APHL Newborn Screening Symposium 2021

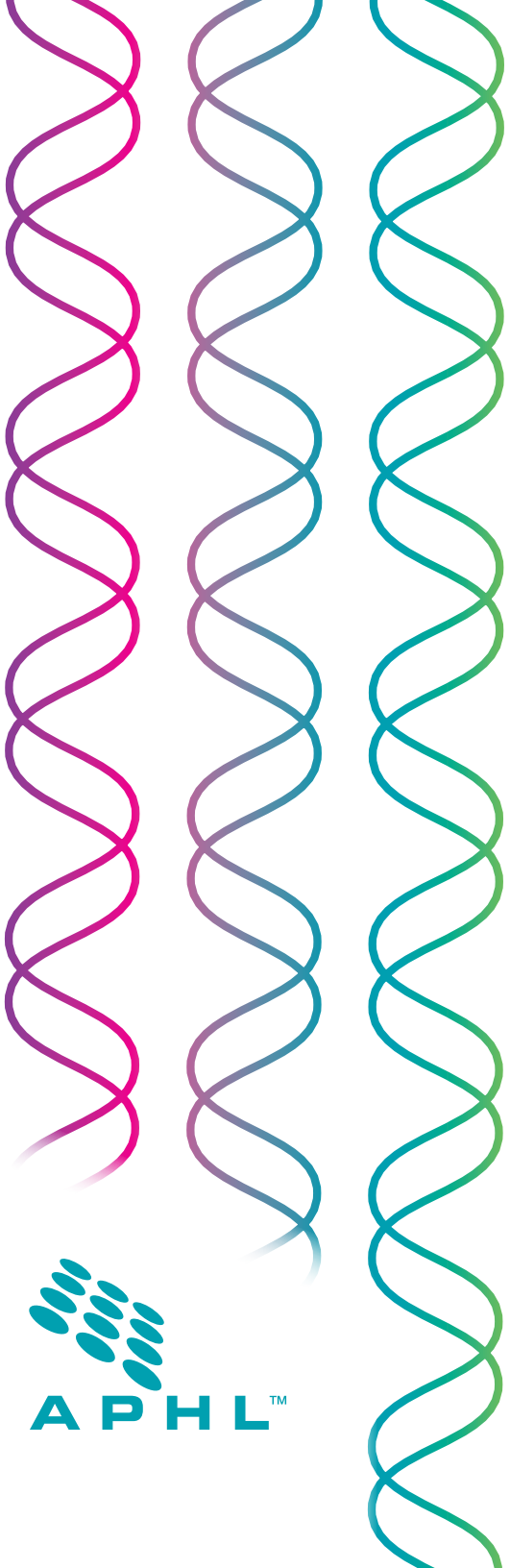
october 5–14 tue wed thu virtual conference

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

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.

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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.

APHL™

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
Tuesday, October 5
Wednesday, October 6
Thursday, October 7
Tuesday, October 12
Wednesday, October 13

12:00–4:00 pm ET
Thursday, October 14

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
5:00–6:00 pm ET

Tuesday, October 12
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

Meet the Manufacturers

Wednesday, October 13 • 5:00–5:30 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.

9:45–11:00 am • Tuesday Oct 12
ISNS Session

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**

Exhibits and Posters • 12:00–5:00 pm

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**
- 5:00–5:30 pm **Break**
- 5:30–6:30 pm **Conversations Around Newborn Screening**

Exhibits and Posters • 12:00–5:00 pm

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**
- 4:45–5:00 pm **Break**
- 5:00–6:00 pm **Innovate! Sessions**
- 6:00–6:30 pm **Break**
- 6:30–7:30 pm **Virtual Social Hour** *sponsored by QIAGEN*

Exhibits and Posters • 12:00–5:00 pm

TUE October 12

- 12:00–1:00 pm **Roundtables**
- 1:00–1:30 pm **Break**
- 1:30–3:00 pm **Public Engagement**
- 3:00–4:00 pm **Break**
- 4:00–5:30 pm **Health Equity**
- 5:30–6:30 pm **Innovate! Sessions**
- 6:30–7:30 pm **NBS Genetic Privacy Concerns**

Exhibits and Posters • 12:00–5:00 pm

WED October 13

- 12:00–1:00 pm **Data Analytics and Bioinformatics**
- 1:00–2:00 pm **Break**
- 2:00–3:00 pm **Quality Improvement and Assurance Activities**
- 3:00–3:30 pm **Break**
- 3:30–4:30 pm **Spinal Muscular Atrophy (SMA)**
- 4:30–5:00 pm **Break**
- 5:00–5:30 pm **Meet the Manufacturers**
- 5:30–6:30 pm **Break**
- 6:30–7:30 pm **Awards Ceremony**

Exhibits and Posters • 12:00–5:00 pm

THU October 14

- 12:00–1:00 pm **Roundtables**
- 1:00–1:30 pm **Break**
- 1:30–3:00 pm **Case Studies**
- 3:00–4:00 pm **Break**
- 4:00–6:00 pm **Parent/Patient Panel**

Exhibits and Posters • 12:00–4:00 pm

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, Calilfornia 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
- **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

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.

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 Kleyn, 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 on 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

12:00–5:00 pm

Exhibit and Poster Hall Open

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.

- **Closing the Gap: Improving Critical Congenital Heart Disease (CCHD) Screening Reporting Rate Among the Homebirth Community**
  Kristen Thompson, MPH, Michigan Department of Health and Human Services
- **Beyond the Fax Machine: Development of a REDCap Tool to Improve Communication on MPS-1 2nd Tier Sequencing Results and Decrease Referrals to Genetics**
  Christen F. Crews, MSN, RN, Virginia Department of Health
- **Interim Results of a Survey Examining the Experiences of Parents After Receiving Newborn Screening Results**
  Anne E. Atkins, MPH, Children's National Hospital

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

3:30–4:45 pm
Training and Education

588-839-21 • 1.25 contact hours

This session will explore improvements and developments to education and communication with the aim of creating a better system of care for all newborn screening stakeholders.

- **Building Prenatal Providers' Capacity to Educate Expectant Mothers About Newborn Screening: Results from Iowa's OTTER Project**
  Jeremy D. Penn, PhD, MPH, University of Iowa College of Education
- **Navigating the Digital Age: Creating Animated Videos for Families to Learn About SCID**
  Jamie Loey, MPH, Expecting Health
- **All for One, Not One for All — Developing and Implementing a Multifaceted Approach to NBS Education Equitable Access to Newborn Screening Education**
  Marianna H. Raia, MS, CGC, Expecting Health
- **Experiences of Uncertainties Among Parents of Children Diagnosed with SCID Through Newborn Screening**
  Melissa Raspa, PhD, RTI International
- **Beyond the Screen: Into a System of Care**
  Molly Martzke, Expecting Health

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.

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

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.

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

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

1:30–3:00 pm

Public Engagement

588-842-21 • 1.5 contact hour

Moderators: Lani Culley, MPH, Washington State Department of Health and Fran Altmaier, BSW, Arizona Department of Health Services

This session will explore how outreach to newborn screening stakeholders through social media, research studies, and other methods of engagement can create positive impacts for recruitment, screening implementation and parental communication.

- **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)**
  - *Amy Gaviglio, MS, CGC, CDC/APHL/Expecting Health*
  - *Aaron J. Goldenberg, PhD, MPH, Case Western Reserve University*
  - *Natasha Bonhomme, Expecting Health at Genetic Alliance*
  - *Beth Tarini, MD, Children's National Hospital*
- **Lack of Documentation of Sickle Cell Trait and Alpha Thalassemia Trait Within the Electronic Health Record: An Issue of Health Equity and Data Integrity**
  Corinna Schultz, MD, Nemours/AI duPont Hospital for Children

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

12:00–5:00 pm

Exhibit and Poster Hall Open

1:00–2:00 pm

Break / Visit the exhibits and posters

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 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.

12:00–4:00 pm

Exhibit and Poster Hall open

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 asympomatic 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.

Innovate! Sessions

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

Presented 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

Presented 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?

Presented 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*

Exhibitors

Thank you to our exhibitors and sponsors for joining us and supporting this symposium!

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Agilent is a leader in life sciences, diagnostics and applied chemical markets. The company provides laboratories worldwide with instruments, services, consumables, applications and expertise, enabling customers to gain the insights they seek. Agilent's expertise and trusted collaboration give them the highest confidence in our solutions.

Assn of Public Health Laboratories

Newborn Screening and Genetics Program

8515 Georgia Ave., Suite 700
Silver Spring, MD 20910
240.485.2726

www.newsteps.org

The APHL Newborn Screening and Genetics Program offers symposiums, workshops, webinars, access to education resources and more. Additionally, the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs) provides data, technical assistance and training to newborn screening programs across the country, with the Continuous Quality Improvement program providing newborn screening programs with the essential tools and techniques needed to successfully implement continuous quality improvement initiatives and champion an improvement culture.

Astoria-Pacific

PO Box 830
Clackamas, OR 97015
503.657.3010

www.astoria-pacific.com/spotcheck/

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

Baebies, Inc.

PO Box 14403
Durham, NC 27709
919.328.8348

www.baebies.com

Baebies, guided by the vision that "everyone deserves a healthy start," delivers innovative products and services for newborn screening and pediatric testing. SEEKER® is the first newborn screening platform authorized by the U.S. FDA for lysosomal storage disorders, and we continue to develop new solutions for the future of children's health everywhere.

Bio-Rad Laboratories

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4000 Alfred Nobel Dr.
Hercules, CA 94547
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.

BSD Robotics

Unit 17, 18 Hinkler Court
Brendale, QLD 4503 Australia
61.07.3881.1834

www.bsdrobotics.com

BSD Robotics proudly designs, manufactures, distributes and services the current BSD Punchers product range, offering 2, 4, 6 and 9 plate solutions for newborn screening laboratories. BSD Punchers are computer-controlled punch instruments used to punch any dried biosample on filter paper into standard 96-well, PCR, deep well plates and tubes in preparation for further downstream processing. BSD Punchers are designed to automate sample preparation and traceability, increase throughput, while ensuring compliance with validated laboratory protocols through worklist files. The BSD product range has an established reputation for high quality and reliability and has provided accurate and fast sample punching to newborn screening laboratories worldwide for over 25 years.

Cambridge Isotope Laboratories, Inc.

3 Highwood Dr.
Tewksbury, MA 01876
978.749.8000

www.isotope.com

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

Clinical and Laboratory Standards Institute

PO Box 633
Annapolis Junction, MD 20701
610.688.0100

www.clsi.org

CLSI is a not-for-profit standards development organization with over 24,000 members, 1,200 volunteers, and 250+ products. CLSI standards are recognized globally by laboratories, accreditors, and government agencies as the best way to improve medical laboratory testing. CLSI facilitates the creation of best practice standards, guidelines, and products for medical laboratories.

Everylife Foundation for Rare Diseases

1012 14th St. NW, Suite 500
Washington, DC 20005
202.697.7273

www.everylifefoundation.org

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

Expecting Health

4301 Connecticut Ave., NW
Suite 404
Washington, DC 20008

www.expectinghealth.org

Born from Genetic Alliance, a nonprofit organization rooted in 33 years of community programs and representing national voices and family-centered experiences, Expecting Health shares science-based and policy-informed information that reflects the lived experiences of individuals and their families. With a focus on pregnancy and newborn health, we utilize principles of community engagement and user driven design to reach diverse audiences. Expecting Health has a rich history of bringing parents, families, and other stakeholders to the table to inform and lead quality projects; empowering people to make the right choices for them using clear, accessible, and accurate information.

Luminex Corporation

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www.luminexcorp.com

Luminex offers flexible solutions for hospitals, reference labs, and researchers, as well as innovative xMAP® and flow cytometry solutions that span a wide variety of applications, including molecular diagnostics, drug discovery, life science research, immunology, and personalized medicine.

OZ Systems

Silver Sustaining Member

2201 East Lamar Blvd., Suite 280
Arlington, TX 76006
214.616.4321

www.ozsystems.com

OZ Systems is an innovator in global screening solutions, bridging crucial information gaps to ensure quality care and timely interventions for patients and those who care for them. We develop end-to-end newborn screening software that reduces human errors and improves patient safety, quality of care and intervention timeliness. We are recognized for interoperability and innovation in design and partnerships. OZ Systems has projects with US and international healthcare providers and public health agencies.

Moderna

200 Technology Square
Cambridge, MA 02139
866.663.3762

www.modernatx.com

Welcome to Moderna. We believe mRNA is the "software of life." Since our founding in 2010, we have worked to build the industry's leading mRNA technology platform, the infrastructure to accelerate drug discovery and early development, a rapidly expanding pipeline, and a world-class team. Our pipeline includes development candidates for mRNA-based vaccines and therapies spanning several therapeutic areas, and we have multiple clinical trials underway with other development candidates progressing toward the clinic. In addition, we have numerous discovery programs advancing toward development.

Exhibitors

PerkinElmer

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710 Bridgeport Ave.
Shelton, CT 06484
203.402.1994

www.perkinelmer.com

PerkinElmer is a global leader committed to innovating for a healthier world. Our dedicated team of about 13,000 employees worldwide is passionate about helping customers work to create healthier families, improve the quality of life, and sustain the wellbeing and longevity of people globally. Our innovative detection, imaging, informatics and service capabilities, combined with deep market knowledge and expertise, help customers gain earlier and more accurate insights to improve lives and the world around us.

QIAGEN

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19300 Germantown Rd.
Germantown, MD 20874
240.805.4806

www.qiagen.com

Every day, QIAGEN serves over 500,000 customers globally, all seeking insights from the building blocks of life — DNA and RNA. Delivering Sample to Insight solutions for molecular testing, we enable scientists and clinicians to achieve breakthroughs in life sciences research, molecular diagnostics and drug development. We make improvements in life possible.

Quantabio

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100 Cummings Center, Suite 407J
Beverly, MA 01915
978.869.1505

www.quantabio.com

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

Travere Therapeutics

3611 Valley Center Dr., Suite 300
San Diego, CA 92130
888.969.7879

www.travere.com

At Travere Therapeutics, we are in rare for life. We are a biopharmaceutical company that comes together every day to help patients, families and caregivers of all backgrounds as they navigate life with a rare disease. On this path, we know the need for treatment options is urgent—that is why our global team works with the rare disease community to identify, develop and deliver life-changing therapies. In pursuit of this mission, we continuously seek to understand the diverse perspectives of rare patients and to courageously forge new paths to make a difference in their lives and provide hope—today and tomorrow.

Ultragenyx Pharmaceuticals, Inc.

60 Leveroni Court
Novato, CA

www.ultragenyx.com

Ultragenyx is a biopharmaceutical company committed to bringing novel therapies to patients for the treatment of serious rare and ultra-rare genetic diseases. The company has built a diverse portfolio of approved medicines and treatment candidates aimed at addressing diseases with high unmet medical need and clear biology, for which there are typically no approved therapies treating the underlying disease. Ultragenyx's strategy is predicated upon time- and cost-efficient drug development, with the goal of delivering safe and effective therapies to patients with the utmost urgency.

Waters Corporation

Platinum Sustaining Member

34 Maple St.
Milford, MA 01757
508.482.4000

www.waters.com

Waters creates business advantages for laboratory-dependent organizations by delivering scientific innovations to enable customers to make significant advancements. Waters helps customers make profound discoveries, optimize laboratory operations, deliver product performance and ensure regulatory compliance with a connected portfolio of separations and analytical science, laboratory informatics, mass spectrometry and thermal analysis.