



A NATIONAL CONVERSATION ON NEWBORN SCREENING RESEARCH AND INFORMED CONSENT

June 1-2, 2015

[Renaissance Washington, DC Dupont Circle Hotel](#)

New Hampshire Ballroom

1143 New Hampshire Avenue NW

Washington, DC 20037-1522

BACKGROUND

The informed consent provisions included in the Newborn Screening Saves Lives Reauthorization Act went into effect on March 16, 2015. The law includes two significant changes to the human subjects regulations as they apply to research with newborn dried blood spots.

- First, the law requires that all research funded pursuant to the Public Health Service Act using newborn dried spots be considered human subjects research regardless of whether the specimens are identifiable. This is contrary to established regulatory approaches in which biological specimens that are not linked to identifying information are not considered identifiable and therefore do not constitute “human subjects” as defined at 45 CFR 46.102(f).
- Second, the law eliminates the ability of the Institutional Review Board (IRB) to approve alterations or waivers of informed consent under 45 CFR 46.116(c) and 116(d) for research involving newborn dried blood spots. This is contrary to long-established IRB practices in which research involving stored identifiable clinical specimens and data could be carried out under a waiver of informed consent if the IRB determined that the ethical and regulatory requirements were satisfied.

The purpose of this meeting is to convene pertinent stakeholders to discuss the informed consent provisions, their interpretation, implications and impact on newborn screening.

MEETING OBJECTIVES

- Understand State concerns and anticipated implications of the Newborn Screening Saves Lives Act Amendment.
- Describe the impact of the Law on Federally funded research activities.
- Discuss the impact of the Law on State Institutional Review Boards (IRBs)
- Outline routine newborn screening laboratory activities that require usage of residual dried blood spots. Discuss whether these activities may be characterized as either research or non-research activities.
- Discuss issues related to broad consent for future use of residual dried blood spots.
- Identify needs and strategies to educate the public about newborn screening, and options to participate in newborn screening research.

LOGISTICS POINT OF CONTACT

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AGENDA DAY 1: MONDAY, JUNE 1, 2015

NEW HAMPSHIRE BALLROOM

HOST: Jelili Ojodu, MPH, Association of Public Health Laboratories

- 8:00** **Breakfast (served New Hampshire Ballroom)**
- 8:30** **Welcome**
- Statement of Newborn Screening Saves Lives Act
 - Statement of broad concerns
 - Scope of meeting
 - Desired outcomes of meeting
- 8:40** **Participant Introductions**
- 9:00** **Introduction to and Background on the 2014 Newborn Screening Saves Lives Act Amendment (Section 12)**
- 9:00 The development of the language for the Newborn Screening Saves Lives Act Amendment | [Peter Kyriacopoulos, Association of Public Health Laboratories](#)
- 9:15 Summary and Highlights of the National Institutes of Health (NIH) Meeting on Informed Consent | [Tiina Urv, PhD, National Institutes of Health, US Department of Health and Human Services](#)
- 9:25 Recommendations from the Secretary's Advisory Committee on Human Research Protections Meeting and Discussion of Implications | [Michelle Huckaby-Lewis, MD, JD, Johns Hopkins University](#)
- 9:45 Recommendations from the Advisory Committee on Heritable Disorders in Newborns and Children Meeting and Discussion of Implications | [Joseph A. Bocchini, Jr., MD, Advisory Committee on Heritable Disorders in Newborns and Children](#)
- 10:00** **Comments and Questions**
- 10:15** **Break**
- 10:30** **State and Federal Considerations Regarding the New Amendment for State Newborn Screening Programs**
This session will feature presentations providing a background and information on state and federal stakeholders.

- 10:30 Overview of routine program activities in newborn screening, as required for CLIA/CAP compliance, quality improvement and program expansion | **Michele Caggana, ScD, FACMG, New York State Department of Health**
- 10:50 Food and Drug Administration (FDA) Laws pertaining to human research protection, the use of investigational devices and considerations for use of Laboratory Developed Tests (LDTs) | **Kellie Kelm, PhD, Food and Drug Administration, US Department of Health and Human Services**
- 11: 15 Office for Human Research Protections (OHRP) role and considerations in the definition of research | **Jerry Menikoff, MD, JD, Office for Human Research Protections, US Department of Health and Human Services**
- 11:45 Comments and Questions
- 12:00 Lunch (served New Hampshire Ballroom)**
- 1:00 What Constitutes Newborn Screening Research: An evaluation of essential program activities for screening of current and new conditions.**
This session will constitute a review of a variety of newborn screening scenarios and discussion regarding research implications. State, OHRP and FDA perspectives will be considered and discussed.
- Newborn Screening Programs Scenario Overview and Proposed Classifications | **Susan Tanksley, PhD, Texas Department of State Health Services**
- 2:30 Break**
- 3:00 (Continued) Newborn Screening Programs Scenario Overview and Proposed Classifications | **Susan Tanksley, PhD, Texas Department of State Health Services****
- 4:30 Adjourn**

AGENDA DAY 2: TUESDAY, JUNE 2, 2015

NEW HAMPSHIRE BALLROOM

HOST: Jelili Ojodu, MPH, Association of Public Health Laboratories

- 8:00** **Breakfast (served New Hampshire Ballroom)**
- 8:30** **Recap of Day 1 and Discussion** | **Dave Orren, JD**, *Minnesota Department of Health*
- 8:45** **Informed Consent for Newborn Screening Research: Examples of a Broad Consent Package**
This session will focus on contents and requirements of a comprehensive informed consent package for States endeavoring to perform newborn screening research.
- 8:45 Experience from Texas | **Susan Tanksley, PhD**, *Texas Department of State Health Services*
- 9:15 Experience from Massachusetts | **Anne Comeau, PhD**, *University of Massachusetts Medical School*
- 9:45 Experience from Michigan | **Carrie Langbo, MS, CGC**, *Michigan Department of Health and Human Services*
- 10:15 Comments and Questions
- 10:30** **Break**
- 10:45** **Open Discussion on items to consider when developing broad informed consent models for newborn screening research**
- Michelle Huckaby-Lewis, MD, JD**, *Johns Hopkins University*
Dave Orren, JD, *Minnesota Department of Health*
Denise Chrysler, JD, *Network for Public Health Law*
- 12:00** **Lunch (served in New Hampshire Ballroom)**
- 1:00** **Educating the Public about Newborn Screening: Current and Proposed Communication Strategies**
This session will focus on public outreach and educational initiatives as they relate to newborn screening and informed consent.
- 1:00 Communication Strategies for Newborn Screening – Where do we go from here? | **Natasha Bonhomme**, *Genetic Alliance*

- 1:20 Saving Babies through Screening Educational Activities and Perspectives on Informed Consent | [Jill Levy-Fisch](#), *Saving Babies through Screening*
- 1:35 Parental preference on research consent: Empirical research | [Aaron Goldenberg, PhD, MPH](#), *Case Western Reserve University*
- 1:55 Engaging the Public/Parents and Enlisting Providers | [Carrie Langbo, MS, CGC](#), *Michigan Department of Health and Human Services*
- 2:15 Awareness, Education, and Training on Newborn Screening | [Amy Gaviglio, MS, CGC](#), *Minnesota Department of Health*
- 2:15** Open discussion on identifying strategies and engaging advocates in targeted organizations: nurses, midwives, childbirth educators, family physicians and other health care professionals | [Natasha Bonhomme](#), *Genetic Alliance*
- 2:45** Next steps and closing
- 3:00** Adjourn

SPEAKER BIOS

Joseph A. Bocchini, Jr., MD

Dr. Bocchini is Professor and Chairman of the Department of Pediatrics at Louisiana State University Health Sciences Center in Shreveport. He is currently serving as Chairman of the Advisory Committee on Heritable Disorders in Newborns and Children. He is also a member of the Advisory Committee on Immunization Practices of the Centers for Disease Control and Prevention and a past Chair of the American Academy of Pediatrics Committee on Infectious Diseases.

Natasha Bonhomme

Natasha Bonhomme has led initiatives in the healthcare field. Since joining Genetic Alliance in 2006, she has worked to improve the state of newborn screening. For the past 6 years, she has overseen maternal and child health initiatives for the organization, with a particular focus on bringing the families perspective into policy setting around newborn screening, and maternal and child health overall. She also supervised five federally funded projects having to do with newborn screening and prenatal diagnoses. As vice president at Genetic Alliance, she launched the nation's center on newborn screening education, [Baby's First Test](#). As director of Baby's First Test, Natasha has testified before the US Senate Health, Education, Labor and Pension Committee's Subcommittee on Children and Families on the importance of public education for newborn screening. Natasha serves on a range of committees including: as a Co-Chair of the Genetics and Bioethics Committee, American Public Health Association; the Association of Public Health Laboratories Committee on Newborn Screening and Genetics in Public Health; and the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.

Michele Caggana, ScD, FACMG

Michele Caggana received her doctoral degree from the Harvard School of Public Health and completed post-doctoral work in molecular virology at the Wadsworth Center and in clinical molecular genetics at the Mt. Sinai School of Medicine in New York City. She has been on staff as a Research Scientist at the Wadsworth Center since 1996 and she is an Assistant Professor in the Department of Biomedical Sciences, School of Public Health. Dr. Caggana is board certified in Clinical Molecular Genetics by the American Board of Medical Genetics and a fellow of the American College of Medical Genetics. Dr. Caggana is the Director of the Newborn Screening Program since 2006 and also directs the Newborn Screening Molecular Laboratory. She is also the Chief of the Laboratory of Human Genetics and the Section Head for the Genetic Testing Quality Assurance Program at the Wadsworth Center. Dr. Caggana's laboratory uses molecular genetics techniques to study frequencies of specific gene mutations in dried blood spots. Her lab also is working towards large scale DNA sequence analyses using NextGen strategies to improve the newborn screening and health outcomes for infants who remain undiagnosed after screening.

Denise Chrysler, JD

Denise Chrysler has 30 years of experience in public health law practice. She joined the University of the Michigan School of Public Health in September 2010 to direct the Mid-States Regional Center of the Network for Public Health Law, which promotes and supports the use of law to protect and improve the

public's health. Previously, she was an attorney to the Michigan Department of Community Health for 27 years, serving as the Public Health Legal Director, the Director of the Office of Legal Affairs, the department's Privacy Officer, a member of the Department's Institutional Review Board, and as an Assistant Attorney General. She provided legal representation and counsel to the Department on an array of public health issues, including the development of the Michigan BioTrust for Health, which makes leftover newborn screening samples more useful and available for medical or public health research. Currently, Denise is a member of the Community Values Advisory Board, which provides guidance to the Michigan BioTrust on ethical issues including what types of research are, or are not, acceptable uses of the newborn screening samples. She also serves on the Bioethics and Legal Issues Workgroup of the Newborn Screening Translational Research Network.

Anne Comeau, PhD

Dr. Comeau earned her PhD at Brandeis University and was a fellow at the Harvard School of Public Health prior to accepting a position as research scientist in the Massachusetts Newborn Screening Program in 1988. Dr. Comeau is now Deputy Director of the New England Newborn Screening Program and Professor of Pediatrics at the University of Massachusetts Medical School. The principal focus of Dr. Comeau's work has been population-based newborn screening, where she directs the molecular laboratory for public health services and research. Dr. Comeau has been an avid advocate of interdisciplinary evidence-based development and evaluation of newborn screening protocols. She has also promoted policies and developed protocols that facilitated the Massachusetts expansion of newborn screening services in 1999 and again in 2009 (newborn screening for biochemical conditions, CF and SCID). Her publications 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 such as cystic fibrosis and SCID as well as considerations for consideration for research use of data held under public health stewardship.

Amy Gaviglio, MS, CGC

Amy Gaviglio has been employed by the Minnesota Department of Health, Newborn Screening Program for the past 8 years. In her current position, Amy supervises the short term follow-up of blood spot, hearing, and pulse oximetry results, aids in statewide educational efforts on newborn screening and genetics, and works on newborn screening-related policy in the state. She is currently the co-chair of the public health special interest group and a member of the public policy committee for the National Society of Genetic Counselors, and a member of the NewSTEPS Short Term Follow-Up workgroup and co-chairs the CCHD Technical Assistance workgroup.

Aaron Goldenberg, PhD, MPH

Aaron Goldenberg is an Assistant Professor of Bioethics at Case Western Reserve University and the Associate Director of the Center for Genetic Research Ethics and Law at Case Western. His work focuses on the ethical, legal and social implications of advances in genomic technology, newborn screening, health disparities, and the ethics of research involving newborns, children, and families. Dr. Goldenberg has a background in bioethics and public health genetics. He is currently the Co-PI of a HRSA funded project to explore the ethical and programmatic challenges of integrating genomics into Newborn Screening Programs. He is also the Principle Investigator on a project funded by the National Human Genome Research Institute to examine parental attitudes regarding the research use of biospecimens collected from newborns.

Michelle Huckaby-Lewis, MD, JD

Michelle Huckaby-Lewis is a pediatrician and an attorney with training in bioethics and health services research. She joined the Berman Institute of Bioethics in 2010. Lewis' current research focuses on the retention and use of residual newborn screening blood samples. Lewis received a BA in English and History from Stanford University, a JD from Vanderbilt University School of Law in 1992 and an MD from Tulane University School of Medicine in 2000. She completed a residency in Pediatrics at the David Geffen School of Medicine at UCLA in 2003.

Kellie B. Kelm, PhD

Kellie 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, Food and Drug Administration). Dr. Kelm joined the 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. She has represented the FDA on several external committees such as the US DHHS Advisory Committee on Heritable Disorders in Newborns and Children, Clinical and Laboratory Standards Institute Subcommittee for Newborn Screening and several CLSI Document Development Committees as well as the APHL NewSTEPS Steering Committee. Dr. Kelm received her AB at Dartmouth College and her PhD from the Johns Hopkins University School of Medicine.

Peter Kyriacopoulos

Into his third decade of work in the realm of federal public policy decision making, Peter Kyriacopoulos is the Senior Director of APHL's Public Policy department. He strives to be current and knowledgeable concerning both health-specific and more general federal policy issues in order to provide solid strategic advice on national policy directions, Administration initiatives, Congressional interests, and other Washington dynamics and how they affect the membership of APHL and its key federal partner, CDC. He is frequently sought out by the senior leadership of state and local governmental laboratories, CDC divisions with public health laboratory activity, and the Washington, DC public health community because of the value of his observations, insight, and forecasting skill. He also shares information on federal rule making that affects state and local laboratory operations, and marials organizational comments on those regulations working collaboratively with the APHL members and program staff while encouraging individual comments.

Carrie Langbo, MS, CGC

Carrie Langbo received her Master of Science in Genetic Counseling from Northwestern University and worked in reproductive genetics prior to her career in public health with the Michigan Department of Health and Human Services where she serves as coordinator of the Michigan BioTrust for Health and also coordinates the Cystic Fibrosis (CF) Newborn Screening Follow-up Program. Ms. Langbo was integrally involved in the development of both programs and continues to facilitate their various advisory boards, programmatic and research activities. Her current role includes monitoring implementation of the BioTrust consent process in birthing hospitals and collaborating with numerous MDHHS programs, the State Laboratory and the Michigan Neonatal Biobank to address policies and procedures for residual newborn screening blood spot use. She facilitates and provides education for

communities, health care professionals and researchers on topics relating to newborn screening in general as well as use of residual dried blood spots for research.

Jill Levy-Fisch

Jill Levy-Fisch began her journey into advocacy after a three-year search for answers about her youngest son's rare medical condition. She discovered that with the benefit of comprehensive newborn screening, her son's disorder could have been identified years earlier. As a result, she decided to lend her voice to a cause that would save the lives of children. In 2003, she began to volunteer for the Save Babies Through Screening Foundation (SBTS) and quickly became an active member. In 2004, Jill joined the Board of Directors as National Director of Education and Awareness. She became President of SBTS in 2006, and continues to hold that position to this day. Jill worked intimately on the writing and passage of the Newborn Screening Saves Lives Act. Her advocacy work led to the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) issuance of a letter to all states recommending that parents be informed of the existence of comprehensive newborn screening. She served for several years as a member of the Treatment and Followup Subcommittee of the SACHDNC during which time she addressed issues surrounding insurance coverage for medical foods for rare diseases. Jill has also represented the parent's perspective as a longtime advisory council member of the New York Mid-Atlantic Regional Collaborative (NYMAC). She is an executive producer of several newborn screening educational films that are in use worldwide and a co-producer of an educational piece for the Newborn Channel. She has also co-authored several articles published in peer-reviewed journals such as *Pediatrics*. Jill lives in Scarsdale, NY with her husband and 3 children.

Jerry Menikoff, MD, JD

Jerry Menikoff is the director of the Office for Human Research Protections in the U.S. Department of Health and Human Services. He previously served as the head of NIH's intramural human subjects protection program, and prior to that, he was Associate Professor of Law, Ethics and Medicine at the University of Kansas.

Dave Orren, JD

Dave Orren is the Chief Legal Counsel for the Minnesota Department of Health (MDH). Dave is responsible for managing the MDH Legal Unit, which provides confidential legal services and advice to the Commissioner, the Department, and MDH staff. He is the Department's liaison with the Attorney General's Office regarding litigation and other MDH legal services performed by the AG's Office. Dave is a member of the Department's Executive Leadership Team. Dave is a graduate of William Mitchell College of Law.

Susan M. Tanksley, PhD

Susan Tanksley is the Laboratory Operations Manager in the Laboratory Services Section of the Texas Department of State Health Services in Austin. She manages the day-to-day operations of Texas' public health laboratory, which encompasses the state newborn screening, clinical chemistry, microbiology, environmental chemistry and emergency preparedness laboratories. These high-volume testing areas process 4,500-5,000 specimens per day. Dr. Tanksley's focus has been on newborn screening program expansion and improvement, including implementation of evidence-based performance measures in the pre-analytical and post-analytical phases of screening. She has chaired the APHL Newborn Screening and

Genetics in Public Health Committee since 2010, co-chaired the Newborn Screening Workgroup for the Mountain States Genetics Regional Collaborative Center from 2009-2015, has served on the Secretary's Advisory Committee for Heritable Disorders in Newborns and Children (DACHDNC) as an organizational representative for APHL since 2013 and has served as a member of the Condition Review Workgroup for DACHDNC since 2012. Dr. Tanksley received a PhD in Genetics from Texas A&M University in 2000 and has been certified as a High Complexity Laboratory Director through the American Board of Bioanalysis since 2005.

Tiina Urv, PhD

Tiina Urv joined NICHD as a Program Director in October 2006. Dr. Urv is a developmental disabilities specialist with a Ph.D. from Columbia University and over 25 years of experience working with individuals with intellectual disabilities in both clinical and research settings. Prior to joining the Branch, she was an assistant professor at University of Massachusetts Medical School's Eunice Kennedy Shriver Center and a research scientist at the New York State Institute for Basic Research in Developmental Disabilities. Dr. Urv's work in the IDD Branch is broad. She serves as coordinator for the research program in newborn screening and manages a diverse portfolio of grants for the IDD Branch related to these efforts. In addition Dr. Urv manages grants with a focus on behavioral, biobehavioral and social sciences research related to developmental disabilities along with overseeing NICHD's Fragile X portfolio.