Final Program

Sponsored by the
Association of Public Health Laboratories

Co-Sponsored by the
International Society for Neonatal Screening
and the California Department of Public Health

www.aphl.org/nbs2021
#APHLNBS
About This Conference

The APHL Newborn Screening Symposium addresses state, national and international newborn screening, genetic testing and policy issues important to public health newborn screening systems. Attendees hear reports from around the globe that emphasize challenges faced and the data generated. Topics include molecular technologies, current and upcoming conditions, quality improvement, communicating with families and the public, and short- and long-term follow-up.

This year’s virtual program includes poster and oral presentations drawn from submitted abstracts, invited oral presentations, exhibits, the Meet the Manufacturers session and more! Input and participation by parents and advocacy organizations are encouraged.

The purpose of this symposium is to enhance participant knowledge of national and international newborn screening and genetics as related to emerging laboratory technologies, follow-up, candidate conditions, quality improvement and clinical outcomes. Join us in October!

The major learning goals of this symposium are:

Discuss and evaluate quality assurance and quality control measures for newborn screening laboratories

Describe state and international experiences with candidate conditions and clinical outcomes in newborn screening

Evaluate the effectiveness of current newborn screening and genetics follow-up programs

Who will benefit from attending this conference?

Newborn screening and genetics laboratory professionals, newborn screening and genetics program personnel and counselors, students, health care practitioners or other maternal and child health service providers, public health nurses, specialists, public health laboratory directors and other public health professionals involved with newborn screening and genetic testing issues and follow-up.

General Information

Registration

Advanced registration through APHL is required; registration is now open on the conference webpage www.aphl.org/nbs2021. If you have any questions, please contact Terry Reamer at terry.reamer@aphl.org or 240.485.2776. Registration fees are: Full conference – $350 | One day – $100 | Student – $100

Continuing Education Credits Available

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 have the opportunity to earn up to 24.75 contact hours by attending the entire conference. Nursing and genetic counseling continuing education units (CEUs), Certified Public Health (CPH), certified health education specialist (CHES) and continuing medical education (CMEs) are in the application process. Up to 20.75 CME, CNE, CGC and CHES credits are available. Full session descriptions and training objectives are posted online at www.aphl.org/nbs2021.

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.
Not to Miss!
All listed times are US Eastern.

**Keynote Session:**
Avoiding Health Disparities: Does Newborn Genome Sequencing Bridge or Widen the Gap?
Tuesday, October 5 • 12:00–2:00 pm ET
This session will begin with an overview of health disparities and then move on to the advantages and disadvantages of genome sequencing in the context of newborn screening. It will conclude with a discussion on the ethical and social implications of sequencing and how emerging technologies have forced us to think critically about equity in newborn screening.

**Baking with Dana of Cookies4Cures**
Tuesday, October 5 • 7:00–7:45 pm ET
Join us for a heart-warming segment featuring Dana, a sixth grader and founder of Cookies4Cures, a nonprofit that raises money for research into rare pediatric diseases. Dana will teach you how to make one of her favorite cookies and will then answer questions about how she started her nonprofit and why this cause means so much to her. As Dana says, “When a child has a rare disease with no cure, research is hope.”

**Exhibits and Poster Hall**
12:00–5:00 pm ET 12:00–4:00 pm ET
Tuesday, October 5 Tuesday, October 14
Wednesday, October 6 Thursday, October 7
Tuesday, October 12 Wednesday, October 13
Exhibitors and posters will be open and available for viewing throughout the virtual conference. Be sure to visit the exhibitors to see the latest products and services and the posters to learn the latest science and practices. Watch videos, see e-posters and chat with exhibitor staff and poster presenters.

**Innovate! Sessions**
Thursday, October 7 Tuesday, October 12
5:00–6:00 pm ET 5:30–6:30 pm ET
Connect with your industry partners and learn of new technologies and services.

**Virtual Social Hour**
Not a Trivial Moment: the Pursuit of a Break sponsored by QIAGEN
Thursday, October 7 • 6:30–7:30 pm ET
Light-hearted presentations from vendors.

**Roundtable Discussions**
Tuesday, October 12
12:00–1:00 pm ET
- The Many Hats of Newborn Screening Bioinformaticians
- What’s in a Word? Discussing Language in Newborn Screening

Thursday, October 14
12:00–1:00 pm ET
- Transfusion Confusion: Managing Its Impact on Screening Results
- CDC’s Modernizing Newborn Screening Through Training and Data Analytics

**Awards Ceremony**
October 13 • 6:30–7:30 pm ET
Celebrate the achievements of your colleagues and friends in the newborn screening community!

**Parent/Patient Panel**
Thursday, October 14 • 4:00–6:00 pm ET
Hear compelling personal accounts of how newborn screening impacts the lives of families. Follow their journeys from screening and diagnosis to treatment and management, and discover the small but powerful community of parents of children with disorders detected by newborn screening.
# Agenda at a Glance

All listed times are US Eastern.

## TUE October 5
12:00–2:00 pm  
Welcome and Keynote: Avoiding Health Disparities: Does Newborn Genome Sequencing Bridge or Widen the Gap?

2:00–2:30 pm Break

2:30–4:00 pm Current RUSP Conditions in State NBS Panels

4:00–5:00 pm Break

5:00–6:30 pm Emergency Preparedness

6:30–7:00 pm Break

7:00–7:45 pm Baking with Dana

## WED October 6
12:00–1:00 pm Around the World in 60 Minutes

1:00–1:30 pm Break

1:30–2:30 pm Adoption and Use of Second-tier Testing

2:30–3:30 pm Break

3:30–5:00 pm Conditions Under Consideration for Addition or Removal from State Panels

4:00–5:00 pm Break

5:00–5:30 pm Conversations Around Newborn Screening

6:00–6:30 pm Break

6:30–7:30 pm Virtual Social Hour sponsored by QIAGEN

## THU October 7
12:00–1:30 pm Health Information Technology (HIT)

1:30–2:00 pm Break

2:00–3:00 pm Follow-up

3:00–3:30 pm Break

3:30–4:45 pm Training and Education

4:00–5:00 pm Break

5:00–6:00 pm Innovate! Sessions

6:00–6:30 pm Break

6:30–7:30 pm NBS Genetic Privacy Concerns

## TUE October 12
12:00–1:00 pm Roundtables

1:00–1:30 pm Public Engagement

2:00–3:00 pm Quality Improvement and Assurance Activities

3:00–3:30 pm Break

3:30–4:45 pm Spinal Muscular Atrophy (SMA)

4:00–5:00 pm Health Equity

4:00–6:00 pm Parent/Patient Panel

5:00–5:30 pm Meet the Manufacturers

## WED October 13
12:00–1:00 pm Data Analytics and Bioinformatics

1:00–2:00 pm Break

2:00–3:00 pm Case Studies

3:00–3:30 pm Break

3:30–4:30 pm Spinal Muscular Atrophy (SMA)

4:00–5:00 pm Break

5:00–5:30 pm Meet the Manufacturers

## THU October 14
12:00–1:00 pm Roundtables

1:00–1:30 pm Parent Patient Panel

1:30–2:00 pm Break

2:00–3:00 pm Case Studies

3:00–3:30 pm Break

3:30–4:30 pm Spinal Muscular Atrophy (SMA)

4:00–5:00 pm Break

5:00–5:30 pm Meet the Manufacturers

6:00–6:30 pm Awards Ceremony

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**9:45–11:00 am • Tuesday Oct 12**

**ISNS Session**

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Agenda of Events
All listed times are US Eastern.

Tuesday, October 5

12:00–2:00 pm
Welcome and Keynote Session: Avoiding Health Disparities: Does Newborn Genome Sequencing Bridge or Widen the Gap?
588-830-21 • 2.0 contact hours
Moderators: Michele Caggana, ScD, FACMG, New York State Department of Health/Wadsworth Center and Richard Olney, MD, California Department of Public Health
Speakers:
• Marsha Treadwell, PhD, Professor, University of California, San Francisco
• Priya Parikh, MD, University of California, San Francisco
• Stephen Kingsmore, MD, DSc, President and Chief Executive Officer, Rady Children’s Institute for Genomic Medicine
• Robert C. Green, MD, MPH, Professor of Medicine (Genetics), Harvard Medical School
• Lainie Friedman Ross, MD, PhD, Carolyn and Matthew Bucksbaum Professor of Clinical Medical Ethics, University of Chicago
This session will begin with an overview of health disparities and then move on to the advantages and disadvantages of genome sequencing in the context of newborn screening. It will conclude with a discussion on the ethical and social implications of sequencing and how emerging technologies has forced us to think critically about equity in newborn screening.

12:00–5:00 pm
Exhibit and Poster Hall open

2:00–2:30 pm
Break / Visit the exhibits and posters

2:30–4:00 pm
Current Recommended Uniform Screening Panel (RUSP) Conditions in State NBS Panels
588-831-21 • 1.5 contact hours
Moderators: Alisha Keehn, MPA, Health Resources and Services Administration and Jillian Chance, BSN, RN, Nebraska Division of Public Health
This session will examine methods of testing in various states for disorders on the current RUSP, including X-linked Adrenoleukodystrophy (X-ALD), Pompe disease, Mucopolysaccharidosis type I (MPS-I), as well as those not on the RUSP including Gaucher and Fabry.
• Screening for X-linked Adrenoleukodystrophy in Texas, A Two-Screen State
  Patricia Hunt, BA, Texas Department of State Health Services
• X-ALD, Pompe Disease and MPS-I Screenings with One Dried Blood Spot: A Tour de Force
  Nicolas Szabo, PhD, Utah Public Health Laboratory

4:00–5:00 pm
Break / Visit the exhibits and posters

5:00–6:30 pm
Emergency Preparedness
88-832-21 • 1.5 contact hours
Moderators: Scott Shone, PhD, HCLD(ABB), North Carolina State Laboratory of Public Health and Brendan Reilly, BS, Texas Department of State Health Services
This session will explore the impact that emergencies have on newborn screening programs, and how hospital staff, parents and newborn screening staff are prepared to handle these cases through education and creative problem solving.

• Comparison of Screening for Four Lysosomal Storage Disorders (Pompe, MPS1, Gaucher and Fabry) Versus Two (Pompe and MPS-1) on the Baebies SEEKER Platform
  Melanie Kessler-Mathieu, MS, PhD, Kansas Department of Health and Environment Laboratory
• Precision Newborn Screening for Lysosomal Disorders and Adrenoleukodystrophy — An Update from Kentucky and Mayo Clinic
  Dietrich Matern, MD, PhD, Mayo Clinic
• Beyond the PKU Test: A Tool to Explore Rare Genetic Diseases in Newborn Screening
  Amy Brower, PhD, American College of Medical Genetics and Genomics
Emergency Preparedness (continued)

- Born to Run... An Explosive Account of Christmas Day!
  M. Christine Dorley, PhD, MT(ASCP), Tennessee Department of Health: Laboratory Services

- Educating Hospital Staff About Newborn Screening During a Pandemic: Michigan’s Experience
  Mary Kley, MS, Michigan Department of Health and Human Services

- Thinking Outside of the Box: Educating Parents About Newborn Screening During a Pandemic
  Kristen Thompson, MPH, Michigan Department of Health and Human Services

- Impact of COVID-19 on Time to Case Closure of Time Critical Disorders
  Sarah Bradley, MS, CGC, New York State Department of Health

7:00–7:45 pm

Baking with Dana: Learn to Bake Sprinkle Cookies

Join us for a heart-warming segment featuring Dana, a sixth grader and founder of Cookies4Cures, a nonprofit that raises money for research into rare pediatric diseases. Dana will teach you how to make one of her favorites, sprinkle cookies, and will then answer questions about how she started her nonprofit, why this cause means so much to her, and what her favorite cookie is, of course!

Wednesday, October 6

12:00–1:00 pm

Around the World in 60 Minutes

588-833-21 • 1.0 contact hour

This session will summarize the state of newborn screening around the world, with an emphasis on modernization and neonatal screening developments in Europe, the Netherlands and the United States.

- The State of Newborn Screening Systems in the United States in 2021
  Sikha Singh, MHS, PMP, Association of Public Health Laboratories

- Neonatal Screening Developments in Europe — An ISNS Report
  Peter Schielen, PhD, International Society for Neonatal Screening

- Evaluation of the First 18 Months of Screening for PA and MMA in The Netherlands: The Findings Are Not as Expected
  Rose Maase, PhD, C(ASCP), MRSC, National Institute for Public Health and the Environment (RIVM)

- Modernizing Newborn Screening to Prepare for a New Generation of Transformative Therapies
  Don Bailey, PhD, RTI International

1:00–1:30 pm

Break / Visit exhibits and posters

1:30–2:30 pm

Adoption and Use of Second-tier Testing

588-834-21 • 1.0 contact hour

This session will evaluate second-tier testing as it relates to timeliness for screening of cystic fibrosis, clinical follow-up to prevent delays and better understand the rate of false positives to bring about improvements.

- Glycosaminoglycan Analysis in Dried Blood Spots: A Critical Second-stage Step in Newborn Screening for MPS-I and Related Disorders
  Michael H. Gelb, PhD, University of Washington

- Wisconsin Redesign of Clinical Follow-up Algorithm Based on Introduction of Second-tier Testing
  Jessica Scott Schwoerer, MD, University of Wisconsin

- A Review of Timeliness of Cystic Fibrosis Diagnosis Post-implementation of Second-tier Testing
  Marina Choi, MPH(c), Washington State Department of Health

- Ten Years of Sending Second Tier Testing for Propionic Acidemia, Methylmalonic Acidemia, and Cobalamin Disorders and the Lessons Learned
  Shawn Moloney, MPH, Michigan Department of Health and Human Services

2:30–3:30 pm

Break / Visit the exhibits and posters
3:30–5:00 pm
Conditions Under Consideration for Addition to State Panels
588-835-21 • 1.5 contact hours
This session will examine various disorders including Guanidinoacetate methyltransferase (GAMT) deficiency, Duchenne muscular dystrophy (DMD) and Ornithine transcarbamylase (OTC) deficiency in order to determine whether they should be added to state panels.

• Infant with Guanidinoacetate Methyltransferase Deficiency Identified Through Newborn Screening
  Kim Hart, MS, CGC, Utah Department of Health

• Identification of an Infant with Guanidinoacetate Methyltransferase (GAMT) Deficiency via Universal Newborn Screening in New York State
  Denise M. Kay, PhD, New York State Department of Health

• A Consented Pilot Study in New York State to Screen Newborns for Duchenne Muscular Dystrophy
  Norma P. Tavakoli, PhD, New York State Department of Health

• Early Check Implementation of Newborn Screening for Duchenne and Related Muscular Dystrophies in North Carolina
  Katerina Kucera, PhD, RTI International

• Newborn Screening of Duchenne Muscular Dystrophy Specifically Targeting Deletions Amenable to Exon-skipping Therapy
  François Boemer, PhD, CHU Liege, Belgium

• Considering Ornithine Transcarbamylase Deficiency for Mandatory Newborn Screening
  Michael Katsuyama, MPHc, Washington State Department of Health

5:30–6:30 pm
Conversations Around Newborn Screening
588-836-21 • 1.0 contact hour
Moderator: Guisou Zarbalian, MS, MPH, Association of Public Health Laboratories
This session will feature two presentations delving into the more controversial side of newborn screening. The first will question whether very rare disorders can—and should—be screened for, while the second will discuss the emerging issue of Neonatal Abstinence Syndrome (NAS) in maternal and child health.

• Some Disorders May Be Too Rare To Be Part of Newborn Screening
  Robert Currier, PhD, University of California, San Francisco

• Exploring the Role of Laboratory Data in Neonatal Abstinence Syndrome (NAS) Surveillance — An Open Dialogue
  Ewa King, PhD, Rhode Island State Health Laboratories

Thursday, October 7

12:00–1:30 pm
Health Information Technology (HIT)
588-837-21 • 1.5 contact hours
This session will evaluate how methods of technology including electronic test orders and results reporting (ETOR), common data models (CDMs) and other health commerce systems can create positive solutions for programs facing challenges with data transfer, analytics, quality improvement and communication.

• ETOR: A Review of Capabilities and Needs
  Willie Andrews, BSMT(ASCP), J Michael Consulting

• ETOR: A Path Forward
  Dari Shirazi, Association of Public Health Laboratories

• Using a Newborn Screening Common Data Model: Implications for Data Transfer and Analytics
  Amy Gaviglio, MS, CGC, CDC/APHL/Expecting Health

• Newborn Screening Hospital Quality Improvement and Communication Application Portal in New York State Health Commerce System
  Kathy Chou, PhD, New York State Department of Health

• Assistance to Newborn Screening Programs to Develop an Interoperability Plan
  Craig Newman, PhD, Altarum

• Understanding Parental Frustrations of NBS Paves Way for Technological Solutions
  Karen Eilbeck, PhD, University of Utah

12:00–5:00 pm
Exhibit and Poster Hall Open

1:30–2:00 pm
Break / Visit the exhibits and posters

2:00–3:00 pm
Follow-up
588-838-21 • 1.0 contact hour
This session will explore ways that newborn screening programs are improving follow-up practices and creating better tools for communicating with different parent communities.
Tuesday, October 12

9:45–11:00 am US ET
15:45–17.00 CET

ISNS Session
The International Society for Neonatal Screening is proud to offer a dedicated ISNS session as part of the APHL 2021 Newborn Screening Symposium.

- Introduction and ISNS Activity 2021
  Prof James Bonham, UK

- CDC QC materials for TSH
  Dr. Joanne Mei, USA

- IFCC/ISNS Global Task Force to Develop NBS
  Dr. Dianne Webster, New Zealand

- Impact of COVID-19 on NBS Worldwide
  Dr. Urh Groselj, Slovenia

- NBS for SCID and the Power of Virtual Meetings
  Dr Peter Schielen, Netherlands

- Closing Remarks
  Prof James Bonham, UK

4:45–5:00 pm
Break / Visit the exhibits and posters

5:00–6:00 pm
Innovate! Sessions (concurrent)
Presented by Agilent Technologies and PerkinElmer. See details on page 12.

6:30–7:30 pm
Virtual Social Hour
Not a Trivial Moment: the Pursuit of a Break
Sponsored by QIAGEN
Take a break with colleagues for a chance to connect with other attendees, take part in a trivia quiz and win prizes.

12:00–1:00 pm
Concurrent Roundtables:

- The Many Hats of Newborn Screening Bioinformaticians
  588-840-21 • 1.0 contact hour
  Gretchen Cote, MS, Virginia Division of Consolidated Laboratory Services
  This roundtable will feature bioinformaticians and fellows from New York, Texas and Virginia who will demonstrate the spectrum of projects
that a bioinformatician can conduct to streamline laboratory algorithms and positively impact babies at a clinical level. It will also discuss the various roles of bioinformaticians within the NBS laboratory, and their potential for helping other programs expand data analytics capabilities.

• **What’s in a Word? Discussing Language in Newborn Screening**

  588-841-21 • 1.0 contact hour

  Brianne Miller, MPH, Expecting Health / Children’s National Hospital

  This roundtable will examine how the language used in different aspects of newborn screening has changed throughout the years, and how improvements can be made as the system evolves. A discussion will be had on how the terms used in newborn screening services and education specifically impacts families and their understanding of the newborn screening system.

• **Bringing Together All Newborn Screening Stakeholders**

  Dylan Simon, MS, EveryLife Foundation for Rare Diseases

• **Cure SMA Outreach to Facilitate Spinal Muscular Atrophy Newborn Screening Implementation**

  Mary Schroth, MD, Cure SMA

• **Use of Social Media Ad Campaigns to Inform Parents and Enroll Newborns in the Early Check Expanded Newborn Screening Research Study**

  Jessica Pikowski, MA, RTI International

• **High and Low-tech Recruitment Strategies in Newborn Screening Research**

  Brianne Miller, MPH, Expecting Health / Children’s National Hospital

3:00–4:00 pm

**Break / Visit the exhibits and posters**

4:00–5:30 pm

**Health Equity**

588-843-21 • 1.5 contact hours

**Moderators:** Erica Wright, MS, CGC, Children’s Hospital Colorado/University of Colorado and Carla Cuthbert, PhD, FACMG, Centers for Disease Control and Prevention

This session will feature a panel of four speakers who will examine equity and disparity as it relates to the newborn screening system and identify challenges in data availability and consider next steps for improvements. There will also be a presentation on the causes and solutions to under-documentation of hemoglobin traits to assure health equity, facilitate reliable communication with families, and ensure integrity of research.

• **Exploring Equity Across the NBS System: From Discourse to Action (Panel)**

5:30–6:30 pm

**Innovate! Session**

Presented by Travere Therapeutics. See details on page 12.

6:30–7:30 pm

**Newborn Screening Genetic Privacy Concerns Persist, Despite the Common Rule**

**Moderators:** Aaron Goldenberg, PhD, MPH, Case Western Reserve University and Kimberly Noble Piper, RN, BS, CPH, CPHG, Iowa Department of Public Health

**Speaker:** Eric Hendricks, JD, Michigan Department of Health and Human Services

This session will provide an overview of the case, Kanuszewski et. al. vs. Michigan Department of Health and Human Services, as well as potential implications of the initial court ruling in this case for newborn screening programs in general. The panel will also discuss how the newborn screening community can address genetic privacy concerns while ensuring that residual dried blood spots remain available for crucial newborn screening quality assurance and improvement purposes.
Wednesday, October 13

12:00–1:00 pm
**Data Analytics and Bioinformatics**
588-844-21 • 1.0 contact hour
This session will evaluate how data analytics and bioinformatics can address challenges to newborn screening programs, specifically as they relate to improving screening for X-linked Adrenoleukodystrophy (X-ALD) and Congenital Adrenal Hyperplasia (CAH) and newborn screening DNA sequencing efforts.

- Using Models to Address Challenges in Newborn Screening Expansion
  Kee Chan, PhD, MBA, Newborn Screening Translational Research Network

- Improving X-linked Adrenoleukodystrophy Screening Performance with Insight from Confirmatory Results
  Hao Tang, PhD, California Department of Public Health

- Data-driven Approach to Revising Screening Cutoffs for Congenital Adrenal Hyperplasia
  Isabel Hurden, MPH, Michigan Department of Health and Human Services

- Applying COVID-19 Next Generation Sequencing Bioinformatics Progress to Newborn Screening DNA Sequencing Efforts
  Samantha Marcellus, MPH, Texas Department of State Health Services

2:00–3:00 pm
**Quality Improvement and Assurance Activities**
588-845-21 • 1.0 contact hour
This session will explore how quality improvement and assurance efforts can enhance timeliness and minimize the unintended consequences of newborn screening programs identifying infants who do not need medical attention.

- California Newborn Screening Specimen Tracking Quality Improvement Initiative
  Luis Cruz, California Department of Public Health

- An Algorithm for Outlier Detection Based Upon the Correlation Between Gestational Age and Birth Weight
  Mary Seeterlin, PhD, Michigan Department of Health & Human Services

- An Additional Analyte Ratio to Better Determine VLCAD Risk and Improve the Positive Predictive Value of the Screen
  Casey Guccione, Kansas Department of Health and Environment Laboratory

- Refinement of Newborn Screening for Cystic Fibrosis Process to Minimize Unintended Outcomes
  Mei Baker, MD, Wisconsin State Laboratory of Hygiene

3:00–3:30 pm
**Break / Visit the exhibits and posters**

3:30–4:30 pm
**Spinal Muscular Atrophy (SMA)**
588-846-21 • 1.0 contact hour
This session will examine the implementation and results of initial screening for SMA in California, Illinois and Wisconsin, as well as evaluate the use of commercial reagents in positively affecting screening for SMA and Severe Combined Immunodeficiency (SCID).

- Comparison of LDT and Commercial Multiplex Assays to Screen for SMA and SCID in Newborns
  Francis K. Lee, MSc, PhD, Centers for Disease Control and Prevention

- SMA Newborn Screening: California’s First Year Experience
  Jamie Matteson, MPH, California Department of Public Health

- Spinal Muscular Atrophy Screening: The Illinois Experience
  Vineet Dhiman, PhD, Illinois Department of Public Health

- Incorporating SMN2 Copy Number Assessment in Routine Newborn Screening for Spinal Muscular Atrophy: The Wisconsin Experience
  Mei Baker, MD, Wisconsin State Laboratory of Hygiene

4:30–5:00 pm
**Break / Visit exhibits and posters**

5:00–5:30 pm
**Meet the Manufacturers**
Light-hearted presentations from Baebies, Inc. and PerkinElmer.

6:30–7:30 pm
**Awards Ceremony**
Thursday, October 14

12:00–1:00 pm
Concurrent Roundtables:

- **Transfusion Confusion: Managing Its Impact on Screening Results**
  588-847-21 • 1.0 contact hour
  Patrick Hopkins, Missouri State Public Health Laboratory
  This roundtable will outline best practices for accurate screening, detection, and reporting of newborn screening results for disorders affected by RBC transfusion including hemoglobinopathy, biotinidase and galactosemia. A discussion of best practices for managing screening results for transfused newborns will also occur.

- **CDC’s Modernizing Newborn Screening Through Training and Data Analytics**
  588-848-21 • 1.0 contact hour
  Cynthia Hinton, PhD, MS, MPH, Centers for Disease Control and Prevention
  This roundtable will feature a presentation from the CDC’s Newborn Screening and Molecular Biology Branch (NSMBB) regarding two online programs created to address concerns around the increasingly complex landscape of testing, training and data handling. A discussion on topics for future modules, such as current and emerging technologies, current and anticipated screened conditions, and policies and issues that impact practice will also occur.

1:00–1:30 pm
Break / Visit exhibits and posters

1:30–3:00 pm
Case Studies
  588-849-21 • 1.5 contact hours
  Moderators: Nathalie Lepage, PhD, Newborn Screening Ontario and Kimberly Noble Piper, RN, BS, CPH, CPHG, Iowa Department of Public Health
  This session will feature five presentations on unique cases and diagnostic dilemmas that have led newborn screening programs to examine current testing methodologies and outreach.
  - Off Target/Unknown Diagnosis Based on Newborn Screening Results
    Debra Freedenberg, MD, PhD, Texas Department of State Health Services
  - C3- Acylcarnitine: A Particularly Tricky Newborn Screening Metabolite. Diagnosis and Management for a Rarely Diagnosed Disorder
    Susan Berry, MD, University of Minnesota Department of Pediatrics
  - X-linked Adrenoleukodystrophy in the Era of Newborn Screening: Cases on the Margin
    Christina Lam, MD, University of Washington and Seattle Children’s Hospital
  - Fatty Acid Oxidation Disorders: Two Atypical Cases Requiring Long-term Follow-up for Diagnostic Confirmation
    Neena Champaigne, MD, Medical University of South Carolina
  - Correct Case Classification in VLCAD Newborn Screening: Additional Evidence for Comprehensive Testing
    Amy Calhoun, MD, University of Iowa

3:00–4:00 pm
Break / Visit exhibits and posters

4:00–6:00 pm
Parent/Patient Panel
  588-850-21 • 2.0 contact hours
  Moderators: Lisa Feuchtbaum, DrPH, MPH, California Department of Public Health and Tracey Bishop, BS, California Department of Public Health
  Speakers:
  - Christina and Daniel Kettler
    Son Fitz (27 months) with Artemis SCID
  - Natalie and Eric Lamb
    Daughter Etta (12 months) with SMA
  - Diane and John Pytel
    Daughter Mia (15 years) with PKU
  - Nicholai and Alison Adler
    Son Lucas (3 ½ years) with asymptomatic ALD
  This live session will feature compelling personal accounts of how newborn screening impacts the lives of families. Follow their journeys from screening and diagnosis to treatment and management and discover the small but powerful community of parents of children with disorders detected by newborn screening.

12:00–4:00 pm
Exhibit and Poster Hall open
Thursday, October 7  
5:00–6:00 pm  
CONCURRENT SESSIONS:

**Improving the Speed and Selectivity of Newborn Screening Through a Combination of Ion Mobility Spectrometry—Mass Spectrometry (IMS-MS) with High Throughput Rapid Fire Automation**  
P resentated by Agilent Technologies  

The development of rapid yet selective analytical methods for diagnosis of congenital disorders is essential to mitigate the detrimental effects of NBS conditions. Current methods utilize flow-injection analysis with tandem MS, however, these methods lack selectivity for isobaric/isomeric systems which complicate disease diagnosis. Here we evaluate the inclusion of ion mobility spectrometry (IMS) prior to MS detection to improve the selectivity and predictive power of NBS and discuss future advancements in IMS research.  

**Speaker:** James Dodds, PhD, North Carolina State University

**PerkinElmer New Innovations—Detecting MPSII and GAMT by Tandem Mass Spectrometry**  
P resentated by PerkinElmer  

The mucopolysaccharidosis type II (MPSII) is a lysosomal storage disorder (LSDs) caused by defects in the metabolic breakdown of glycosaminoglycans (GAGs) and Guanidinoacetate Methyltransferase Deficiency (GAMT) is a severe but treatable Cerebral Creatine Deficiency Syndrome (CCDS). Both disorders are currently under the nomination for Recommended Uniform Screening Panel (RUSP) in US. Currently PerkinElmer is developing methods for measuring I2S enzymatic activities and guanidinoacetate (GUAC) and creatine (CRE) analyte levels using Tandem Mass Spectrometry technology. The results provide supporting data for detecting MPSII and GAMT on dried blood spots.  

**Speaker:** Tao He, Global Business Manager, PerkinElmer

Tuesday, October 12  
5:30–6:30 pm  

**An Evaluation of Confirmatory Practice in X-ALD Newborn Screening: Are We Doing Enough?**  
P resentated by Travere Therapeutics  

Currently most states screening for X-ALD are only confirming for the ABCD1 gene leaving other conditions that present with very long chain fatty acid elevations such as Zellweger Spectrum Disorder to be missed. Here we discuss the science behind the need for a more comprehensive confirmatory practice in X-ALD NBS and the potential impact this could have on identification of these other conditions for patients and their families.  

**Speakers:** Elisa Seeger, ALD Alliance and Christy Tise, MD, PhD, Stanford University
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