

APHL Newborn Screening Symposium 2020



october 20 – november 12 *virtual conference*

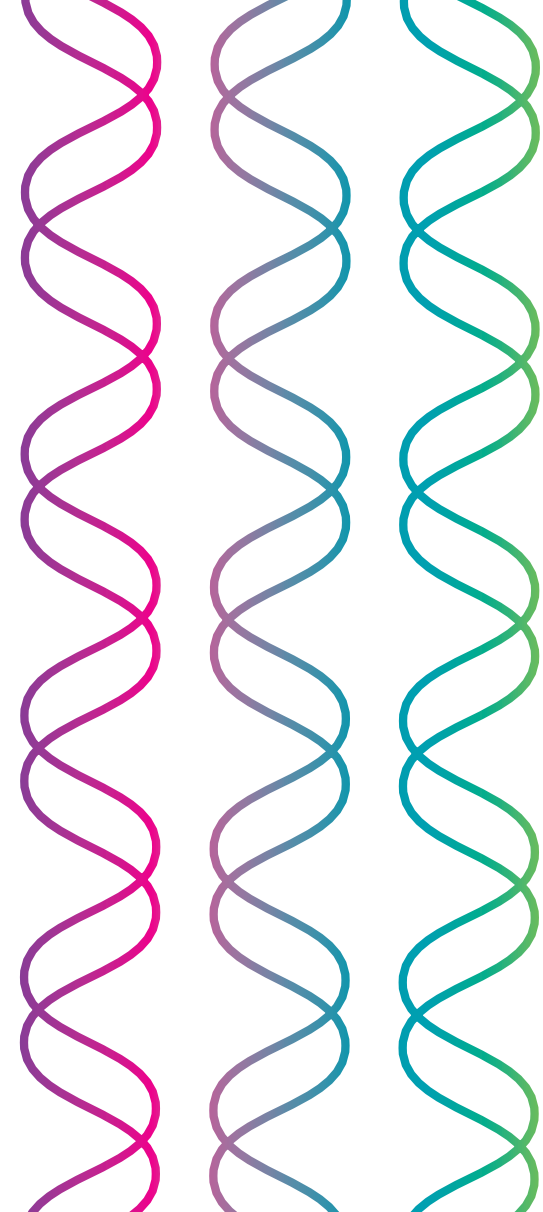
Final Program



Sponsored by the
Association of Public Health Laboratories

Co-Sponsored by the
International Society for Neonatal Screening and the
State Hygienic Laboratory at The University of Iowa

www.aphl.org/NBS2020
[#APHLNBS](https://twitter.com/APHLNBS)



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About This Conference

The 2020 APHL Newborn Screening Virtual Symposium (APHL NBS 2020) will address state, national and international newborn screening, genetic testing and policy issues important to public health newborn screening systems. The meeting will emphasize reports from around the globe, the challenges they face, and the data they have generated, as done in previous years. Topics include molecular technologies, current and upcoming conditions, quality improvement, communicating with families and the public and short- and long-term follow-up.

The program includes poster and platform presentations drawn from the submitted abstracts, invited oral presentations, exhibits and a Meet the Manufacturers session. Input and participation by parents and advocacy organizations is 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.

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. Go to the conference webpage www.aphl.org/NBS2020 for details. If you have any questions, please contact Terry Reamer at terry.reamer@aphl.org or 240.485.2776. *After registering you will receive a second email with log in information for the virtual symposium platform. The platform works best in the Chrome browser.*

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 30 contact hours by attending the entire conference.

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 20 hours of credit by attending the entire conference. APHL will not issue certificates of attendance.

APHL has been approved to offer continuing medical education (CME), nursing (CNE) and health education (CHES) credits and genetic counseling continuing education units (CEUs). Click for [more information](#).

Consent to Use Photographic Images

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Posters

More than 50 posters will be displayed on the symposium platform where live contact with the presenters will be available. Poster abstracts are available for previewing on the symposium website.

APHL 2020 Newborn Screening Virtual Symposium
October 20 – November 12, 2020
www.aphl.org/NBS2020

Not to Miss!

All listed times are Eastern.



Exhibits and Poster Hall

11:00 am – 4:30 pm ET

Tuesday, October 20

Thursday, October 22

11:00 – 4:00 pm ET

Tuesday, October 27

Thursday, October 29

Monday, November 2

Wednesday, November 4

Tuesday, November 10

Exhibitors and posters will be open and available for viewing throughout the virtual conference. Exhibitors will be available during both afternoon breaks; be sure to visit them 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.

For listings, go to page 15.

Connection Contest

Visit the exhibitors and win prizes like air pods, gift cards and airline tickets. See this [flyer](#) for the rules and details!

Meet the Manufacturers

Tuesday, October 27

3:30 pm – 4:00 pm ET

Light-hearted presentations from vendors.

Dance Party with DJ Keelez

Thursday, October 29

4:00 pm – 5:00 pm ET

Join us for a virtual dance party with DJ Keelez! You'll have the option to be on camera for a dance off or simply sit back, relax and enjoy some fun tunes with friends. Family members, roommates and pets welcome!

Innovate! Sessions

Monday, November 2

3:30 pm – 4:30 pm ET

Connect with your industry partners and learn of new technologies and services. See page 11 for details.

Trivia Showdown!

Monday, November 2

4:30 pm – 5:15 pm ET

“Wine” down with your favorite beverage and play a game – or two – of trivia with your newborn screening friends!

Roundtable Discussions

(concurrent)

Thursday, October 22

3:30 pm – 4:30 pm ET

- Towards Uniformity in the Screening and Reporting of α - and β -Thalassemia: A Discussion of Current Variations in Practice in the Era of Clinical and Laboratory Standards Institute Guidelines
- Introducing the Newborn Screening Follow-up Learning EXchange (NBS FLEX) Program
- New Disorder Implementation: Finding Solutions to Old and New Challenges

Wednesday, November 4

3:30 pm – 4:30 pm ET

- Harmonization Needs in Newborn Screening Programs: From Lab to Follow-Up
- Online Training for Communicating Initial Newborn Screening Results

11:00 am • Tuesday Oct 20
Opening Keynote Session

This live session features current and future implementation activities of newborn screening in the context of new disorders, new technology and bioethics.

Agenda at a Glance

All listed times are Eastern.

Tuesday October 20	Thursday October 22	Tuesday October 27	Thursday October 29	Monday November 2	Wednesday November 4	Tuesday November 10	Thursday November 12
11:00 am Session 1	11:00 am Session 1	11:00 am Session 1	11:00 am Session 1	11:00 am Session 1	11:00 am Session 1	11:00 am Session 1	11:00 am Session 1
12:30 pm Break	12:30 pm Break	12:30 pm Break	12:00 pm Break	12:30 pm Break	12:30 pm Break	12:30 pm Break	12:30 pm Break
1:30 pm Session 2	1:30 pm Session 2	1:30 pm Session 2	1:00 pm Session 2	1:30 pm Session 2	1:30 pm Session 2	1:30 pm Session 2	1:30 pm Session 2
3:00 pm Break	3:00 pm Break	3:00 pm Break	2:30 pm Break	3:00 pm Break	3:00 pm Break	3:00 pm Break	2:30 pm Break
3:30 pm Session 3	3:30 pm Roundtables	3:30 pm Meet the Manufacturers	3:00 pm Session 3	3:30 pm Innovate! Sessions	3:30 pm Roundtables	3:30 pm Session 3	3:30 pm Parent/Patient Panel
			4:00 pm Dance Party	4:30 pm Trivia Showdown!			

Exhibits and Posters • 11:00 am – 4:30 pm

Exhibits and Posters • 11:00 am – 4:30 pm

Exhibits and Posters • 11:00 am – 4:00 pm

Exhibits and Posters • 11:00 am – 4:00 pm

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Exhibits and Posters • 11:00 am – 4:00 pm

Exhibits and Posters • 11:00 am – 4:00 pm (Last Day)

Exhibits and Posters • 11:00 am – 4:00 pm

Exhibits and Posters • 11:00 am – 4:00 pm

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Exhibits and Posters • 11:00 am – 4:00 pm

Exhibits and Posters • 11:00 am – 4:00 pm

10:00–10:55 am • Thursday Oct 22
Restorative Yoga

Relax and unwind while you learn how to set up your space at home for optimal relaxation.

11:00 am – 4:30 pm
Exhibit Hall and Posters Open

11:00 am – 12:30 pm

Keynote Session

P.A.C.E. #588-825-20 • 1.5 contact hours

Moderators:

- Michele Caggana, ScD, FACMG, New York State Department of Health
- Susan Tanksley, PhD, Texas Department of State Health Services

Newborn Screening: Where Are We Now and How Did We Get There?

- Susan Berry, MD, Genetics and Metabolism, Department of Pediatrics, University of Minnesota

Compulsory Population-based Newborn Screening: Challenges for the Future

- Shawn McCandless, MD, Children's Hospital Colorado Anschutz Medical Campus

New Screening, Old Ethics: Is It Time to Re-examine the Ethical Justification for Compulsory Newborn Screening?

- Aaron Goldenberg, PhD, MA, MPH, Department of Bioethics & Medical Humanities, Case Western Reserve University School of Medicine

This live session features current and future implementation activities of newborn screening in the context of new disorders, new technology and bioethics.

12:30 pm – 1:30 pm
Break / Visit the Exhibit Hall and Posters

1:30 pm – 3:00 pm

COVID-19 Update

P.A.C.E. #588-826-20 • 1.0 contact hour

Moderators:

- Pranesh Chakraborty, MD, FRCPC, FCCMG, Newborn Screening Ontario
- John Thompson, PhD, MPH, MPA, Washington State Public Health Laboratories

Speakers:

- Michele Caggana, ScD, FACMG, New York State Department of Health – Wadsworth Center
- Rachel Lee, PhD, Texas Department of State Health Services
- Carol Johnson, University of Iowa Hospitals and Clinics
- Ashley Ramirez, State Hygienic Laboratory at the University of Iowa
- Adrienne Manning, Connecticut Department of Public Health Laboratory
- Mary Kleyn, MS, Michigan Department of Health & Human Services
- Ashleigh Ragsdale, MPH, Washington State Department of Health

This live session will highlight the activities that several newborn screening programs have undertaken to ensure that essential operations continue, and newborns are continually screened and treated during these extraordinary times.

3:00 pm – 3:30 pm
Break / Visit the Exhibit Hall and Posters

3:30 pm – 5:00 pm

The Future Is Now!

P.A.C.E. #588-827-20 • 1.5 contact hours

Moderators:

- Chenelle Norman, MPH, Association of Public Health Laboratories
- Cindy Hinton, PhD, MS, MPH, Centers for Disease Control and Prevention

The State of Newborn Screening Systems in the United States in 2020

- Careema Yusuf, MPH, Association of Public Health Laboratories

The Newborn Screening Translational Research Network (NBSTRN): A Program of the National Institute of Child Health and Human Development to Expand Newborn Screening and Improve the Understanding of Rare Disease

- Amy Brower, PhD, American College of Medical Genetics and Genomics

Bringing Together Newborn Screening Stakeholders

- Dylan Simon, MS, EveryLife Foundation for Rare Diseases

I Screened, You Screened, We All Screened: A Newborn Screening Student Event at the Virginia State Lab

- Jennifer Macdonald, BSN, RN, MPH, Virginia Department of Health

This session will examine the state of newborn screening (NBS) in the United States today, how the Newborn Screening Translational Research Network (NBSTRN) is expanding NBS and improving the understanding of rare disease, as well as how students can learn about the complexities of the NBS system.

10:00 am – 10:55 am

Restorative Yoga

Relax and unwind during this restorative yoga session. You will learn about the benefits of restorative yoga and how to set up your space at home for optimal relaxation. Come prepared with a few blankets and books and get ready to embrace your inner Zen!

11:00 am – 4:30 pm

Exhibit Hall and Posters Open

11:00 am – 12:30 pm

Molecular Technology

P.A.C.E. #588-828-20 • 1.5 contact hours

Moderators:

- Rachel Lee, PhD, Texas Department of State Health Services
- Travis Henry, PhD, State Hygienic Laboratory at the University of Iowa

Genetic Sequencing Detects Two Phenylalanine Hydroxylase Variants in Over Half of Infants with False Positive MS/MS Screens for Phenylketonuria

- Aashish Adhikari, PhD, University of California, San Francisco

Discordant Results for a Variant's Genotype and Investigation of Allelic Dropout: Experience with Newborn Screening for MPS-I

- Binod Kumar, PhD, University of Massachusetts Medical School

Improvements to the New York State IRT-DNA-SEQ Cystic Fibrosis Newborn Screening Algorithm: Results from the First Two Years

- Denise Kay, PhD, New York State Department of Health

Whole-exome Sequencing Pipeline for Newborn Screening Disorders: Targeted, but Disorder and Gene Agnostic

- Nicole Ruiz-Schultz, MS, Utah Department of Health

This session will summarize the types of testing — including gene sequencing for Cystic Fibrosis (CF) and next generation sequencing (NGS) — used to decrease the likelihood of false positives and improve testing for newborn screening and beyond.

12:30 pm – 1:30 pm

Break / Visit the Exhibit Hall and Posters

1:30 pm – 3:00 pm

Education and Communication

P.A.C.E. #588-829-20 • 1.5 contact hours

Moderators:

- Carol Johnson, University of Iowa Hospitals & Clinics
- Erica Wright, MS, CGC, Children's Hospital Colorado

Survey of Prenatal Care Provider Practices Regarding Newborn Screening Education

- Mary Kleyn, MS, Michigan Department of Health and Human Services

Charting a Course for Family Education: How We Created Navigate Newborn Screening

- Natasha Bonhomme, Chief Strategy Officer at Genetic Alliance

Beyond the Fax Machine: Alternative Tools for Improved Communication and Timeliness in Follow-up

- Christen Crews, MSN, RN, Virginia Department of Health

Using AAP's Maintenance of Certification Program to Improve Communication of Newborn Screening Results to Parents

- Amy Gaviglio, MS, CGC, G2S/CDC/APHL

Evaluating Consumer Questions to Improve Communication and Collaboration in Newborn Screening

- Brianna Miller, MPH, Expecting Health at Genetic Alliance

This session will explore how to better educate and engage parents within the newborn screening system as well as ways to improve communication and timeliness around follow-up.

3:00 pm – 3:30 pm

Break / Visit the Exhibit Hall and Posters

3:30 pm – 4:30 pm

Roundtable Discussions

(3 concurrent sessions)

See next page for details.

Thursday, October 22

(continued)

3:30 pm – 4:30 pm

Roundtable Discussions

(concurrent)

Towards Uniformity in the Screening and Reporting of α - and β -Thalassemia: A Discussion of Current Variations in Practice in the Era of Clinical and Laboratory Standards Institute Guidelines

P.A.C.E. #588-830-20 • 1.0 contact hour

- M. Bender, MD, University of Washington

This roundtable will present data from surveys of all US newborn screening (NBS) programs regarding methodology and reporting of α - and β -thalassemia. This interactive roundtable will focus on defining barriers to, interest in, and resources/partners for working towards more uniform screening and reporting, and to discuss if the newly released Clinical and Laboratory Standards Institute (CLSI) guidelines for NBS for Hemoglobinopathies will lead to alterations in their practices.

Introducing the Newborn Screening Follow-up Learning EXchange (NBS FLEX) Program

P.A.C.E. #588-831-20 • 1.0 contact hour

- John Thompson, PhD, MPH, MPA, Washington State Public Health Laboratories
- Carol Johnson, University of Iowa Hospitals and Clinics

Roundtable presentation to present the NBS FLEX program, a grant from the NewSTEPS program to facilitate virtual and in-person technical assistance between follow-up staff in different programs. The goal is to match programs experiencing challenges with mentors from programs with relevant expertise.

New Disorder Implementation: Finding Solutions to Old and New Challenges

P.A.C.E. #588-832-20 • 1.0 contact hour

- George Dizikes, PhD, Tennessee Department of Health

The roundtable will provide attendees with a brief overview of disorders recently added to the Recommended Uniform Screening Panel (RUSP). The remainder of the roundtable will ask participants to choose one of the following topics (Molecular Testing, Tiered Testing and Timeliness, Result Notification and Pre-Diagnostic Follow-Up) for in-depth conversations focused on learning about or brainstorming solutions to common challenges programs face when implementing and maintaining laboratory-based screening for disorders recently added to the RUSP.

11:00 am – 4:00 pm
Exhibit Hall and Posters Open

11:00 am – 12:30 pm

Spinal Muscular Atrophy (SMA)

P.A.C.E. #588-833-20 • 1.5 contact hours

Moderators:

- Suzanne Cordovado, PhD, Centers for Disease Control and Prevention
- Kshea Hale, MPH, Association of Public Health Laboratories

Spinal Muscular Atrophy: Pilot Screening Project in Georgia

- Angela Wittenauer, BSN, RN, Emory University

Michigan's Experience with Implementing Spinal Muscular Atrophy by Multiplexing with the Established Severe Combined Immunodeficiency Assay

- Becky Shaulis, MS, Michigan Department of Health and Human Services

Implementation of Population-Based Newborn Screening for Spinal Muscular Atrophy (SMA) in New York State

- Colleen Stevens, PhD, New York State Department of Health

Newborn Screening for Spinal Muscular Atrophy: Wisconsin Experience

- Mei Baker, MD, FACMG, Wisconsin State Laboratory of Hygiene

This session will examine the implementation and results of initial newborn screening for Spinal Muscular Atrophy (SMA) in multiple states.

12:30 pm – 1:30 pm
Break / Visit the Exhibit Hall and Posters

1:30 pm – 3:00 pm

Current Recommended Uniform Screening Panel (RUSP)

P.A.C.E. #588-834-20 • 1.5 contact hours

Moderators:

- Funke Akinsola, MPH, Association of Public Health Laboratories
- Scott Shone, PhD, HCLD(ABB), North Carolina State Laboratory of Public Health

Age of Intervention and Diagnosis for Cystic Fibrosis at CF Care Centers following Newborn Screening in the US

- Sarah McKasson, MPH, Center for Public Health Innovation at CI International

SUAC All Over the Place: The Kansas Experience for Determining a Suitable Cutoff for Succinylacetone to Screen for Tyrosinemia Type I

- Michelle Mills, MSFS, Kansas Health and Environmental Laboratories

30 Seconds per Specimen Mass Spectrometry Based First Tier Newborn Screening with Intelligent Reflex to Second Tier Screening

- Samantha Isenberg, PhD, Centers for Disease Control and Prevention

Long-term Follow-up of Children with Central Hypothyroidism

- Beth Vogel, MS, CGC, New York State Department of Health

This session will explore different methods of testing for disorders on the current Recommended Uniform Screening Panel (RUSP), including cystic fibrosis (CF), Tyrosinemia Type I and Central Hypothyroidism.

3:00 pm – 3:30 pm

Break / Visit the Exhibit Hall and Posters

3:30 pm – 4:00 pm

Meet the Manufacturers!

Light-hearted presentations from:

3:30 pm Baebies
3:45 pm PerkinElmer

11:00 am – 4:00 pm
Exhibit Hall and Posters Open

11:00 am – 12:00 pm

New Disorders

P.A.C.E. #588-835-20 • 1.0 contact hour

Moderators:

- George Dizikes, PhD, Tennessee Department of Health: Laboratory Services
- Amy Gaviglio, MS, CGC, G2S/CDC/APHL

Screening for Lysosomal Storage Disorders in Virginia – A Year in Review

- Leigh-Emma Lion, Virginia Division of Consolidated Laboratory Services

Newborn Screening for X-ALD in Texas

- Debra Freedenberg, MD, Texas Department of State Health Services

Newborn Screening for X-ALD: Massachusetts Experience

- Inderneel Sahai, MD, New England Newborn Screening Program

The session will examine the challenges and solutions of screening for new disorders, including Lysosomal Storage Disorders and X-Linked Adrenoleukodystrophy (X-ALD).

12:00 pm – 1:00 pm

Break / Visit the Exhibit Hall and Posters

1:00 pm – 2:30 pm

Quality Improvement

P.A.C.E. #588-836-20 • 1.5 contact hours

Moderators:

- Joanne Mei, PhD, Centers for Disease Control and Prevention
- Ashley Ramirez, State Hygienic Laboratory at the University of Iowa

Method Validation of Mass Spectrometry Based Newborn Screening Assays

- Samantha Isenberg, PhD, Centers for Disease Control and Prevention

Evaluation of MSMS Algorithm and Cutoff Value Changes in Georgia

- Patricia Hall, PhD, Emory University

New York State Newborn Screening Pilot Quality Improvement (QI) Initiative for Endocrine Specialty Care Centers

- Kathy Chou, PhD, New York State Department of Health

Multi-state Harmonization Study: Efforts to Harmonize the Cut-offs Used in Newborn Screening for Adrenoleukodystrophy

- Joseph Orsini, PhD, New York State Department of Health

This session will explore the importance of implementing different screening methods in order to continuously ensure accuracy, timeliness and quality within the newborn screening system.

2:30 pm – 3:00 pm

Break / Visit the Exhibit Hall and Posters

3:00 pm – 4:00 pm

Quality Assurance Systems

P.A.C.E. #588-837-20 • 1.0 contact hour

Moderators:

- Cindy Hinton, PhD, MS, MPH, Centers for Disease Control and Prevention
- Fran Altmaier, BSW, Arizona Department of Health Services

For You By You: An Update on CLSI Products for Newborn Screening Programs

- Amy Gaviglio, MS, CGC, G2S/CDC/APHL

The Newborn Screening Quality Assurance Program Participant Portal: Modernization and Improvements for Proficiency Testing and Quality Control Results and Evaluations

- Joanne Mei, PhD, Centers for Disease Control and Prevention

This session will summarize the Clinical and Laboratory Standards Institute's (CLSI) guidelines for professionals and examine how the Newborn Screening Quality Assurance Program (NSQAP) has created efficiencies within newborn screening laboratories, specifically in light of changing quality assurance needs.

4:00 pm – 5:00 pm

Virtual Dance Party with DJ Keelez

Join us for a virtual dance party with DJ Keelez! You'll have the option to be on camera for a dance off or simply sit back, relax and enjoy some fun tunes with friends. Family members, roommates and pets welcome!



11:00 am – 4:00 pm
Exhibit Hall and Posters Open

11:00 am – 12:30 pm

Congenital Cytomegalovirus (cCMV)

P.A.C.E. #588-838-20 • 1.5 contact hours

Moderators:

- Kimberly Noble Piper, BSN, RN, CPH, CPHG, Iowa Department of Public Health
- Carrie Wolf, MBS, Minnesota Department of Health

Panel: Congenital Cytomegalovirus Universal Newborn Screening

Planning and Implementation Challenges of Including Congenital Cytomegalovirus in Universal Newborn Screening

- Lisa Gehtland, MD, RTI International

Universal Screening for Permanent Hearing Loss Risk Factors: Policy Considerations Leading to Implementation in Ontario

- Pranesh Chakraborty, MD, FRCPC, FCCMG, Newborn Screening Ontario

Universal Newborn Screening for Congenital Cytomegalovirus and Genetic Variants Associated with Permanent Hearing Loss: Preliminary Findings from Initial 5 Months of Screening in Ontario, Canada

- Lauren Gallagher, MS, Newborn Screening Ontario

Improvements in the Analytic Sensitivity of Dried Blood Spot (DBS) PCR for the Diagnosis of Congenital Cytomegalovirus (cCMV) Infection: Results from a Minnesota Universal Screening Study

- Mark Schleiss, MD, University of Minnesota Medical School

Education of Providers and Parents on the Prevention, Detection and Treatment of Congenital Cytomegalovirus

- Marcia Fort, AuD, CCC-A, North Carolina Division of Public Health

This session will explore the challenges that Congenital Cytomegalovirus (cCMV) poses to the newborn screening system and summarize ways to educate healthcare providers and the public on the importance of early detection and treatment of cCMV.

12:30 pm – 1:30 pm

Break / Visit the Exhibit Hall and Posters

1:30 pm – 3:00 pm

Health Services Research

P.A.C.E. #588-839-20 • 1.5 contact hours

Moderators:

- Beth Tarini, MD, Children's National Hospital
- Sikha Singh, MHS, PMP, Association of Public Health Laboratories

The Value of Cognitive Testing of a NBS Survey: Revealing Blind Spots and Improving Validity

- Norma-Jean Simon, MPH, Children's National Hospital

Panel: Partnering with NBS Programs to Conduct Health Services Research: Programmatic Considerations, Research Regulations, and Study Design

Building Study Partnerships with NBS Programs: Key Considerations for Successful Collaborations

- Amy Gaviglio, MS, CGC, G2S/CDC/APHL

How to Successfully Navigate Research Regulatory Requirements When Conducting NBS Health Services Research

- Anne Atkins, MPH, Children's National Hospital

Integrating Professional and Public Input into Health Services Research Study Design: Challenges and Lessons Learned

- Brianne Miller, MPH, Children's National Hospital

This live session will illustrate how to construct a survey for maximum usability and examine the effectiveness of the data results. Ways in which researchers can identify gaps in knowledge by partnering with newborn screening programs will also be addressed.

3:00 pm – 3:30 pm

Break / Visit the Exhibit Hall and Posters

3:30 pm – 4:30 pm

Innovate! Sessions

Vendor presentations from PerkinElmer and Agilent Technologies. See next page for details.

4:30 pm – 5:15 pm

Trivia Showdown!

“Wine” down with your favorite beverage and play a game — or two — of trivia with your Newborn Screening friends!

Monday, November 2
3:30–4:30 pm

New Advances in Metabolomics for Cystic Fibrosis: Mechanistic Insights of Affected Infants Early in Life

Presented by Agilent Technologies

Cystic fibrosis (CF) is among the most common life-shortening genetic disorders that benefits from early detection. Newborn screening programs for CF rely on genetic testing that is suboptimal as it identifies unaffected carriers in the population. We will discuss new advances in clinical metabolomics research using high throughput capillary electrophoresis-mass spectrometry technology for the identification of biomarkers of CF from dried blood spot and sweat specimens for new insights into disease mechanisms early in life.

For Research Use Only. Not for use in diagnostic procedures.

Speaker: Philip Britz-McKibbin, Professor and Cystic Fibrosis Canada Researcher, Department of Chemistry and Chemical Biology, McMaster University, Hamilton, ON Canada

Monday, November 2
3:30–4:30 pm

Dried Blood Spot Sample Newborn Screening and Beyond – New Assay for Duchenne Muscular Dystrophy and Understanding the Spread of SARS-CoV-2 in Populations

Presented by PerkinElmer

Dried blood spot sample enables large population based screening due to ease of sampling, sample process and shipment. PerkinElmer's high-throughput GSP Instrument is widely used in newborn screening for a variety of disorders including, most recently Duchenne Muscular Dystrophy. Now SARS-CoV-2 IgG antibodies can be easily measured from dried blood spot samples on the GSP instrument in order to understand how the COVID-19 epidemic is spreading in populations and to help with global vaccine development.

Products may not be licensed in accordance with the laws in all countries. Please check with your local representative for availability.

Speaker: Hanna Polari, Global Business Manager, PerkinElmer



11:00 am – 4:00 pm
Exhibit Hall and Posters Open

11:00 am – 12:30 pm

Follow-up

P.A.C.E. #588-840-20 • 1.5 contact hours

Moderators:

- Sarah Bradley, MS, CGC, New York State Department of Health
- Jennifer Macdonald, BSN, RN, MPH, Virginia Department of Health

Making the Case for Newborn Screening Follow-Up Guidelines

- Carol Johnson, University of Iowa Hospitals & Clinics

Assessing Primary Care Physician's Use of the High Immunoreactive Trypsinogen Letter

- Isabel Hurden, MPH, Michigan Department of Health and Human Services

Implementation of Pompe and MPS-I in Virginia: One-Year Review

- Christen Crews, MSN, RN, Virginia Department of Health

Outcomes of Newborn Screening for Adrenoleukodystrophy in New York State

- Beth Vogel, MS, CGC, New York State Department of Health

Facilitating Rare Genetic Disease Research: Methods for CDE Development to Support the Longitudinal Follow-Up of Individuals Diagnosed Through Newborn Screening

- Mike Hartnett, American College of Medical Genetics and Genomics

This session will critique follow-up practices within newborn screening, present the results of a survey sent to access primary care physician's use of high immunoreactive trypsinogen (IRT) notifications

as well as assess the outcomes of testing for Adrenoleukodystrophy (ALD), Pompe and MPS I in various states.

12:30 pm – 1:30 pm
Break / Visit the Exhibit Hall and Posters

1:30 pm – 3:00 pm

International Perspectives

P.A.C.E. #588-841-20 • 1.5 contact hours

Moderators:

- James Bonham, MSc, PhD, CSci, FRCPath, UK Newborn Screening
- Joanne Mei, PhD, Centers for Disease Control and Prevention

Reporting of Newborn Screening Results to International, Low-resource Settings from Canada

- Monica Lamoureux, MS, Newborn Screening Ontario

Implementation of SCID Screening in Denmark

- Marie Baekvad-Hansen, MS, Statens Serum Institut

Including Classical Galactosaemia in the Expanded Newborn Screening Panel Using Tandem Mass Spectrometry for Galactose-1-phosphate

- Arieh Cohen, PhD, Statens Serum Institut

Reduced Sensitivity for Congenital Hypothyroidism Screening in the NICU: A Question of Case Definition?

- Graham Sinclair, PhD, British Columbia Newborn Screening Program

Evaluation of Changes Implemented in 2016 in Cystic Fibrosis Newborn Screening Protocol in the Netherlands

- Marelle Bouva, MS, The Netherlands Newborn Screening Laboratory

This session will examine newborn screening systems with an international lens: the results of tests performed in Canada on babies in low-resource settings, the implementation of Severe Combined Immunodeficiency (SCID) screening in Denmark and changes made to screening for Cystic Fibrosis (CF) in the Netherlands, among others.

3:00 pm – 3:30 pm
Break / Visit the Exhibit Hall and Posters

3:30 pm – 4:30 pm

Roundtable Discussions

(2 concurrent sessions)

Harmonization Needs in Newborn Screening Programs: From Lab to Follow-Up

P.A.C.E. #588-842-20 • 1.0 contact hour

- Amy Gaviglio, MS, CGC, G2S/CDC/APHL

As newborn screening is state-based, a number of discrepancies exist in pre-analytical, analytical, and post-analytical processes and outcomes. This roundtable will be a launching point for discussion on how we can harmonize practices, especially analyte results.

Online Training for Communicating Initial Newborn Screening Results

P.A.C.E. #588-843-20 • 1.0 contact hour

- Natasha Bonhomme, Chief Strategy Officer at Genetic Alliance

This roundtable will focus on the practical application of newborn screening results communication training in states and health systems. Attendees will have the opportunity to view the training, hear data from preliminary testing, and provide feedback on the format and content. Attendees will leave with an understanding of where to find the training and what content the training covers.

11:00 am – 4:00 pm

Exhibit Hall and Posters Open (Last Day)

11:00 am – 12:30 pm

Newborn Screening Pilot Studies

P.A.C.E. #588-844-20 • 1.5 contact hours

Moderators:

- Joseph Orsini, PhD, New York State Department of Health
- Mei Baker, MD, FACMG, Wisconsin State Laboratory of Hygiene

Panel: Consented Newborn Screening Pilot Studies: Issues, Opportunities and Case Examples

Pilot Screening Studies for Newborn Screening

- Jeffrey Botkin, MD, MPH, University of Utah

Early Check: Findings and Lessons Learned from Two Years of Implementation

- Don Bailey, PhD, RTI International

ScreenPlus: A Comprehensive, Multi-Disorder, Consented Pilot Newborn Screening Program

- Melissa Wasserstein, MD, Albert Einstein College of Medicine

Duchenne Muscular Dystrophy Newborn Screening: Protocol for a Pilot in New York State

- Norma Tavakoli, PhD, New York State Department of Health

This session will explore what the future of pilot studies might look like in the United States and examine the challenges of conducting population based screening and recruiting for newborn screening pilot studies. The benefits and risks of conducting such studies will be discussed.

12:30 pm – 1:30 pm

Break / Visit the exhibit Hall and Posters

1:30 pm – 3:00 pm

Homocystinuria

P.A.C.E. #588-854-20 • 1.5 contact hours

Moderators:

- Patricia Hunt, Texas Department of State Health Services
- Kostas Petritis, PhD, Centers for Disease Control and Prevention

Homocystinuria Deserves Better Newborn Screening

- Harvey Levy, MD, Boston Children's Hospital/ Harvard Medical School

Homocystinuria Screening: Michigan's Experience

- Shawn Moloney, MPH, Michigan Department of Health and Human Services

Newborn Screening for Homocystinurias Using Methionine as a Marker: New England Experience

- Inderneel Sahai, MD, New England Newborn Screening Program

Opportunities and Challenges of Multiplexing Homocystinuria into Newborn Screening Assays

- Austin Pickens, PhD, Centers for Disease Control and Prevention

Wisconsin's Screening Algorithm for Homocystinuria: Incorporation of Second-Tier Testing

- Patrice Held, PhD, Wisconsin State Laboratory of Hygiene

This session will examine the challenges associated with screening for Homocystinuria (HCU) and why many newborns are often missed. It will present the experiences of multiple states and the solutions implemented to better identify at risk patients.

3:00 pm – 3:30 pm

Break / Visit the exhibit Hall and Posters

3:30 pm – 4:30 pm

Health Information Technology and Bioinformatics

P.A.C.E. #588-845-20 • 1.0 contact hour

Moderators:

- Ashleigh Ragsdale, MPH, Washington State Department of Health
- Brendan Reilly, Texas Department of State Health Services

Bioinformatics: The Next Newborn Screening Frontier

- Samantha Marcellus, MPH, Texas Department of State Health Services

Correlation Analysis for CAH Screening: Identification of Additional Variables for Improved Risk Assessment

- Alankar Kampoowale, MS, State Hygienic Laboratory at the University of Iowa

Electronic Data Exchange: Needs and Solutions for Public Health Laboratories

- Frank Delin, State Hygienic Laboratory at the University of Iowa

This session will defend the use of bioinformatics in newborn screening and examine solutions to data exchange to create better communication across states. An evaluation of ways to improve the risk assessment for Congenital Adrenal Hyperplasia (CAH) will also be discussed.

11:00 am – 12:30 pm

Financial, Legal, Ethical, Policy and Social Implications (FLEPSI)

P.A.C.E. #588-856-20 • 1.5 contact hours

Moderators:

- Kimberly Noble Piper, RN, BSN, CPH, CPHG, Iowa Department of Public Health
- Aaron Goldenberg, PhD, MA, MPH, Department of Bioethics & Medical Humanities, Case Western Reserve University School of Medicine

Newborn Screening and Genomics: Ethics Resources and Tools to Support Patients, Parents, Public Health Partners, Clinicians and Researchers

- Kee Chan, PhD, MBA, American College of Medical Genetics and Genomics

Transitioning from Two Screens to One: Delaware's Experience

- Michael Cellucci, MD, Nemours Al DuPont Hospital for Children

The Michigan BioTrust for Health and Pregnancy Risk Assessment Monitoring System (PRAMS) Linkage: Associations Between Consent Decisions and Knowledge of the Michigan BioTrust, 2012–2015

- Shelby Atkinson, MPH, Michigan Department of Health and Human Services

This session will critique the healthcare cost burden of newborn screening on states and present solutions to communicating with families about research consent. There will also be a discussion on how genome sequencing can impact clinical and family decision-making.

12:30 pm – 1:30 pm

Break

1:30 pm – 2:30 pm

Second-tier Testing

P.A.C.E. #588-857-20 • 1.0 contact hour

Moderators:

- Kostas Petritis, PhD, Centers for Disease Control and Prevention
- Denise Kay, PhD, New York State Department of Health

Identification of Newborns with Congenital Adrenal Hyperplasia Using Principal Component Analysis

- Patrice Held, PhD, Wisconsin State Laboratory of Hygiene

A Closer Look at the Introduction of a Second-Tier Assay Used in Newborn Screening for Congenital Adrenal Hyperplasia in Texas

- Patricia Hunt, Texas Department of State Health Services

A Second-tier Newborn Screening LC-MS/MS Method for MSUD, HCY, MMA, PA, IVA, GA-I, GA-II, SCAD, GAMT, X-ALD, CAH and Pompe Disease

- Matthew Kilgore, PhD, Centers for Disease Control and Prevention

This session will cover the use of Hydrophilic Interaction (HILIC) chromatography as well as evaluate the effectiveness of using a second-tier assay and principal component analysis for identifying newborns at risk for Congenital Adrenal Hyperplasia (CAH).

2:30 pm – 3:30 pm

Break

3:30 pm – 5:00 pm

Parent/Patient Panel

P.A.C.E. #588-858-20 • 1.0 contact hour

ModeratorS:

- Amy Calhoun, MD, University of Iowa
- Erin Darby, MPH, MCHES, Association of Public Health Laboratories

Parental Experience of Medium-chain Acyl-CoA Dehydrogenase Deficiency in Colorado

- Erica Wright, MS, CGC, Children's Hospital Colorado

Panel: Parents/Patients Discuss Their Experience

- Corrine Hurst and Tyler Birkenholz
- Stephanie and Steven McDonough
- Karen and Kade Hammes
- Ashlee and Oladapo Oguntoyinbo

This live session features stories from families and their experiences with newborn screening. There will also be a presentation on the parental experience of Medium-chain Acyl-CoA Dehydrogenase Deficiency in Colorado.

Exhibitors

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