Gene Sequencing in Public Health Newborn Screening Meeting

February 16 - 17, 2017
The Westin Buckhead Atlanta
Buckhead I Ballroom
3391 Peachtree Road NE
Atlanta, GA 30326
AGENDA

The purpose of this meeting is to convene pertinent stakeholders to discuss the current status of gene sequencing in newborn screening, and identify barriers and solutions for the successful incorporation of gene sequencing into newborn screening.

MEETING OBJECTIVES

- Discuss the current status of gene sequencing in newborn screening and identify second tier and future applications.
- Outline the differences between mutation panel and sequencing data.
- Discuss laboratory and follow up needs, barriers and solutions for the incorporation of gene sequencing into newborn screening.
- Explain how gene sequencing information is used in newborn screening education, follow up and patient care.
- Outline implementation considerations in new gene sequencing technologies and newborn screening disorders.
- Provide state experiences in implementing gene sequencing.
- Develop plans of action for the incorporation of gene sequencing into newborn screening.

POINT OF CONTACT
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This project was also supported by the Health Resources and Services Administration (HRSA) under grant # U22MC24078 Heritable Disorders $950,000. This information or content and conclusions are those of the authors and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the US Government.
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<tr>
<td>8:00</td>
<td>Breakfast and Registration (served in Buckhead I Ballroom)</td>
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<tr>
<td>8:30</td>
<td>Welcome and Introductions</td>
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<td><strong>Jelili Ojodu, MPH, Association of Public Health Laboratories</strong></td>
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<td><strong>Carla Cuthbert, PhD, FCCMG, FACMG, Centers for Disease Control and Prevention (CDC)</strong></td>
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<tr>
<td>8:40</td>
<td>Session One: Current Status of Gene Sequencing in Newborn Screening (NBS)</td>
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<td>8:40 – 9:00 Introduction and Background of Gene Sequencing in NBS</td>
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<td><strong>Suzanne Cordovado, PhD, CDC</strong></td>
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<td>9:00 – 9:45 Current Second Tier and Future Applications of Gene Sequencing in NBS</td>
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<td><strong>Michele Caggana, ScD, FACMG, New York State Department of Health</strong></td>
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<td><strong>Colleen Stevens, PhD, New York State Department of Health</strong></td>
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<td>9:45</td>
<td>Morning Break</td>
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<td>10:00</td>
<td>Mutation Panel vs. Sequencing Data Pros and Cons</td>
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<td><strong>Anne Comeau, PhD, University of Massachusetts Medical School</strong></td>
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<td>10:30</td>
<td>Breakout Session One: Case Studies with Molecular Analysis</td>
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<td>11:45</td>
<td>Report out from Breakout Session</td>
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<td>12:15</td>
<td>Lunch (served in Buckhead I Ballroom)</td>
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<td>1:00</td>
<td>Session Two: What Does the NBS Program Need to Make this Happen?</td>
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<td>Instrumentation and Informatics Requirements</td>
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<td><strong>Chris Greene, PhD, CDC</strong></td>
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<td><strong>Scott Sammons, MPH, CDC</strong></td>
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<td>2:00</td>
<td>Implementation Considerations for an Emerging NBS Technology</td>
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<td><strong>Mei Baker, MD, Wisconsin State Laboratory of Hygiene</strong></td>
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<td>Afternoon Break</td>
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<td>Program Planning</td>
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<td><strong>Yvonne Kellar-Guenther, PhD, Colorado School of Public Health, NewSTEPs</strong></td>
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<td>Breakout Session Two: Identify Laboratory and Follow-Up Barriers to Incorporating Complex</td>
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<td>Molecular Testing into the NBS Workflow</td>
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<td>Report out from Breakout Session</td>
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AGENDA DAY 2: FRIDAY, FEBRUARY 17, 2017
BUCKHEAD I BALLROOM

7:30 – 8:00  Breakfast (served in Buckhead I Ballroom)

8:00 – 8:15  Recap of Day 1
Rachel Lee, PhD, Texas Department of State Health Services

8:15 – 12:15  Session Two: What Does the NBS Program Need to Make this Happen?
8:15 – 9:00  Sequencing Results Interpretation and Reporting
Rachel Lee, PhD, Texas Department of State Health Services
9:00 – 9:45  Short-Term and Long-term Follow Up Needs
Amy Gaviglio, MS, CGC, Minnesota Department of Health

9:45 – 10:00  Morning Break

10:00 – 10:30  Process to Brainstorm Solutions
Yvonne Kellar Guenther, PhD, Colorado School of Public Health, NewSTEPs

10:30 – 11:30  Breakout Session Three: Identify Laboratory and Follow-Up Solutions to Barriers

11:30 – 12:00  Report out from Breakout Session

12:00 – 12:15  Afternoon Break

12:15 – 2:30  Session 3: Experience from NBS Lab and Follow Up
(Working Lunch served in Buckhead I Ballroom)

12:15 – 12:25  Introduction for Programs New to Sequencing
Michele Caggana, ScD, FACMG, New York State Department of Health

12:25 – 1:00  Programs New to Sequencing
Scott Shone, PhD, New Jersey Division of Public Health & Environmental Laboratories

1:00 – 1:15  Choosing the Starting Point
Yvonne Kellar-Guenther, PhD, Colorado School of Public Health, NewSTEPs

1:15 – 2:00  Breakout Session Four: Identifying a Plan of Action

2:00 – 2:30  Report out from Breakout Session

2:30 – 3:00  Meeting Wrap Up and Review of Action Items
Rachel Lee, PhD, Texas Department of State Health Services

3:00  Meeting Adjourn
Mei Baker, MD, FACMG, is a professor in the Department of Pediatrics, and Co-Director in the Newborn Screening Laboratory at the University of Wisconsin School of Medicine and Public Health. Dr. Baker practiced medicine before being trained in both biochemical and molecular genetics, obtaining a clinical biochemical genetics certification from the American Board of Medical Genetics and Genomics in 2009. She has more than 10 years of experience in routine newborn screening (NBS) with specific interest in, and a successful track record of, applying emerging technologies to implement new screening tests for disorders and to improve ongoing screening tests. She is one of the leading scientists who made Wisconsin the first state in the nation to implement universal NBS for severe combined immunodeficiency (SCID) in 2008. She is a principal investigator for an ongoing NIH/NICHD funded project for “Establishing a Newborn Screening Process for Early Identification and Treatment of Infants with Pompe Disease”. She is also a principal investigator in an ongoing project aimed at improving newborn screening for cystic fibrosis by using next generation sequencing technology. Dr. Baker is currently a member of the Advisory Committee on Heritable Disorders in Newborns and Children, Co-chair of the Newborn Screening Technical Assistance and Evaluation Program (NewSTEPs) Steering Committee, and a member of the APHL NBS Molecular Subcommittee.

Michele Caggana, ScD, FACMG

Michele Caggana, ScD, FACMG received her doctoral degree from the Harvard School of Public Health and completed post-doctoral work in clinical molecular genetics at the Mt. Sinai School of Medicine. She is board certified in clinical molecular genetics by the American Board of Medical Genetics and a fellow of the American College of Medical Genetics and Genomics. Dr. Caggana has been employed by the Wadsworth Center since 1996, where she is Deputy Director of the Division of Genetics, Chief of the Laboratory of Human Genetics, and Director of the Newborn Screening Program. She is involved in many national newborn screening efforts, including the national pilot for Pompe disease implementation and works with CDC and APHL. She is a member of the APHL NBS Molecular Subcommittee and Co-chair of the APHL Newborn Screening and Genetics in Public Health Committee. Dr. Caggana is also a consultant to the FDA. Her laboratory has developed several new newborn screening tests and uses DNA technology to study frequencies of specific gene mutations in dried blood spots in the context of newborn screening.
Anne Comeau, PhD, University of Massachusetts Medical School

Anne Marie Comeau, PhD, is a Professor in the Department of Pediatrics at the University of Massachusetts Medical School and Deputy Director of the New England Newborn Screening Program. Dr. Comeau earned her Ph.D. from Brandeis University in 1985 where she studied yeast genetics and molecular biology. She turned her attention to public health, was awarded an NIAID Individual National Research Service Award for her post-doctoral fellowship at the Harvard School of Public Health and in 1988 joined the New England Newborn Screening Program. Technical advances from Dr. Comeau’s laboratory have yielded molecular assays for high-throughput population-based applications and multiplex mutation analyses for second tier testing. Experience from the Massachusetts’ pilot programs implementing CF and SCID newborn screening contributed to the national level recommendations to include CF and SCID in state newborn screening panels.

Dr. Comeau has been an avid advocate of interdisciplinary evidence-based development and evaluation of newborn screening protocols. She is a member of the APHL NBS Molecular Subcommittee. She is a member of the Working Group on Pilot Studies for the Federal Secretary of Health’s Advisory Committee on Heritable Diseases in Newborns and Children (ACHDNC) and continues as an active member of the Condition Review Group for External Reviews submitted to the SACHDNC.

Dr. Comeau has promoted policies and developed protocols that facilitate Massachusetts’ expansion of newborn screening services while respecting human subjects protections. Her publication topics include evaluations of molecular applications in newborn screening, implications of expanded newborn screening on the healthcare community, recommendations for successful implementation of newborn screening programs and considerations for research use of data held under public health stewardship.

Suzanne Cordovado, PhD

Suzanne Cordovado, PhD, is the Chief of the Molecular Quality Improvement Program (MQIP) in the Newborn Screening and Molecular Biology Branch at CDC in Atlanta, GA. MQIP provides quality assurance for newborn screening laboratories engaged in molecular testing and assists states with the incorporation of molecular tests. In collaboration with the APHL NBS Molecular Subcommittee, MQIP also supports NBS laboratories by providing resources through training and education, research on molecular work specific for dried blood spots and Molecular Assessment Program site visits to evaluate and provide assistance for molecular testing. Dr. Cordovado’s previous work at CDC involved managing a genetic testing laboratory which studied the genetics of type 1 diabetes and diabetic nephropathy as well as other diseases of public health importance. Dr. Cordovado has worked at CDC since 1997 in the Division of Laboratory Sciences after earning her doctorate in Cell and Developmental Biology from Emory University.
Carla Cuthbert, PhD, FACMG, FCCMG

Carla Cuthbert, PhD, FACMG, FCCMG, 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.

Amy Gaviglio, MS, CGC, Minnesota Department of Health

Amy Gaviglio is a genetic counselor and the follow-up supervisor for the Minnesota Department of Health, Newborn Screening Program. She oversees the follow-up of newborn screening results from bloodspot, EHD1, and CCHD screening and aids in statewide educational efforts on newborn screening and genetics and works on genetics-related policy in the state. She has special interest in ELSI issues surrounding newborn screening. Professional services include co-chairing the Public Health Special Interest Group for the National Society of Genetic Counselors, co-chairing the APHL NewSTEPs CCHD Technical Assistance Workgroup, and membership on a host of other newborn screening related workgroups.

Chris Greene, PhD, Centers for Disease Control and Prevention

Chris Greene, PhD, is a member of CDC’s Molecular Quality Improvement Program. He is the lead for the Molecular Assessment Program and for congenital adrenal hyperplasia molecular methods development. At the CDC, he has conducted high-throughput genotyping projects and the evaluation of emerging technologies for studies of diseases of public health importance. Dr. Greene earned his doctorate in Genetics and Molecular Biology at Emory University and was a post-doctoral fellow in the Department of Biochemistry at Emory where he led biochemical and genetic research projects of DNA damage and repair mechanisms.

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

Yvonne Kellar-Guenther, PhD, is the program evaluator for NewSTEPS. She has been conducting program evaluations for over 15 years and teaching research and evaluation design for the past 3 years. Dr. Kellar-Guenther is an associate professor of Community and Behavioral Health at the Colorado School of Public Health. She holds a PhD in Communication and Health.
Rachel Lee, PhD, Texas Department of State Health Services

Rachel Lee, PhD, is the Biochemistry and Genetics Branch Manager in the Laboratory Services Section of the Texas Department of State Health Services in Austin. She oversees the operations of Biochemistry and Genetics Branch, which encompasses the state newborn screening and clinical chemistry laboratories. Dr. Lee currently serves as the chair of the APHL NBS Molecular Subcommittee. Prior to joining the Texas Newborn Screening Program, Dr. Lee worked at the University of Texas and private sector in research and development of various molecular and immunological assays. She received her doctorate in Food Microbiology from University of Wisconsin – Madison.

Jelili Ojodu, MPH

Jelili Ojodu is the director of the Newborn Screening and Genetics Program at APHL. He is also the project director for NewSTEPs. Prior to joining APHL, he worked at Georgetown University Medical Center on an NIH initiative to reduce infant mortality in DC as a research associate. He received his Master’s in Public Health from The George Washington University and a Bachelor of Science degree in Biological Sciences from University of Maryland, College Park.

Scott Sammons, MPH, Centers for Disease Control and Prevention

Scott Sammons, MPH, is a Science Officer for the Office of Advanced Molecular Detection (AMD) at CDC. The Office of AMD supports CDC’s research scientists by providing a large scientific computing infrastructure, bioinformatics workforce development and intramural funding for CDC programs involved in using bioinformatics and next generation sequencing to support their public health mission. Mr. Sammons provides technical expertise and project management for the infrastructure and bioinformatics teams within the Office. Prior to joining AMD Office, Mr. Sammons was the Lead of the Bioinformatics Team within the Biotechnology Core Facility Branch providing bioinformatics support for next generation sequencing projects involved in outbreak investigations and surveillance of public health related pathogens including cholera, salmonella, legionella, smallpox, and influenza. Prior to joining the CDC, Mr. Sammons served as the Director of the Biomolecular Computing Resource (BIMCORE) at Emory University in Atlanta. This core facility provided training and bioinformatics support for the graduate school programs for the University and Medical School. Mr. Sammons received his Bachelor’s and Master’s degrees from the University of Georgia in 1985 and 1987, respectively.

Scott Shone, PhD, New Jersey Department of Public Health

Scott Shone, PhD, received his bachelor’s degree in Biological Sciences from Rutgers, the State University of New Jersey, and subsequently received his PhD in Molecular Microbiology and Immunology 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 Co-Chair of the NewSTEPs Steering Committee. 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.
Colleen Stevens, PhD, New York State Department of Health

Colleen Stevens, PhD, is a research scientist with the New York State Department of Health Newborn Screening Program. Her primary role is in the development and validation of new molecular tests and quality assurance within the NYS NBS DNA lab. Dr. Stevens has participated in a number of site visits with the NBS Molecular Assessment Program (MAP) to provide guidance to newborn screening programs bringing on new molecular tests. In addition, Dr. Stevens works with the NYS Clinical Laboratory Evaluation Program (CLEP) which provides regulatory oversight of all laboratories that perform clinical testing for New York State residents. In that position, Dr. Stevens is responsible for the review of method validations for laboratory developed tests submitted by NYS-regulated labs. Dr. Stevens received her doctorate in biomedical sciences with a concentration in molecular genetics from the State University of New York at Albany School of Public Health.