Chiles, DHPE, MA, MCHES, Cincinnati Children's Hospital Medical Center, lisa.shook@cchmc.org*

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Evaluating methods for process improvements to allow for further growth in Colorado Newborn Screening

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From heel prick to recommendation statements: CF Foundation Newborn Screening Consensus Recommendations

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Presenters: *Marissa Taylor, MPH, Cystic Fibrosis Foundation, mataylor@cff.org and Marci Sontag, PhD, Center for Public Health Innovation, marci.sontag@cphinnovation.org*

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digitalMLPA: A new, multiplexable, DNA based method for Screening Newborn Conditions

T. Fabella[1], E. Sistermans[1], L. Henneman[2], G. Pals[1], J. den Hoed[3], C. Padilla[4], E. Cutiongco-de la Paz[5], J. Schouten[3], [1]Department of Human Genetics, Amsterdam UMC, Vrije Universiteit Amsterdam, [2]Amsterdam Reproduction and Development Research Institute, [3]MRC Holland, [4]Newborn Screening Reference Center, [5]Institute of Human Genetics, National Institutes of Health, University of the Philippines

Presenter: *Terence Diane Fabella, MS, PhD candidate, Department of Human Genetics, Amsterdam UMC, Vrije Universiteit Amsterdam, t.d.fabella@amsterdamumc.nl*

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digitalMLPA for confirmatory testing of Congenital Adrenal Hyperplasia (CAH), Glucose 6 Phosphate Dehydrogenase (G6PD) Deficiency and Hemoglobinopathies in newborn screening

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Presenter: *Terence Diane Fabella, MS, PhD candidate, Department of Human Genetics, Amsterdam UMC, Vrije Universiteit Amsterdam, t.d.fabella@amsterdamumc.nl*

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Case Study: A positive screen for Pompe Disease leads to early diagnosis of Duchenne Muscular Dystrophy

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

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Mother to child transmission rate of HBV and its associated risk factors

G. Gutema[1], C. Bashea[1], D. Bikila[1], B. Bejiga[1], D. Leta[1], [1]Ethiopian Public Health Institute

Presenter: *Gadissa Gutema Jebessa, PhD candidate, Ethiopian Public Health Institute, gadissagutema@gmail.com*

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Pretreatment HIV drug resistance among treatment-naive infants newly diagnosed with HIV in Ethiopia: Multicenter Cross-Sectional Study

C. Hailu[1], D. Bakila[1], T. Lejisa[1], G. Gutema[1], [1]Ethiopian Public Health Institute

Presenter: *Chala Bashea Hailu, MS, Ethiopian Public Health Institute, chala.basha@gmail.com*

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Biotinidase Deficiency: Prevalence and Correlation between Enzyme Activity and Molecular Findings in a Population of Newborns in Mexico

H. Cruz-Camino[1], C. Cantú-Reyna[1], H. Guerrero-Márquez[1], R. Gómez-Gutiérrez[1], [1]Genomi-K

Presenter: *Hector Cruz-Camino, MSc, Genomi-K, hcruz@genomi-k.com*

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AI cloud-based end-to-end technology for accurate, fast & affordable Newborn Screening of genetic disorders / rare diseases towards precision treatments

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Presenter: *Chris Kyriakidis, PhD, MSc, MBA, gMendel, chris@g-mendel.com*

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N-acetyl tyrosine: Semiquantitative marker for Total Parenteral Nutrition in newborn screening by tandem mass spectrometry

I. Khneisser[1], D. Platis[2], [1]HVD Life Sciences Vertriebs-GmbH, [2]Institure of Child Health

Presenter: *Issam Khneisser, MS, MBA, HVD Life Sciences Vertriebs-GmbH, issam.khneisser@usj.edu.lb*

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Newborn Screening for Fabry Disease in Illinois and Tennessee — Two States' Experiences

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Presenter: *George Dizikes, PhD, HCLD/CC(ABB), Illinois Department of Public Health, george.dizikes@illinois.gov*

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Towards an accessible, high-quality and innovative newborn screening solution for SCID, XLA, SMA, and SCD

D. Silva[1], E. Svedin[1], M. Cornago[1], M. Baroncelli[1], E. Rzeszutek[1], F. Leijon[1], [1]ImmunoIVD AB

Presenter: *Diogo Silva, PhD Biochemistry, ImmunoIVD AB, diogo.silva@immunoivd.com*

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Performance Evaluation of NeoMAP® 4plex AT Kit for Simultaneous Detection of Total Antibodies against Syphilis, Chagas, HTLV ½, and Hepatitis C.

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Presenter: *Rainara Almeida, MBA, Intercientifica, rainara@intercientifica.com.br*

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Thematic Analysis of Questions from Parents during Family Contact Calls

G. Younger[1], J. Ginter[1], J. Zamzow[1], [1]Iowa Newborn Screening Program

Presenter: *Georgianne Younger, MS, LGC, Iowa Newborn Screening Program, georgianne-younger@uiowa.edu*

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Retrospective Review of 2023 Iowa Newborn Screening Phone Genetic Counseling Service for Hemoglobinopathy Traits and Diseases

J. Zamzow[1], E. Phillips[2], J. Ginter[1], G. Younger[1], [1]Iowa Newborn Screening Program, [2]University of Iowa

Presenter: *Jaclyn Zamzow, MS, LGC, Iowa Newborn Screening Program, jaclyn-zamzow@uiowa.edu*

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Engagement for data exchange/clinical ETOR in SC DHEC

M. Yost-Daljev[1], E. Jordanov[1], [1]J Michael Consulting

Presenter: *Mary Kate Yost-Daljev, PhD, J Michael Consulting, myostdaljev@jmichael-consulting.com*

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Implementation of clinical ETOR in SC DHEC

M. Yost-Daljev[1], E. Jordanov[1], [1]J Michael Consulting

Presenter: *Mary Kate Yost-Daljev, PhD, J Michael Consulting, myostdaljev@jmichael-consulting.com*

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Charting a Path: Advancing from a LIMS Evaluation to an RFP and LIMS Replacement

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Presenter: *Mary Kate Yost-Daljev, PhD, J Michael Consulting, myostdaljev@jmichael-consulting.com*

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Identification of Gaps in NBS Laboratory Informatics Management

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Presenter: *Mary Kate Yost-Daljev, PhD, J Michael Consulting, myostdaljev@jmichael-consulting.com*

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Obtaining Sickle Cell Status in the State of Kansas

M. Mills[1], [1]Kansas Health and Environmental Laboratories

Presenter: *Michelle Mills, MS, Kansas Health and Environmental Laboratories, Michelle.J.Mills@ks.gov*

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Implementation of MPS II Screening to the Kansas Newborn Screening Program

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Presenter: *Hannah Radke, Kansas Health and Environmental Laboratories, hannah.m.radke@ks.gov*

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Development of a flexible 56-plex Second Tier LC-MS/MS method in Newborn Screening Program

M. Cassonello[1], M. Maghnie[1], L. Alberti[2], C. Cereda[2], [1]Laboratory for the Study of Inborn Errors of Metabolism and Newborn Screening UNIGE – DINOGMI IRCCS G. Gaslini Children Hospital, [2]Regional Newborn Screening Laboratory, Children's Hospital V. Buzzi

Presenter: *Michela Perrone Donnorso, PhD, Laboratory for the Study of Inborn Errors of Metabolism and Newborn Screening UNIGE – DINOGMI IRCCS G. Gaslini Children Hospital, michelaperronedonnorso@gaslini.org*

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Eliminating Biases and Improvement in Education Advances Health Equity

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Two-tier Hemoglobinopathy Screening and Reporting of Hemoglobin Traits: 10 Years Experience from the Maritimes, Canada

C. Filiaggi[1], A. Rideout[2], Z. Liu[1], [1]Maritime Newborn Screening, IWK Health, [2]Maritime Medical Genetics Service, IWK Health

Presenter: *Corey Filiaggi, CGC, CCGC, Maritime Newborn Screening, IWK Health, corey.filiaggi@iwk.nshealth.ca*

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Genetic Providers' Views on Trauma-Informed Care in Genetics Clinics

J. Scott Schwoerer[1], A. Gaviglio[2], K. Sala-Hamrick[3], L. Stone[3], K. Justice[3], M. Edick[3],[1]Medical College of Wisconsin, [2]Connetics Consulting, LLC, [3]Michigan Public Health Institute

Presenter: *Jessica Scott Schwoerer, MD, Medical College of Wisconsin, jscottschwoerer@mcw.edu*

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Improving communication with a positive newborn screen

J. Scott Schwoerer[1], E. Singh[1], G. Heckel[1], [1]Medical College of Wisconsin

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Redesigning the Newborn Screening Results Report

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Withdrawn

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Going Back to the Basics: Michigan's Experience Screening for Classic Galactosemia

S. Moloney[1], [1]Michigan Department of Health and Human Services

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Congenital CMV Universal Newborn Screening and Statewide Public Health Surveillance: A Confusing Venn Diagram

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Presenter: *Lexie Barber, MPH, Minnesota Department of Health, lexie.barber@state.mn.us*

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Utilizing an Extended Incubation Time for MPS I, Pompe, and Krabbe with Revvity's NeoLSD™ MSMS Kit

C. Lord[1], A. Hietala[1], J. Orsini[2], L. Sorenson[2], [1]Minnesota Department of Health, [2]New York State Department of Health

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Sorry for the delay. A new option for completing newborn screening

K. Tricas[1], T. Kaye[1], A. Dahle[1], M. Johnson[1], M. Gourneau[1], [1]Minnesota Department of Health

Presenter: *Sondra Rosendahl, MS, LCGC, Minnesota Department of Health, sondra.rosendahl@state.mn.us*

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digitalMLPA as a DNA-based method to detect copy number variants in genes associated with 20 screened newborn conditions in a single assay

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Presenter: *Joery den Hoed, PhD, MSc, MRC Holland, j.denhoed@mrcholland.com*

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Bioinformatics and Data Science: Data Cleaning and Coordination for Improving the Newborn Screening Data Infrastructure in Nebraska

T. Mills[1], [1]Nebraska Newborn Screening

Presenter: *Tyra Mills, MPH, Nebraska Newborn Screening, Tyra.mills@nebraska.gov*

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Molecular Prognosis for Spinal Muscular Atrophy using Multiplex Ligation Dependent-Probe Amplification Assay for SMN2 Copy Number

K. Jolanta[1], S. Barton[1], J. Thompson[1], J. Hale[1], A. Counihan[1], B. Kumar[1], A. Comeau[1], [1]New England Newborn Screening, UMASS Medical School

Presenter: *Binod Kumar, PhD, New England Newborn Screening, UMASS Medical School, binod.kumar@umassmed.edu*

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Lot-to-lot comparison of GSP® Neonatal hTSH kit reveals a significant negative bias attributed to a manufacturer change in reagent formulation

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Presenter: *Laxmi Nayak, MS, New Jersey Department of Health, Laxmi.Nayak@doh.nj.gov*

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Continuity of Operations Plan: The Mass Spectrometry Perspective

P. Patel[1], V. Floriani[1], M. Carayannopoulos[1], M. Schachter[1], [1]New Jersey Department of Health

Presenter: *Pallavi Patel, New Jersey Department of Health, pallavi.patel@doh.nj.gov*

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A Framework for Continuous Quality Improvement for Hemoglobinopathy Specialty Care Centers in New York State

R. Wilson[1], S. Gagnon[1], M. Erickson[1], M. Mortimer[1], M. Caggana[1], K. Chou[1], [1]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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Electronic vs. Manual Demographic Data Entry by Birth Hospitals: Which method produces fewer missing data for newborn screening?

N. Guilfoyle[1], R. Wilson[1], M. Caggana[1], K. Chou[1], [1]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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Assessing Eligibility of CFTR Modulator Therapies in Infants with Cystic Fibrosis Identified Through Newborn Screening in New York State

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

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Are We Speaking the Same Language? Improving Readability of and Access to Educational Parent Brochures about Sickle Cell Trait

S. Gagnon[1], R. Wilson[1], K. Chou[1], M. Erickson[1], V. Sack[1], M. Caggana[1], [1]New York State Department of Health

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Withdrawn

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A Screening Challenge Unique to CMV: Specimens Collected at >21 Days Old

V. Sack[1], S. Bradley[1], L. Steen[1], N. Tavakoli[1], M. Caggana[1], E. Laitinen,[1] J. Patel[1], [1]New York State Department of Health

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Withdrawn

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Modification and Comparison of Two Revvity qPCR Assays for the Detection of Congenital Cytomegalovirus in Newborns

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

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Biochemical and Molecular Profile of Confirmed Hyperphenylalaninemia Screened at NSC Mindanao

C. Abarquez[1], [1]Newborn Screening Center Mindanao

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Comparison of Three Commercially Available Newborn Screening Kits for Amino Acids, Acylcarnitines, Succinylacetone and Lyso-phosphatidylcholine Quantitation in Dried Blood Spots

N. McIntosh[1], B. Wong[1], M. Henderson[1], [1]Newborn Screening Ontario

Presenter: *Nathan McIntosh, MLT, BSc, Newborn Screening Ontario, nmcintosh@cheo.on.ca*

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Implementing an interface to connect infant hearing screening program to newborn screening in Ontario

B. Milne[1], C. O'Sullivan[1], L. Gallagher[1], M. Kowalski[1], J. Milburn[1], [1]Newborn Screening Ontario

Presenter: *Bailey Milne, MPH, PhD, Newborn Screening Ontario, bmilne@cheo.on.ca*

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Genetic Testing as a Confirmatory Test for Fatty Acid Oxidation Defects (FAODs) in the Philippine Expanded Newborn Screening (ENBS) Program

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Presenter: *Michelle Abadingo, MD, FPPS, Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila, meabadingo@up.edu.ph*

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The Philippine Newborn Screening Portal: Making Data More Accessible to Local Planners, Stakeholders and Decision Makers

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Presenter: *Ma Elouisa Reyes, Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila, mlreyes9@up.edu.ph*

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Evaluation of Capillary Electrophoresis for Newborn Hemoglobinopathy Screening in the Philippines

M. Canlas[1], M. Escoreal[1], C. Jomento[1], A. Sombong[1], M. Abadingo[1], T. Vesagas[1], B. Mendoza[2], J. Comelio[2], C. Samia[2], [1]Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila, [2]Newborn Screening Center – Central Luzon

Presenter: *Margarita Aziza Canlas, Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila, mecanlas@up.edu.ph*

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Mucopolysaccharidosis type I (MPS I) and Glycogen Storage Disease Type II (Pompe) Screening in North Carolina: A Year in Review

J. Mills[1], L. Percenti[2], K. Clinard[3], S. Freeman[1], M. Fort[2], D. Petit[2], S. Shone[1], K. Blake[1], [1]North Carolina State Laboratory of Public Health, [2]North Carolina Division of Child and Family Well-Being, Whole Child Health Section, [3]Department of Pediatrics, The University of North Carolina at Chapel Hill

Presenter: *Jamie Mills, MLS(ASCP), North Carolina State Laboratory of Public Health, jamie.mills@dhhs.nc.gov*

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Dried blood spot steroids profiling for using Revvity QSight® 225MD

T. Au Yeung[1], V. Simonian[1], J. Trometer[1], [1]Revvity

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C16 Sulfatide Analysis in Dried Blood Spots on a Revvity QSight® 225MD Mass Spectrometer

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NeoBase™ 2 Non-derivatized MSMS assay validation on SCIEX Triple Quad™ 4500MD system

A. Tuominen[1], S. Talha[1], L. Koskimäki[1], T. Leppänen[1], S. Poikonen[1], H. Polari[1], J. Brozinski[1], [1]Revvity

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

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A streamlined newborn sequencing research workflow for detecting genomic variants using a curated gene panel

R. Kaukonen[1], L. Wang[1], K. Fura[1], M. Niemelä[1], M. Siitonen[1], E. Chin[1], Z. Ma[1], M. Hegde[1], [1]Revvity

Presenter: *Riina Kaukonen, PhD, Revvity, riina.kaukonen@revvity.com*

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Simple and flexible solution for SMA, SCID and XLA newborn screening with dry qPCR for all sample throughputs

M. Sjöqvist[1], M. Mäkinen[1], H. Savela[1], S. Vidbäck[1], R. Rantanen[1], T. Helenius[1], [1]Revvity

Presenters: *Maari Mäkinen, MS, Revvity, maari.makinen@revvity.com and Marika Sjöqvist, PhD, MSc, Revvity, marika.sjoqvist@revvity.com*

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Comparison of NeoLSD™ MSMS flow injection method with the new LC-MSMS method

H. Appelblom[1], A. Kiviniemi[1], J. Suvanto[1], C. Rautanen[1], T. Hanses[1], T. Leppänen[1], T. Au Yeung[1], V. Simonian[1], J. Trometer[1], [1]Revvity

Presenter: *Hanna Polari, MS, Revvity, hanna.polari@revvity.com*

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NeoLSD™ MSMS assay feasibility study with SCIEX Triple Quad™4500MD LC-MS/MS System

R. Bozic[1], L. Bacci[1], F. Ferron[1], F. Settanni[2], T. Sauro[2], L. Leoni[2], M. Lucchiari[1], F. Ponzetto[2], G. Mengozzi[2], [1]Revvity Italy, [2]Clinical Biochemistry Laboratory of Città della Salute e della Scienza University Hospital, Turin, Italy

Presenter: *Hanna Polari, MS, Revvity, hanna.polari@revvity.com*

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GSP® Neonatal Anti-Toxoplasma IgM kit for the determination of IgM antibodies to Toxoplasma gondii from Newborn DBS Samples

G. Hayashi[1], M. Dos Santos[2], A. Zani[2], H. Polari[2], L. Koskimäki[2], C. Rautanen[2], T. Heikkilä[2], T. Leppänen[2], T. Pölönen[2], [1]Instituto Jô Clemente (Reference Service in Neonatal Screening), [2]Revvity

Presenter: *Gustavo de Sá Schiavo Matias, The Center for Teaching, Research, and Innovation (CEPI-IJC), gustavo.matias@ijc.org.br and Penélope Vieira Camille, The Center for Teaching, Research, and Innovation (CEPI-IJC), penelope.camille@ijc.org.br*

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Detection of cytomegalovirus from DBS samples using crude DNA extraction and dry chemistry qPCR – The Eonis™ CMV qPCR assay

V. Veikkolainen[1], M. Aaltoranta[1], S. Adams[2], H. Paybe[3], [1]Revvity, [1]Great Ormond Street Hospital for Children, [3]Imperial College

Presenter: *Ville Veikkolainen, PhD, MS, Revvity, ville.veikkolainen@revvity.com*

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Alternative workflows for Eonis™ CMV qPCR assay – saliva samples, urine samples, DNA extraction volume and 384-well dry chemistry

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Presenter: *Ville Veikkolainen, PhD, MS, Revvity, ville.veikkolainen@revvity.com*

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Development of an LC-MS/MS Assay for Arylsulfatase A Activity in Dried Blood Spots for Second-Tier Metachromatic Leukodystrophy Testing

R. Bearden[1], A. Kaib[1], S. Smith[1], T. Donti[1], P. Borandi[1], M. Hegde[1], [1]Revvity Omics

Presenter: *Rebecca Bearden, PhD, Revvity Omics, Rebecca.Bearden@revvity.com*

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Improved IEF workflow by using Migele Gel Analyzer

A. Lichtner[1], Y. Pan[1], L. Lira[1], [1]Revvity Omics

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Newborn screening for the haemoglobinopathies using Isoelectric Focusing Gel Electrophoresis with high resolution imaging and interpretative algorithm (Migele)

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Presenter: *Lynn Lira, BS, MT(ASCP), Revvity Omics, lynn.lira@revvity.com*

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Assessing Family Outcomes in Newborn Screening

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Presenter: *Elizabeth Reynolds, PhD, RTI International, erreynolds@rti.org*

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A Newborn Screening Assay for Angelman, Prader-Willi, and Dup15q Syndromes for Implementation as Part of the Early Check Newborn Screening Research Program in North Carolina

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Presenter: *Katerina Kucera, PhD, RTI International, kkucera@rti.org*

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Evaluation of the Newborn Screening Information Center Website

M. Raspa[1], M. Lynch[1], R. Wright[1], A. Corbo[1], K. Baker[1], C. Gieck[1], [1]RTI International

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

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PEDI CAMPAIGN – A Newborn Screening Working Group US-wide Pediatricians Education Initiative on Newborn Screening for Lysosomal Storage Diseases

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Evaluation of the Sebia CAPILLARYS 3 DBS Instrument for Newborn Hemoglobinopathy Screening in Arizona

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Presenter: *Trung Huynh, PhD, MB(ASCP), Arizona Department of Health Services, trung.huynh@azdhs.gov*

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Simple and cost effective screening of 22q11.2 deletion syndrome in DBS samples

A. Kubar[1], S. Temel[2], S. Beken[3], G. Onder[3], O. Hatirnaz[3], A. Korkmaz[3], Y. Alanay[3], U. Ozbek[3], S. Ozemri-Sag[2], E. Kubar[4], C. Sonmezalp[1], O. Dogan Cosar[1], [1]SNP Biotechnology, Ltd., [2]Bursa Uludag University, [3]Acibadem Mehmet Ali Aydınlar University, [4]Ege University Faculty of Medicine

Presenter: *C. Zeynep Sönmezalp, MS, SNP Biotechnology, Ltd., zeynep@snp.com.tr*

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Improving the Quality of Newborn Screening Specimens through Outreach and Education

C. Harrelson[1], [1]South Carolina Department of Public Health

Presenter: *Christine Harrelson, MS, MLS(ASCP)cm, South Carolina Department of Public Health, harrelcl@dhec.sc.gov*

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Improving Newborn Screening (NBS) Data Quality Through LIMS and Vital Statistics Data Matching

J. Vasquez[1], R. Ryland[2], [1]Ruvos, [2]Florida Department of Health

Presenter: *Juan Vasquez, MHA, State of Florida/Ruvos, jvasquez@ruvos.com*

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Psychosocial aspects of positive newborn screening and systemic therapy approaches

B. Lindner-Fingerhut[1], R. Fingerhut[2], [1]Comitatus pro anima, Practice for Systemic Individual, Couples, and Family Therapy, [2]SYNLAB MVZ Weiden GmbH

Presenter: *PD Dr.rer.nat. Ralph Fingerhut, SYNLAB MVZ Weiden GmbH, ralph.fingerhut@synlab.com*

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A Rapid and Simple 2nd-Tier LC-MS/MS Methode for Newborn Screening for Isovaleric acidaemia and gltaric aicuria type 1

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Evaluation of the Labsystems AAAC 3.0 test kit for the determination of amino acids and acylcarnitine, and comparison to the Chromsystems test kit

R. Fingerhut[1], [1]SYNLAB MVZ Weiden GmbH

Presenter: *PD Dr.rer.nat. Ralph Fingerhut, SYNLAB MVZ Weiden GmbH, ralph.fingerhut@synlab.com*

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Nurturing Expertise: Newborn Screening Nurse Competency Modules

A. Keyton[1], H. Fryman[1], A. Ingram[1], B. Radowick[1], [1]Tennessee Department of Health

Presenter: *Angela Keyton, RN, Tennessee Department of Health, angela.keyton@tn.gov*

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Quality Improvement: Impact of OZ Systems on Timeliness of Newborn Screening

A. Porter[1], M. Dorley[1], A. Ingram[1], Y. Li[1], H. Fryman[1], H. Peeples[1], V. Ragland[1], S. Roberts[1], [1]Tennessee Department of Health

Presenter: *Ashley Porter, BSN, RN, Tennessee Department of Health, ashley.m.porter@tn.gov*

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Tennessee's Journey of Steroid Profiling for Congenital Adrenal Hyperplasia

L. Tyler[1], G. McKee[1], M. Dorley[1], A. Morgan[1], A. Bowman[1], S. Lampson[1], M. Finlay[1], [1]Tennessee Department of Health

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

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Withdrawn

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How the Advancement in Medical Treatment Impacts Hemoglobinopathy Screening and Clinical Care Coordination Approach to Communicating

O. Ordonez[1], J. Ybarbo[1], V. McNeely[1], A. Schlabach[1], E. Fitch[1], S. Tanksley[1], [1]Texas Department of State Health Services

Presenter: *Omar Ordonez, BS Microbiology, Texas Department of State Health Services, omar.ordonez@dshs.texas.gov*

P-114

Characterize novel biochemical marker associated with Biotinidase deficiency (BTD) to improve diagnostic accuracy and disease monitoring

A. Jamil[1], [1]The Aga Khan Hospital

Presenter: *Azeema Jamil, The Aga Khan Hospital, azeema.jamil@aku.edu*

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Quantitation of Lysosphingolipids in Dried Blood Spots and Plasma Using Targeted High-Resolution Mass Spectrometry for the Screening of Sphingolipidoses

F. Ducatez[1], A. Tebani[1], S. Bekri[1], J. Guo[2], K. Hassell[2], [1]CHU Rouen, [2]Thermo Fisher Scientific

Presenter: *Jingshu Guo, PhD, Thermo Fisher Scientific, jingshu.guo@thermofisher.com*

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Enhanced Detection of Hemoglobin Variants in Clinical Research using DBS and High-Resolution Accurate Mass (HRAM) Mass Spectrometry

Y. Song[1], K. Hassell[1], E. Goucher[1], J. Guo[1], T. Correa[1], [1]Thermo Fisher Scientific

Presenter: *Jingshu Guo, PhD, Thermo Fisher Scientific, jingshu.guo@thermofisher.com*

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Parallel biochemical and genetic testing informs a timely and accurate diagnosis of MPS VII: Findings from 5 years of sponsored testing programs

R. Giugliani[1], V. Rangel Miller[2], S. Daughtery[2], R. Naik Gupta[2], M. Hegde[3], J. Johnson[4], D. Marsden[2], J. Merritt II[2], L. Pollard[5], E. Peng[2], M. Scarpa[6], K. Simmons[2], N. Smith[4], R. Wang[7], N. Miller[2], [1]Department of Genetics of the Federal University of Rio Grande do Sul, HCPA, DASA Genomics, Casa dos Raros, [2]Ultragenyx Pharmaceutical, Inc., [3]Revvity Omics, [4]BioAgilytix, [5]Greenwood Genetic Center, [6]Regional Coordinator Centre for Rare Diseases, University Hospital of Udine, [7]Children's Hospital of Orange County, Department of Pediatrics, University of California-Irvine

Presenter: *Nicole Miller, PhD, Ultragenyx Pharmaceutical, Inc., NMiller@ultragenyx.com*

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Phenotype Associations from a Comprehensive Database of Long-Chain Fatty Acid Oxidation Disorder Gene Variants

V. Rangel Miller[1], H. Richbourg[1], O. Japalaghi[1], J. Vockley[2], M. AlSayed[3], P. Baker II[4], S. Daugherty[1], T. Ekstein[5], S. Grünert[6], M. Kiel[7], A. Khan[8], L. Korngut[9], D. Marsden[1], S. Monteleone[7], I. Schwartz[10], N. Miller[1], [1]Ultragenyx Pharmaceutical, Inc., [2]Division of Medical Genetics and Center for Rare Disease Therapy, UPMC Children's Hospital of Pittsburgh, [3]College of Medicine, Al-Faisal University, Riyadh, Kingdom of Saudi Arabia, [4]University of Colorado, Anschutz Medical Campus, [5]Invitae Corporation,[6]Center for Pediatric and Adolescent Medicine, University Medical Center, [7]Genomenon, [8]M.A.G.I.C. (Metabolics and Genetics in Canada) Clinic Ltd., [9]Department of Clinical Neurosciences, University of Calgary, [10]Hospital de Clinicas

Presenter: *Nicole Miller, PhD, Ultragenyx Pharmaceutical, Inc., NMiller@ultragenyx.com*

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Long-chain fatty acid oxidation disorders: A review of newborn screening around the globe for LC-FAOD

I. Schwartz[1], J. Audi[2], S. Grünert[3], P. Kainth[2], D. Kasper[4], D. Marsden[2], T. Nalin[2], M. Pasquali[5], V. Rangel Miller[2], I. Sosova[6], G. Tajima[7], K. Tobita[2], N. Miller[2], [1]Hospital de Clinicas, [2]Ultragenyx Pharmaceutical, Inc., [3]Center for Pediatric and Adolescent Medicine, University Medical Center, [4]ARCHIMEDlife Laboratories, [5]University of Utah School of Medicine, [6]Alberta Newborn Screening and Biochemical Genetics Laboratory, University of Alberta Hospital, [7]National Center for Child Health and Development

Presenter: *Nicole Miller, PhD, Ultragenyx Pharmaceutical, Inc., NMiller@ultragenyx.com*

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Newborn sequencing: Approaches taken by programs around the globe

M. Bailey[1], M. Baker[2], H. Cope[3], T. DeFay[4], L. Faivre[5], M. Hegde[6], S. Kingsmore[7], S. Terry[8], P. Tsipouras[9], N. Miller[10], [1]BioMarin Pharmaceutical Inc., [2]Wisconsin State Laboratory of Hygiene & Department of Pediatrics, University of Wisconsin, [3]RTI International, [4]Alexion, AstraZeneca Rare Disease (BeginNGS), [5]Université de Bourgogne-Franche Comté, [6]Revvity Omics, [7]Rady Children's Institute for Genomic Medicine, Rady Children's Hospital, [8]Genetic Alliance, [9]Plumcare RWE LLC, [10]Ultragenyx Pharmaceutical, Inc.

Presenter: *Nicole Miller, PhD, Ultragenyx Pharmaceutical, Inc., NMiller@ultragenyx.com*

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Very Long-Chain and Medium chain Acyl-CoA Dehydrogenase Deficiency: High Incidence of Detected Patients With Newborn Screening Program

Z. Remec[1], D. Trampuz[1], D. Perko[1], V. Čuk[1], E. Kozjek[1], A. Drole Torkar[1], M. Mlinaric[1], B. Repic Lampret[1], U. Groselj[1], [1]University Medical Centre Ljubljana

Presenter: *Žiga Iztok Remec, BSc Biology, University Medical Centre Ljubljana, ziga.remec@kclj.si*

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Opting Out of Newborn Screening in Iowa – Process Improvement Plan to Increase Documentation of Refusals

E. Phillips[1], C. Johnson[1], K. Noble Piper[2], [1]University of Iowa, [2]Iowa Department of Health and Human Services

Presenter: *Emily Phillips, BSN, RN, CCRC, University of Iowa, emily-phillips@uiowa.edu*

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Newborn Screening for Lysosomal Storage Diseases including Metachromatic Leukodystrophy

M. Gelb[1], [1]University of Washington

Presenter: *Michael Gelb, PhD, University of Washington, gelb@uw.edu*

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Successful newborn screening for Thalassemias in the Philippines

C. Padilla[1], M. Alcausin[1], J. Caneba[1], [1]University of the Philippines Manila

Presenter: *Carmencita Padilla, MD, MAHPS, University of the Philippines Manila, cdpadilla1@up.edu.ph*

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Expansion of the Utah Newborn Screening LC-MS/MS platform with the addition of I2S enzymatic assay for the screening of MPS II

F. Mesmar[1], H. Khaledi[2], H. Golsan[1], M. Gelb[3], N. Szabo[1], [1]Utah Public Health Laboratory, [2]GelbChem, LLC, [3] University of Washington

Presenter: *Fahmi Mesmar, PhD, Utah Public Health Laboratory, fmesmar@utah.gov*

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Overcoming Barriers: Virginia's Journey to Improve Timeliness of Follow-up Through Direct Access to Electronic Medical Records

M. Lowe[1], C. Seckner[1], V. Goyne[1], [1]Virginia Department of Health

Presenter: *Mary Lowe, BSN, RN, Virginia Department of Health, mary.lowe@vdh.virginia.gov*

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Supporting Best Outcomes Through Outreach: Improving Timeliness of Care Through Collaboration and Education

M. Lowe[1], V. Goyne[1], C. Seckner[1], [1]Virginia Department of Health

Presenter: *Mary Lowe, BSN, RN, Virginia Department of Health, mary.lowe@vdh.virginia.gov*

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UHPLC-MS/MS Analysis of Amino Acids in Dried Blood Spot using Waters Kairos™ Amino Acid Kit for Clinical Research

R. McBrinn[1], P. Rossiter[1], L. Calton[1], G. Hammond[1], [1]Waters Clinical Business Unit

Presenter: *Anahi Santoyo Castelazo, PhD, Waters Clinical Business Unit, Anahi_Santoyo_Castelazo@waters.com*

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Quantitative analysis of four long-chain lysophosphatidylcholines in dried-blood spot using Liquid Chromatography Tandem Mass Spectrometry for clinical research

A. Santoyo Castelazo[1], R. McBrinn[1], G. Hammond[1], J. Nisbet-Fahy[1], [1]Waters Clinical Business Unit

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Notes

Symposia History

National Neonatal Screening Symposium (1981-1999)

Newborn Screening & Genetic Testing Symposium (2001-2019)

Newborn Screening Symposium (2020 onward)

1981	Austin, TX
1982	Chicago, IL
1984	Orlando, FL
1985	Columbus, OH
1986	Austin, TX
1988	Portland, OR
1989	New Orleans, LA
1991	Saratoga, NY
1992	Raleigh, NC
1994	Seattle, WA
1995	Corpus Christi, TX
1996	Boston, MA
1998	San Diego, CA
1999	St. Louis, MO
2001	Raleigh, NC
2002	Phoenix, AZ
2004	Atlanta, GA
2005	Portland, OR
2007	Minneapolis, MN
2008	San Antonio, TX
2010	Orlando, FL
2011	San Diego, CA
2013	Atlanta, GA (with ISNS)
2014	Anaheim, CA
2016	St. Louis, MO
2017	New Orleans, LA
2019	Chicago, IL
2020	Virtual
2021	Virtual
2022	Tacoma, WA
2023	Sacramento, CA (with ISNS)
2024	Omaha, NE

2025 APHL Meetings

ID Lab Con
Pasadena, California
March 25 – 27, 2025

APHL 2025 Annual Conference
Portland, Oregon
May 5 – 8, 2025

2025 APHL Newborn Screening Symposium
Providence, Rhode Island
October 5 – 9, 2025

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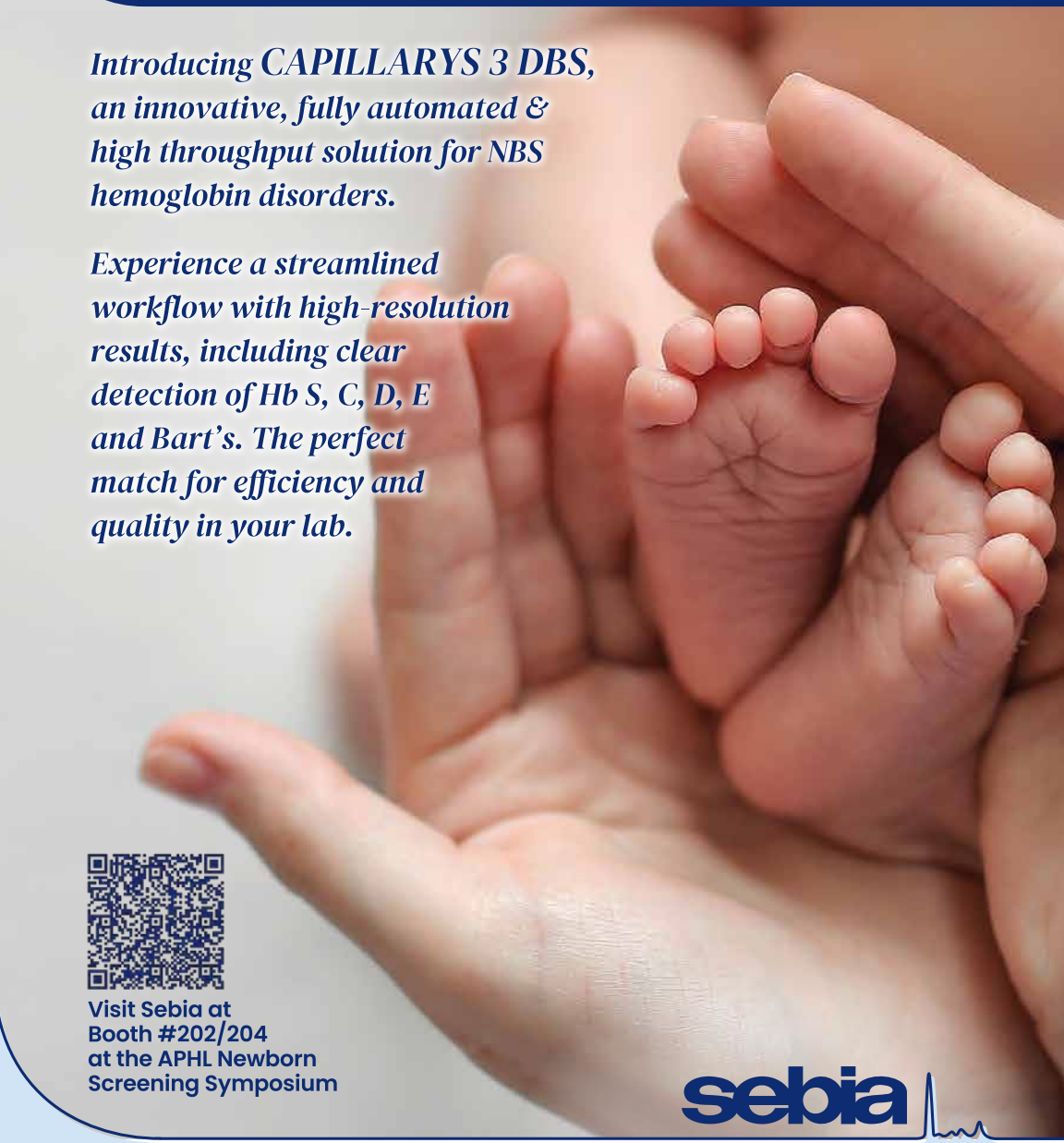

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