2017 APHL Newborn Screening and Genetic Testing Symposium

September 10 – 13, 2017
New Orleans, LA
About This Conference

The 2017 APHL Newborn Screening & Genetic Testing Symposium (NBSGTS) will address state, national and international newborn screening, genetic testing and policy issues important to public health newborn screening systems. The meeting will emphasize reports from around the globe, the challenges they face, and the data they have generated, as done in previous years. Topics include molecular technologies, current and upcoming conditions, quality improvement, communicating with families and the public and short and long-term follow-up.

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

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

The major learning goals of this symposium are:

• Discuss and evaluate quality assurance and quality control measures for newborn screening laboratories
• 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, students, health care practitioners or other maternal and child health service providers, public health nurses, specialists, public health laboratory directors, and other healthcare professionals involved with newborn screening & genetic testing issues and follow-up.

General Information

The 2017 APHL NBSGTS begins with two roundtables at 7:30am, Sunday, September 10 followed by the Welcome Session at 8:30am.

Posters will be displayed from Sunday afternoon, September 10 through 4:00pm on Tuesday, September 12. Dedicated viewing times (with authors present) will be during Sunday’s Welcome Reception from 6:00pm-6:30pm and during Tuesday’s lunch from 12:30pm-1:00pm.

The Exhibit Hall is open Sunday, September 10 from 3:30pm-7:00pm, Monday, September 11 from 10:00am-4:30pm and Tuesday, September 12 from 10:00am-4:00pm.

All general sessions will take place in the Grand Ballroom. The exhibit hall, posters, breaks and lunch will take place in the Grand Salon just across the hall.
Optional Tour of the Louisiana State Newborn Screening Laboratory

An optional tour of the Louisiana State Newborn Screening Laboratory has been arranged for Wednesday afternoon, September 13. The lab is in Baton Rouge which is a 90 minute drive from New Orleans. The tour will run from 12:30pm-7:00pm with lunch provided. You must sign up for this tour at the same time as you register for the symposium. Space is limited.

Registration

Advance registration through APHL is required.

Early registration (until August 7, 2017) is $500. After that date, registration is $550.

If you are paying by credit card you may register online by clicking the “Online Registration” link on the APHL NBSGTS conference page. If you are paying for your registration by check you may click the “Registration Form” link and download and mail or fax the completed form to APHL.

The 2017 APHL NBSGTS conference page, with more details, is [www.aphl.org/nbsgts](http://www.aphl.org/nbsgts).

Hotel

The 2017 APHL NBSGTS will be held at the Hilton New Orleans Riverside Hotel at Two Poydras St., New Orleans, LA 70103, 504.561.0500. The hotel is located in downtown New Orleans right on the Mississippi River. It is just steps from the French Quarter with easy access to numerous restaurants and shops.

APHL has negotiated a block of rooms split between $150 and the at the 2017 federal per diem rate, currently $128, single/double per night plus tax. These rates will be filled on a first come, first served basis.

To make your reservation, please call the hotel at 800.774.1500 or 504.584.3999 and provide the code “PHL” for the conference rate and “PHLG” for the per diem rate. You may also make your reservation online. These rates are valid until August 8, 2017 or until the block is filled. This room block may fill up early so be sure to register as soon as possible. Rooms cancelled less than 72 hours before arrival will be subject to a one night cancellation fee.

Reservation Online

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 23.5 P.A.C.E.® credits by attending the entire conference. CPH recertification credits will be available. Florida, nursing and genetic counseling continuing education units (CEUs) and continuing medical education (CMEs) are also being applied for.
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.

Conference 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

The 2017 NBSGTS app will be available on the iPhone and Android platforms. Look for it at the end of August in the Apple and Google Play stores!
Visit These Exhibitors in the Exhibit Hall
(As of July 7, 2017)

GOLD CIRCLE:
• PerkinElmer

SILVER CIRCLE:
• Baebies

BRONZE CIRCLE:
• Astoria-Pacific, Inc.  • EBF, Inc.  • Microelectronic Systems
• Bio-Rad Laboratories  • Illumina, Inc.  • OZ Systems
• Cambridge Isotope Laboratories  • Integrated Software Solutions  • Quantabio
• Clinical and Laboratory Standards Institute  • Luminex Corporation  • Waters Corporation

EXHIBITORS:
• Baby’s First Test  • Greenwood Genetic Center  • Newborn Screening Translational Research Network/National Coordinating Center for the Regional Genetics Networks

Thanks to Our Sponsors!

PerkinElmer
  Tote bags, hotel key cards, general conference support

Baebies
  Pad folios

Astoria-Pacific, Inc.
  Welcome Reception bar
SATURDAY, SEPTEMBER 9
3:00 pm – 7:00 pm
Registration
1st Floor Registration

SUNDAY, SEPTEMBER 10
7:00 am – 5:45 pm
Registration
1st Floor Registration

7:00 am – 9:00 am
Coffee
Chemin Royale

7:30 am – 8:15 am
Roundtables
Navigating the Legal, Policy, and Ethical Landscape of Adding Conditions to the State Newborn Screening Panel
Grand Ballroom A
• Kimberly Piper, RN, BSN, CPH, CPHG, Iowa Department of Public Health

Working with Advocates: Breaking Down Barriers and Building Bridges
Grand Ballroom B
• Amy Gaviglio, MS, CGC, Minnesota Public Health Laboratory Division

8:30am – 9:00am
Welcome Session
Grand Ballroom CD
Moderators: Stephen J. Martin, PhD, Louisiana Public Health Laboratory
Patrick Hopkins, Missouri State Public Health Laboratory
9:00am – 10:30am

Session 1 - Current Recommended Uniform Screening Panel (RUSP) Conditions in State Newborn Screening Panels

Grand Ballroom CD
Moderators: Joanne Mei, PhD, Centers for Disease Control and Prevention
            Eric Blank, DrPH, Association of Public Health Laboratories

Detecting and Reporting Alpha Thalassemia in Newborns
• Tim Davis, Washington State Public Health Laboratory

Connecticut’s Experience with Validation and Screening for X-linked Adrenoleukodystrophy Using a Negative Ion-Mode LC-MS/MS Analysis Method
• Adrienne Manning, Connecticut Department of Public Health Laboratory

Newborn Screening for Pompe Disease in New York State Identifies a Wide Spectrum of Variants in the GAA Gene
• Colleen Stevens, PhD, New York State Department of Health

Newborns with Hypoxic Ischemic Encephalopathy Treated with Therapeutic Hypothermia Have Elevated TSH Levels on Newborn Screening
• Richard Parad, MD, MPH, Brigham and Women’s Hospital

10:30am – 11:00am

Break
Chemin Royale

11:00am – 12:30pm

Session 2 – Quality Improvement & Assurance Activities in Newborn Screening

Grand Ballroom CD
Moderators: Patrick Hopkins, Missouri State Public Health Laboratory
            Patricia Hunt, Texas Department of State Health Services

Implementation of a New Electronic Shipping System to Improve Timeliness
• Scott Shone, PhD, RTI International

Infographics and Outreach Used to Improve Newborn Screening Timeliness
• Ashley Comer, State Hygienic Laboratory at the University of Iowa
SCID Screening and Diagnostic Uncertainty: The California Experience Since 2010  
• Stanley Sciortino, PhD, California Department of Public Health

Analyzing Patterns in NewSTEPs Site Review Recommendations: The Big Picture for Newborn Screening Programs  
• Yvonne Kellar-Guenther, PhD, University of Colorado, Denver

The Relationship Between Newborn Screening Unsatisfactory Specimens and Unsatisfactory Results  
• Miriam Schachter, PhD, New Jersey Department of Health

12:30pm – 2:00pm  
Lunch (on your own)

2:00pm – 3:30pm  
Keynote Session - Panel Discussion  
Grand Ballroom CD  
The Big Picture for Population Screening: Lessons from Leaders  
Moderator: Susan Tanksley, PhD, Texas Department of State Health Services  
• Joseph Bocchini, Jr., MD, Louisiana State University Health Center  
• Alex Kemper, MD, MPH, MS, Nationwide Children’s Hospital

3:30pm – 4:00pm  
Break in the Exhibit Hall  
Grand Salon

3:30pm – 7:00pm  
Exhibit Hall Open  
Grand Salon  
Posters Available for Viewing
4:00pm – 5:30pm
**Session 3 – Past, Present & Future Newborn Screening Conditions**
Grand Ballroom CD
Moderators: Rodney Howell, MD, University of Miami
Michele Caggana, ScD, FACMG, New York State Department of Health

**Removing Short-chain Acyl-CoA Dehydrogenase Deficiency and Isobutyryl-CoA Dehydrogenase Deficiency from the Newborn Screening Panel: Michigan’s Experience**
• Kristy Karasinski, MPH, Michigan Department of Health and Human Services

**Development of a Multiplex Real-time PCR Newborn Screening Assay to Simultaneously Identify Spinal Muscular Atrophy, Severe Combined Immunodeficiency, and X-linked Agammaglobulinemia**
• Mei Baker, MD, FACMG, Wisconsin State Laboratory of Hygiene

**Implementation of Newborn Screening for Duchenne Muscular Dystrophy (DMD)**
• Michele Lloyd-Puryear, MD, PhD, Parent Project Muscular Dystrophy

**Congenital CMV – A Pilot study**
• Maggie Dreon, MS, CGC, Minnesota Public Health Laboratory Division

5:30pm – 7:00pm
**Welcome Reception in the Exhibit Hall**
Grand Salon

6:00pm – 6:30pm
**Poster authors available to discuss their posters**
Grand Salon

7:30pm – 9:00pm
**Short-term Follow-up Mixer**
Grand Ballroom A
MONDAY, SEPTEMBER 11

7:00am – 5:45pm  
**Registration**  
1st Floor Registration

7:00am – 8:00am  
**Coffee**  
Marlborough Foyer

7:30am – 8:15am  
**Innovate!**  
(formerly Industry Workshops) – 4 vendor presentations

8:00am – 9:00am  
**Coffee**  
Chemin Royale

8:30am – 10:00am  
**Joint Follow-up and Quality Assurance/Quality Control Session – Cut-offs**  
Grand Ballroom CD  
**Moderators:** Adrienne Manning, Connecticut Department of Public Health Laboratory  
Mike Ramirez, State Hygienic Laboratory at the University of Iowa

- A False Negative CPTII Case: Using the (C16+C18:1)/C2 Ratio to Improve both FN & FP Metrics  
  • Mary Seeterlin, PhD, Michigan Department of Health & Human Services

- Impact of Post-analytical Interpretive Tools on Newborn Screening for Three Lysosomal Disorders: First Year Prospective Experience in Kentucky  
  • Dietrich Matern, MD, PhD, Mayo Clinic Minnesota

- Comparison of Traditional Fixed Cutoffs to Use of Collaborative Laboratory Integrated Reports (CLIR) Tools in Screening for Pompe Disease and Krabbe Disease  
  • Joseph Orsini, PhD, New York Department of Health
North Carolina Mucopolysaccharidosis Type I (MPS I) Pilot Study: Screening for a Single Lysosomal Storage Disorder
• Jennifer Taylor, PhD, RTI International

Median Normalization of Newborn Screening Analyte Data to Improve Screening Performance
• Travis Henry, PhD, State Hygienic Laboratory at the University of Iowa

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

10:00am – 4:30pm
Exhibit Hall Open
Grand Salon
Posters Available for Viewing

10:30am – 12:00pm
Joint Follow-up and Quality Assurance/Quality Control Session – Timeliness in Newborn Screening
Grand Ballroom CD
Moderators: Inderneel Sahai, MD, New England Newborn Screening Program
Sara Denniston, Oregon State Public Health Laboratory

Partnering with a University Data Science Department to Develop Innovative Tools for the Analysis and Reporting of Newborn Screening Data
• Rhonda West, Virginia Division of Consolidated Laboratory Services

It’s Time to be On Time
• Sulay Rivera, PhD, MS, Puerto Rico Newborn Screening Program

The Impossible is Possible: Improving Timeliness in Alaska’s Newborn Bloodspot Screening
• Sabra Anckner, Alaska State Division of Public Health

Towards Improved Timeliness, Accuracy, and Operational Efficiency: Utah Newborn Screening Program’s Journey
• Andreas Rohrwasser, PhD, MBA, Utah Department of Health
CQI Texas – Improving Timeliness through a Systematic Investigation of Process Workflows
• Brendan Reilly, Texas Department of State Health Services

12:00pm – 2:00pm
Awards Lunch and Ceremony
Grand Ballroom A

2:00pm – 3:30pm
Session 4 – Follow-up Activities in Newborn Screening
Grand Ballroom CD
Moderators: Carol Johnson, University of Iowa Hospitals and Clinics
John Thompson, PhD, MPH, MPA, Washington State Public Health Laboratories

When You Can’t Find a Baby and the Clock is Ticking: Partnering with Home Visiting Nurses to Improve Follow-up Outcomes
• Sondi Aponte, Arizona Bureau of State Laboratory Services

High Presumptive Positive Rates in Neonatal ICU Babies - The Texas Experience and How to Fix It!
• Lori Fitzgerald, MSN, Texas Department of State Health Services

North Carolina Mucopolysaccharidosis I (MPS I) Pilot Study: How Incorporating Sequencing Data Impacts Follow-up
• Lisa Gehtland, MD, RTI International

An Update on Newborn Screening for Adrenoleukodystrophy in New York State: A Review of Management Protocol Changes and Confirmed Cases
• Beth Vogel, MS, CGC, New York State Department of Health

Long-Term Follow-Up of Sickle Cell Disease in California
• Patricia McLendon, MPH, California Department of Public Health

3:30pm – 4:00pm
Break in the Exhibit Hall
Grand Salon
4:00pm – 5:30pm

**Session 5 - Financial, Legal, Ethical, Policy and Social Implications (FLEPSI)**

Grand Ballroom CD
Moderators: Richard Olney, MD, MPH, California Department of Public Health
Sue Berry, MD, University of Minnesota

- **Benefit-Cost Analysis for Adding Newborn Screening for X-linked Adrenoleukodystrophy**
  - John Thompson, PhD, MPH, MPA, Washington State Public Health Laboratory

- **Development of a State Condition-readiness Tool for Disorders Not on the Recommended Uniform Screening Panel**
  - Kristy Karasinski, MPH, Michigan Department of Health and Human Services

- **Updating the National Newborn Screening Contingency Plan to Facilitate Preparedness for Newborn Screening**
  - Scott Shone, PhD, RTI International

- **A Systematic Review of the Research Uses of Residual Newborn Screening Dried Blood Spots: A Scoping Protocol**
  - Erin Rothwell, PhD, CTRS, University of Utah

- **Equality and Equity in Newborn Screening**
  - Amy Gaviglio, MS, CGC, Minnesota Department of Health
  - Aaron Goldenberg, PhD, Case Western Reserve University

6:00pm – 7:15pm

**Meet the Manufacturers Session**

Grand Ballroom B

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

7:30pm – 9:00pm

**Follow-up forum from Gene Sequencing Meeting**

Grand Ballroom A
TUESDAY, SEPTEMBER 12

7:00am – 5:15pm

Registration
1st Floor Registration

7:00am – 9:00am

Coffee
Chemin Royale & Marlborough Foyer

7:30am – 8:15am

Roundtables

Is a Voluntary Panel of Non-RUSP Conditions a Viable Future for Newborn Screening?
Grand Ballroom A
• Don Bailey, PhD, RTI International

What’s Next? Follow Up on Best Practices from the Beyond the Bloodspot: Education and Engagement Summit
Grand Ballroom B
• Jackie Seisman, MPH, Genetic Alliance

Newborn Screening for Alpha Thalassemias
Marlborough AB
• M. Christine Dorley, MSP, BS, MT(ASCP), Tennessee Department of Health

8:30am – 10:00am

Session 6 – International Perspectives in Newborn Screening
Grand Ballroom CD
Moderators: Brad Therrell, PhD, National Newborn Screening & Global Research Center
Enzo Ranieri, PhD, Women’s & Children’s Hospital, Australia

Neonatal Screening of Duchenne Muscular Dystrophy In Zhengjiang, China
• Guilig Qian, Zhejiang University School of Medicine, China
**Session 7 – Communicating with Families and the Public**

**Grand Ballroom CD**

**Moderators:** Natasha Bonhomme, Genetic Alliance
Amy Gaviglio, MS, CGC, Minnesota Department of Health

**MinneStories: The Importance of Newborn Screening Storytelling**

• Sondra Rosendahl, MS, CGC, Minnesota Department of Health

**Improving Communication of Negative Newborn Screening Results**

• Whitney Thompson, University of Minnesota

**Assessing Parental Attitudes about Newborn Screening for Late Onset Disorders**

• Sylvia Mann, MS, CGC, Western States Genetic Services Collaborative
Redefining Public Engagement in Newborn Screening and its Impact on Education and Policy

- Aaron Goldenberg, PhD, Case Western Reserve University

12:00pm – 1:30pm
Box Lunch (in the Exhibit Hall)
Grand Salon
Visit the exhibitors and posters

12:30pm – 1:00pm
Poster authors available to discuss their posters
Grand Salon

1:30pm – 3:00pm
Session 8 - Parent/Patient Panel
Grand Ballroom CD
Moderators: Hans Andersson, MD, Hayward Genetics Center
Jackie Seisman, MPH, Genetic Alliance

- Speakers - TBD

3:00pm – 4:00pm
Break in the Exhibit Hall with raffle drawing at 3:30pm
Grand Salon

4:00pm – 5:00pm
Session 9 – Health Information Technology
Grand Ballroom CD
Moderators: Brendan Reilly, Texas Department of State Health Services
Willie Andrews, Virginia Division of Consolidated Laboratory Services

Understanding an HL7 Implementation Guide – A Primer on IT Nerd-dom for Newborn Screening Nerds
- Brendan Reilly, Texas Department of State Health Services
Challenges and Benefits of Implementing Electronic Messaging in Newborn Screening Laboratories
• Ashleigh Ragsdale, MPH, Washington State Public Health Laboratory

Baby’s First Message: Next Steps and Lessons Learned After Achieving Statewide Implementation of Electronic Demographics and Point-of-Care Result Reporting
• Amy Gaviglio, MS, CGC, Minnesota Department of Health

6:00pm – 10:00pm
Off-site Evening Social
Sponsored by PerkinElmer, Inc.
WEDNESDAY, SEPTEMBER 13

7:30am – 12:30pm

Registration
1st Floor Registration

7:00am – 9:00am

Coffee
Chemin Royale

7:30am – 8:30am

International Society for Neonatal Screening Membership Meeting
Marlborough B

8:30am – 10:00am

Session 10 – Molecular Technology in Newborn Screening
Grand Ballroom CD
Moderators: Travis Henry, PhD, State Hygienic Laboratory at the University of Iowa
Rachel Lee, PhD, Texas Department of State Health Services

A Targeted Next Generation Sequencing (tNGS) Screening Assay for Menkes Disease and its Implications for Primary DNA-based Newborn Screening
• Richard Parad, MD, MPH, Brigham and Women’s Hospital

Design and Validation of a 2nd Tier Next Generation Sequencing (NGS) Panel for Newborn Screening for Severe Combined Immunodeficiency Disease
• Colleen Stevens, PhD, New York State Newborn Screening Program

Implementing Next Generation Sequencing as a Third-Tier Newborn Screen for Cystic Fibrosis in New York State
• Denise Kay, PhD, New York State Department of Health

Challenges of Severe Combined Immunodeficiency Screening in a Two Screen State
• Adam Coleman, PhD, Maryland Laboratories Administration
10:00am – 10:30am
Break
Chemin Royale

10:30am – 12:00pm
Session 11 – Spinal Muscular Atrophy
Grand Ballroom CD
Moderators: Art Hagar, PhD, Georgia Public Health Laboratory
Colleen Stevens, PhD, New York State Department of Health

Spinal Muscular Atrophy Screening in New York State: Feasibility and Prospective Pilot Study
• Denise Kay, PhD, New York State Department of Health

Evaluation of Multiplexing Spinal Muscular Atrophy Identification with a Laboratory Developed Severe Combined Immunodeficiency Assay in Minnesota
• Carrie Wolf, MBS, Minnesota Department of Health

Newborn Screening for Spinal Muscular Atrophy: Current and Alternative Assay Methods
• Kristina Mercer, Centers for Disease Control and Prevention

Clinical Challenges of Providing Nusinersen for Spinal Muscular Atrophy and the Implications for Newborn Screening
• Jennifer Kwon, MD, MPH, FAAN, University of Rochester Medical Center

12:00pm
Adjournment

12:30pm – 7:00pm
Optional tour of the Louisiana State Newborn Screening Laboratory
(Pre-registration is required)

Please keep in mind that this schedule is a work in progress and is subject to change.