FINAL PROGRAM

2014 APHL Newborn Screening and Genetic Testing Symposium
Newborn Screening: Re-Assessing Business as Usual

October 27–30, 2014
Hyatt Regency Orange County
Anaheim, CA

Sponsored by the Association of Public Health Laboratories
Cosponsored by the International Society for Neonatal Screening and the California State Public Health Laboratory

#nbsgts
www.aphl.org/2014nbsgts
AGENDA OF EVENTS

SUNDAY, OCTOBER 26
3:00 pm – 7:00 pm
Registration | Royal Ballroom Lobby

MONDAY, OCTOBER 27
7:00 am – 5:30 pm
Registration | Royal Ballroom Lobby

7:00 am – 8:30 am
Coffee | Royal Ballroom Lobby

7:00 am – 8:00 am
Roundtables

Making the NewSTEPs Repository Data Work for You | Garden 3

NewSTEPs is designed to improve programmatic outcomes and the overall quality of newborn screening systems by providing the necessary tools and skills to implement quality improvement. Participants will have the opportunity to see the functionality of the NewSTEPs Data Repository.

Yvonne Kellar-Guenther, PhD, University of Colorado, Denver

Roundtable Discussion of Short-Term Follow-Up Processes in Newborn Screening | Garden 4

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.

Carol Johnson, University of Iowa Hospitals and Clinics
Day One
588-848-14, 5.5 contact hours

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

- Describe the historical and future directions of newborn screening from the laboratory perspective and the clinical perspective
- Name and describe two main challenges in newborn screening laboratory methods
- Describe new research findings relating to improved clinical outcomes, and morbidity and mortality for infants identified by newborn screening
- Summarize DNA extraction methods and ways to improve efficiency

8:30 am – 10:00 am
Welcome and Keynote Session | Royal Ballroom

8:30 am – 8:45 am
Welcome to Orange County and California

Moderator: Leslie Gaffney, California Department of Public Health

- Cheryl Hermerath, MBA, DLM(ASCP), RR(NRCM), Oregon State Public Health Laboratory
- Leslie Gaffney, California Department of Public Health
- Jelili Ojodu, MPH, Association of Public Health Laboratories

8:45 am – 10:00 am
Keynote Session

Newborn Screening: Where We’ve Been and Where We Think We’re Going

This session will provide a summary of the history of newborn screening including laboratory and clinical perspectives, as well as a look to future directions of newborn screening.

Moderator: Cheryl Hermerath, MBA, DLM(ASCP), RR(NRCM), Oregon State Public Health Laboratory

- Edward R.B. McCabe, MD, PhD, Senior Vice-President and Medical Director, March of Dimes
- Michael Glass (retired), MS, Director, Office of Newborn Screening, Washington State Public Health Laboratories

10:00 am – 10:30 am
Break | Royal Ballroom Lobby
**10:30 am – 12:00 pm**  
**Session 1 – Laboratory Updates** | Royal Ballroom  

*This session will focus on methods and validation techniques used for newborn screening for Primary Immune Deficiency Syndromes (PIDS) and Lysosomal Storage Disorders (LSDs).*

**Moderators:** Victor De Jesus, PhD, Centers for Disease Control and Prevention and Cheryl Hermerath, MBA, DLM(ASCP), RR(NRCM), Oregon State Public Health Laboratory

- Newborn Screening of Premature Infants: Clinical Trial Data Suggests Gestational Age and Chronological Age Are Key to Interpretation  
  Donald Chace, PhD, Pediatrix Medical Group

- IRT Cut-off Levels Related to Age of Sampling  
  Toni Torresani, PhD, Swiss Newborn Screening Laboratory

  Heather Wood, MS, Michigan Department of Community Health

- Multiplex Mass Spectrometric Newborn Screening and Diagnosis of Lysosomal Storage Diseases  
  Michael Gelb, PhD, University of Washington

- A Five-Minute Extraction Protocol for MSMS: STAT Reporting of Medical Emergency MSMS Profiles  
  Mary Seeterlin, PhD, Michigan Department of Community Health

**12:00 pm – 1:30 pm**  
**Lunch** (on your own)

**1:30 pm – 3:00 pm**  
**Session 2 – Clinical Outcomes** | Royal Ballroom

*As an increasing number of treatable disorders are added to newborn screening panels worldwide, clinical outcomes for these infants continue to improve as does follow-up programs responsible for early detection. This session will provide an overview of the recent improvements made in clinical outcomes with reports from several states.*

**Moderators:** Erica Wright, MS, CGC, Children’s Hospital of Colorado and Neena Champaigne, MD, Greenwood Genetic Center
SCID Secondary Targets Identified by the Texas Newborn Screening Program
Debra Freedenberg, MD, PhD, Texas Department of State Health Services

Homozygosity for a Carnitine Palmitoyltransferase 1A Genetic Variant Is Associated with an Increased Risk for Infant Mortality: Implications for Newborn Screening
David Koeller, MD, Oregon Health & Science University

VLCADD Pacific Island Mutation? A Study of Hawaii Newborn Screening Dried Blood Spots
Kirsty McWalter, MS, CGC, Hawaii Department of Health

Assuring Access to Nutritional Treatment for Inborn Errors of Metabolism: Michigan’s Diet for Life Work Group Process and Experience
Janice Bach, MS, Michigan Department of Community Health

3:00 pm – 6:30 pm
Exhibit Hall Open | Grand Ballroom
Posters Available for Viewing

3:00 pm – 3:30 pm
Break in the Exhibit Hall | Grand Ballroom

3:30 pm – 5:00 pm
Session 3 – Molecular Advances | Royal Ballroom

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) and other T-cell and B-cell disorders. Next generation sequencing will also be highlighted.

Moderators: Suzanne Cordovado, PhD, Centers for Diseases Control and Prevention and Michele Caggana, ScD, New York State Department of Health

Evaluation of Stored Newborn Screening Specimens From Children Diagnosed With Conditions That May Be Identifiable in the Newborn Period by Molecular Testing for Measures of T and B Cell Development
Anne Marie Comeau, PhD, New England Newborn Screening Program

Determination of TREC Copy Numbers From Dried Blood Spots Using Digital PCR
Travis Henry, PhD, State Hygienic Laboratory at the University of Iowa
A Multiplex Assay for Concurrent Newborn Screening of Spinal Muscular Atrophy (SMA) and Severe Combined Immunodeficiency (SCID)
Francis Lee, PhD, Centers for Disease Control and Prevention

Next Generation Sequencing of CFTR From Dried Blood Spots Using the ION Torrent PGM™
Miyono Hendrix, Centers for Disease Control and Prevention

NC NEXUS: North Carolina Newborn Exome Sequencing for Universal Screening Project
Cynthia Powell, MD, University of North Carolina, Chapel Hill

5:00 pm – 6:30 pm
Welcome Reception in the Exhibit Hall | Grand Ballroom
Visit the exhibitors and posters

5:30 pm – 6:00 pm
Poster authors available to answer questions | Grand Ballroom

6:30 pm – 8:00 pm
Short-term Follow-up Mixer | Garden 3
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 and John Thompson, PhD, Washington State Public Health Laboratories
**Industry Workshops**  October 28, 2014 – 7:00 am – 8:00 am

**PerkinElmer, Inc. | Garden 3**

**Partnering in a New Solution for Automated Newborn Hemoglobin Screening**

This workshop will outline the existing challenges in Hb testing for Newborn Screening labs. It will review a packaged solution to these challenges that is a result of a collaboration of Sebia (provider of capillary electrophoresis systems and analysis software) and PerkinElmer (provider of the Panthera-Puncher™ 9 Instrument and Specimen Gate® Software).

Speakers:  
**Aigars Brants**, PhD, Scientific Affairs Manager, Sebia, Inc.

**Justin Anderle**, Global Products Manager, PerkinElmer, Inc.

1.0 hour of P.A.C.E. credit is available for this workshop

**Thermo Fisher Scientific | Garden 4**

**Streamline Post-analytical Data Analysis of Tandem Mass Spectrometry for Inborn Error Metabolism Research**

- Overview of technology trend in the research of inborn error metabolism (IEM)
- Common errors in pre-analytical, analytical and post-analytical aspects for IEM research
- Introduction of tandem mass spectrometry in IEM research
- Challenge of handling huge quantities of data generated from TMS analysis
- Discuss research results of using a meta calculation software to streamline data processing of 480 donor samples, 9,120 analytes and 15,360 calculations.

Speaker:  
**Jason Lai**, PhD, Thermo Fisher Scientific
TUESDAY, OCTOBER 28
7:00 am – 5:30 pm
Registration | Royal Ballroom Lobby

7:00 am – 8:30 am
Coffee | Royal Ballroom Lobby

7:00 am – 8:00 am
Industry Workshops | Garden 3 & 4
• PerkinElmer
• Thermo Fisher Scientific

8:30 am – 10:00 am
Concurrent Sessions

Short-Term and Long-Term Follow-Up 1: Reassessing Business as Usual in Follow-up | Royal Ballroom CDEF
588-849-14, 1.5 contact hours

This workshop will be an interactive opportunity to explore some of the innovative ways of enhancing outcomes for babies with abnormal screening results. Educational efforts and patient management will be emphasized.

At the conclusion of this session, the participant will be able to:
• Describe issues that may arise when implementing change in follow-up programs.

Moderators: Carol Johnson, University of Iowa Hospitals and Clinics and Lisa Feuchtbaum, DrPH, California Department of Public Health

Evaluating and Implementing Change in Follow-Up
Sara Denniston, Oregon State Public Health Laboratory

The Controversy on Mild (Compensated) Congenital Hypothyroidism: The Path We Took to Resolve the Dilemma in Washington NBS
Patricia Fechner, MD, Seattle Children’s Hospital
Feasibility of Providing Long-Term Care and Follow-Up for Patients With Congenital Hypothyroidism by Primary Care Providers in California and Hawaii
Ning Rosenthal, MD, PhD, California Department of Public Health

Reducing Time From Referral to Treatment: Strengthening the Weak Links in the Newborn Screening Chain of Events

QA/QC 1: Strategies to Reduce False Positives and False Negatives (Part 1)
Royal Ballroom AB
588-850-14, 1.5 contact hours

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 false negatives. Participants will view a demonstration of an online tool to record laboratory-observed analyte interferences, and methods to reduce false negatives in Cystic Fibrosis (CF) screening will be discussed in detail.

At the conclusion of this session, the participant will be able to:
• List the reasons for maintaining an online interferences list and how to add or edit an entry.

Moderators: Fizza Gulamali-Majid, PhD, Maryland Department of Health and Mental Health and Joseph Orsini, PhD, New York State Department of Health

A New Comprehensive and Ongoing Newborn Screening Interference List Maintained by the APHL Quality Assurance/Quality Control Subcommittee
Patrick Hopkins, Missouri State Public Health Laboratory

Incidental G-6-PD Identification Using the PerkinElmer GSP Neonatal GALT Kit
Cheryl Hermerath, MBA, DLM(ASCP), RR(NRCM), Oregon State Public Health Laboratory

Cystic Fibrosis Screening: Attempts to Reduce False Negatives
Gary Hoffman, Wisconsin State Laboratory of Hygiene

Does IRT/IRT/DNA Really Work? Review of Cystic Fibrosis Newborn Screening in Texas
Rachel Lee, PhD, Texas Department of State Health Services

Establishing High Performance Analyte Cutoffs in Metabolic Disorders Screened by Mass Spectrometry Through Understanding of False Negative Risk
Konstantinos Petritis, PhD, Arizona State Public Health Laboratory
10:00 am – 5:00 pm
Exhibit Hall Open  |  Grand Ballroom
Posters Available for Viewing

10:00 am – 10:30 am
Break in the Exhibit Hall  |  Grand Ballroom

10:30 am – 12:00 pm
Concurrent Sessions

**Short-Term and Long-Term Follow-Up 2: Emerging Conditions (Get Ready!)**
Royal Ballroom CDEF
588-851-14, 1.5 contact hours
This workshop 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:
- Describe how to develop a framework for follow-up of Pompe Disease or X-ALD patients.


- **Low Citrulline in Newborn Screening Specimens: The Proximal Urea Cycle Defects and Beyond**
  Inderneel Sahai, MD, New England Newborn Screening Program

- **Missouri’s Full Population Pilot Screening for Fabry Disease and the Implications for Families**
  Jami Kiesling, BSN, Missouri State Department of Health & Senior Services

- **Developing a Newborn Screening Follow-Up Framework for Pompe Disease**
  Sarah Bradley, MS, New York State Department of Health

- **Developing Short- and Long-Term Follow-Up for X-linked Adrenoleukodystrophy**
  Beth Vogel, MS, New York State Department of Health

- **Diagnostic Follow-Up of 41 Infants With a Positive Newborn Screen for Hurler Syndrome (MPS-I): Identification of Four Recurrent IDUA Sequence Changes That Significantly Reduce Enzyme Activity**
  Laura Pollard, Greenwood Genetic Center
QA/QC 2: Strategies to Reduce False Positives and False Negatives (Part 2)
Royal Ballroom AB
588-852-14, 1.5 contact hours

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 false negatives. The Region 4 Post Analytical Tool will be discussed in detail, as well as the use of cutoffs to reduce false positive and negative results of various metabolic disorders.

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

• List ways in which states have improved their false negative and false positive rates in newborn screening
• Describe how the Region 4 Post Analytical Tool can be used to conduct quality improvement of follow-up strategies

Moderators: Patricia Hunt, Texas Department of State Health Services and Bob Currier, California Department of Public Health

Variant MSUD: A Strategy to Increase Detection Through Newborn Screening
Ashleigh Ragsdale, MPH, Washington State Public Health Laboratories

Analysis of False Positive and False Negative MSUD Cases: Using Age-Specific Cutoffs to Reduce Both
Mary Seeterlin, PhD, Michigan Department of Community Health

Quality Improvement of Follow-Up Strategies Using Region 4 Post-Analytical Tools to Evaluate VLCADD and CACT/CPTII Newborn Screening Results
Angela Wittenauer, BSN, Emory University

Retrospective Analysis of the Region 4 Post-Analytical Tool and Confirmatory Testing for Long Chain Fatty Acid Oxidation Disorders Screened in the State of Iowa
Alvaro Serrano Russi, MD, University of Iowa Hospitals and Clinics

The Compelling Benefits of Routine Second Newborn Screens: A Fifteen-Year Review in Washington State
Caroline Nucup-Villaruz, MD, Washington State Public Health Laboratories
12:00 pm – 1:30 pm
Lunch provided in the Exhibit Hall  |  Grand Ballroom
Visit the exhibitors and posters

12:30 pm – 1:00 pm
Poster authors available to answer questions  |  Grand Ballroom

Day Two Afternoon
588-853-14, 3.0 contact hours

At the conclusion of today, the participant will be able to:
• Review methods to conduct an economic evaluation of a SCID screening
• Discuss the impact recent changes to newborn screening legislation in Minnesota
• Discuss the role and impact of health information technology in CCHD screening and in improving short-term and long-term follow-up efficiency

1:30 pm – 3:00 pm
Session 4 – Financial, Legal, Ethical, Policy and Social Implications (FLEPSI)  |  Royal Ballroom

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: Mark McCann, Minnesota Public Health Laboratory Division and Susan Tanksley, PhD, Texas Department of State Health Services

Feasibility of a Population-Based Newborn Screening Study for Spinal Muscular Atrophy in Colorado
Joshua Miller, MPH, Colorado School of Public Health

Economic Evaluation for SCID Screening: Methods on Cost-Effectiveness Analysis and Budget Impact Analysis
Kee Chan, PhD, Boston University
Review of Best Practices in Documenting Newborn Screening Refusals for States
Jeremy Penn, PhD, North Dakota State University

Impact of Lawsuit Settlement and Recent Legislation to the Minnesota Newborn Screening Program
Carrie Wolf, MBS, Minnesota Department of Health

3:00 pm – 3:30 pm
Break in the Exhibit Hall | Grand Ballroom

3:30 pm – 5:00 pm
Session 5 – Health Information Technology | Royal Ballroom

Electronic data transfer is on the horizon for all newborn screening programs. This session will provide perspectives on implementing data transfer as well as an overview of Ontario’s experience using the Better Outcomes and Registry Network (BORN).

Moderators: Robin Thomas, BSN, MPA, California Department of Public Health and Patricia Scott, MT(BT), Delaware Public Health Laboratory

Newborn Screening for CCHD: The First Six Months of Data Reporting in Michigan Using Multiple Electronic Options
Kristy Tomasko, Michigan Department of Community Health

CCHD Screening: Screening Interpretation and Data Sharing Between Providers and Public Health to Improve Outcomes
Annamarie Saarinen, MA, Newborn Coalition
Lura Daussat, MPH, OZ Systems

Improving Short- and Long-Term Follow-Up Efficiency Through Implementation of an Internet Case Management System
Beth Vogel, MS, New York State Department of Health

Performance Feedback and Proactive Alerts From the BORN Ontario Registry Have Contributed to More Comprehensive and Timely Newborn Screening
Jennifer Milburn, MHA, Newborn Screening Ontario, Canada

NewSTEPs Data Repository: A Resource for the Newborn Screening Community
Careema Yusuf, MPH, Association of Public Health Laboratories
5:30 pm – 6:30 pm

Meet the Manufacturers Session | Grand Ballroom

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

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<tr>
<th>Time</th>
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<tr>
<td>5:30 pm</td>
<td>Illumina, Inc.</td>
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<td>Cambridge Isotope Laboratories, Inc.</td>
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<td>Astoria-Pacific, Inc.</td>
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6:30 pm – 8:00 pm

Concurrent Sessions

Genomic Sequencing and Newborn Screening Disorders | Royal Ballroom

In 2013, the National Human Genome Research Institute and the *Eunice Kennedy Shiver* National Institute of Child Health Development funded four grantees (Brigham and Women’s and Boston Children’s Hospitals, Boston, University of North Carolina, Chapel Hill, Mercy Children’s Hospital, Kansas City and University of California, San Francisco) to investigate whether sequencing of newborns’ genomes can provide useful medical information beyond current newborn screening practices. The projects at each site were required to investigate technical, clinical and ethical aspects of genome sequencing in the newborn period.

*This session will provide an overview and rationale from each of the four studies, study designs and a current status report.*

**Moderator:** Richard B. Parad, MD, MPH, Department of Pediatric Newborn Medicine, Brigham and Women’s Hospital, Boston, MA

**Speakers:**
- Richard B. Parad, MD, MPH, Department of Pediatric Newborn Medicine, Brigham and Women’s Hospital, Boston, MA
- Cynthia Powell, MD, Department of Pediatrics, University of North Carolina, Chapel Hill, Chapel Hill, NC
- Laurie Smith, MD, PhD, Department of Pediatrics, Children’s Mercy Hospital, Kansas City, MO
- Robert Currier, PhD, Genetic Disease Screening Program, California Department of Public Health
Current Status of Critical Congenital Heart Disease Screening in the United States | Garden 3

Hot topics including state-by-state implementation of screening across the U.S., examples of data collection involving public health departments, NICU populations, and fee collection practices by newborn screening programs; also, other cost issues will be discussed during this session.

Moderators: Amy Gaviglio, MS, CGC, Minnesota Public Health Laboratory Division and Richard Olney, MD, MPH, Centers for Disease Control & Prevention

Free CEs for Laboratory and Health Professionals Involved in Newborn Screening

**HOW?**

1. **Read** MMWR Recommendation and Reports: Good Laboratory Practices for Biochemical Genetic Testing and Newborn Screening for Inherited Metabolic Disorders

2. **Take and pass a test** to document knowledge gained and earn your CEs!

For more information, go to www.cdc.gov/TCEOnline and search for course number WB2010.

This announcement was supported by Cooperative Agreement # U60HM000803 funded by the Centers for Disease Control and Prevention (CDC). The language used in this announcement is solely the responsibility of the authors and does not necessarily represent the official views of CDC or the DHHS.
WEDNESDAY, OCTOBER 29

7:00 am – 5:30 pm

Registration | Royal Ballroom Lobby

7:00 am – 8:30 am
Coffee | Royal Ballroom Lobby

7:00 am – 8:00 am

Roundtables

Cost-Effectiveness of Screening for Severe Combined Immunodeficiency (SCID) | Garden 3

This roundtable will present a user-friendly spreadsheet model that allows states to specify their own parameters and to estimate the cost effectiveness of universal newborn screening for SCID and, in addition, the expected budget impact and financial Return on Investment (ROI) for state Medicaid programs and other healthcare payers.

- Yao Ding, MSc, Association of Public Health Laboratories

Supplemental Findings From Newborn Screening—Where Do We Stand?

Garden 4

This roundtable will present findings from a 50-state survey of how newborn screening programs report supplemental or incidental findings during routine newborn screening.

- Cynthia F. Hinton, PhD, MS, MPH, Centers for Disease Control and Prevention
- Patrick Hopkins, Missouri State Public Health Laboratory
- Kimberly Psaltis RN, CDE, University of Colorado School of Public Health
- Marci Sontag, PhD, University of Colorado, Denver

Day Three

588-854-14, 6.0 contact hours

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

- Explain parent perspectives on the storage and use of residual blood spots
- Share parents’ concerns and questions about their child’s treatment with laboratory/follow-up staff to encourage solutions
- Discuss the feasibility of implementing methods for screening new markers
- Describe methods to improve CCHD detection by exploring false negative results
8:30 am – 10:00 am

**Session 6 – Teach and Preach: Education Initiatives** | Royal Ballroom

*Many state programs are dealing with educational issues surrounding parental consent for 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 these issues and will provide an overview of NewSTEPs’ second year of activities.*

**Moderators:** Natasha Bonhomme, Genetic Alliance and Amy Gaviglio, MS, CGC, Minnesota Public Health Laboratory Division

*The State of Newborn Screening Systems in the US*
Marci Sontag, PhD, University of Colorado, Denver

*What Parents Want to Know About the Storage and Use of Residual Newborn Bloodspots*
Erin Rothwell, PhD, The University of Utah

*Developing and Maintaining Parent Fact Sheets for Newborn Screening: A Decade of Multi-State Collaboration*
Sylvia Mann, MS, Hawaii Department of Health

*A Comprehensive Resource for US Newborn Screening Programs: NewSTEPs Year Two*
Sikha Singh, MHS, PMP, Association of Public Health Laboratories

10:00 am – 4:00 pm

**Exhibit Hall Open** | Grand Ballroom

Visit the exhibitors and posters

10:00 am – 10:30 am

**Break in the Exhibit Hall** | Grand Ballroom

10:30 am – 12:00 pm

**Session 7 – Newborn Screening: Personal Experiences Parent/Patient Panel** | Royal Ballroom

*This session will feature families affected by genetic disorders. They will present their stories. The disorders represented come from the Recommended Uniform Screening Panel (RUSP), secondary targets, and disorders under investigation.*
**Moderators:** Leslie Gaffney, California Department of Public Health and Jelili Ojodu, MPH, Association of Public Health Laboratories

- Jordann Coleman (parent of child with MSUD)
- Idario Santos (parent of child with MSUD)
- Korissa Olson (parent of child with Galactosemia)
- Brianna Capers (patient with sickle cell disease)

**12:00 pm – 1:30 pm**

**Awards Luncheon | Grand Ballroom**

**Moderators:** Joanne Mei, PhD, Centers for Disease Control and Prevention/International Society for Neonatal Screening and Scott Becker, MS, Association of Public Health Laboratories

**1:30 pm – 3:00 pm**

**Session 8 – Candidate Conditions | Royal Ballroom**

This session will discuss new methods currently being developed for the detection of secondary markers. Feasibility of adding candidate conditions will also be discussed.

**Moderators:** Joan Scott, MS, CGC, Health Resources and Services Administration and Scott Shone, PhD, New Jersey Division of Public Health & Environmental Laboratories

**New York’s Experience: Summary of the First Four Months of ALD Screening**
Joseph Orsini, PhD, New York State Department of Health

**Missouri’s Experience With Full Population Pilot Screening for Pompe, Gaucher, Fabry and Hurler Disorders Using Digital Microfluidics Methodology**
Patrick Hopkins, Missouri State Public Health Laboratory

**Enzymatic Assays for Newborn Screening of Mucopolysaccharidoses With Improved Performance**
Michael Gelb, PhD, University of Washington

**A Three-Year Pilot Study for Guanidinoacetate Methyltransferase (GAMT) Deficiency in British Columbia**
Graham Sinclair, PhD, University of British Columbia, Canada
3:00 pm – 4:00 pm
Break in the Exhibit Hall with raffle drawing (3:30 pm) | Grand Ballroom

4:00 pm – 5:30 pm
**Session 9 – Point of Care for Critical Congenital Heart Disease (CCHD)** | Royal Ballroom

*With the recent inclusion of CCHD (Critical Congenital Heart Disease), many programs in the US are grappling with issues on how to best implement CCHD screening into their programs. This session will provide guidance from several perspectives.*

Moderators: Julie Luedtke, Nebraska Department of Health and Human Services and Brad Therrell, PhD, National Newborn Screening & Global Resource Center

**Implementing Universal Pulse Oximetry Screening: From Pilot to Policy to Practice**
Amy Gaviglio, MS, CGC, Minnesota Public Health Laboratory Division

**CCHD Screening in Maryland—Year 1 Results**
Debbie Badawi, MD, Maryland Department of Health and Mental Hygiene

**Exploring False Negative Pulse Oximetry Screens to Improve Critical Congenital Heart Disease Detection**
Lisa Hom, RN, Children’s National Medical Center

**Modification of Critical Congenital Heart Disease Screening Practices at Moderate Altitude**
Marci Sontag, PhD, University of Colorado, Denver

**Critical Congenital Heart Disease Resource Center**
Thalia Wood, MPH, Association of Public Health Laboratories

6:00 pm – 10:00 pm
**Evening Social – Haunted Halloween event (wear a costume!)**
Sponsored by PerkinElmer Inc.
THURSDAY, OCTOBER 30

7:30 am – 12:30 pm

Registration | Royal Ballroom Lobby

7:00 am – 8:30 am

Coffee | Royal Ballroom Lobby

7:00 am – 8:00 am

ISNS Membership Meeting | Salon 1

Day Four

588-855-14, 3.0 contact hours

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

• Explain how other countries’ newborn screening programs differ from those in the U.S.
• Discuss why newborn screening must occur in a timely manner to be effective
• Describe one state’s newborn screening timeliness improvements

8:30 am – 10:00 am

Session 10 – International Perspectives | Royal Ballroom

This session will showcase international newborn screening efforts underway in Palestine, the Middle East, Mexico and India, as well as an overview of global implementation efforts for CCHD and newborn screening in general.

Moderators: Joanne Mei, PhD, Centers for Disease Control and Prevention and Issam Khneisser, MS, MBA, St. Joseph University, Lebanon

A Pilot Study on an Expanded Newborn Screening Program in Palestine, Phase II
Samir Khatib, PhD, Al-Quds University Medical School, Palestine

Challenges in Implementation of Newborn Screening Programs in the Middle East: An Initiative to Create a Central Institute of Newborn Screening Regulating NBS Programs Across the Region
Jamal Golbahar, PhD, Al-Jawhara Centre for Genetic Diagnostics and Research, Bahrain
Congenital Hypothyroidism Detected In Newborn Screening: Three Years of Data From a Tertiary Care Centre in India
Sumaira Khalil, DNB, Maulana Azad Medical College & Associated L N Hospital, India

First Experience With a Fully Automated Analyzer System in the Newborn Screening Central Laboratory of the Ministry of Public Health in Mexico
Erika Paola Garcia Flores, MD, Ministry of Health, Mexico

Screening for Critical Congenital Heart Disease: Global Implementation Efforts
Gerard Martin, MD, Children’s National Medical Center

Further Expansion of Newborn Screening in European Countries: Horse Carriages Versus Bullet Trains
Gerard Loeber, PhD, International Society for Neonatal Screening

10:00 am – 10:30 am
Break | Royal Ballroom Lobby

10:30 am – 12:00 pm
Session 11 – No Time to Lose:
Re-assessing Timeliness | Royal Ballroom

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

Moderators: Carla Cuthbert, PhD, Centers for Disease Control and Prevention and Patrice Held, PhD, Wisconsin State Laboratory of Hygiene

Timeliness of Newborn Screening: Secretary’s Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) Guidance to Newborn Screening Systems
Susan Tanksley, PhD, Texas Department of State Health Services

On Time/Every Time: A Partnership of Safety and Reliability for Newborn Screening
Neena Champaigne, MD, Greenwood Genetic Center

Improving Newborn Screening Program: A Systematic Approach
Mei Baker, MD, Wisconsin State Laboratory of Hygiene
Achieving Dramatic Improvements in Sample Transit Time — The Arizona Model  
Celia Nabor, MPA, Arizona Department of Health Services

John Thompson, PhD, Washington State Public Health Laboratories

Leveraging the Attention — How Texas Took Advantage of Heightened National Awareness to Expedite Ongoing Quality Improvement Efforts  
Brendan Reilly, Texas Department of State Health Services

Improving Quality Indicators Associated With Specimen Collection and Transport  
Scott Shone, PhD, New Jersey Division of Public Health and Environmental Laboratories

12:00 pm  
Adjournment

1:00 pm – 3:30 pm  
Optional Tour of the Long Beach Newborn Screening Laboratory  
Meet in the hotel lobby by 12:45 pm. The bus will leave promptly at 1:00 pm. Pre-registration required.

See you in 2016  
Newborn Screening & Genetic Testing Symposium  
February 29 – March 3, 2016  
St. Louis, MO