NewSTEPs New Disorders Implementation Meeting

Date: June 19, 2018

Location:

Washington Marriott at Metro Center
775 12th Street NW,
Washington, DC 20005-3901

Background

Pompe newborn screening was added to the RUSP in March 2015 and X-Linked Adrenoleukodystrophy (X-ALD) and Mucopolysaccharidosis I (MPS I) were added in February 2016. As of June 1, 2018, 12 states are screening for Pompe, 11 states are screening for MPS I and 10 states are screening for X-ALD. Current challenges faced by states newborn screening programs in implementing newborn screening include: (1) integration of additional instrumentation, space and capacity within the lab, (2) laboratory staffing to conduct tests, (3) clinical follow-up capacity and resources, (4) funding for personnel, equipment, education, and (5) gaining legislative or statutory approval.

Meeting Purpose

The purpose of this meeting is to convene APHL New Disorder Implementation subawardees to discuss activities toward achieving project goals. Subawardees will provide a status update regarding where they are in the implementation of new disorder (Pompe, MPS I, x-ALD) newborn screening and discuss challenges and successes associated with implementing new disorders as well as share resources.

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Tuesday, June 19, 2018	
Time	**Agenda**
8:30 am – 9:00 am	Arrival and Continental Breakfast (*Salons A/B- foyer*)
9:00 am – 9:10 am	Welcome: *Jelili Ojodu, NewSTEPs* (*Salons A/B)*
9:10 am – 9:20 am	Overview and Goals of the Day: *Yvonne Kellar-Guenther, NewSTEPs*
9:20 am –10:05 am	Cut-off and Validation Discussions: *Michael Sarzynski, Michigan Department of Health* *Amy Gaviglio, Minnesota Department of Health* *George J Dizikes and Christine Dorley, Tennessee Department of Health*
10:05 am – 10:25 am	Cut-off and Validation Q & A
10:25 am – 10:40 am	Break (*Salons A/B- foyer*)
10:40 am – 11:55 am	Second-Tier Screening Panel: *Joe Orsini, New York State Department of Health (moderator)* *Patrick Hopkins, Missouri Department of Health* *Dietrich Matern, Mayo Clinic* *Michele Caggana, New York State Department of Health*
11:55 am – 12:15 pm	Second-Tier Screening/Confirmatory Testing Q & A
12:15 pm – 1:15 pm	Lunch *(Salon C)*
1:15 pm – 2:00 pm	Workflow Models for Laboratory: *Michael Sarzynski, Michigan Department of Health* *George J Dizikes and Christine Dorley, Tennessee Department of Health*
2:00 pm – 2:15 pm	Lab Workflow Model Q & A
2:15 pm – 3:15 pm	Workflow Models for Follow-Up: *Beth Vogel, New York State Department of Health* *Sharmini Rogers, Missouri Department of Health* *Karen Eveans, Nebraska Department of Health*
3:15 pm – 3:30 pm	Follow-Up Workflow Models Q & A
3:30 pm – 3:45 pm	Break (*Salons A/B- foyer*)
3:45 pm – 4:00 pm	Activity: *Yvonne Kellar-Guenther*
4:00 pm – 4:45pm	Team Updates
4:45 pm – 5:00 pm	Next Steps

Biographies

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.

Christine Dorley

Christine completed her undergraduate studies in medical technology from Western Kentucky University in Bowling Green, Kentucky and obtained a Master's degree in public service management from Cumberland University in Lebanon, Tennessee. She joined the Tennessee Department of Health Laboratory Services in 1994 and worked performing testing in Serology and Special Microbiology before moving to Newborn Screening. Christine is currently serving as the Assistant Director for the State of Tennessee Newborn Screening Division and has been in this role since 2007. In this position she oversees screening of all Tennessee newborns for metabolic and genetic diseases.

George J Dizikes, PhD, HCLD/CC(ABB)

George Dizikes joined the Tennessee Department of Health Knoxville Regional Laboratory as its Director in September, 2015. Prior to coming to Knoxville, Dr. Dizikes spent nineteen years with the Illinois Department of Public Health laboratory at Chicago, where he had been the laboratory manager, supervised the molecular diagnostics and newborn screening sections, and served as CLIA director. Dr. Dizikes received his PhD in microbiology from the University of Minnesota, and he completed a post-doctoral fellowship at UCLA on the genetics of metabolic diseases. Dr. Dizikes serves on the Association of Public Health Laboratories Committee on Newborn Screening and Genetics in Public Health where he participates on workgroups dealing with legal and legislative issues and new conditions. He is also a member of the Laboratory Procedures & Standards Workgroup of the US Department of Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children. Since coming to Tennessee, he has advised the newborn screening program, particularly on issues of timeliness and the addition of screening for lysosomal storage disorders.

Karen A. Eveans, MD

Dr. Eveans graduated from Rush Medical College in Chicago, Illinois. She completed a residency in Pediatrics at Butterworth Hospital/MSU in Grand Rapids, Michigan. Dr. Eveans practiced Pediatrics in both a staff model HMO and private practice settings. Dr. Eveans has worked for 12+ years in newborn screening follow-up. She has primary responsibility for following up on abnormal CF, Hemoglobinopathy and SCID results and post-transfusion specimens. As of

July, 2018 she will also be responsible for abnormal X-ALD results.

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.

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.

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. Yvonne is an Associate Professor of Community and Behavioral Health at the Colorado School of Public Health. She holds a PhD in Communication and Health.

Dietrich Matern, M.D., PhD
Dietrich Matern, M.D., Ph.D., is a Professor of Laboratory Medicine, Medical Genetics and Pediatrics, and serves as Chair of Mayo Clinic's Division of Laboratory Genetics and Genomics, as well as co-director of Mayo Clinic's Biochemical Genetics Laboratory. His primary research interest is the development and improvement of laboratory assays for the effective and efficient screening, diagnosis and follow-up of patients with inborn errors of metabolism. Dr. Matern authored or co-authored more than 140 peer-reviewed papers and 20 textbook chapters. His research has been funded by the National Institute of Child Health and Human Development (NICHD), the Newborn Screening Translational Research Network (NBSTRN), the Legacy of Angels Foundation, and other not-for-profit organizations.

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 George Washington University and a Bachelor of Science degree in Biological Sciences from the University of Maryland, College Park.

Joseph J. Orsini, PhD
Joseph James Orsini, PhD, is the Director of Operations, Lysosomal Storage Disorder Screening Laboratory Manager and Follow-up Unit Manager for the Newborn Screening Program at the New York State Department of Health. Dr. Orsini received his PhD in Analytical Chemistry from the University of Vermont, and a master's degree in Laboratory Sciences program development. He is a member of the Quality Assurance and Quality Control Subcommittee at the Association of Public Health Laboratories, as well as the Wadsworth Center QA Council. He has also been a member of the Lysosomal Storage Disease Screening Work Group headed by the CDC. He continues to be a journal reviewer for the Molecular Genetics and Metabolism, Clinical Chemistry Clinica and Chimica Acta. Dr. Orsini received grants from both the Health Resources and Services Administration (HRSA) and the National Institute of Health (NIH) for effective follow-up in Newborn Screening and an LSD pilot study. He has received professional honors & recognition such as the Wadsworth Center Above and Beyond Award and the Commissioner's Recognition Award.

Sharmini Rogers, M.B.B.S, MPH
Sharmini Rogers received a Bachelor of Medicine and Bachelor of Surgery (M.B.B.S.) from Kasturba Medical College, University of Mysore, Karnataka State, India in 1985 and a Master's in Public Health from the University of Michigan, Ann Arbor in 1998. Dr. Rogers has held the position of Chief in the Bureau of Genetics and Healthy Childhood overseeing the newborn screening and other maternal and child health programs within the DHSS since 2003, as well as other leadership positions within DHSS since 1999. Including her work as a physician in Malaysia, she has over 26 years of experience within public health.

Michael Sarzynski, M.S
Mike Sarzynski has worked as a scientist at Michigan NBS for 6 years and is currently the lead LSD scientist. He enjoys writing procedures, troubleshooting, reporting results and actually running an assay from time to time. When not at work, Mike likes playing Ultimate Frisbee, eating his wife's Instant Pot creations, and having light saber duels and nerf gun battles with his kids.

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.