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

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POSTER PRESENTATIONS

The full poster and oral abstracts may be found on the symposium website www.aphl.org/2014nbsgts and on the mobile app.

Posters may be viewed in the Grand Ballroom during exhibit hall hours on Monday, Tuesday and Wednesday, Oct. 27-29.

P-01
Implementation of a Single-Wash, In-Situ, Real-time PCR Screening Procedure for Severe Combined Immunodeficiency in Oklahoma
T. McCallister, R. Thompson and S.T. Dunn, Oklahoma State Department of Health, Oklahoma City, OK

Presenter: Tonya McCallister, Oklahoma State Department of Health, Public Health Laboratory, Oklahoma, City OK, Phone: 405.271.5070, Email: tonyaj@health.ok.gov

P-02
Pulse Oximetry Screening for Critical Congenital Heart Disease in Oklahoma: Collaboration between the Newborn Screening Program and the Oklahoma Birth Defects Registry
L. Caton, S. Vaz and D. Christie, Oklahoma State Department of Health, Oklahoma City, OK

Presenter: Lisa Caton, BSN, Oklahoma State Department of Health, Newborn Screening Program, Oklahoma City, OK, Phone: 405.271.6617, Email: lisarc@health.ok.gov

P-03
Ten Years of MS/MS Screening at the Northwest Regional Newborn Screening Program [NWRNBS]
D. Sesser, Oregon State Public Health Laboratory, Hillsboro, OR

Presenter: David Sesser, Oregon State Public Health Laboratory, Newborn Screening, Hillsboro, OR, Phone: 503.693.4187, Email: David.e.sesser@state.or.us
P-04
NW Regional NBS Program - Disorders Detected Over 50 Years of Screening
D. Sesser, S. Willis, S. Denniston, and C. Hermerath, Oregon State Public Health Laboratory, Hillsboro, OR

Presenter: Cheryl Hermerath, MBA, Oregon State Public Health Laboratory, Newborn Screening, Hillsboro, OR, Phone: 503.693.4172, Email: cheryl.a.hermerath@state.or.us

P-05
Colorado’s Strategies to Improve the Timeliness of the Newborn Screening Process Statewide
L. Gillim-Ross and D. Wright, Colorado Department of Public Health and Environment, Denver, CO

Presenter: Daniel Wright, Colorado Department of Public Health and Environment, Laboratory Services Division, Denver, CO, Phone: 303.692.3673, Email: daniel.wright@state.co.us

P-06
From Supportive to Enforcement: Public Health Newborn Screening Strategies to Address Batching & Specimen Delays
J. Luedtke, Nebraska Department of Health and Human Services, Lincoln, NE

Presenter: Julie Luedtke, Nebraska Department of Health and Human Services, Newborn Screening & Genetics Program, Lincoln, NE, Phone: 402.471.6733, Email: julie.luedtke@nebraska.gov

P-07
Maryland’s Experience of Implementing Quality Assurance Initiative
F. Gulamali-Majid and M.D. Celiz, Maryland Laboratory Administration, Baltimore, MD

Presenter: Fizza Gulamali-Majid, PhD, Maryland Laboratories Administration, Baltimore, MD Phone: 410.767.6099, Email: Fizza.Majid@maryland.gov
P-08
Achieving Dramatic Improvements in Sample Transit Time – The Arizona Model
C. Nabor and S. Aponte, Arizona Department of Health Services, Phoenix, AZ

Presenter: Celia Nabor, MPA, Arizona Department of Health Services, Office of Newborn Screening, Phoenix, AZ, Phone: 602.364.2579, Email: celia.nabor@azdhs.gov

P-09
Improving Newborn Screening Sample Delivery Time in Arkansas
P. Scott and L. Himstedt, Arkansas Department of Health, Little Rock, AR

Presenter: Leslie Himstedt, MT(ASCP), Arkansas Public Health Laboratory, Little Rock, AR, Phone: 501.661.2445, Email: leslie.himstedt@arkansas.gov

P-10
The Discrepancy between the Concentration of Phenylalanine Measured in Dried Blood Spots Compared to Plasma in Individuals with Phenylketonuria
S. Van Calcar, M. Clayton, B. Stroup and M. Baker, University of Wisconsin School of Medicine and Public Health, Madison, WI

Presenter: Sandra Van Calcar, RD, PhD, University of Wisconsin School of Medicine and Public Health, Madison, WI, Phone: 608.263.5981, Email: vancalcar@pediatrics.wisc.edu

P-11
VLCADD Screening: Wisconsin’s Experience
J. Scott Schwoerer, S. Van Calcar, M. Baker and G. Rice, University of Wisconsin School of Medicine and Public Health, Madison, WI

Presenter: Jessica Scott Schwoerer, MD, University of Wisconsin School of Medicine and Public Health, Pediatrics, Madison, WI, Phone: 608.263.5993, Email: jscottschwoerer@pediatrics.wisc.edu
P-12
Wisconsin’s Approach to Addressing Quality Assurance within the Pre-Analytical Testing Phase of Newborn Screening

P. Held, S. Dawe, M. Anderson, M. Baker and C. Brokopp, Wisconsin State Laboratory of Hygiene, Madison, WI

Presenter: Patrice Held, PhD, Wisconsin State Laboratory of Hygiene, Madison, WI, Phone: 608.265.5968, Email: heldpk@slh.wisc.edu

P-13
Improving Newborn Screening Rates within the Michigan Homebirth Community

L. Turbett, M. Kleyn, K. Urquhart, J. Bach, Michigan Department of Community Health, Lansing, MI

Presenter: Lois Turbett, MSN, Michigan Department of Community Health, Lansing, MI, Phone: 517.335.1966, Email: turbettl@michigan.gov

P-14
A DNA Stability Study for the T-cell Receptor Excision Circle (TREC) Real-time PCR Assay Screen for Newborn Immunodeficiency

H. Woods, C. Burns, H. Hawkins and B. Taffe, Michigan Department of Community Health, Lansing, MI

Presenter: Heather Wood, MS, Newborn Screening Program, Michigan Department of Community Health, Lansing, MI, Phone: 517.335.9381, Email: WoodH@michigan.gov

P-15
Development and Implementation of an Automated DNA Extraction Protocol with Selective Sample Transfer, or ‘Cherry Picking’ Capabilities for Cystic Fibrosis (CF) Screening

K. TenEyck, H. Wood, L. Foster and E. Muth, Michigan Department of Community Health, Lansing, MI

Presenter: Kelly TenEyck, MS, Newborn Screening Program, Michigan Department of Community Health, Lansing, MI, Phone: 517.335.8070, Email: teneyckk@michigan.gov
P-16
**Pulse Oximetry Screening for Homebirths: A Survey of Midwives and Follow-up to Address Barriers**

_**K. Urquhart, M. Kleyn and J. Bach, Michigan Department of Community Health, Lansing, MI**_

**Presenter:** Janice Bach, MS, Michigan Department of Community Health, Lansing, MI, Phone: 517.335.8497, Email: bachj@michigan.gov

P-17
**Use of Hospital-specific Cutoffs to Evaluate NBS Specimen Transit Time**

_**M. Kleyn, L. Turbett, W. Young, K. Tomasko and J. Bach, Michigan Department of Community Health, Lansing, MI**_

**Presenter:** Lois Turbett, MSN, Michigan Department of Community Health, Lansing, MI, Phone: 517.335.1966, Email: turbettl@michigan.gov

P-18
**Evaluating Timeliness of Newborn Screening Result Availability on a Web-based Care Improvement Registry, Michigan, 2013**

_**M. Kleyn, K. Tomasko and J. Bach, Michigan Department of Community Health, Lansing, MI**_

**Presenter:** Kristy Tomasko, Michigan Department of Community Health, Lansing, MI, Phone: 517.335.9296, Email: tomaskok@michigan.gov

P-19
**Newborn Screening Social Media Strategies in Minnesota**

_**P. Constant, B. Roby, G. Caldwell and M. McCann, Minnesota Department of Health, St. Paul, MN**_

**Presenter:** Patti Constant, MPH, Minnesota Department of Health, Newborn Screening, St. Paul, MN, Phone: 651.201.4517, Email: patti.constant@state.mn.us

P-20
**Clarification and Branding of Newborn Screening in Minnesota**

_**P. Constant, B. Roby, S. Rosendahl and M. McCann, Minnesota Department of Health, St. Paul, MN**_

**Presenter:** Patti Constant, MPH, Minnesota Department of Health, Newborn Screening, St. Paul, MN, Phone: 651.201.4517, Email: Patti.Constant@state.mn.us
P-21

SCID Screening in Minnesota: Program Changes after Implementation

B. Warman and C. Wolf, Minnesota Department of Health, St. Paul, MN

Presenter: Carrie Wolf, MBS, Minnesota Department of Health, Newborn Screening Laboratory, St. Paul, MN, Phone: 651.201.5458, Email: carrie.wolf@state.mn.us

P-22

California Sickle Cell Disease Provider Survey: Assessment of Patient Management Approaches, Barriers to Care and Educational Needs

L. Feuchtbaum, N. Rosenthal and C. Hutchinson, California Department of Public Health, Richmond, CA

Presenter: Lisa Feuchtbaum, DrPH, MPH, California Department of Public Health, Genetic Disease Screening Program, Richmond, CA, Phone: 510.412.1455, Email: lisa.feuchtbaum@cdph.ca.gov

P-23

California Newborn Screening Program Quality Control Sample Production and Application

Y. Hou and T. Ho, California Department of Public Health, Richmond, CA

Presenter: Yu Hou, PhD, Genetic Disease Laboratory Branch, California Department of Public Health, Richmond, CA, Phone: 510.231.1735, Email: yhou@cdph.ca.gov

P-24

Identification of Galactose-1-Phosphate Uridyl Transferase Gene Common Mutations in Dried Blood Spots

M. Sartippour, R. Doroudian, G. Frampton, F. Lorey and T. Ho, California Department of Public Health, Richmond, CA

Presenter: Maryam Sartippour, PhD, Genetic Disease Laboratory, California Department of Public Health, Richmond, CA, Phone: 510.231.1718, Email: msartipp@cdph.ca.gov
P-25
What Factors Influence the Decisions of Parents Regarding Residual Dried Blood Spot Storage and Use in an Opt in System
B. Reilly, J. Wallace, C Bresette and S. Arshadmansab, Texas Department of State Health Services, Austin, TX

Presenter: Brendan Reilly, Texas Department of State Health Services, Austin, TX, Phone: 512.776.2477, Email: brendan.reilly@dshs.state.tx.us

P-26
Newborn Screening for Severe Combined Immunodeficiency: The First 17 Months of the Texas Experience
R. Lee, M. Nolen, C. Corrigan, D. Moren and D. Freedenberg, Texas Department of State Health Services, Austin, TX

Presenter: Rachel Lee, PhD, Texas Department of State Health Services, Austin, TX, Phone: 512.776.7158, Email: rachel.lee@dshs.state.tx.us

P-27
Affordable Care Act Web-based Resource Tool
S. Mann, L. Hasegawa and J. Boomsma, Hawaii Department of Health, Honolulu, HI

Presenter: Sylvia Mann, MS, Hawaii Department of Health Genomics Section, Honolulu, HI, Phone: 808.733.9063, Email: sylvia@hawaiigenetics.org

P-28
Technology Transfer Among NBS Programs within SCID NBS Pilots
J. Gerstel-Thompson, J. Hale, R. Lee, S. Tanksley; 1New England Newborn Screening Program, Jamaica Plain, MA, 2Texas Department of State Health Services, Austin, TX

Presenter: Anne Marie Comeau, PhD, New England Newborn Screening Program, University of Massachusetts Medical School, Jamaica Plain, MA, Phone: 617.983.6324, Email: anne.comeau@umassmed.edu or jacalyn.gerstel-thompson@umassmed.edu
P-29
The 15-year Experience of Newborn Screening for Medium Chain Acyl-CoA Deficiency in Massachusetts
I. Sahai, J. Bailey, J. Hale and T. Zytkovicz, New England Newborn Screening Program, Jamaica Plain, MA

Presenter: Inderneel Sahai, MD, New England Newborn Screening Program, University of Massachusetts, Jamaica Plain, MA, Phone: 617.983.6300, Email: inderneel.sahai@umassmed.edu

P-30
Newborn Screening for Galactosemia: The New England Newborn Screening Program Experience
I. Sahai, A.M. Comeau, J. Hale, T. Zytkovicz and D. Britton, New England Newborn Screening Program, Jamaica Plain, MA

Presenter: Inderneel Sahai, MD, New England Newborn Screening Program, University of Massachusetts Medical School, Jamaica Plain, MA, Phone: 617.983.6300, Email: inderneel.sahai@umassmed.edu

P-31
Influence of Breast vs. Formula Feeding on Newborn Screening Results
H.-W. Hsu, I. Sahai, T. Zytkovicz and R. Eaton, New England Newborn Screening Program, Jamaica Plain, MA

Presenter: Ho-Wen Hsu, MD, New England Newborn Screening Program, University of Massachusetts Medical School, Jamaica Plain, MA, Phone: 617.983.6300, Email: ho-wen.hsu@umassmed.edu

P-32
Evaluation of a New Automated Biotinidase Assay for Newborn Screening
N. Breen and J. Borneo, Indiana Newborn Screening Program, Indianapolis, IN

Presenter: Nancy Breen, Indiana Newborn Screening Laboratory, Pathology, Indianapolis, IN, Phone: 317.491.6675, Email: nbreen@iupui.edu
P-33
Impact of Seasonal Changes and Kit Lot Changes on Immunoreactive Trypsinogen Values Using the ImmuchemTM Blood Spot Trypsin-MW ELISA Kit

B. Maloney, R. Hamel, M. Caggana and N.P. Tavakoli, Newborn Screening Program, Wadsworth Center, New York State Department of Health, Albany, NY

Presenter: Norma Tavakoli, PhD, Newborn Screening Program, Wadsworth Center, New York State Department of Health, Albany, NY, Phone: 518.474.4897, Email: norma.tavakoli@wadsworth.org

P-34
Comparison of the ImmuchemTM Blood Spot Trypsin-MW ELISA kit (MP Biomedical) and the AutoDELFIA Neonatal IRT Kit (Perkin Elmer) for Measuring Immunoreactive Trypsinogen for the Detection of Cystic Fibrosis in Newborns

R. Hamel, B. Maloney, M. Pearce, L. DeMartino and E. McGrath, New York State Department of Health, Albany, NY

Presenter: Norma Tavakoli, PhD, Newborn Screening Program, Wadsworth Center, New York State Department of Health, Albany, NY, Phone: 518.474.4897, Email: norma.tavakoli@wadsworth.org

P-35
Expression of GALC Activity in Common Screening Referral Haplotypes in NYS


Presenter: Carlos Saavedra-Matiz, MD, New York State Department of Health, Wadsworth Center, Albany, NY, Phone: 518.486.1641, Email: carlos.saavedra@health.ny.gov

P-36
A Review of Plans to Improve Specimen Transit Time for Hospitals in New York State

B. Vogel, M. Caggana, M. Graziano, J. Orsini, K. Fiato, New York State Department of Health, Albany NY

Presenter: Beth Vogel, MS, New York State Department of Health, Wadsworth Center, Newborn Screening, Albany, NY, Phone: 518.474.7945, Email: bmh06@health.state.ny.us
P-37
A High-throughput Multiplex Newborn Screening Assay for Six Lysosomal Storage Disorders (LSDs) Using Dried Blood Spots and UPLC-Tandem Mass Spectrometry

K. Basheeruddin¹, R. Shao¹, M. Gelb², P. Gardley¹; ¹Illinois Department of Public Health, Chicago, IL, ²University of Washington, Seattle, WA

Presenter: Khaja Basheeruddin, PhD, Illinois Department of Public Health, Laboratories, Chicago, IL, Phone: 312.793.1712, Email: khaja.basheeruddin@illinois.gov

P-38
Evaluation of a New Automated Assay to Measure Total Galactose for Newborn Screening

S. Shone¹, L. Nayak¹, E. Leachko¹, S. Patel¹, D. McCourt¹, P. Furu²; ¹New Jersey Department of Health, Trenton, NJ, ²PerkinElmer, Turku, Finland

Presenter: Scott Shone, PhD, New Jersey Department of Health, Newborn Screening Laboratory, Ewing NJ, Phone: 609.530.8361, Email: scott.shone@doh.state.nj.us

P-39
Modification of an On-Card Quantitative PCR Assay for T-cell Receptor Excision Circles (TREC) to Screen Newborns for Severe Combined Immune Deficiency (SCID)

S. O’Leary¹, R. Mathes¹, E. Leachko¹, L. Misiakowski¹, J. Taylor², L. Hancock², G. Yazdanpanah², F. Lee², R. Vogt², S. Shone¹; ¹New Jersey Department of Health, Ewing, NJ, ²Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Shawn O’Leary, New Jersey Department of Health, Newborn Screening Laboratory, Ewing NJ, Phone: 609.530.8371, Email: shawn.oleary@doh.state.nj.us

P-40
Equivocals on the Move

T. Ahmad, F. Santos, I. Margolin and P. Patel, New Jersey Department of Health, Trenton, NJ

Presenter: Tariq Ahmad, MBBS, MPH, Newborn Screening & Genetic Services, New Jersey Department of Health, Trenton, NJ, Phone: 609.292.1582, Email: tariq.ahmad@doh.state.nj.us
P-41
Iowa SCID Newborn Screening Pilot – Follow-up Perspective
E. Phillips1, M.B. Fasano1, P. Ferguson1, C. Johnson1, N. Dennler1, T. Hatland1, C. Kremer1, S. Rumelhart1, K. Piper2; 1University of Iowa Hospitals and Clinics, Pediatrics, Iowa City, IA, 2Iowa Department of Public Health, Des Moines, IA

Presenter: Emily Phillips, BSN, University of Iowa Hospitals and Clinics, Pediatrics, Iowa City, IA, Phone: 319.356.7970, Email: emily-phillips@uiowa.edu

P-42
Developing a Robust TREC Assay with a Low False Positive Rate
T. Davis, J. Thompson, S. Weiss and S. Shaunak, Washington State Department of Health, Shoreline, WA

Presenter: Tim Davis, Newborn Screening, Washington State Department of Health, Shoreline, WA, Phone: 206.291.4778, Email: tim.davis@doh.wa.gov

P-43
Analysis of Sulfatides in Dried Blood and Urine Spots: Toward a Method for Newborn Screening of Metachromatic Leukodystrophy
M. Barcenas, A.B. Kumar, S. Massi, F. Turecek, C.R. Scott and M.H. Gelb, University of Washington, Seattle, WA

Presenter: Michael Gelb, PhD, University of Washington, Chemistry and Biochemistry, Seattle WA, Phone: 206.543.7142, Email: gelb@chem.washington.edu

P-44
Evaluating Awareness, Understanding, and Utilization of Good Laboratory Practices in Biochemical Genetic Testing and Newborn Screening for Inherited Metabolic Diseases
B. Su1, K. Breckenridge1, B. Chen2, J. Ojodu1; 1Association of Public Health Laboratories, Silver Spring, MD, 2Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Bertina Su, Association of Public Health Laboratories, Silver Spring, MD, Phone: 240.485.2729, Email: bertina.su@aphl.org
P-45
Newborn Screening and Hemoglobin Barts (HbBarts)
S. Carlisle, E. Lam, D. Holstein and C. Simmons, University of Colorado Hospital and Colorado Center for Laboratory Sciences, Aurora, CO

Presenter: Scot Carlisle, University of Colorado Hospital and Colorado Center for Laboratory Sciences, Aurora, CO, Phone: 480.734.6386, Email: scarlis1@msudenver.edu

P-46
CPT1A Diagnosis Five Years after a Presumptive Positive Newborn Screen
J. Bartley, Loma Linda University Children’s Hospital, San Bernardino, CA

Presenter: James Bartley, MD, PhD, Loma Linda University Children’s Hospital, Pediatrics, Loma Linda, CA, Phone: 909.633.6578, Email: jamesbartley1@msn.com

P-47
Development of Newborn Screening Connect (NBS Connect): A Self-reported Patient Registry and its Role in Improvement of Care for Inherited Metabolic Disorders
Y. Osara, A. Devarajan, K. Coakley and R. Singh, Emory University, Decatur, GA

Presenter: Yetsa Osara, Emory University, Human Genetics, Decatur, Ga, Phone: 404.778.3353, Email: yadadev@emory.edu

P-48
Evidence Analyst’s Role in Creating Nutrition Management Guidelines
Y. Osara, K. Coakley, A. Aisthorpe and A. Stembridge, Emory University, Decatur, GA

Presenter: Yetsa Osara, Emory University, Human Genetics, Decatur, Ga, Phone: 404.778.3353, Email: yadadev@emory.edu

P-49
Utilizing Technology to Facilitate Analysis and Synthesis of Evidence and Consensus-based Data in Developing and Disseminating Nutrition Management Guidelines
A. Stembridge, Y. Osara, A. Aisthorpe, F. Rohr and R. Singh, Emory University, Decatur, GA

Presenter: Adrya Stembridge, Emory University, Department of Human Genetics, Decatur, GA, Phone: 404.778.8535, Email: astembr@emory.edu
P-50
Newborn Screening for Duchenne Muscular Dystrophy: Rationale, Options, and Laboratory Challenges

J. Kwon¹, P. Farrell², E. Ciafaloni³; ¹University of Rochester Medical Center, Rochester, NY, ²University of Wisconsin, Madison, WI

Presenter: Jennifer Kwon, MD, MPH, University of Rochester Medical Center, Neurology & Pediatrics, Rochester, NY, Phone: 585.273.5264, Email: jennifer_kwon@urmc.rochester.edu

P-51
Arkansas Newborn Screening Long-Term Follow-up Cohort Study - Year 2

J. Bolick, University of Arkansas for Medical Sciences/Arkansas Children’s Hospital, Little Rock, AR

Presenter: Jo Ann Bolick, BSN, MA, University of Arkansas for Medical Sciences/Arkansas Children’s Hospital, Department of Pediatrics, Division of Genetics, Little Rock, AR, Phone: 501.364.1906, Email: jabolick@uams.edu

P-52
Arkansas Newborn Screening Short- and Long-term

T. Moore and J. Ketcher, University of Arkansas for Medical Sciences, Little Rock, AR

Presenter: Tiffany Moore, BSN, RN, University of Arkansas for Medical Sciences, Little Rock, AR, Phone: 501.364.1608, Email: tpmoore@uams.edu

P-53
Novel Techniques for Preparing Dried Blood Spot Proficiency Testing Specimens for Sickle Cell Disease Newborn Screening Tests

B. Adam, M. Azimi and M. Earley, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Victor De Jesus, PhD, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 770.488.7963, Email: foa5@cdc.gov
P-54

The Stability of Hexacosanoyl Lysophosphatidylcholine in Dried-Blood Spot Quality Control Materials for X-linked Adrenoleukodystrophy Newborn Screening

C. Haynes and V. De Jesus, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Christopher Haynes, PhD, Centers for Disease Control and Prevention, NSMBB, Atlanta, GA, Phone: 770.488.7459, Email: cph7@cdc.gov

P-55

Important Factors in Designing Accurate and Reliable Next-generation Sequencing (NGS) Assays

P. Mueller, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Patricia Mueller, PhD, Centers for Disease Control and Prevention, NSMBB, Atlanta, GA, Phone: 770.488.4015, Email: pwm2@cdc.gov

P-56—Withdrawn

P-57

Long Term Stability of Long-Chain Acylcarnitines in Enriched Dried Blood Spots Stored at Various Temperatures

P. Pickens, T. Lim, V. De Jesus, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Patrick Pickens, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 678.230.2976, Email: xgl0@cdc.gov

P-58

Proficiency Testing for Lysosomal Storage Disorders in Dried Blood Spots to Detect Krabbe and Pompe Diseases

J. Mei, H. Zhou, F. Lee, I. Williams and S. Zobel, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Joanne Mei, PhD, Centers for Disease Control and Prevention, Newborn Screening Quality Assurance Program, Atlanta, GA, Phone: 770.488.7945, Email: jmei@cdc.gov
P-59
Development of Dried Blood Spot Reference Materials for Fragile X

J. Lyons, P. Mueller and G. Kerr, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Justine Lyons, PhD, Centers for Disease Control and Prevention, NSMBB, Atlanta, GA, Phone: 770.448.7704, Email: vro9@cdc.gov

P-60
Improved Method for the Detection of Hemoglobinopathies via an Enhanced Trypsin Digestion/Extraction and Sample Analysis via HPLC-ESI-MS/MS

D. Foreman, C. Haynes, V. De Jesus, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: David Foreman, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 770.488.7147, Email: xgb1@cdc.gov

P-61
Second Tier Proficiency Testing for Congenital Adrenal Hyperplasia: Laboratory and Method Performance from February 2012 to February 2014

S. Klass, T. Myers, J. Mei and V. De Jesus, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Sarah Klass, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 678.813.1292, Email: xhb4@cdc.gov

P-62
The Newborn Screening Molecular Assessment Program (MAP) for Molecular Screening Quality Improvement

C. Greene, S. Cordovado and C. Cuthbert, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Christopher Greene, PhD, Centers for Disease Control and Prevention, Newborn Screening and Molecular Biology Branch, Atlanta, GA, Phone: 770.488.4845, Email: crg0@cdc.gov
P-63

New Automation Resources on the NBS Molecular Resource Website

L. Hancock¹, T. Davis², T. Henry³, J. Isabelle⁴; ¹Centers for Disease Control and Prevention, Atlanta, GA, ²Washington State Department of Health, Shoreline, WA, ³State Hygienic Laboratory at the University of Iowa, Coralville, IA, ⁴New York State Department of Health, Albany, NY

Presenter: Laura Hancock, Centers for Disease Control and Prevention, Newborn Screening and Molecular Biology Branch, Atlanta, GA, Phone: 770.488.4617, Email: lhancock@cdc.gov

P-64

Next-Generation Sequencing (NGS) Assay for Pompe Disease

P. Mueller¹, G. Kerr¹, J. Lyons¹, R. Isett²; ¹Centers for Disease Control and Prevention, Atlanta, GA, ²Emory University, Decatur, GA

Presenter: Patricia Mueller, PhD, Centers for Disease Control and Prevention, NSMBB, Atlanta, GA, Phone: 7707.488.4015, Email: pwm2@cdc.gov

P-65

Digital PCR in Newborn Screening: A New Methodology to Characterize Calibrators and Reference Materials for Quantitative Molecular Assays

J. Taylor, F. Lee, G. Yazdanpanah and R. Vogt, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Jennifer L. Taylor, PhD, Centers for Disease Control and Prevention, Newborn Screening and Molecular Biology Branch, Atlanta, GA, Phone: 770.488.7919, Email: vjf6@cdc.gov

P-66

Dried Blood Spot Microsatellite Marker Sample Identification Using the QIAxcel

Z. Detwiler, M. Hendrix, C. Cuthbert and S. Cordovado, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Zachary Detwiler, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 770.488.0420, Email: wtp4@cdc.gov
P-67

Using Epstein-Barr Virus to Transform B-Lymphocytes for the Creation of Molecular Quality Assurance Materials

T. Gilliland, S. Foster, C. Cuthbert and S. Cordovado, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Travis Gilliland, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 770.488.4373, Email: xic5@cdc.gov

P-68

Evaluation of Hologic Inplex® CF Molecular Test Performance Using Two DNA Extraction Methods for Dried Blood Spots

S. Foster, S. Nikolova, C. Cuthbert and S. Codovado, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Stephanie Foster, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 770.488.7348, Email: xji4@cdc.gov

P-69

Examining Factors that Influence Blood Absorption Time on Newborn Screening Blood Collection Devices

E. Hall, W.H. Hannon and V. De Jesus, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Elizabeth M. Hall, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 770.488.7853, Email: erm5@cdc.gov

P-70

Pilot Distribution of Galactose-1-Phosphate Uridyltransferase Quality Control Dried Blood Spot Materials

S. Flores, B. Adam, C. Cuthbert and V. De Jesus, Centers for Disease Control and Prevention, Atlanta, GA

Presenter: Sharon Flores, MS, Centers for Disease Control and Prevention, Atlanta, GA, Phone: 770.488.4182, Email: fil4@cdc.gov
P-71
Conjoint Survey of Parental Preferences to Receive Incidental Information from Genetic Testing

Presenter: Megan Lewis, PhD, RTI International, Research Triangle Park NC, Phone: 919.541.6834, Email: melewis@rti.org

P-72
The Implementation of a Sebia CAPILLARYS 2 Neonat Fast as a First Line Screening Method for Sickle Cell Disease in a United Kingdom Newborn Screening Centre
S. Brown and D. Hawden, Epsom and St Helier University Hospitals NHS Trust, Surrey, England

Presenter: Sarah Brown, Fellowship of IBMS, Epsom and St Helier University Hospitals NHS Trust, Blood Sciences (Haematology), Surrey, England, Phone: 44.208.296.2812, Email: sarah.brown@esth.nhs.uk

P-73
The Feasibility of Using an MSMS-based Method with Perkin Elmer Lysosomal Storage Disease (LSD) Reagents to Implement a Newborn Screening Test for Six LSD
S. Stark, M. Fietz, E. Ranieri, Women’s & Children’s Hospital, Adelaide, SA, Australia

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P-74
An Experience of 4 Years in a First Step Strategy on the CAPILLARYS 2 Neonat Fast for Sickle Cell Disease
J.M. Perini and V. Huin, Lille University Hospital, Lille, France

Presenter: Jean-Marc Périni, MD, Lille University Hospital, Lille, France, Phone: 33.6.7953.3075, Email: jean-marc.perini@chru-lille.fr
P-75
Impact of Age at Collection on Performance Indicators in Screening for Primary Congenital Hypothyroidism Using Thyroid Stimulating Hormone in Normal Birth Weight Neonates
T. Wotton and V. Wiley, The Children’s Hospital at Westmead, Wentworthville, NSW, Australia

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P-76
Pitfalls in Diagnostic Laboratory Results of Lysosomal Storage Disorders: An Institutional Experience
M. Serteser, Acibadem University, Istanbul, Turkey

Presenter: Mustafa Serteser, MD, Acibadem University, School of Medicine, Department of Medical Biochemistry, Atasehir Kampus, Istanbul, Turkey, Phone: 90.5.32.674.2413, Email: mserteser@acibademlabmed.com.tr

P-77 – Withdrawn

P-78
MENA Newborn Screening Status: 8 Years After the Marrakech Declaration
I. Khneisser, Saint Joseph University, Beirut, Lebanon

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P-79
Development of Amino Acid, Acylcarnitine and Total Protein Assays from Dried Milk Spots
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Presenter: Donald Chace, PhD, Pediatrix Medical Group, Pediatrix Analytical, Centers for Research, Education and Quality, Sunrise FL, Phone: 954.608.9463, Email: donald_chace@pediatrix.com
P-81

**Novel Automated Assay for the Determination of Total Galactose (TGal)**

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P-82

**Six-plex FIA-MS/MS Assay to Screen for ABG, ASM, GAA, GALC, GLA and IDUA Activities in Neonatal Dried Blood Spots**

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P-83

**Diagnostic Odyssey in Spinal Muscular Atrophy: A Systematic Literature Review**

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