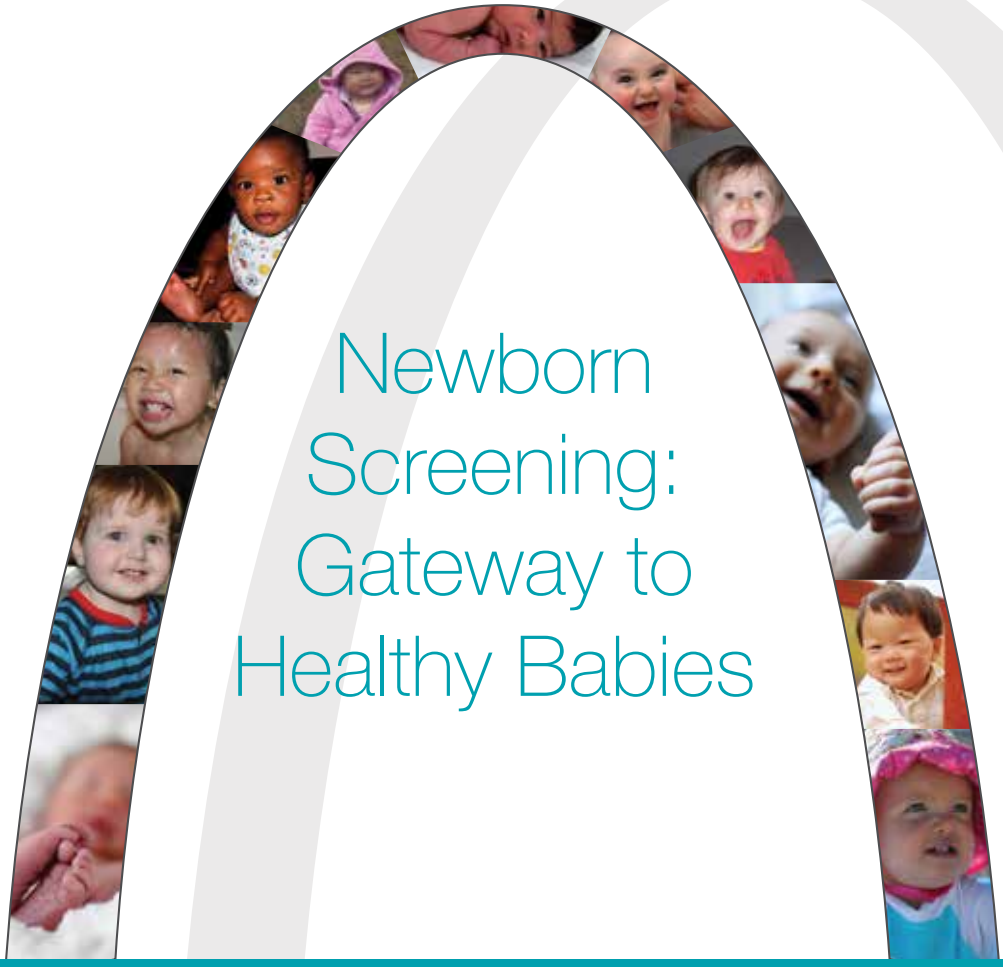


Newborn Screening: Gateway to Healthy Babies

Final Program

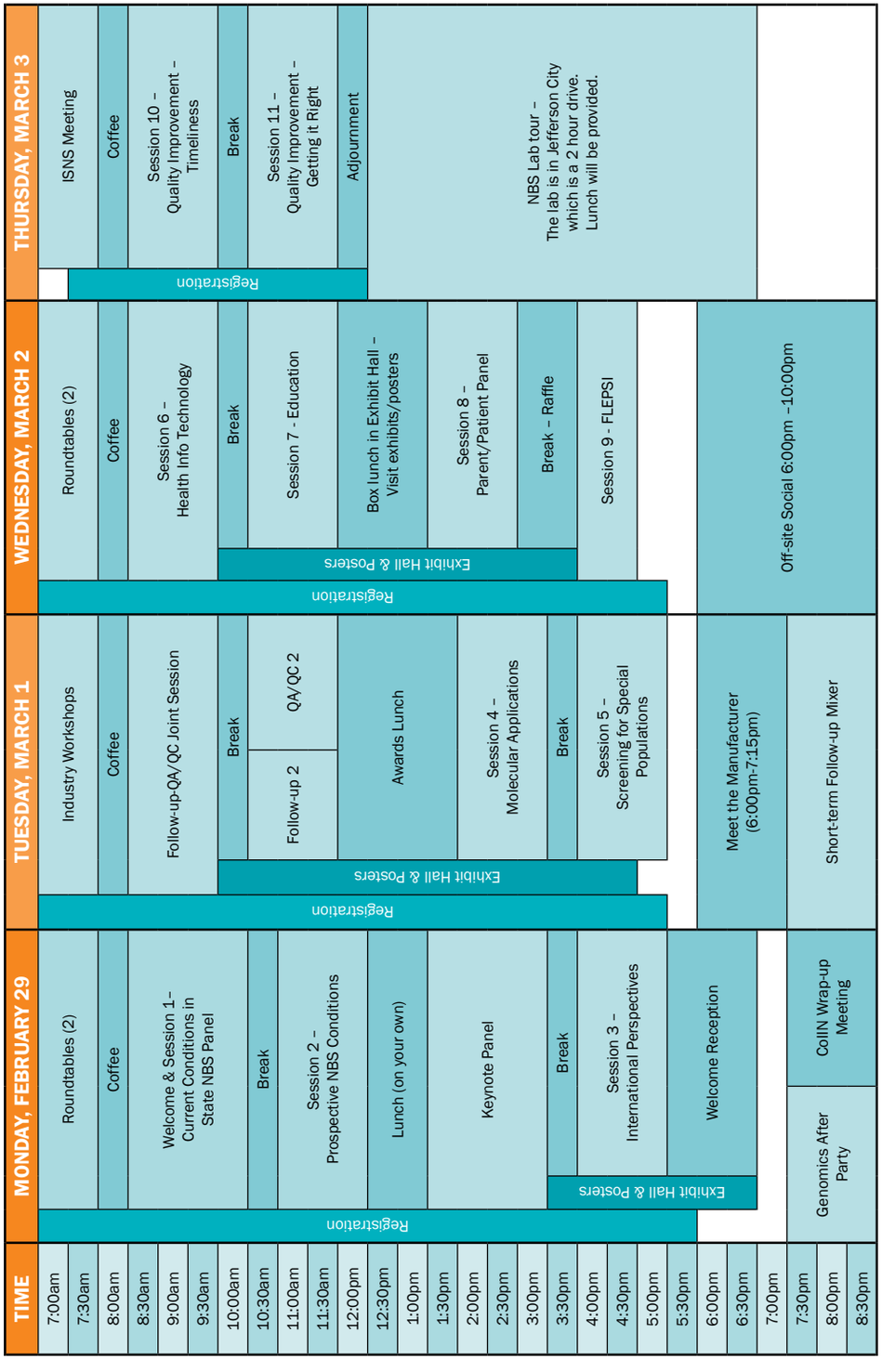
2016 APHL Newborn Screening and Genetic Testing Symposium

February 29 – March 3, 2016
St. Louis, MO

APHL

#nbsgts
www.aphl.org/nbsgts

Meeting at a Glance



Dear Friends, Colleagues and Attendees,

On behalf of the Association of Public Health Laboratories (APHL), the Missouri State Public Health Laboratory, the International Society for Neonatal Screening, and the Symposium Planning Committee, it is our great pleasure to welcome you to the 2016 APHL Newborn Screening and Genetic Testing Symposium (NBSGTS).

Welcome to the beautiful State of Missouri and to the great City of St. Louis, called the "Gateway to the West" with its magnificent Gateway Arch by the Mississippi River welcoming travelers. It is fitting that this year's theme for the symposium is "Newborn Screening: Gateway to Healthy Babies" as that is truly what newborn screening is all about. Newborn screening not only saves babies, but literally saves entire families from the devastation of many treatable conditions through early intervention. With its continual advances, expansions and successful outcomes, the entire newborn screening system continues to improve each year.

The NBSGTS provides many golden opportunities to stay abreast of the field by keeping up with new and innovating practices and discoveries, and provides many golden opportunities to keep up with new and innovating opportunities to network with others. The conference opens on Leap Day this year. Following morning roundtables and platform sessions on current and prospective conditions on state panels, we will look to the future in the afternoon with a provocative Keynote Session on current and prospective applications of genomic technologies and precision medicine in the world of newborn screening, with an ethical perspective and feedback from our audience, as well as an invited guest from National Public Radio: Dr. Joe Palca. The Exhibit Hall also opens on Monday for three days of thought-provoking poster presentations and opportunities to interact with innovative exhibitors, as well as a Meet the Manufacturers session scheduled for Tuesday.

Each day of the conference will feature original platform presentations, with two sessions on Quality Improvement as well as Quality Assurance/Quality Control and Follow-up sessions, more roundtables with opportunities to discuss cutting-edge issues in smaller groups, and other platform sessions on timely topics such as Education and Health Information Technology. Tuesday features an Awards Luncheon while the always esteemed Parent/Patient Panel takes place on Wednesday. The conference will conclude on Thursday with an opportunity to tour the Newborn Screening Laboratory Section of the Missouri State Public Health Laboratory.

We hope that you will learn useful new things while you are here that you can take back and share with your coworkers at home, and that you will meet many new colleagues from across the country and around the world, who will be part of a long lasting network of support for you throughout your career in newborn screening. We would like to thank the members of the Planning Committee for contributing their time on conference calls and abstract reviews, and we are especially thankful for the work that every one of our colleagues contributes every day and the passion that you all share in these efforts. You are the Gateway to Healthy Babies!

Sincerely,

Patrick V. Hopkins
Chief, Newborn Screening Unit
Missouri State Public Health Laboratory
MO Department of Health and Senior Services
Jefferson City, MO
Co-Chair, 2016 APHL NBSGTS Planning Committee

Richard Olney, MD, MPH
Division Chief
Genetic Disease Screening Program
California Department of Public Health
Richmond, CA
Co-Chair, 2016 APHL NBSGTS Planning Committee

The Association of Public Health Laboratories

VISION: A healthier world through quality laboratory systems.

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

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

APHL IS A NATIONAL LEADER IN:

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

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Susan Tanksley, PhD, Texas Department of State Health Services

John Thompson, PhD, MPH, MPA, Washington State Department of Health

Beth Vogel, MS, CGC, Wadsworth Center

Veronica Wiley, PhD, The Children's Hospital at Westmead, Australia

About This Conference

The 2016 APHL Newborn Screening and Genetic Testing Symposium addresses state, national and international newborn screening, genetic testing and policy issues relevant to public health. The meeting will emphasize reports from around the globe, the challenges documented and the data they have generated. Topics include new and emerging technologies, candidate conditions, common issues and solutions in newborn screening, clinical outcomes and short- and long-term follow-up.

The program will include poster and platform presentations drawn from the submitted abstracts, invited oral presentations, industry workshops, exhibits and a Meet the Manufacturers session. Input from 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 it relates to emerging laboratory technologies, follow-up, candidate conditions, quality improvement and clinical outcomes.

The major learning goals of this symposium are to:

- Discuss and evaluate quality assurance and quality control measures for newborn screening laboratories
- Evaluate the effectiveness of current newborn screening and genetics follow-up programs
- Describe state and international experiences with candidate conditions and clinical outcomes in newborn screening

Who will benefit from attending this conference?

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

General Information

The 2016 APHL NBSGTS begins with two roundtables at 7:00 am, Monday, February 29 followed by the Welcome Session at 8:30 am.

All the posters will be displayed from Monday afternoon, February 29 through 4:00 pm on Wednesday, March 2. Dedicated viewing times (with authors present) will take place during Monday's Welcome Reception from 6:00 pm – 6:30 pm and during Wednesday's lunch from 12:30 pm – 1:00 pm.

The Exhibit Hall is open Monday, February 29 from 3:30 pm – 7:00 pm, Tuesday, March 1 from 10:00 am – 5:00 pm and Wednesday, March 2 from 10:00 am – 4:00 pm.

All general sessions will take place in the Grand Ballroom EF. The exhibit hall, posters, breaks and lunches will take place in the Midway and Pegram. Both rooms are located on the lower level of the hotel in close proximity to one another.

Location/Hotel

The 2016 APHL NBSGTS will be held at the St. Louis Union Station Hotel at 1820 Market St., St. Louis, MO 63103, 314.231.1234. The hotel is located in downtown St. Louis in the old train station. It is on the red Metrolink (light rail) line with easy access to numerous restaurants, with several in the station complex and within easy walking distance.

Optional Tour of the Missouri State Newborn Screening Laboratory

An optional tour of the Missouri State Newborn Screening Laboratory has been arranged for Thursday afternoon, March 3. It is currently full. Registered attendees are requested to please meet in the Midway near the symposium registration desk by 12:20pm. The bus will leave promptly at 12:30pm and return no later than 7:00pm. Lunch will be provided. Last minute replacements may occur if those with existing reservations are unable to attend. Tour bus and lunch sponsored by Baebies, Inc.

Registration

The registration fee is $550.

APHL's Federal ID Number is 52-1800436. Cancellation policy: Registrations cancelled 30 days prior to the symposium will be refunded minus a $100 administrative fee. Registrations cancelled less than 30 days prior to the symposium will not be refunded.

Consent to Use Photographic Images

Registration and attendance at or participation in APHL meetings 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, videotapes, electronic reproductions and audiotapes of such events and activities.

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

APHL is an approved provider of continuing education programs in the clinical laboratory sciences through the American Society of Clinical Laboratory Science (ASCLS) P.A.C.E.® program. Attendees may earn up to 21 P.A.C.E.® credits by attending the entire conference. Florida and genetic counselor CEUs are also in process.

CPH Recertification Credits

APHL is an approved provider of Certified in Public Health (CPH) Recertification Credits through the National Board of Public Health Examiners (NBPHE). Attendees have the opportunity to earn up to 19 hours of credit by attending the entire conference. APHL will not issue certificates of CPH credits earned. The attendee is responsible for keeping track of the hours earned.

CMEs/CNEs

CMEs and CNEs (up to 20 credit hours) will be provided. See website or tote bag insert for more information.

2016 NBSGTS Mobile App

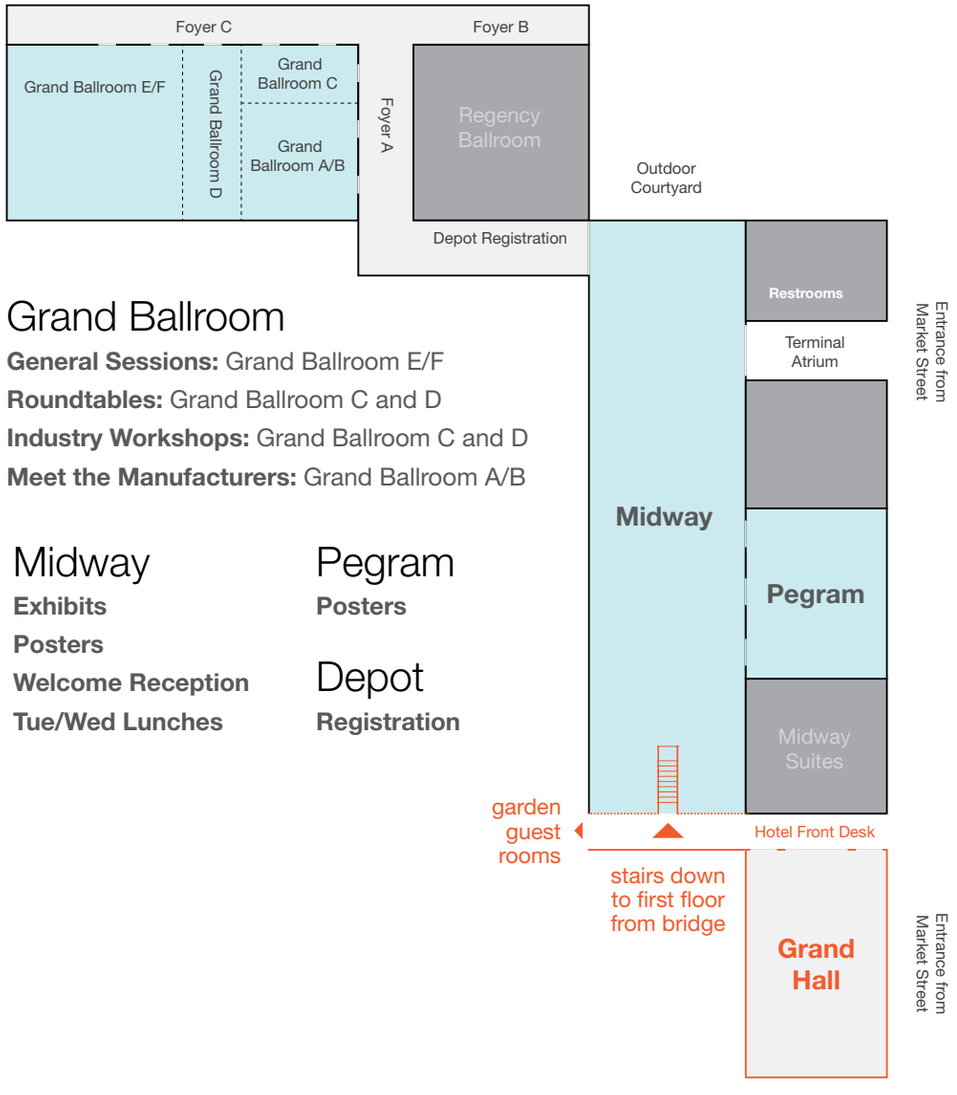
Access all the detailed information about sessions, exhibitors and poster abstracts at your fingertips before the meeting and onsite. Plan your experience with the My Show feature. Receive alerts and reminders onsite.

The app includes these useful features:

- Interactive floor plans of the hotel and exhibit for easy navigation
- My Show, where one can personalize their experience by tagging sessions, exhibitors, city destinations and create notes
- Alerts, reminders or changes about the conference
- APHL Blog daily conference summary and other social media
- Speaker and sponsor profiles
- Play the APHL Challenge! Exhibitors will challenge you to a unique task you must complete at their booth, such as taking a selfie with one of their staff. Complete all the tasks in the challenge and be entered to win a $100 gift card.

The 2016 NBSGTS mobile app is available on the iPhone and Android platforms and can be found in the Apple and Google Play stores.

Hotel Floor Plan



Grand Ballroom

General Sessions: Grand Ballroom E/F
Roundtables: Grand Ballroom C and D
Industry Workshops: Grand Ballroom C and D
Meet the Manufacturers: Grand Ballroom A/B

Midway

Exhibits
Posters
Welcome Reception
Tue/Wed Lunches

Pegram

Posters

Depot

Registration

Final Program

SUNDAY, FEBRUARY 28

3:00 pm – 7:00 pm

Registration

Depot Registration

MONDAY, FEBRUARY 29

7:00 am – 6:00 pm

Registration

Depot Registration

7:00 am – 8:30 am

Coffee

Grand Ballroom Foyer

7:00 am – 8:00 am

Roundtables

Legislative Fact Sheet: Adding Conditions to State NBS Panels

Grand Ballroom C

The APHL Legal and Legislative Issues in Newborn Screening Workgroup published a legislative fact sheet which provides background on newborn screening, the factors considered by the Advisory Committee on Heritable Disorders in Newborn and Children in adding conditions to the Recommended Uniform Screening Panel (RUSP), and a state specific section that can be modified by your state to inform legislators about the process by which new conditions are added to your state newborn screening panel.

- **Kimberly Piper**, Iowa Department of Public Health, The APHL Legal and Legislative Issues in Newborn Screening

Short-Term Follow-up Roundtable: Successes and Challenges

Grand Ballroom D

Short-term follow-up is an integral part of the newborn screening process. This roundtable will serve as an information forum where program staff can discuss barriers, success stories and ideas for strengthening their follow-up programs.

- **John D. Thompson**, PhD, MPH, MPA, Washington State Newborn Screening Program

Day One

588-831-16, 6.5 contact hours (PACE)

At the conclusion of today, the participant will be able to:

- Interpret a modified screening algorithm for Critical Congenital Heart Disease in moderate altitude settings
- Describe unique challenges with implementing Adrenoleukodystrophy and Pompe disease screening
- Describe new assays to detect Krabbe, Mucopolysaccharidosis types II, IVA and VI and Duchenne Muscular Dystrophy
- Discuss and evaluate assays to screen for Spinal Muscular Atrophy in patient dried blood spots
- Discuss future directions of genomics and precision medicine and implications for state NBS programs
- Describe newborn screening expansion efforts in Australia, the Netherlands and Germany
- Evaluate available screening assays for SCID in the Dutch NBS program

8:30 am – 9:00 am

Welcome Session

Grand Ballroom EF

Moderators: Patrick Hopkins, Missouri Department of Health & Senior Services
Richard Olney, MD, MPH, California Department of Public Health

- **Richard Olney**, MD, MPH, California Department of Public Health
- **Patrick Hopkins**, Missouri Department of Health & Senior Services
- **Mike Massman**, Missouri State Public Health Laboratory
- **Veronica Wiley**, PhD, The International Society for Neonatal Screening
- **Jelili Ojodu**, MPH, Association of Public Health Laboratories

9:00 am – 10:30 am

Session 1: Current Conditions in State Newborn Screening Panels

Grand Ballroom EF

This session will provide a nationwide review of screening implementation of disorders recently added to the Recommended Uniform Screening Panel (RUSP). Best laboratory practices for hemoglobinopathy screening and diagnosis will also be highlighted.

Moderators: Patrick Hopkins, Missouri Department of Health & Senior Services
Richard Olney, MD, MPH, California Department of Public Health
Veronica Wiley, PhD, The Children's Hospital at Westmead, Australia

Status of Screening for Recommended Disorders in the United States

- **Careema Yusuf**, MPH, Association of Public Health Laboratories

Building and Enhancing Laboratory Capacity for Screening and Diagnosis of Hemoglobinopathies

- **M. Christine Dorley**, MSP, MT(ASCP), Tennessee Department of Health

Evaluation of Modified Newborn Screening Algorithms for Critical Congenital Heart Disease at Moderate Altitude

- **Leilani Russell**, MPH, Colorado School of Public Health

Screening for ALD and Pompe in New York, Expected the Unexpected

- **Michele Caggana**, ScD, FACMG and **Joseph Orsini**, PhD, New York State Department of Health

10:30 am – 11:00 am

Break

Grand Ballroom Foyer

11:00 am – 12:30 pm

Session 2: Prospective Newborn Screening Conditions

Grand Ballroom EF

This session will discuss experiences in screening for, and the feasibility of adding, conditions not currently on the Recommended Uniform Screening Panel (RUSP).

Moderators: Leslie Gaffney, California Department of Public Health
Don Bailey, PhD, RTI International

Missouri's Experience with Krabbe Screening Using a Simple Bench Fluorometric Assay

- **Patrick Hopkins**, Missouri State Public Health Laboratory

Development of a 3-plex Assay by MS/MS to Detect the Lysosomal Storage Diseases MPS-II, MPS-IVA and MPS-VI

- **Susan Elliott**, University of Washington

Development of a New Bloodspot Screening Assay for Duchenne Muscular Dystrophy (DMD)

- **Stuart Moat**, PhD, FRCPath, University Hospital of Wales, United Kingdom

Progress of Newborn Screening for Spinal Muscular Atrophy

- **Shu-Chuan Chiang**, National Taiwan University Hospital, Taiwan

Inter-Laboratory Comparison of Assays to Measure SMN2 Copy Number in Dried Blood Spots from Patients with Spinal Muscular Atrophy

- **Robert Vogt**, PhD, Centers for Disease Control and Prevention

12:30 pm – 1:30 pm

Lunch (on your own)

1:30 pm – 3:30 pm

Keynote Session: Expanding the Newborn Screening Gateway: Considerations, Applications, and Future Implications for Genomics and Precision Medicine

Grand Ballroom EF

This session will provide various perspectives on the considerations, applications and future implications for genomics and precision medicine.

Moderator: Michele Caggana, ScD, FACMG, New York State Department of Health

- **Michele Caggana**, ScD, FACMG, New York State Department of Health
- **Jeffrey Botkin**, MD, MPH, University of Utah
- **Joe Palca**, PhD, National Public Radio Science Desk
- **Cynthia M. Powell**, MD, MS, University of North Carolina at Chapel Hill

3:30 pm – 4:00 pm

Break in the Exhibit Hall

Midway

3:30 pm – 7:00 pm

Exhibit Hall Open

Midway/Pegram

Posters Available for Viewing

4:00 pm – 5:30 pm

Session 3: International Perspectives

Grand Ballroom EF

This session will showcase international newborn screening efforts underway in Australia, the Netherlands, Germany and Belgium.

Moderators: Joanne Mei, PhD, Centers for Disease Control and Prevention
Fred Lorey, PhD, International Society for Neonatal Screening

Toward National Newborn Screening in Australia

- **Veronica Wiley**, PhD, The Children's Hospital at Westmead, Australia

Further Expansion of the Neonatal Screening Panel in the Netherlands

- **J. Gerard Loeber**, PhD, International Society for Neonatal Screening, The Netherlands

The Prevalence of Hereditary Hemoglobin Disorders and Its Implications for Newborn Screening in Germany

- **Zoltan Lukacs**, PhD, Hamburg University Medical Center, Germany

Integration of SCID Screening into the Dutch Newborn Screening Program: Benefits and Shortcomings of the Available Screening Assays

- **Peter Schielen**, RIVM, The Netherlands

Multiplex Screening for Treatable Lysosomal Storage Diseases (LSDs)

- **Francois Eyskens**, MD, PhD, PCMA vzw, Belgium

5:30 pm – 7:00 pm

Welcome Reception in the Exhibit Hall

Midway/Pegram

6:00 pm – 6:30 pm

Poster Authors Available to Answer Questions

Pegram/Midway

7:30 pm – 9:00 pm

The Genomics After Party

Grand Ballroom D

This will be an interactive discussion on the integration of genomics into newborn screening.

Hosts:

- **Aaron Goldenberg**, PhD, Case Western Reserve University
- **Beth Tarini**, MD, MS, University of Michigan
- **Amy Gaviglio**, MS, CGC, Minnesota Department of Health
- **Jelili Ojodu**, MPH, Association of Public Health Laboratories

7:30 pm – 9:00 pm

Collaborative Improvement & Innovation Network (CoIIN) on Timeliness in NBS Systems Wrap Up Meeting

Grand Ballroom C

For the past 18 months, 8 states have worked to improve their timeliness with regards to the quality indicator on timeliness for newborn systems. This meeting will serve as the final meeting of the project and we will share the lessons learned and accomplishments over the project period.

Monday, February 29

TUESDAY, MARCH 1

7:00 am – 5:30 pm

Registration

Depot Registration

7:00 am – 8:30 am

Coffee

Grand Ballroom Foyer

7:00 am – 8:00 am

Industry Workshops

PerkinElmer, Inc. | Grand Ballroom C

PerkinElmer: New Solutions for Emerging Disorders

The workshop will discuss PerkinElmer's newly launched products and services, emerging newborn screening disorders and product development pipeline. The emerging disorder discussion will focus on Lysosomal Storage Disorders (LSDs) and X-ALD. Other disorders (i.e., SMA and XL) and technology (i.e., sequencing) will also be discussed.

STACS DNA | Grand Ballroom D

You Tell Us: Could this Simple Tracking Software Improve your NBS Timeliness?

Check out brand new software that tracks newborn screening samples from collection to reporting, with portals specifically designed for the laboratory policy center, birthing centers, physicians and parents. Would this software help reduce missed or delayed tests, expedite testing, reduce errors and increase proficiency? Will lab staff, birthing centers, midwives, parents and physicians benefit from better communications? This is a terrific opportunity for you to give us your input so we can help improve NBS timeliness.

Speakers: **Heather Macintosh**, VP Marketing and **Kyle Kipp**, Quality Assurance Analyst, STACS DNA

8:30 am – 10:00 am

Joint Follow-up and QA/QC Session

Grand Ballrom EF

588-832-16, 1.5 contact hours (PACE)

This session will cover presentations relating to both follow-up and quality assurance/quality control issues. Participants will view a demonstration of a training webinar for proper NBS specimen collection and hear other presentations on cross-cutting issues.

At the conclusion of this session, the participant will be able to:

- Describe findings from a needs assessment on communicating incidental findings of SC trait

Moderators: John D. Thompson, PhD, MPH, Washington State Newborn Screening Program
Eleanor Stanley, MT(ASCP), Michigan Department of Health and Human Services

Updating the National Newborn Screening Contingency Plan: Addressing Gaps in the System

- **Scott Shone**, PhD, New Jersey Department of Health

Communicating Incidental Findings of Sickle Cell Trait on the Newborn Screen: A Community Needs-based Assessment

- **Maggie Dreon**, MS, CGC, Minnesota Department of Health

Interactive Training Webinar for Newborn Screening Specimen Collection

- **Patrice Held**, PhD, FACMG, Wisconsin State Laboratory of Hygiene

Follow-up Status During the First Five Years of Life for Select Primary RUSP Disorders Diagnosed Through California Newborn Screening

- **Lisa Feuchtbaum**, DrPH, MPH, California Department of Public Health

10:00 am – 10:30 am

Break in the Exhibit Hall

Midway

Sponsored by the Bronze Circle exhibitors

10:00 am – 5:00 pm

Exhibit Hall Open

Midway/Pegram

Posters Available for Viewing

Tuesday, March 1

10:30 am – 12:00 pm

Concurrent Sessions

Improving Short- and Long-term Follow-up

Grand Ballroom C

588-833-16, 1.5 contact hours (PACE)

This session will be an interactive opportunity to explore some of the innovative ways of enhancing outcomes for babies with abnormal screening results. A variety of programs and disorders will be included.

At the conclusion of this session, the participant will be able to:

- Identify considerations in building a long-term sustainable strategy for assisting families with Inborn Errors of Metabolism management
- Describe one state's strategies to improve the newborn hearing screening program

Moderators: Carol Johnson, University of Iowa Hospitals and Clinics
Rachael Montgomery, RN, Alabama Department of Public Health

Quality Improvement Strategies for a State Newborn Hearing Screening Program
- **Sylvia Mann**, MS, CGC, Hawaii Department of Health Genetics Program

Using State Birth Defects Registries to Evaluate Outcomes of Critical Congenital Heart Disease Newborn Screening
- **Monica McClain**, MS, PhD, University of New Hampshire

Minnesota Medical Foods Initiative: Building a Long-term Strategy for Assisting Families in Obtaining Medical Foods and Dietary Supplements for Inborn Errors of Metabolism Management
- **Susan Berry**, MD, University of Minnesota

Congenital Hypothyroidism in Newborn Infants with Borderline Thyroid Stimulating Hormone Screening Values
- **Conchita G. Abarquez**, MD, Newborn Screening Center Mindanao, Philippines

Arkansas Newborn Screening Long-term Follow-up Cohort Study — Year 3
- **Jo Ann Bolick**, BSN, MA, University of Arkansas for Medical Sciences/Arkansas Children's Hospital

QA/QC Session: Factors That Can Improve Screening Outcomes

Grand Ballroom EF

588-834-16, 1.5 contact hours (PACE)

This session will discuss methods from within and outside of the laboratory to newborn screening outcomes. Effects of gestational age and weight on newborn enzyme activity will be discussed in detail, as well as the normalization of MS/MS cutoffs using CDC quality control materials.

At the conclusion of this session, the participant will be able to:

- Evaluate variabilities in filter paper lots and their effects on biotinidase screening results.

Moderators: Patricia Hunt, Texas Department of State Health Services
Art Hagar, PhD, Georgia Public Health Laboratory

The Effects of Gestational Age and Birth Weight on the Newborn Screening Activities of Enzymes Associated with Lysosomal Storage Disorders
- **Rong Shao**, MD, Illinois Department of Public Health

Normalization of Laboratory MS/MS Cutoffs Using CDC NSQAP Quality Control Materials
- **Mary Seeterlin**, PhD, Michigan Department of Health & Human Services

Variability in Biotinidase Screening Results Between Filter Paper Lots
- **Roger Eaton**, PhD, New England Newborn Screening Program, University of Massachusetts Medical School

Factors to Consider in Improving the Screening Algorithm for Congenital Adrenal Hyperplasia
- **Norma Tavakoli**, PhD, New York State Department of Health

Propionic Acidemia Screening in the Amish and Mennonite Populations
- **Zineb Ammous**, MD, The Community Health Clinic

12:00 pm – 2:00 pm

Awards Luncheon

Midway

Host:
- **Scott Becker**, MS, Association of Public Health Laboratories

Day Two Afternoon

588-835-16, 3.0 contact hours (PACE)

At the conclusion of this afternoon, the participant will be able to:

- Describe a multiplex genotyping assay for screening of CAH
- Discuss the benefits of full gene sequencing for newborn screening conditions
- Identify methods to improve newborn screening for out-of-hospital births
- Describe several unique considerations when screening special populations

2:00 pm – 3:30 pm

Session 4: Molecular Application

Grand Ballroom EF

There has been a recent increase in collaborations to better understand molecular testing for newborn screening. The focus of this session is on national efforts to improve molecular testing for several disorders, including Severe Combined Immune Deficiency (SCID), Cystic Fibrosis and Congenital Adrenal Hyperplasia. Second tier full gene sequencing for Very long-chain acyl-CoA dehydrogenase and Glutaric Aciduria Type 1 will also be highlighted.

Moderators: Rachel Lee, PhD, Texas Department of State Health Services
Suzanne Cordovado, PhD, Centers for Disease Control and Prevention

Development of a Multiplex CYP21A2 Genotyping Assay for Congenital Adrenal Hyperplasia Screening

- **Christopher Greene**, PhD, Centers for Disease Control and Prevention

Automation of the in situ Dried Blood Spot Screening Assay for Severe Combined Immunodeficiency

- **Laura Hancock**, MS, Centers for Disease Control and Prevention

Overview of the First Eight Years of Newborn Screening for Cystic Fibrosis: The California Experience

- **Tracey Bishop**, California Department of Public Health

Second Tier Full Gene Sequencing for Follow-up of Positive Newborn Screening for Very Long-chain Acyl-CoA Dehydrogenase Deficiency and Glutaric Aciduria Type 1

- **Neena Champaigne**, MD, Greenwood Genetic Center

Sequence Coverage of Genes for Inborn Errors of Metabolism by DNA Prepared from Residual Newborn Screening Dried Blood Spots

- **Robert Currier**, PhD, California Department of Public Health

3:30 pm – 4:00 pm

Break in the Exhibit Hall

Midway

4:00 pm – 5:30 pm

Session 5: Screening for Special Populations

Grand Ballroom EF

This session will focus on special considerations for newborn screening of special populations, including out-of-hospital births and low birth weight infants. Screening of newborns with CCHD and SCID will also be discussed.

Moderators: Christine Dorley, MSP, MT(ASCP), Tennessee Department of Health: Laboratory Services
Kimberly Piper, RN, BSN, CPH, CPHG, Iowa Department of Public Health

Improving the Entire Newborn Screening Process for Out of Hospital Births

- **Amy Gaviglio**, MS, CGC, Minnesota Department of Health

Newborn Screening in the NICU: Colorado's Experience with Screening Low Birth Weight Infants

- **Erica Wright**, MS, CGC, Colorado Department of Public Health and Environment

Comprehensive Screening for Severe Combined Immunodeficiency in Manitoba, Canada

- **J. Robert Thompson**, RT, DBT, Cadham Provincial Laboratory, Canada

Catching Blue Babies: Critical Congenital Heart Defects in Newborns

- **Merlin Ariefdjohan**, PhD, Colorado School of Public Health, University of Colorado

6:00 pm – 7:15 pm

Meet the Manufacturers Session

Grand Ballroom AB

Lighthearted presentations from vendors along with munchies and beverages (not a meal)

6:00 pm	STACS DNA
6:15 pm	Illumina, Inc.
6:30 pm	Baebies, Inc.
6:45 pm	Astoria-Pacific, Inc.
7:00 pm	PerkinElmer

7:30 pm - 9:00 pm

Short-term Follow-up Mixer

Grand Ballroom D

Calling all short-term follow-up folks! This informal session will allow us to meet one another, share some of the unique challenges we face in our respective programs and network for future collaboration.

Moderators: Carol Johnson, University of Iowa Hospitals and Clinics
John Thompson, PhD, MPH, Washington State Department of Health

WEDNESDAY, MARCH 2

7:00 am – 6:00 pm

Registration

Depot Registration

7:00 am – 8:30 am

Coffee

Grand Ballroom Foyer

7:00 am – 8:00 am

Roundtables

Grand Ballroom C

Strategies for Implementing Newborn Screening for Adrenoleukodystrophy (ALD)

- **Lisa Feuchtbaum**, DrPH, MPH, California Department of Public Health

This roundtable session will involve participants working in small groups to brainstorm strategies to address program challenges of implementing screening of ALD.

Grand Ballroom D

Solutions for Implementing Newborn Screening Surveillance Case Definitions

- **Louis Bartoshesky**, MD, Christiana Care

This roundtable session will provide an overview of the public health surveillance case definitions and presentations from states who have implemented the case definitions.

Day Three

588-836-16, 5.5 contact hours (PACE)

At the conclusion of today, the participant will be able to:

- Identify factors to consider in implementing statewide electronic ordering, reporting and birth notification systems
- Identify the achievements and efforts of the Newborn Screening Clearinghouse
- Describe health communication strategies for newborn screening
- Identify parental concerns and considerations when weighing participation in research, including genomic sequencing of healthy newborns

8:30 am – 10:00 am

Session 6: Health Information Technology

Grand Ballroom EF

Electronic data transfer is on the horizon for all newborn screening programs. This session will provide perspectives from Minnesota and Texas on implementing data transfer, as well as an overview of Indiana's experience integrating the Healthcare Enterprise's (IHE) Newborn Admission Notification Information (NANI) technical framework.

Moderators: Dariush Shirazi, State Hygienic Laboratory at the University of Iowa
Andrew Richardson, Florida Department of Health

HIT the Ground Running: Statewide Implementation of Electronic Demographics and Reporting of Point-of-Care Newborn Screening Results

- **Amy Gaviglio**, MS, CGC, Minnesota Department of Health

Past, Present, Future: Health Information Technologies in the Texas Newborn Screening Laboratory

- **Brendan Reilly**, Texas Department of State Health Services

Increasing Newborn Screening Health Information Interoperability: A Systems Approach

- **Heather Brand**, Minnesota Department of Health

Proof of Concept Project Using Electronic Birth Notification to Improve Surveillance and Quality of Newborn Screening Pre-Analytic Processes: Implementation of NANI in Select Indiana Hospitals

- **Victoria Buchanan**, Indiana State Department of Health

10:00 am – 10:30 am

Break in the Exhibit Hall

Midway

Sponsored by the Bronze Circle exhibitors

10:00 am – 4:00 pm

Exhibit Hall Open

Midway/Pegram

Posters Available for Viewing

10:30 am – 12:00 pm

Session 7: Education

Grand Ballroom EF

Many state programs encounter issues surrounding educating parents on newborn screening as well as policy and ethical issues regarding the use and storage of residual dried blood spots. This session will provide perspectives from state programs on experiences with parental education and will provide an overview of the Newborn Screening Clearinghouse activities.

Moderators: Amy Gaviglio, MS, CGC, Minnesota Department of Health
Brad Therrell, PhD, National Newborn Screening and Global Resource Center

Baby's First Test: 5 Years of a National Educational Initiative

- **Jaclyn Seisman**, MPH, Genetic Alliance

Impact of Continuing Medical Education on Primary Care Providers' Knowledge and Confidence in Caring for Patients with Congenital Hypothyroidism

- **Emily Bezar**, MA, Public Health Foundation Enterprises

Health Communication Strategies for Newborn Screening

- **Patti Constant**, MPH, Minnesota Department of Health

Development of the NC NEXUS Decision Aid: Implications for Parental Education and Newborn Screening

- **Megan Lewis**, PhD, RTI International

12:00 pm – 1:30 pm

Lunch Provided in the Exhibit Hall

Midway

Visit the Exhibitors and Posters

12:30 pm – 1:00 pm

Poster Authors Available to Answer Questions

Pegram/Midway

1:30 pm – 3:00 pm

Session 8: Parent/Patient Panel

Grand Ballroom EF

This session will feature families affected by genetic disorders. They will present their stories as they choose and will not have a script. The disorders represented come from the Recommended Uniform Screening Panel (RUSP), secondary targets and disorders under investigation.

Moderators: Sharmini Rogers, MPH, MBBS, Missouri Department of Health & Senior Services
Natasha Bonhomme, Genetic Alliance

- **Felicia Smith** (parent of child with Cystic Fibrosis)
- **Tina Jarrell** (parent of child with severe Hurler)
- **Kari Jacobsen** (parent of child with infantile Pompe)
- **Rosemary Britts** (parent of child with Sickle Cell Disease)

3:00 pm – 4:00 pm

Break in the Exhibit Hall (Raffle drawing at 3:30 pm)

Midway

Sponsored by the Silver Circle exhibitors

4:00 pm – 5:00 pm

Session 9: Financial, Legal, Ethical, Policy and Social Implications (FLEPSI)

Grand Ballroom EF

This session will provide perspectives from state programs on issues surrounding parental consent for newborn screening as well as a discussion of economic and feasibility studies for certain conditions.

Moderators: Alisha Keehn, MPA, American College of Medical Genetics and Genomics
Aaron Goldenberg, PhD, MPH, Case Western Reserve University

Parental Interest in Genomic Sequencing (GS) of Healthy Newborns: Experiences from the BabySeq Project

- **Richard Parad**, MD, MPH, Brigham and Women's Hospital

The Michigan BioTrust for Health: Impact of Parental Consent Process on Robust Population-based Research Using Residual Newborn Screening Blood Spots

- **Mary Kleyn**, MSc, Michigan Department of Health and Human Services

Analysis of Four Lysosomal Storage Disorders Within the Context of Newborn Screening in Washington State

- **Megan McGrillis**, MPH, Washington State Department of Health

6:00 pm – 10:00 pm

Evening Social

Sponsored by PerkinElmer

THURSDAY, MARCH 3

7:30 am – 12:30 pm

Registration

Depot Registration

7:00 am – 8:30 am

Coffee

Grand Ballroom Foyer

7:00 am – 8:00 am

ISNS Membership Meeting

New York Central

Day Four

588-837-16, 3.0 contact hours (PACE)

At the conclusion of this morning, the participant will be able to:

- Identify factors that may impede or promote timeliness of NBS specimen collection
- Describe two strategies for improving timeliness of newborn screening
- Describe mechanisms to increase rates of newborns screened
- Discuss challenges in implementing and improving upon national quality measures

8:30 am – 10:00 am

Session 10: Quality Improvement — Timeliness

Grand Ballroom EF

This session will feature presentations from various states regarding recent newborn screening timeliness improvements, highlighting their challenges, lessons learned and accomplishments.

Moderators: **Susan Tanksley,** PhD, Texas Department of State Health Services
Cheryl Hermerath, MBA, DLM(ASCP), RR(NRCM), Portland, Oregon

Meet Your Match: The Importance of Vital Record Matching in the Realm of Timely Newborn Screening

- **Amy Dahle**, MPH, Minnesota Department of Health

What Predicts NBS Specimen Timeliness in a State-based Cohort of Birthing Hospitals?

- **Beth Tarini**, MD, MS, University of Michigan

A Lean Six Sigma Approach to Continuous Quality Improvement in Texas Newborn Screening Program

- **Patricia Hunt**, Texas Department of State Health Services

Collaborating Across States to Improve NBS Timeliness: An Overview of the CoIIN (Collaborative Improvement and Innovation Network) for Timeliness

- **Yvonne Kellar-Guenther**, PhD, Colorado School of Public Health

Collaborative Improvement and Innovation Network (CoIIN) for Timeliness in Newborn Screening in Colorado and Wyoming

- **Erica Wright**, MS, CGC, Colorado Department of Public Health and Environment

10:00 am – 10:30 am

Break

Grand Ballroom Foyer

10:30 am – 12:00 pm

Session 11: Quality Improvement — Getting it Right

Grand Ballroom EF

This session will discuss methods from within and outside of the laboratory to achieve greater accuracy in newborn screening results by reducing false positives and making other improvements in NBS. Participants will learn about use of lean process to make improvements in NBS, and methods to reduce false positives in Cystic Fibrosis (CF) screening will be discussed in detail.

Moderators: **Rasoul Koupaei,** PhD, DABCC, FACB, California Department of Public Health
Susan Berry, MD, University of Minnesota School of Medicine

Does Every Baby Get Screened? Overhauling Birth Monitoring in Washington State

- **Ashleigh Ragsdale**, MPH, Washington State Newborn Screening Program

Multi-analyte Data Analysis Reduces False Positives in Cystic Fibrosis

- **Travis Henry**, PhD, State Hygienic Laboratory at the University of Iowa

Use of Quality Initiative to Increase CCHD Screening

- **Jamey Kendall**, RN, Kansas Department of Health and Environment

Using Lean Process for Improvement in Newborn Screening

- **Sarah Hasselbalch**, Washington State Newborn Screening Program

The Wisconsin Experience Getting In-Step with NewSTEPs Quality Indicators

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

12:00 pm

Adjournment

12:30 pm – 7:00 pm

Optional Tour of the Missouri State Newborn Screening Laboratory

Lunch and travel sponsored by Baebies, Inc.

Exhibit Hall Schedule

Monday, February 29	3:30 pm – 7:00 pm	Hall Open
	3:30 pm – 4:00 pm	Break
	5:30 pm – 7:00 pm	Welcome Reception
Tuesday, March 1	10:00 am – 5:00 pm	Hall Open
	10:00 am – 10:30 am	Break
	12:00 pm – 2:00 pm	Awards Lunch
	3:30 pm – 4:00 pm	Break
	6:00 pm – 7:15 pm	Meet the Manufacturers Grand Ballroom AB
Wednesday, March 2	10:00 am – 4:00 pm	Hall Open
	10:00 am – 10:30 am	Break
	12:00 pm – 1:30 pm	Box Lunch
	3:00 pm – 4:00 pm	Break (Raffle Drawing 3:30 pm)

Meet the Manufacturers Schedule

Tuesday, March 1

6:00 pm – 7:15 pm

Grand Ballroom AB

6:00 pm	STACS DNA
6:15 pm	Illumina, Inc.
6:30 pm	Baebies, Inc.
6:45 pm	Astoria-Pacific, Inc.
7:00 pm	PerkinElmer

Visit the Exhibitors! Win Prizes!

Visit with the vendors and win prizes! Take the exhibit raffle card from your totebag and visit each exhibit booth. Ask one of the booth personnel to sign their initials in the box corresponding to their booth number. When all of the boxes are filled, turn in the card at the registration desk. Cards must be turned in at the registration desk no later than 3:00 pm on Wednesday, March 2. Be sure to include your name on the card. Winners of the prizes will be announced starting at 3:30 pm on Wednesday in the exhibit hall. You must be present to win.

PRIZES

- Two coach airline tickets on Delta Airlines to anywhere in the continental United States (compliments of APHL)
- Complimentary registration to the 2017 Newborn Screening & Genetic Testing Symposium
- Go Pro Camera (compliments of Baebies, Inc.)
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- ASUS MeMO Pad 8 Tablet (compliments of STACS DNA)
- One coach airline ticket on American Airlines to anywhere in the continental United States
- $100 Amazon.com gift card (compliments of Cambridge Isotope Laboratories)
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- Garmin – nuvi2597LMT GPS (compliments of APHL)
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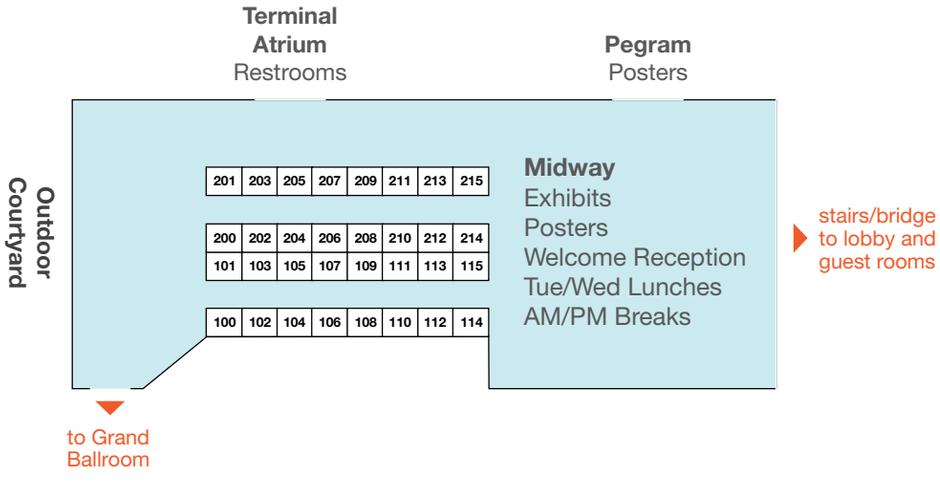
Mobile App Challenge

Click to get even more prizes! Download the mobile app to play the "APHL Challenge." Vendors will challenge you to a unique task you must complete at their booth, such as taking a selfie with one of their staff. Complete all the tasks in the challenge no later than 3:00 pm on Wednesday, March 2, and be entered to win a $100 gift card (compliments of APHL). You may enter to win both the exhibit raffle and the mobile app challenge!

EXHIBIT HALL



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101	EBF, Inc.
102/104	Baebies, Inc.
103	Labsystems Diagnostics
105	Luminex Corporation
106	Mead Johnson Nutrition
108	NBSTRN/NCC
109	EGL Genetic Diagnostics
110/112/114	PerkinElmer
111	Natus Medical, Inc./ Neometics
113	STACS DNA
115	QuantaBio
200	Illumina
201	RTI International
202	QIAGEN
203	UPS
204	CLSI
205	APHL NBSG Programs
208/210	Bio-Rad Laboratories
209	Baby's First Test
211/213	Astoria-Pacific, Inc.
212	Cambridge Isotope Laboratories
214	Integrated Software Solutions
215	OZ Systems

LIST OF EXHIBITORS

APHL Newborn Screening and Genetics Program

Booth 205

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

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The APHL Newborn Screening and Genetics Program offers conferences, workshops, webinars, access to molecular and quality improvement 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. NewSTEPs is the comprehensive national resource center for state newborn screening programs and stakeholders.

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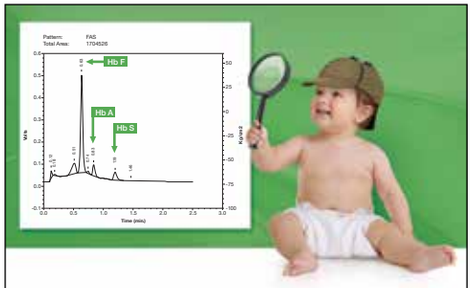

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www.nbstrn.org and www.nccrcg.org

The Newborn Screening Translational Research Network (NBSTRN) and the National Coordinating Center for the Regional Genetic Service Collaboratives (NCC) will be sharing excellent resources available for researchers, healthcare providers, public health professionals and consumers.

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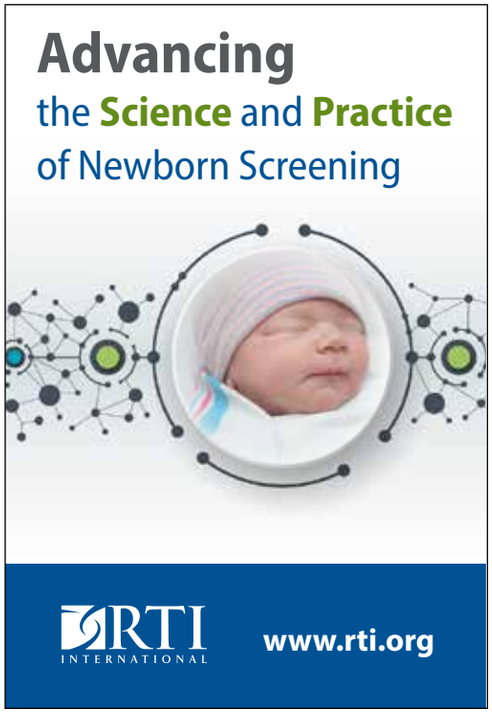

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RTI International is one of the world's leading research institutes, dedicated to improving the human condition by turning knowledge into practice. Our expertise includes newborn screening research pilot projects, statewide implementation of pilots in partnership with a state newborn screening program, evaluation of state newborn screening programs, and extensive health communication expertise that encompasses targeted health communication, public health communication campaigns, communication around informed consent, and decision aids for potential study participants.

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- UPS

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Special thanks to Baebies, Inc. for sponsoring the bus and lunch for the Missouri Newborn Screening Lab tour.

Mead Johnson Nutrition

Special thanks to Mead Johnson Nutrition for a generous educational grant supporting the 2016 NBSGTS.

POSTER ABSTRACTS

The full poster and oral abstracts may be found on the symposium website www.aphl.org/nbsgts and on the mobile app.

Posters may be viewed in Pegram and the Midway during exhibit hall hours on Monday, Tuesday and Wednesday, Feb. 29 – Mar. 2.

P-1

High Throughput Newborn Screening for Severe Combined Immunodeficiency (SCID) Using the Automated T-cell Receptor Excision Circle (TREC) in situ Assay

R. Haughton[1], S. Dever[1], K. Turner[1], L. Hancock[2], J. Taylor[3], Golriz Yazdanpanah[2], F. Lee[2]; [1]Virginia Division of Consolidated Laboratory Services, Richmond, VA, [2]Centers for Disease Control and Prevention, Atlanta, GA, [3]RTI International, Research Triangle Park, NC

Presenter: Richard Haughton, MA, Virginia Division of Consolidated Laboratory Services, Richmond, VA, Email: Richard.Haughton@dgs.virginia.gov

P-2

Saving Virginia's Babies – The Virginia NBS Transit Time Project

K. Turner, W. Andrews, J. Macdonald and R. West, Virginia Division of Consolidated Laboratory Services, Richmond, VA

Presenter: Kim Turner, MT(ASCP), MSQA, Virginia Division of Consolidated Laboratory Services, Richmond, VA, Email: Kim.Turner@dgs.virginia.gov

P-3

To Mandate or Not to Mandate? That is the Question...A Collaborative Decision Making Approach in Virginia

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P-4

Sequence Analysis of TREC δRec-ΨJα Signal Joint Region of the most common ethnicities in New York State Newborn Population

C.A. Saavedra-Matiz, A. Brown, L. DiAntonio, A. Parker, B. Vogel and M. Caggana, Wadsworth Center, New York State Department of Health, Albany, NY

Presenter: Carlos A. Saavedra-Matiz, MD, Research Scientist, Wadsworth Center, DAI 5072, NY State Dept. of Health, Albany, NY 12208, Email: carlos.saavedra@health.ny.gov

P-5

Evaluation of Birth Hospital Compliance with CLSI Guidelines to Collect a Specimen Prior to Transfusion

A. McGeoch, B. Vogel, M. Caggana and J. Orsini, New York State Department of Health, Albany, NY

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P-6

An Exploration of the Roles and Responsibilities of Genetic Counselors Working With Newborn Screening Programs

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P-7

Revisiting the Follow-up Protocol for Initial Positive Results for Phenylketonuria (PKU)

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P-8

California Goals for Timeliness in Newborn Screening Specimen Collection and Transit Time

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P-9

California Thalassemia Fact Sheet & Provider Survey

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P-10

Findings from a Survey of Michigan Birth Hospitals: What Happens After a Failed Screen for Critical Congenital Heart Disease?

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P-11

50 Years of Newborn Screening in Michigan: A Year-long Celebration

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Use of State Administrative Data Sets for Newborn Screening Long-term Follow-up: Michigan's Experience

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P-13

Use of Quarterly Quality Assurance Reports to Improve Hospital Performance

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A False Positive for Severe Combined Immune Deficiency (SCID) Caused by a Mutation for the MI T-cell Receptor Excision circle (TREC) Assay

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Glutaric Acidemia Type II and LCHAD Deficiency Cases Identified by Newborn Screening Short-term Outcome

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2 Year Clinical Follow up of a Patient with Methylbutyrylglycinuria Identified by Newborn Screening

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Cross-Sectional Survey on Newborn Screening in Wisconsin Amish and Mennonite Communities

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Collaborative Improvement and Innovation Network (COIIN) for Timeliness in Newborn Screening - the Iowa Experience to Date

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Beyond Implementation: Developing Collaborative Partnerships for SCID Follow-up, Treatment, and Management

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P-21

Developing Data Collection for Critical Congenital Heart Disease Newborn Screening

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Screening for SCID: The Colorado Experience

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Pulse Oximetry Screening for Homebirths in Oklahoma: A Collaboration between the Newborn Screening Program, the University of Oklahoma College of Medicine, and the Oklahoma Midwives Alliance

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The Oklahoma "Every Baby Counts" Quality Improvement Program

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A Picture Worth 1000 Words: The Use of Infographics to Convey Complex Information

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Newborn Screening Education in the Prenatal Period: New Parent Experiences and Desires

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Medical Provider Practices and Desired Assistance: Newborn Screening Education in the Prenatal Period

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Pre-Analytic Operational Improvements: Minnesota's Experience with UPS' CampusShip®

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Poster Abstracts

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Collaborative Partners Project: A Regional Approach to Dissemination of Newborn Screening Information

A. Herrera Morales, Heartland Genetics Services Collaborative, Little Rock, AR, Arkansas Children's Hospital Research Institute, Little Rock, AR

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Identification of Rare Folate and Remethylation Defects Due to Elevated C4 Due to FIGLU in Newborn Screening

I. Sahai[1], J. Bailey[1], T. Zytkovicz[1], R. Eaton[1], D. Britton[1], G. Berry[2], [1]New England Newborn Screening Program, University of Massachusetts Medical School, Jamaica Plain, MA, [2]Childrens Hospital Boston, Boston, MA

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Quality Assurance Highlights and Educational Efforts in Washington State

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Utilization of Lean Six Sigma to Expedite the Initiation of Testing Processes in Texas

B. Reilly, P. Trevino Gonzales, T. Odoms, A. Vinyard and R. Lee, Texas Department of State Health

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The Road to 95% – Texas and the NewSTEPs CoIIN

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Second-tier Newborn Screening DNA Testing for VLCAD Deficiency in Texas

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GALT Mutation Analysis in New Jersey: Development, Implementation, and a Year of Babies

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Newborn Screening for Critical Congenital Heart Defects (CCHD) in New Jersey

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NewSTEPs Site Visits Have Tangible Impacts

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Timely Newborn Screening Status Assessment

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Assessing Residual Blood Spot Volumes in the Midst of Newborn Screening Expansion

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Timeliness of Metabolic Screening in Tennessee - A Progress Report

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Improvement in Transit Times for Newborn Screening Specimens in North Carolina

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Quality Improvement Efforts in Arkansas

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P-45

Integrated Bloodspot-Hearing Newborn Screening Hospital Scorecards

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Regional Newborn Screening Laboratory Backup Planning: 2015 Snapshot

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Diagnosing Prader-Willi Syndrome in Infancy Reduces Obesity and Associated Co-morbidities

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Newborn Screening for Mucopolysaccharidoses: Determination of Sensitivity, Specificity and Cutoff Values

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Current Status of Newborn Screening in Puerto Rico

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Bridging the Language Gap: Creating Culturally Competent Newborn Screening Education for Spanish-Speaking Communities

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Couple Dynamics in Decisions About Elective Whole Exome Sequencing for Newborn Screening

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Values and Beliefs Important for Parental Decisions to Have Genetic Screening for a Child

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Proficiency Testing for T-Cell Receptor Excision Circles (TREC) in Blood Spots to Detect Severe Combined Immunodeficiency (SCID)

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CDC Reference Methods and Materials to Support Expanded Newborn Bloodspot Screening for Pompe Disease and Other Lysosomal Storage Disorders

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Poster Abstracts

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Assessing DNA Extraction Methods Commonly Used in Newborn Screening Labs for the Detection of Cytomegalovirus in Dried Blood Spots

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Validating CDC's Cystic Fibrosis Molecular Dried Blood Spot Repository for Use with Next Generation Sequencing Methods

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Monitoring Method Performance Through the Use of External QC Materials for Newborn Screening Analytes

I. Williams, S. Brown, J. Lauterbach, L. Shockley, C. Nguyen, S. Zobel, V. De Jesus and J. Mei, Centers for Disease Control and Prevention, Atlanta, GA

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Quantifying the Effectiveness and Cost-Effectiveness of Early Detection of Cystic Fibrosis Through Newborn Screening: A Review

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Minnesota Population Spectrum of Congenital Adrenal Hyperplasia Causing Mutations in the CYP21A2 Gene

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Long-term Stabilities of 17-Hydroxyprogesterone, 4-Androstenedione, Cortisol, 11-Deoxycortisol and 21-Deoxycortisol in Dried Blood Spots at Various Conditions

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Dried Blood Spot DNA Extraction Efficiency and Quantification Varies Greatly by Extraction and Measurement Method

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Stability of Glucose-6-Phosphate Dehydrogenase Proficiency Testing Dried Blood Spot Materials

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Poster Abstracts

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Growth of CDC's Newborn Screening Quality Assurance Program Since 1978

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Assessing Challenges to Fragile X Syndrome Newborn Screening

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Use of Newborn Dried Blood Spots for Detection of Prenatal Alcohol Exposure

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A Novel ETHE1 Mutation Identified in a First Nation Canadian Patient, Ascertained Following a Positive Newborn Screen for Isovaleric academia

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Newborn Screening for Cystic Fibrosis in Nuevo León, México

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Feasibility Study Using the Six-Plex PerkinElmer LSD Reagents Utilising Tandem Mass Spectrometry (MSMS) for Newborn Screening and Diagnostic Testing for Six Lysosomal Storage Diseases (LSD)

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Newborn Screening in Slovakia after Starting Tandem Mass Spectrometry: New Epidemiological Data in Slovak Newborns Population

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P-71 – Withdrawn

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The Philippine Expanded Newborn Screening-The First Year of Implementation

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Newborn Screening Incidental Findings: Variability in State Practices

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The State of Newborn Screening Systems

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P-75 – Withdrawn

P-76 – Withdrawn

P-77 – Withdrawn

P-78 – Withdrawn

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Severe Combined Immunodeficiency (SCID) Newborn Screening Implementation: Grantee Experience

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Impact of a Marketing Campaign to Raise Awareness of a Continuing Education Activity

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Results from the 2014 APHL Newborn Screening Timeliness Survey

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Homogeneous Single-point PCR Detection of TREC and Beta-Actin DNA in Dried Blood Spot Samples

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Results of Prototype Automated Creatine Kinase Muscle Isozyme Immunoassay for Potential Duchenne Muscular Dystrophy Identification

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Developing Improvements to a Non-derivatized Mass Spectrometry Method

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Improvement in the Extraction Efficiency of Succinylacetone

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Systematic Stability Test of Markers of Inborn Errors of Metabolism (IEM) in Dried Blood Spots

D. Shah[1], L. Goncalves[1], J. DiPerna[2], A. Grushecky[2], M. Schermer[1]; [1]PerkinElmer, Waltham, MA, [2]PerkinElmer Genetics, Bridgeville, PA

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Throughput Optimization of Two NBS Assays on a Single MS Analysis Platform

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Long-term Stability of ABG, ASM, GAA, GALC, GLA and IDUA in Frozen DBS

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Contrived Enzyme-specific Deficiency in Dried Blood Spots

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Using an Amino Acid and Acylcarnitines Metabolic Profile as a Potential Tool for Premature Baby Assessment

D. Chace[1], A. Spitzer[1], R. Clark[1], D. Shah[2], M. Schermer[2], M. Kuracina[2]; [1]Pediatrix Medical Group, Sunrise, FL, [2]PerkinElmer, Waltham, MA

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A Novel Five-Plex Real-Time PCR Assay that Detects SMN1, SMN2, TREC, KREC, and RPP30

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Evaluation of an Enzymatic Method for Screening of Biotinidase Deficiency from Newborns

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New NeoMass™ AAAC Kit for Expanded Newborn Screening of Multiple Metabolic Disorders by LC-MS/MS

M. Halme[1], N. Kivi[1], J. Wakkinen[1], R. Fingerhut[2], G. Carrad[1]; [1]Labsystems Diagnostics Oy, Vantaa, Finland, [2]University Children's Hospital, Zurich, Switzerland

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The State of Newborn Screening for Hereditary Tyrosinemia Type 1 (HT-1) in the United States

M. Yeh[1], P. Hillan[2], T. Hendirkson[2]; [1]Sobi North America, Waltham, MA, [2]Sixsense Strategy Group, Toronto, ON, Canada

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ELSI Advantage- A Technology Solution for Ethical, Legal, and Social Issues in Newborn Screening

V. High[1], I. Butler[2]; [15]AM Solutions, Inc./NBSTRN, Rockville, MD, [2]ACMG, Bethesda, MD

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Improve Newborn Screening Pre-Analytical Timeliness with Sample-Tracking Software

H. MacIntosh, STACS DNA, Ottawa, ON, Canada

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Treatable Causes of Intellectual Disability: An Indian Newborn Screening Experience

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Presenter: Sudhesna Mohapatra, MD, Neogen Labs, Lingarajapuram, Bangalore, 560084, India, Email: sudhesna.moh@gmail.com

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Benefits of a Comprehensive NextGen Sequencing (NGS) Panel for Supplemental Newborn Screening and Confirmatory Testing: Surprise Findings beyond the Primary Diagnosis!

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Poster Abstracts

P-100 – Withdrawn

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The LC-MS/MS Assay of Leukocyte Acid α-Glucosidase Activity Reliably Differentiates Early-onset and Late-onset Pompe Disease

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Comparison of Tandem Mass Spectrometry and Digital Microfluidics Fluorimetry for Newborn Screening of Lysosomal Storage Diseases: Results of Large Scale Pilot Studies

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Notes

Symposia History

National Neonatal Screening Symposium (1981-1999)

Newborn Screening & Genetic Testing Symposium (2001 onward)

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

Future APHL Meetings

2016 APHL Annual Meeting and Tenth Government Environmental Conference

Albuquerque, NM

June 6-9, 2016

2017 APHL Newborn Screening & Genetic Testing Symposium

New Orleans, LA

September 10-13, 2017

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