National Newborn Screening Meeting on New Disorders [Pompe, MPS I, X-ALD]

June 20-21, 2018

Washington Marriott at Metro Center

Washington, DC

NewSTEPs

A Program of the Association of Public Health Laboratories™

This meeting is sponsored by the Association of Public Health Laboratories

About NewSTEPs:

NewSTEPs (Newborn Screening Technical assistance and Evaluation Program) is a program of the Association of Public Health Laboratories in collaboration with the Colorado School of Public Health. It is a national newborn screening program designed to provide data, technical assistance, and training to newborn screening programs across the country and to assist states with quality improvement initiatives. NewSTEPs is a comprehensive resource center for state newborn screening programs and stakeholders.

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.

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under grant number # UG9MC30369 New Disorders Implementation Project for $4,000,000. This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

BACKGROUND

Pompe was added to the Recommended Uniform Screening Panel (RUSP) in March 2015, and X-Linked Adrenoleukodystrophy (X-ALD) and Mucopolysaccharidosis I (MPS I) were added in February 2016. State newborn screening programs pursuing universal implementation of these three new disorders encounter laboratory, staffing, clinical follow-up, personnel, equipment, education and legislative challenges.

PURPOSE

The purpose of this meeting is to convene newborn screening personnel (both APHL awardees and non-awardees) who support the implementation of new disorders added to the RUSP, as well as pertinent partners and stakeholders who have experience with implementing new disorders.

OBJECTIVES

1. Discuss current status of as well as future for MPS I, x-ALD and Pompe newborn screening in the United States.
2. Discuss decision points, barriers, unintended consequences to address in preparing for and implementing population screening for new disorders.
3. Identification and sharing of tools/resources used by newborn screening programs to prepare for and conduct screening for new disorders.

CONTACT

Kshea Hale, MPH Specialist

Email: Kshea.Hale@aphl.org
Phone: 240.485.3842

DAY 1

AGENDA

8:00 – 8:30 am	Arrival/Continental Breakfast *(Salons AB- Foyer)*
8:30 – 8:45 am	Welcome *(Salons AB)*
8:45 – 9:15 am	Lessons Learned- New Disorders Project Meeting: *Yvonne Kellar-Guenther (NewSTEPs)*
9:15 – 10:15 am	Building a Cost Analysis • *Scott Grosse (Centers for Disease Control and Prevention)* • *Andreas Rohrwasser and Robert Paul (Utah Public Health Laboratory)*
10:15 – 10:30 am	APHL Public Policy Tools: *Jelili Ojodu and Nisha Quasba (Association of Public Health Laboratories)*
10:30 – 10:45 am	Questions/Discussion
10:45 – 11:00 am	Break *(Salons AB- Foyer)*
11:00 – 11:10 am	Surveillance Case Definitions around Pompe: *Careema Yusuf and Marci Sontag (NewSTEPs)*
11:10 - 11:40 am	Long Term Follow-Up Clinical Guidelines for Pompe: *Austin Hamm (East Tennessee Children's Hospital)*
11:40 - 11:55 am	Questions/Discussion
11:55 am – 12:55 pm	Lunch (provided) *(Salons CD)*
12:55 – 1:05 pm	Surveillance Case Definitions around MPS I: *Careema Yusuf and Marci Sontag (NewSTEPs)*
1:05 – 1:35 pm	Long Term Follow-Up Clinical Guidelines for MPS I: *Tomi Toler (Washington University School of Medicine)*
1:35 – 1:50 pm	Questions/Discussion
1:50 – 2:30 pm	Breakout Session: Readiness Scale Phases • Group 1: Phase 1- Authority to Screen *(Salon C)* • Group 2: Phase 2- Lab & Follow-Up Logistics *(Salons AB)* • Group 3: Phase 3- Education *(Salons AB)* • Group 4: Phase 4- Full Implementation *(Salon E)*
2:30 – 3:00 pm	Group Discussion
3:00 – 3:15 pm	Break *(Salons AB- Foyer)*
3:15 – 3:25 pm	Surveillance Case Definitions around X-ALD: *Careema Yusuf and Marci Sontag (NewSTEPs)*
3:25 – 3:55 pm	Long Term Follow-Up Clinical Guidelines for X-ALD: *Gerald Raymond (Penn State Children's Hospital)*
3:55 – 4:10 pm	Questions/Discussion
4:10 – 4:45 pm	Facilitated Group Discussion: *Yvonne Kellar-Guenther (NewSTEPs)*
4:45 – 5:00 pm	Wrap Up and Adjourn

DAY 2

AGENDA

7:30 – 8:00 am	Arrival/Continental Breakfast *(Salons AB- Foyer)*
8:00 – 8:15 am	Overview of Day 1: *Sikha Singh and Kshea Hale (Association of Public Health Laboratories) (Salons AB)*
8:15 – 9:00 am	Educating Newborn Screening Staff: *Mei Baker (Wisconsin Department of Health)* and *Suzanne Canuso (New Jersey Department of Health)*
9:00 – 9:15 am	Educating Clinicians, Genetic Counselors, & Providers: *Amy Gaviglio (Minnesota Department of Health)*
9:15 – 9:45 am	Family Education: *Amelia Mulford (Baby's First Test) and Kimberly Piper (Iowa Department of Public Health)*
9:45 – 10:15 am	Questions/Discussion
10:15 – 10:30 am	Break *(Salons AB- Foyer)*
10:30 – 11:30 am	Educating Providers: Clinician and Families Panel • *Austin Hamm (Clinician- Pompe)* • *Amanda Java (Parent- Pompe)* • *Tomi Toler (Clinician- MPS I)* • *Kenneth Jarrell (Parent- MPS I)* • *Gerald Raymond (Clinician- X-ALD)* • *Kerri DeNies (Parent- X-ALD)*
11:30 am - 12:00 pm	Questions/Discussion
12:00 – 1:00 pm	Working Lunch *(Salons CD)*
1:00- 1:30 pm	*Next Steps/Wrap Up*
1:30 pm	Adjourn

NOTES

Biographies

Mei Baker, MD

Mei Baker, MD, is an associate 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 received her medical education in China, where she practiced medicine before receiving biochemical and molecular genetics training in the United States. By the time she joined the Wisconsin Newborn Screening Laboratory as an Associate Scientist in 2003, Dr. Baker had been working in the molecular genetics field for over 10 years, both in academic and industrial settings. Dr. Baker obtained her American Board Medical Genetics Certification in Clinical Biochemical Genetics in 2009. She serves as the co-chair of the NewSTEPs Steering Committee.

Michele Caggana, ScD, FACMG

Michele Caggana, Sc.D., 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 a member and past Chair of the Molecular Subcommittee and Chair of the Newborn Screening and Genetics in Public Health Committee for the Association of Public Health Laboratories. 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.

Suzanne Canuso, MSN, RN

Suzanne Canuso is a Public Health Nurse Consultant for the NJ Department of Health, Newborn Screening and Genetic Services program. She earned her bachelor's and master's of science in nursing degrees from Thomas Jefferson University school of Nursing in Philadelphia. Suzanne has been with the Newborn Screening program for the last four years, out of her eight years at the NJ Department of Health. Her primary role is to manage grants for newborn screening services in NJ that support an infrastructure of specialty care and ensure access to care for children and families impacted by newborn screening.

Kerri DeNies

Kerri DeNies was born and raised in Manchester, CT, along with her four sisters. She received her classical ballet training at Connecticut Concert Ballet and went on to become a ballet teacher at the young age of 16. Kerri moved to San Diego, CA in 2007 and has since continued as a Ballet Instructor and Ballet Mistress at the San Diego Civic Youth Ballet. She particularly enjoys teaching her "Ballet For Me" class, which focuses on children with unique physical needs. She co-developed this program to make it possible for all children to have the joyful experience of dance.

After her son was diagnosed with Adrenoleukodystrophy (ALD) in 2016, (in thanks to California adding the test to the newborn screening panel), Kerri has become dedicated to sharing her story to help other families with their diagnosis. Along with her family, she is working to spread the word about this disease to bring more awareness, and in hopes that the test will be added universally for all newborns. She is actively raising funds for research through various efforts.

Kerri lives with her partner, Eddie, her two-year-old son, Gregory, and her two rescue dogs, Pippa and Winter.

Adrianna Evans, MPH
Adrianna (Annie) Evans serves as Program Coordinator at Genetic Alliance for both the PCORnet Coordinating Center and Expecting Health teams. In this role, she works to engage different stakeholders in several Genetic Alliance programs through outreach and the development of practical tools. Annie has previously volunteered with families with children experiencing challenging health conditions and wants to continue improving the family experience in seeking healthcare.

Annie earned her Bachelor's degree in Biology from Penn State University in 2015 and her Master of Public Health from George Washington University in 2017. Her focus was on Global Health Program Design, Monitoring, and Evaluation. She is passionate about providing good quality non-communicable disease healthcare that transcends cultural and language barriers.

Amy Gaviglio, MS, CGC
Amy Gaviglio is a certified genetic counselor and has been employed by the Minnesota Department of Health, Newborn Screening Program for the past 10 years. Amy currently oversees follow-up of blood spot, hearing, and pulse oximetry results for the program and provides oversight of informatics and policy related initiatives. She is currently the co-chair of the CCHD Technical Assistance workgroup and a member of the Short Term Follow-Up, Legal and Legislative Issues in Newborn Screening, and New Conditions workgroups for APHL. She also serves as Vice Chair of the NBS Expert Panel for CLSI, APHL's Newborn Screening and Genetics in Public Health Committee, Baby's First Test Clearinghouse Steering Committee, and the ACHDNC Education and Training workgroup.

Scott Grosse, PhD
Scott Grosse trained in population and development economics at the University of Michigan. Since 1996 he has been employed at the Centers for Disease Control and Prevention (CDC) in Atlanta, where he currently serves as a health economist within the National Center for Birth Defects and Developmental Disabilities. He has a particular interest in the impact and cost-effectiveness of newborn screening for disorders such as PKU, congenital hypothyroidism, congenital adrenal hyperplasia, cystic fibrosis, and sickle cell disease.

Kshea Hale, MPH
Kshea Hale is a Specialist for the Newborn Screening and Genetics Program at the Association of Public Health Laboratories working with New Disorders. Kshea has over 10 years' experience working in public health in a variety of areas including infectious disease and maternal and child health. Ms. Hale holds a Master of Public Health from Boston University.

J. Austin Hamm, MD, FACMG
J. Austin Hamm, MD, FACMG completed his medical degree and general pediatrics residency through the University of Tennessee and completed additional training in clinical genetics and medical biochemical genetics at the University of Alabama at Birmingham. He is dual boarded by the American Board of Pediatrics and the American Board of Genetics and Genomics. He presently serves as Medical Director of Pediatric Genetics at East Tennessee Children's Hospital and provides clinical follow up for patients in the region who are identified through newborn screening. He lives in Knoxville, TN with his wife and two children.

Patrick Hopkins
Patrick Hopkins is the retired Chief of the Missouri Newborn Screening Laboratory after working for the Missouri State Public Health Laboratory for over 33 years, with 27 years in the Newborn Screening Laboratory. Patrick serves on APHL's NBSGPH Committee and was a former Chair of the APHL QA/QC Subcommittee. He has made great contributions over the years in NBS emergency preparedness across the Heartland States and with the implementation of LSD screening in Missouri. Patrick continues to work part-time in the Missouri newborn screening laboratory as a Project Specialist leading Missouri's Peer Network Resource Center made possible by NewSTEPs to assist other state programs with their implementation efforts for Pompe and MPS I.

Kenneth Jarrell
Kenneth Jarrell lives in Dexter, Missouri. He graduated from Bloomfield High School in 2000. Kenneth married his wife, Tina (Strickland) Jarrell, on July 3, 2008. They welcomed their first child, Josiah, on November 8, 2010 and their second child, Elizabeth, on August 28, 2015. Kenneth graduated Three Rivers Community College with an Associate Degree in Nursing in December 2014. He is currently employed at WW Wood Products in Dudley Missouri. When he's not at work, Kenneth enjoys spending time with his family outdoors and watching sports with his son. He also enjoys spending time with friends and attending church.

Amanda Java
Amanda Java is a home nurse that lives in Wisconsin with her husband Jon Java and their four children. They were all diagnosed with Pompe disease. The Java's learned of their children's Pompe status after giving birth to their fourth child through the Wisconsin Newborn Screening program. She is honored to be getting the opportunity to discuss her experience with Pompe disease and the treatment it needs.

Yvonne Kellar-Guenther, PhD
Dr. Yvonne Kellar-Guenther is the program evaluator for NewSTEPs. She has conducted program evaluation for over 20 years and teaching research and evaluation design for the past 10 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.

Amelia Mulford
As Program Coordinator at Genetic Alliance, Amelia provides support to the Expecting Health team and to Baby's First Test, which houses the Newborn Screening Clearinghouse. In her role, she creates and manages content for the English and Spanish Baby's First Test websites, coordinates collaborative activities with partners, shares educational materials at national conferences, and contributes articles on newborn screening to Exceptional Parent magazine. In addition, she leads the condition-specific parent education work groups as part of the New Disorders Implementation Project and previously presented at the National New Disorders Meeting in June 2017. Amelia holds a B.A. in Psychology and Hispanic Studies and will be entering the Johns Hopkins/NHGRI Genetic Counseling Master's program this fall.

Kimberly Noble Piper, RN, BS, CPH, CPHG
Kimberly Noble Piper is the state genetics coordinator for Iowa and the executive officer for the Center for Congenital and Inherited Disorders at the Iowa Department of Public Health. She is certified in public health and public health genomics. Ms. Noble Piper started her nursing career as a maternity nurse and manager of maternity services in a large hospital in Des Moines.

Jelili, Ojodu, MPH
Jelili Ojodu, MPH, is the Director for Newborn Screening and Genetics Program at the Association of Public Health Laboratories (APHL). He is also the Project Director for the Newborn Screening Technical assistance and Evaluation Programs (NewSTEPs). Mr. Ojodu is responsible for providing guidance and direction for the Newborn Screening and Genetics in Public Health Program. Prior to joining APHL, he spent four years at Georgetown University Medical Center on a National Institutes of Health initiative to reduce infant mortality in the District of Columbia 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 the University of Maryland, College Park.

Robert Paul
Robert Paul is the financial manager at the Utah Public Health Laboratory.

Nisha Quasba, MPH
Nisha Quasba is an associate specialist in Public Policy at the Association of Public Health Laboratories (APHL). For our membership to stay alert of new NBS legislation in their state, she worked closely with the NewSTEPS staff to develop a State Legislative Tracker that continuously updates with any NBS-specific bills that may impact PHLs. Currently she is working on understanding the

financial models of NBS programs nation-wide by contacting states and collating data into a policy analysis. She occasionally goes to the Hill with the Policy Team to advocate for PHL and has written Lab Matters articles reflecting on those conversations and discussions. Internally, she has assisted in moving NBS position statements through the development process and to publication. She holds a MPH in Health Policy and Management from the Milken School of Public Health at George Washington University.

Gerald Raymond, MD

Dr. Gerald Raymond is a neurologist and clinical geneticist at Penn State Children's Hospital where he is also a professor of pediatrics and neurology. He obtained his MD at the University of Connecticut and was trained in pediatrics at Johns Hopkins Hospital and Neurology at the Mass. General Hospital. He subsequently received training in clinical genetics at the Harvard Longwood Program. Dr. Raymond's research has been at the overlap of genetics and neurology with specific focus on peroxisomal disorders including adrenoleukodystrophy (ALD). He has worked extensively in the past 10 years in developing and expanding newborn screening for ALD.

Andy Rohrwasser, PhD, MBA

Dr. Andy Rohrwasser is the director of the Newborn Screening Laboratory and serves as the Deputy Director of Utah Public Health Laboratory. Andy is trained in human genetics and holds an MBA. Andy is passionate about newborn screening and cares deeply about patients with rare and undiagnosed disorders.

Sikha Singh, MHS, PMP

Ms. Singh is the Manager of Operations within the Newborn Screening and Genetics program at the Association of Public Health laboratories, where she serves as program manager for the NewSTEPs program. She joined APHL in 2009 and has significant experience in high-throughput genomic sequencing. Ms. Singh holds a Master of Health Science degree from the Johns Hopkins University and a Bachelor of Science degree from Temple University. She is a member of the Delta Omega Honorary Society as well as the recipient of a Project Management Professional (PMP) certificate.

Marci Sontag, PhD

Marci Sontag is an Associate Professor of Epidemiology and Pediatrics at the Colorado School of Public Health and the University of Colorado Anschutz Medical Center, and the associate director of NewSTEPs and the director of NewSTEPs 360. She has a PhD in Epidemiology and an MS in Biometrics from the University of Colorado Health Sciences Center. Dr. Sontag has studied clinical outcomes and newborn screening in cystic fibrosis since 1995.

Tomi L. Toler, MS, CGC

Tomi L Toler, MS, CGC is a genetic counselor at Washington University School of Medicine (WUSM) in St. Louis. Tomi graduated from the University of North Carolina at Greensboro Genetic Counseling Program in 2009 and has been at WUSM since 2015. At WUSM, Tomi is one of the genetic counselors primarily involved in the follow-up of newborn screening for lysosomal storage diseases (LSDs) and the coordination of care for patients with LSDs. Tomi has a personal connection to LSDs as her husband has Fabry disease.

Beth Vogel, MS, CGC

Beth Vogel, a board certified genetic counselor, is a research scientist within the New York State Newborn Screening Program. She is the Director of Operations for the NYS Newborn Screening Program and is the Project Manager for the New York-Mid-Atlantic Regional Genetics Network. She serves on the APHL New Steps Steering Committee. Prior to joining the Newborn Screening Program, Vogel was a pediatric genetic counselor at Albany Medical Center. She received a Bachelor of Science degree in Psychobiology from the State University of New York at Binghamton and a Master of Science Degree in Medical Genetics from Indiana University.

Careema Yusuf, MPH

Careema Yusuf is a Manager for the Newborn Screening, Technical assistance and Evaluation Program (NewSTEPs) at the Association of Public Health Laboratories (APHL) where she manages the

overall activities related to the website and data repository system for NewSTEPs. Careema provides technical assistance and support related to quality practices, data analysis and reporting. She has worked in a variety of public health settings that have included training in outbreak investigation policies and procedures to health personnel in Ghana, working at the State of Maryland on HIV/AIDS prevention program performance measures and with Johnson, Bassin & Shaw International working on quality improvement activities within substance use treatment programs in the US. Ms. Yusuf holds a Master of Public Health from the George Washington University in Washington, DC.