National Conversation:
Tandem Mass Spectrometry in Newborn Screening

February 5–6, 2015
International Salon 6–8
Marriott Marquis Hotel
Atlanta, GA

www.aphl.org/nbs
AGENDA

DAY 1

8:00–8:45 am  Breakfast Served (International Salon 6-8)

8:45–9:00 am  Welcome (C. Cuthbert, J. Ojodu)

9:00–9:30 am  Adoption of MS/MS Technology in newborn screening (W. Harry Hannon)
   • History (impetus for the shift)
   • Expansion of screening targets

9:30–10:00 am  Identification of laboratory challenges to MS/MS screening — overview (QA/QC Subcommittee — represented by A. Hagar)

10:00–10:15 am  Identification of laboratory challenges to MS/MS screening: Breakout group instructions (QA/QC Subcommittee — represented by V. De Jesús and A. Hagar)
   Breakout group discussion topics will be:
   • Missed case or delayed diagnosis?
   • Barriers to changing markers
   • Barriers to method selection
   • Cutoff reviews
   • Use of ratios / enhanced interpretation schemes
   • Quality assurance / Corrective and Preventive Action (CAPA) procedures

10:15–10:30 am  Break

10:30–11:15 am  Laboratory Challenges Breakout Groups — Discussion

11:15–12:30 pm  Laboratory Challenges Breakout Groups — Reports

12:30–1:30 pm  Lunch Served (International Salon 6-8)

1:30–3:00 pm  Enhancing specificity: the case for additional markers/methods for RUSP disorders
   A. The Issues
      1. Case review: missed cases and other disorders detected with MS/MS in Texas, with emphasis on Tyrosinemia Type 1 (P. Hunt)
      2. Clinical aspects of Tyrosinemia Type 1 (N. Champaigne)
   B. The (possible) solutions
      3. State experiences with second-tier tests (M. Pasquali, P. Hunt)
      4. SUAC addition: evidence review (D. Matern)

3:00–3:15 pm  Break

3:15–4:00 pm  Laboratory-Developed Tests (LDTs) and proposed FDA regulation (K. Kelm)

4:00–4:20 pm  Lysosomal Storage Disorders MS/MS reagent discussion — CDC Newborn Screening Translation Research Initiative (R. Vogt)

4:20–4:50 pm  Vendor discussion, part 1 — Cambridge Isotope Laboratories (W. Wood)

4:50–5:00 pm  Day 1 Wrap-up (V. De Jesús)
**AGENDA**

**DAY 2**

8:00–8:30 am  
Breakfast Served (International Salon 6-8)

8:30–8:45 am  
Overview of Day 1 (V. De Jesús)

8:45–9:15 am  
Vendor Discussion, part 2 — PerkinElmer (M. Kuracina)

9:15–10:15 am  
Quality assurance and quality improvement issues for MS/MS (V. De Jesús, S. Shone)

10:15–10:30 am  
Break

10:30–11:45 am  
MS/MS assays for additional disorders: State experiences  
1. Lysosomal Storage Disorders — (G. Dizikes – IL)  
2. X-linked Adrenoleukodystrophy — (J. Orsini – NY)

11:45–12:45 pm  
Lunch Served (International Salon 6-8)

12:45–2:00 pm  
Additional MS/MS Application in Newborn Screening (20 min per topic, 15 min Q&A)  
1. Hemoglobinopathies (C. Haynes)  
2. ADA – SCID (C. Haynes)  
3. GAMT (M. Pasquali)

2:00–2:30 pm  
Meeting Wrap-Up (V. De Jesús)

2:30 pm  
Meeting Adjourned

**SPEAKERS**

**Neena L. Champaigne, MD**  
Dr. Neena Champaigne is a board-certified pediatrician and clinical geneticist with additional certification in biochemical genetics. She joined the Greenwood Genetic Center (GGC) in 2008 and has served as director of the Center’s Metabolic Treatment Program since 2012 and the Medical Genetics Training Program since 2013. Her primary responsibilities include the diagnosis and management of children and adults with inborn errors of metabolism, as well as providing newborn screening short-term and long-term follow-up for the state of South Carolina. In addition, Dr. Champaigne has served as a member of the APHL Newborn Screening Subcommittee on Quality Assurance/Quality Control.

**Carla Cuthbert, PhD, FACMG, FCCMG**  
Dr. Carla Cuthbert is the chief of the Newborn Screening and Molecular Biology Branch, at the National Center for Environmental Health at CDC. Prior to that, Dr. Cuthbert was a biochemical genetics laboratory director at the University of Miami. Her fellowship in Biochemical Genetics at the Hospital for Sick Children in Toronto sparked an early interest in small molecule method development using tandem mass spectrometry for the diagnosis of inborn errors of metabolism. During fellowships at the Mayo Clinic in Rochester, MN, her projects included mass spectrometry method development of steroid markers for Congenital Adrenal Hyperplasia and development of second-tier assays for newborn screening disorders. Dr. Cuthbert is board-certified in Clinical Laboratory Biochemical Genetics from the ACMG and the Canadian College of Medical Geneticists.

**Victor Raúl De Jesús, PhD**  
Dr. Victor Raúl de Jesús is the chief of the Biochemical Mass Spectrometry Laboratory at the Newborn Screening and Molecular Biology Branch at CDC. He leads the mass spectrometry program at CDC’s Newborn Screening Quality Assurance Program (NSQAP). Dr. De Jesús led the programmatic undertaking to achieve complete NSQAP coverage for the congenital disorders detectable by mass spectrometry, identified in the US Recommended Uniform Screening Panel. He also established a highly-recognized quality assurance program for lysosomal storage disorders (LSD) newborn screening. Dr. De Jesús has served on several committees of professional organizations, published over 50 peer-reviewed papers and given over 30 formal presentations at domestic and international professional meetings, with several by special invitation.

**George Dizikes, PhD**  
Dr. George Dizikes is the section chief of the Illinois Department of Public Health (IDPH) Newborn Screening Laboratory. Dr. Dizikes is also the CLIA laboratory director for the three public health laboratories that comprise the IDPH Division of Laboratories. In these roles, he is responsible for
overseeing all aspects of testing and quality assurance for newborn screening in Illinois. Prior to joining IDPH, Dr. Dizikes spent nine years as director of the Molecular Pathology Laboratory at Loyola University Medical Center. He received his PhD in microbiology from the University of Minnesota at Minneapolis and did postdoctoral research in inborn errors of metabolism at University of California, Los Angeles.

Art Hagar, PhD
Dr. Art Hagar has been the director of the newborn screening section of the Georgia Public Health Laboratory (GPHL) since August 2009. Prior to this position, Hagar was manager of the newborn screening section of the Louisiana Office of Public Health Laboratory (LOPHL) for six years (1996–2002) and assistant director of LOPHL for seven years (2002–2009). During his position as manager of the OPHL newborn screening section, three diseases (Biotinidase Deficiency, Galactosemia, and Congenital Adrenal Hyperplasia) were added to the newborn screening battery, and the implementation of tandem mass spectrometry began. As assistant director of LOPHL, Hagar was responsible for transferring newborn screening from Louisiana to the University of Iowa Hygienic Laboratory within seven days after hurricane Katrina permanently closed the LOPHL building. Hagar also oversaw the re-establishment of LOPHL’s newborn screening section two years later in a new facility.

W. Harry Hannon, PhD
In 2009, Dr. W. Harry Hannon retired from CDC with over 40 years of service. He was chief of CDC’s Newborn Screening Branch for over 25 years and retired as the acting branch chief of the recently organized, Newborn Screening and Molecular Biology Branch. Dr. Hannon received his PhD (1972) from the University of Tennessee in Biochemistry and did post-doctoral training at the Oak Ridge National Laboratories. In 1978, Dr. Hannon created the acclaimed Newborn Screening Quality Assurance Program at CDC, which now provides services to all US newborn screening laboratories and numerous laboratories worldwide. He has over 275 scientific publications and has served on over 35 national and international committees for a variety of laboratory issues. He has received many awards for his scientific contributions. Since retirement, Dr. Hannon has worked to expand and improve newborn screening worldwide. Dr. Hannon has continued his work with Clinical and Laboratory Standards Institute (CLSI), presently serving as Chair of the Consensus Committee on Newborn Screening.

Christopher Haynes, PhD
Dr. Christopher Haynes is the supervisor of amino acid and succinylacetone quality control and proficiency testing specimens for the Newborn Screening Quality Assurance Program (NSQAP) at CDC. He is also the supervisor of X-linked adrenoleukodystrophy specimen production and hemoglobinopathy analysis by tandem mass spectrometry. Prior to joining CDC, he was a participant in the Lipid MAPS lipidomics initiative supported by the NIH. He received his Doctorate in Biology from the Georgia Institute of Technology and a Bachelor of Science degree in Biology from the University of Miami.

Patricia R. Hunt
Patricia Hunt is the manager of the Texas Newborn Metabolic Screening Laboratory of the Department of State Health Services in Austin, TX. Her more than 20 years of laboratory experience include time in Clinical Chemistry and Newborn Screening. In 1999, Hunt joined the Newborn Screening Program as the supervisor of the Metabolic Screening Team, where she oversaw testing procedures, instrument performance, quality assurance of the testing area and implemented a change from bioassay screening methods for PKU and Galactosemia to fluorometric procedures including validation of new instrumentation, methodology and reporting ranges. In July 2006, Hunt was promoted to manager of the Newborn Tandem Mass Spectrometry Screening Laboratory and was instrumental in the successful implementation of screening for 19 additional amino acid, fatty acid oxidation and organic acid disorders in December 2006. Her current duties include overseeing the tandem mass spectrometry, galactosemia and biotinidase deficiency test areas, including oversight of instrument performance, quality assurance and personnel. Hunt is a graduate of the University of Texas at Austin where she received her Bachelor’s in Microbiology.

Kellie B. Kelm, PhD
Dr. Kellie B. Kelm is the chief of the Cardio-Renal Diagnostic Devices Branch in the Division of Chemistry and Toxicology Devices in the Office of In Vitro Diagnostics and Radiological Health (Center for Devices and Radiological Health, FDA). She joined FDA in 2006 and was a lead reviewer of premarket submissions and Investigational Device Exemption (IDE) applications for clinical studies for chemistry, toxicology, genetic/genomic and newborn screening devices for eight years before becoming branch chief in 2015. Dr. Kelm has represented FDA on several external committees such as the Clinical and Laboratory Standards Institute Subcommittee for Newborn Screening and several CLSI Document Development Committees as well as the APHL NewSTEPs Steering Committee. She received her BA at Dartmouth College and her PhD from the Johns Hopkins University School of Medicine.

Mark Kuracina, MBA
Mark Kuracina is director of the mass spect based newborn screening and diagnostics business at PerkinElmer. He is responsible for managing the global mass spect based business including new product development. Prior to joining PerkinElmer, he spent two years as director of marketing & commercial operations as part of a new venture within Abbott. Kuracina also served as director of business development for molecular devices and applied markets global marketing manager at Applied Biosystems/ABSCIEX. He received his MBA in Marketing and Strategic Management from York University and an Honors Degree in Biochemistry and Chemistry from the University of Toronto.

Dietrich Matern, MD, PhD
Dr. Dietrich Matern is a professor of laboratory medicine, medical genetics and pediatrics at the Mayo Clinic College of Medicine. Prior to this, he completed a pediatric residency in his native Germany (Albert-Ludwigs-University, Freiburg) and genetics fellowships at Duke University. He serves as chair of the Division of Laboratory Genetics and co-director of the Biochemical Genetics Laboratory in the Department of Laboratory Medicine and Pathology and holds joint appointments in the Department of Pediatric & Adolescent Medicine and in the Department of Medical Genetics. Dr. Matern’s work focuses on the early diagnosis of inborn errors of metabolism. He has a special interest to reduce false positive results in newborn screening by second-tier assays using the original blood spot sample. A current project aims to find the most efficient and effective way to expand newborn screening to include lysosomal storage and other disorders.
developing meeting content.

Assurance Quality Control Subcommittee of the Newborn Screening and Genetics in Public Health Committee for assisting with corporate and federal partners for participating in this national meeting. APHL also thanks its Newborn Screening Quality Assurance Quality Control Subcommittee of the Newborn Screening and Genetics in Public Health Committee for assisting with developing meeting content.

Scott M. Shone, PhD

Dr. Scott Shone received his bachelor’s degree in Biological Sciences from Rutgers, the State University of New Jersey, and subsequently received his PhD from the Johns Hopkins Bloomberg School of Public Health. Since 2008, he has served as the program manager for the Newborn Screening Laboratory at the New Jersey Department of Health. Dr. Shone is a member of the Advisory Council for the New York Mid-Atlantic Consortium for Genetic and Newborn Screening Services (NYMAC) and is chair of the collaborative’s Newborn Screening and Emergency Preparedness Workgroup. He is a member of APHL’s Newborn Screening and Genetics in Public Health committee and serves as Chair of the Steering Committee for the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). In 2013, Dr. Shone received the Jean Dussault Medal for young investigators from the International Society for Neonatal Screening, and in 2014 he received the Emerging Leader Award from APHL. Dr. Shone is contributing to his own job security as his wife is currently pregnant with their second child.

Robert Vogt, Jr., PhD

Dr. Robert Vogt is primary investigator for the Newborn Screening Translation Research Initiative, a cooperative partnership with the CDC Foundation. He was trained in immunology, toxicology and experimental pathology at the Johns Hopkins Bloomberg School of Public Health under Dr. Arthur M. Dannenberg, Jr. His laboratory interests involve quantitative fluorescence measurements of cellular biomarkers. Since 1999, his public health interests involved translating laboratory research methods into newborn screening and other public health applications. Dr. Vogt is a consensus committee member of the Clinical and Laboratory Sciences Institute (CLSI) and a lifetime member of the Alpha Chapter, Delta Omega Honorary Society in Public Health.

William W. Wood, PhD

Dr. William W. Wood is the director of chemistry at Cambridge Isotope Laboratories (CIL), the world’s premier stable isotope company, based in Andover, MA. In addition to directing the activities of 40 scientists working on the synthesis and formulation of materials labeled with $^{13}$C, $^{15}$N, $^{18}$O and deuterium, Dr. Wood is also the subject matter expert at CIL for products used in neonatal screening. Prior to joining CIL in 2001, he worked for 13 years in discovery chemistry in the agricultural chemical business. Dr. Wood also taught organic chemistry at the University of Sheffield in the UK for three years. He received his PhD (organic chemistry) and BSc (chemistry) degrees from King’s College, University of London.

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