Newborn Screening: Current Landscape and Future Directions – Meeting 3

May 16th, 2024 | 9:00am–5:00pm ET

A Hybrid Event

ATTENDEE PACKET

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NEWBORN SCREENING: CURRENT LANDSCAPE AND FUTURE DIRECTIONS

STATEMENT OF TASK

An ad hoc committee of the National Academies of Sciences, Engineering, and Medicine will examine the current landscape of newborn screening (NBS) systems, processes, and research in the U.S. The committee will make recommendations for future improvements that help modernize NBS to be adaptable, flexible, coordinated, communicative, capable of efficient and sustainable adoption of screening for new conditions using new technologies, and an equitable public health program from which all infants benefit. The committee's work will focus on the following tasks:

1) Examine state and federal capacities to strengthen current screening processes and implement screening for new conditions, including considerations for future conditions added to the Recommended Uniform Screening Panel (RUSP).
2) Review existing and emerging technologies that would permit screening for new categories of conditions and describe:
    - how these new technologies may impact states;
    - changes to public health infrastructure needed to incorporate new technologies while upholding and implementing the required components of NBS;
    - options for incorporating new technologies to allow for screening of additional conditions;
    - research, technological, and infrastructure needs to improve diagnosis, follow-up, and public health surveillance.
3) Review NBS data collection processes for tracking disease prevalence, improving health outcomes, conducting longitudinal follow-up, ensuring health equity, defining the natural history of conditions that can be screened for, and measuring quality of life.
4) Examine the RUSP review and recommendation processes, including the process of selecting new conditions that could be added to the RUSP; conducting review of the evidence to support adding new condition; scaling up these review and recommendation processes to efficiently handle the review of potentially hundreds of conditions; and considering whether additional factors should be included in the analysis of harms and benefits (e.g., societal harms such as financial cost or opportunity costs, and family benefits such as avoiding the “diagnostic odyssey”).

The committee's final report will describe (a) short-term options that could be implemented at the state and/or federal level over the next 2-3 years to help strengthen existing NBS programs and address the current challenges facing state programs, and (b) a vision for the future of NBS and a roadmap for how to implement and achieve that vision over the next 5-15 years. The report will include options for how to implement longitudinal follow-up data collection to improve understanding of the impact of NBS on infant health outcomes (including morbidity and mortality, and quality of life for screen-positive infants). The committee will consider the resources required for implementation, such as changes to the current NBS system that will need to occur, the feasibility of implementing the future vision, and the challenges and barriers that may arise when trying to implement the roadmap.

Newborn Screening: Current Landscape and Future Directions – Meeting 3

Hybrid Meeting

THURSDAY, MAY 16TH, 2024

Through this virtual information-gathering session, the Committee on Newborn Screening: Current Landscape and Future Directions will gather input from experts and interested parties on the current landscape of newborn screening (NBS) systems in the United States and discuss issues informing a future vision for this system. The committee will deliberate and prepare a report providing short-term options to strengthen existing programs and a roadmap for the future. This information-gathering session focuses particularly on forward-looking needs and challenges, reflected in bullets 2 and 3 of the study's statement of task. Future information gathering sessions may explore other issues to be addressed in the study.

9:00am **Welcome and Opening Remarks**

Jewel Mullen (she/her), University of Texas at Austin, Dell Medical School; *Committee Chair*

Session I: Reflecting on Emerging Challenges for Newborn Screening and the Role of Public Trust

- Reflect on emerging challenges in the newborn screening system that could threaten public trust.
- Share opening perspectives on potential paths forward to strengthen the newborn screening system and the public's trust in it.

9:05am **Overview**

Faith Fletcher (she/her), Baylor College of Medicine, *Committee Member, Session Moderator*

9:10am **Panel Reflections and Discussion on Public Trust**

Ellen Wright Clayton (she/her), Vanderbilt University Medical Center

Sonia S. Hassan (she/her), Wayne State University

Susan Tanksley (she/her), Texas Department of State Health Services

Heidi Wallis (she/her), Association for Creatine Deficiencies

10:00am **BREAK**

Session II: Discussing Challenges and Opportunities for Building Newborn Screening Research Infrastructure

- Discuss opportunities for synergy between newborn screening research and newborn screening public health programs.
- Identify research, technological, and infrastructure needs and potential solutions for addressing them.
- Examine lessons learned from coordination between research programs and public health newborn screening.

10:15am **Session Overview**

Don Bailey (he/him), RTI International; *Committee Member; Session Moderator*

10:20am **Opportunities for Research Infrastructure to Inform and Prepare NBS Programs**

Stephen Kingsmore (he/him), Rady Children's Institute for Genomic Medicine

Building Research Infrastructure to Support NBS Preparedness

Kee Chan (she/her), ValueMinded LLC

Lessons Learned from Coordination between Research Programs and Public Health Newborn Screening

Denise M. Kay (she/her), New York State Department of Health

11:00am **Moderated Discussion with Speakers and Q&A**

11:30am **BREAK**

Session III: Considering How Higher Throughput Technologies Might Impact Newborn Screening Programs

For a deeper discussion on the potential uses and implications of genomics, please see the 2023 NASEM workshop on The Promise and Perils of DNA Sequencing of Newborns at Birth.

- Discuss challenges and opportunities related to the potential incorporation of different higher-throughput technologies into newborn screening programs.
- Envision solutions for addressing the increased burdens that screening for more conditions could place on the newborn screening system if this were to occur.
- Examine opportunities to maximize efficiency and eliminate redundancy in the newborn screening system.

11:50am **Session Overview**

Wendy Chung (she/her), Boston Children's Hospital; *Committee Member; Session Moderator*

11:55am **Genomics Is Not the Only Way to be Higher Throughput**

Expansion of Newborn Screening by Consolidated Biomarker Analysis and Genomic Sequencing to Find Highly Penetrant Genotypes

Michael H. Gelb (he/him), University of Washington

Artificial Intelligence / Machine Learning

Curt Scharfe (he/him), Yale University

12:20pm **Genetic Counselor Workforce Development and Diversification**

Michelle Takemoto (she/her), Alliance for Genomic Justice

12:30pm **Moderated Discussion with Speakers and Q&A**

1:00pm **LUNCH**

Session IV: Envisioning the Future of Data Collection Processes in Newborn Screening

- Review current state and federal data collection processes related to newborn screening.
- Discuss options for building a data infrastructure for the newborn screening system.
- Consider future-looking options for data collection processes related to longitudinal follow-up.
- Identify barriers and opportunities for collecting data across the system.

1:50pm **Session Overview**

Jochen Profit (he/him), Stanford University; *Committee Member; Session Moderator*

1:55pm **Challenges and Opportunities for Data Collection in Newborn Screening**

Marci Sontag (she/her), Center for Public Health Innovation

2:10pm **Future Looking Options for Longitudinal Data collection**

Ines Maria Vigil (she/her), Independent

Julia Skapik (she/her), National Association of Community Health Centers

2:35pm **Moderated Discussion and Q&A**

3:05pm **BREAK**

Session V: Navigating the Storage and Secondary Use of Newborn Dried Blood Spots

- Examine current processes for storage and secondary research with dried blood spots, ongoing challenges to systems in place, and potential paths forward.
- Reflect on the role of informed consent and other protections in research involving human participants and consider whether they might apply.
- Explore diverse perspectives on the storage and secondary use of blood spots, and potential procedures and protections that could be considered moving forward.

3:25pm **Session Overview**

Krystal Tsosie (she/her), Arizona State University; *Committee Member, Session Moderator*

3:30pm **Lessons Learned from the Michigan BioTrust for Health**

Dominic Smith (she/her), Michigan Department of Health and Human Services

Eric Hendricks (he/him), Michigan Department of Health and Human Services

Considering Protections

Jerry Menikoff (he/him), National University of Singapore

3:55pm **Panel Reflections**

Ellen Wright Clayton (she/her), Vanderbilt University Medical Center

Susan Tanksley (she/her), Texas Department of State Health Services

Heidi Wallis (she/her), Association for Creatine Deficiencies

4:20pm **Moderated Q&A and Discussion**

4:55pm **Closing Remarks**

Jewel Mullen (she/her), University of Texas at Austin, Dell Medical School; *Committee Chair*

5:00pm **ADJOURN**

Biosketches of Speakers

Session 1 Speakers

Ellen Wright Clayton, MD, JD (she/her)

Ellen Wright Clayton, MD, JD, is the Craig-Weaver Professor of Pediatrics, Professor of Health Policy, and Co-Founder of the Center for Biomedical Ethics and Society at Vanderbilt University Medical Center and Professor of Law at Vanderbilt School of Law. An internationally known scholar, her research has focused on addressing ethical, legal, and social issues in genomics and its translation to clinical care. She has written many articles, in particular, on newborn screening and has served on Tennessee's Genetics Advisory Committee for more than 25 years. She is an elected member of the National Academy of Medicine, where she served in numerous leadership roles, the American Society of Human Genetics, an affiliate member of the American College of Genetics and Genomics, as well as the American Association for the Advancement of Science. She was recently elected as Fellow of the American College of Medical Informatics.

Sonia S. Hassan, MD, MBA (she/her)

Sonia S. Hassan, MD, MBA is Associate Vice-President at Wayne State University (WSU), a Professor of Obstetrics and Gynecology and Maternal-Fetal Medicine, and the Founding Director of the Office of Women's Health at WSU. She leads the SOS Maternity Network in Michigan, a statewide implementation science program to reduce preeclampsia and preterm birth, the leading causes of maternal and infant mortality. The SOS Maternity Network is composed of 14 leading universities, hospital systems, and private practices that care for over 50,000 deliveries annually. Dr. Hassan has authored or co-authored over 400 publications and several patents and was the lead author of a landmark international trial, which demonstrated an intervention that decreases preterm birth by 45%. Dr. Hassan was an NIH-funded researcher for over 17 years and previously served as the Project Site Manager for the $165 million contract for the Perinatology Research Branch of the National Institute of Child Health and Human Development of the National Institutes of Health, on behalf of WSU. Dr. Hassan is Co-Chair of Michigan's Mother Infant Health and Equity Collaborative. Dr. Hassan has received many honors, including the Michiganian of the Year Award by the Detroit News, the March of Dimes Michigan Prematurity Prevention Hero Award, and the Service Award & Samaritas Ambassador Award for her work as the First Lady of the City of Detroit and Co-Chair of the Detroit Refugee Network in partnership with Samaritas. She is an ambassador of the Smithsonian Women's History Museum's Michigan Women Making History Committee. Dr. Hassan serves on the Boards of the International Women's Forum, Detroit Institute of Arts, and United Way of Southeastern Michigan. She completed her Bachelor of Arts at the University of Michigan and Medical Degree at Wayne State University. Dr. Hassan completed her MBA at the University of Michigan's Ross School of Business. Dr. Hassan is a Fellow of the third class of the Aspen Institute's Health Innovators Fellowship and a member of the Aspen Global Leadership Network.

Susan Tanksley, PhD (she/her)

Susan M. Tanksley, PhD is the Deputy Associate Commissioner and Deputy Laboratory Director for the Public Health Laboratory Division at the Texas Department of State Health Services in Austin. Texas. Dr. Tanksley oversees public health laboratory informatics, grants, legislative affairs and special projects. She assists the Laboratory Director in managing 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 public health career started in newborn screening in 2001. Since then, her focus has been on newborn screening program expansion, quality improvement, retention and recruitment of staff, and improving communication and transparency. She chaired the APHL Newborn Screening and Genetics in Public Health Committee from 2011-2017, co-chaired the Newborn Screening Workgroup for the Mountain States Genetics Regional Collaborative Center from 2009-2015, has served on the Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC) as an organizational representative for APHL since 2013 and has served as a member of the Evidence Review Workgroup for ACHDNC 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,

Heidi Wallis (she/her)

Heidi Wallis is the Executive Director of the Association for Creatine Deficiencies and the parent of two children with GAMT Deficiency, a rare creatine synthesis disorder. Prior to her current role, Heidi worked as a grant analyst and project manager in the Utah Public Health Lab Newborn Screening Informatics program. Heidi is a member of the Utah Newborn Screening Advisory Committee, the ClinGen CCDS Variant Curation Expert Panel, and the ClinGen DAPC Working Group. Heidi and collaborators nominated GAMT deficiency for inclusion on the RUSP three times. After nearly 7 years, GAMT was added in January 2023.

Session 2 Speakers

Stephen Kingsmore, MD, DSc (he/him)

Stephen Kingsmore, MD, DSc is President/CEO of Rady Children's Institute for Genomic Medicine (RCIGM) which is implementing pediatric genomic medicine across North America. The major applications are rapid diagnostic genome sequencing of hospitalized children, Genome-to-Treatment electronic management guidance for newly diagnosed children, and BeginNGS, artificial intelligence-based genome screening of healthy infants to detect and treat rare genetic diseases at or before symptom onset. He is also the David F. Hale Chair at Rady Children's Hospital, San Diego. Previously he was the Dee Lyons Chair in Genomic Medicine at the University of Missouri-Kansas City, President of the National Center for Genome

Resources, Chief Operating Officer of Molecular Staging Inc., Vice President of Research at CuraGen Corporation, and Assistant Professor at the University of Florida. Dr. Kingsmore received BSc, MB, ChB, BAO, and DSc degrees from the Queen's University of Belfast. He did residency in internal medicine and fellowship at Duke University Medical Center. He is a fellow of the Royal College of Pathologists. He was a MedScape Physician of the year in 2012, received the 2013 Scripps Genomic Medicine award, 2013 ILCHUN prize of the Korean Society for Biochemistry and Molecular Biology, and 2022 Precision Medicine World Conference Luminary Award. TIME magazine ranked his rapid genome diagnosis one of the top 10 medical breakthroughs of 2012. RCIGM has held two Guinness World Records for fastest time to genome-based diagnosis of genetic diseases.

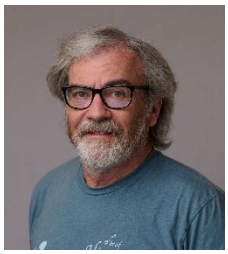
Kee Chan, PhD (she/her)

Kee Chan, PhD is the Founder of ValueMinded LLC, a boutique public health and population health consulting firm interested in addressing maternal and child health issues and newborn screening. Her PhD dissertation research focused on developing a screening test to detect T-cell receptor excision circles (TRECs) in newborns, in which the absence of TRECs indicates severe combined immunodeficiency (SCID). As of 2024, all the states in the U.S. have implemented newborn screening for SCID to screen for over 3 million babies, and many international countries have adopted newborn screening for SCID. Dr. Chan has over 15 years of experience in newborn screening, strategic management, and cost-effectiveness analysis. She has taught in academia, researched at the National Institutes of Health (NIH), and worked as a scientific strategy manager for the American College of Medical Genetics and Genomics, a non-profit organization. From 2020 to 2024, she led the workgroup on Researcher Needs for the Newborn Screening Translational Research Network (NBSTRN), organized their virtual summits, and hosted the Newborn Screening SPOTlight Podcast. She also has years of experience in Higher Education Administration management and leadership as a Program Director and Certified Instructional Designer.

Denise M. Kay, PhD (she/her)

Denise M. Kay, PhD has worked as a Research Scientist with the New York State Newborn Screening Program at the Wadsworth Center, New York State Department of Health since 2009. Her background is in genetics and genetic epidemiology. Her work is focused on addressing challenges in newborn screening, including improving current methods and algorithms, and developing and implementing assays including expanded genetic analysis for existing and 'new' conditions. Dr. Kay was responsible for onboarding full gene sequence analysis for cystic fibrosis screening, resulting in an 80% reduction in referrals. She is active in the national newborn screening community working with the Centers for Disease Control and Prevention, the Eunice Kennedy Shriver National Institute for Child Health and Human Development and the Association of Public Health Laboratories (APHL). She is Co-Chair of the Molecular Subcommittee for APHL. She has managed several pilot studies in New York State, including implementing screening for Spinal Muscular Atrophy (SMA) and the GUARDIAN study.

Session 3 Speakers



Michael H. Gelb, PhD (he/him)

Michael H. Gelb, PhD is the Boris and Barbara L. Weinstein Endowed Chair in Chemistry in the Department of Chemistry and Department of Biochemistry at the University of Washington. He holds a Ph.D. from Yale University, and was an American Cancer Society Postdoctoral Fellow at Brandeis University with Professor Robert H. Abeles. In 1985, he joined the faculty of chemistry at the University of Washington. The Gelb research laboratory combines chemical and molecular and cellular biochemistry techniques to study enzymatic processes of medical importance. Major achievements include: 1) discovery of protein prenylation; 2) development of Isotope-Coded Affinity Tags (ICAT) for quantitative proteomics; 3) development of assays for newborn screening of metabolic diseases; 4) development of drugs to treat tropical parasite diseases; 5) discovery of phospholipases A2 and understanding their mode of action. In the area of newborn screening, assays for approximately half of the diseases added to the Recommended Newborn Screening Panel (RUSP) over the past decade have been developed in the Gelb laboratory. He has received a number of awards including the Repligen Award and the Pfizer Award in Biological Chemistry (American Chemical Society), the ICI Pharmaceuticals Award for Excellence in Chemistry (AstraZeneca, Inc.), the Gustavus John Esselen Award (Harvard University), Merit Award (National Institutes of Health), Medicines for Malaria Venture Project of the Year (MMV, Geneva), Guthrie Award (International Society of Neonatal Screening), and the University of Washington Faculty Lecture Award. He is a Fellow of the American Association for the Advancement of Sciences and a Fellow of the Alfred P. Sloan Foundation.

Curt Scharfe, MD, PhD (he/him)

Curt Scharfe, MD, PhD, an Associate Professor of Genetics at Yale School of Medicine, holds a medical degree from the University of Frankfurt and a PhD in biomedical sciences from the University of Wurzburg. His residency in pediatrics and medical genetics was completed at the University of Munich Children's Hospital, followed by a fellowship in clinical molecular genetics at Stanford University, where he obtained board certification from the American Board of Medical Genetics and Genomics (ABMGG) in 2017. As a DFG postdoctoral fellow at Stanford University with Ronald Davis, Dr. Scharfe pioneered technology combining functional genomics, transcriptomics, and mass-spectrometry proteomics to uncover hundreds of mitochondrial protein genes. This breakthrough facilitated the identification of rare DNA variants in patients with mitochondrial disorders. Currently, Dr. Scharfe serves as a CLIA Section Director at the Yale Center for Genome Analysis (YCGA), specializing in DNA diagnostic testing for a wide range of genetic diseases. His primary research focus lies in neonatal and maternal health, with a particular emphasis on enhancing molecular diagnostic tools for screenable disorders. Collaborating with the California newborn screening (NBS) program, he led efforts in the development of next-generation sequencing assays tailored for DNA testing from newborn dried blood spots, thereby improving screening accuracy for cystic fibrosis and inborn metabolic disorders. Dr. Scharfe has employed long-read sequencing technology to determine the haplotype phase of DNA variants from dried blood spot specimens, eliminating the need for additional parental testing. Furthermore, his laboratory has pioneered data

mining applications utilizing artificial intelligence and machine learning techniques for tandem-mass spectrometry data analysis. This innovative approach is aimed at minimizing false positive results and enhancing the precision of metabolic disease screening, ultimately reducing diagnostic delays in newborn screening programs.

Michelle Takemoto, MS, CGC (she/her)

Michelle Takemoto, MS, CGC is the founder of the Alliance for Genomic Justice (AGJ), which stives to advance health equity and workforce diversity in genetic and genomic healthcare. These goals are accomplished through community building and mentoring relationships among a diverse network of practicing professionals, trainees, and prospective students in clinical genomics. Members of the network recruit diverse candidates into the field and support one another along the career pipeline. Michelle began her career in 2015 with the Hawaii Department of Health Genetics Program and the Western States Regional Genetics Network (WSRGN). It was there that she established the Minority Genetic Professionals Network (MGPN) in 2018, laying the groundwork for her current work with AGJ. Future projects of the AGJ network will be to a) systematize outreach efforts to prospective students and b) provide education to marginalized community groups about family health history and ways to access to genomic healthcare services.

Session 4 Speakers

Marci Sontag, PhD, MS (she/her)

Marci Sontag, PhD, MS is the Executive Director of the Center for Public Health Innovation. She has a Ph.D. in Epidemiology and an M.S. in Biometrics from the University of Colorado Health Sciences Center. Dr. Sontag has studied pediatric clinical outcomes and newborn screening systems since 1995, with a particular focus on cystic fibrosis newborn screening and long-term follow-up of newborn screen disorders. Her passion is to help improve the public health field by applying innovative solutions to common problems and partnering with public health stakeholders at the local, regional, and national levels.

Ines Maria Vigil, MD, MPH, MBA (she/her)

Passionate about creating a more effective and sustainable healthcare system, Ines Vigil, MD, MPH, MBA is dedicated to empowering healthcare professionals and consumers with data and insights that facilitate informed decisions and improved outcomes. Her commitment to advancing healthcare technology, population health, and data science with a health equity mindset further exemplifies her dedication to shaping a more progressive and inclusive healthcare landscape. As a physician of the American College of Preventative Medicine, co-author of the first of its kind textbook, "Population Health Analytics," published in 2021, and contributor to published works like "Both/And: Medicine & Public Health Together", Dr. Vigil has significantly contributed to shaping the landscape of healthcare. With over 18 years of leadership

experience, Dr. Vigil has held pivotal roles across healthcare sectors. She successfully led the application of Clarify Health's technology and data-driven solutions, improving health outcomes and reducing cost across payer, provider, and life sciences customers in her role as Senior Vice President of Transformation. In the role of Senior Vice President and General Manager, Provider, she made their provider business vertical their most profitable to-date. As Vice President of Advanced Analytics at Priority Health, Dr. Vigil led provider organizations into successful full-risk VBC arrangements within an integrated delivery system. Additionally, as Vice President of Patient- Centered Medical Home (PCMH) at CareFirst BlueCross BlueShield, she spearheaded primary care practice and workflow transformation. Educationally, Dr. Vigil holds a Master of Business Administration (MBA) from the Carey Business School at Johns Hopkins University, a Master of Public Health (MPH) from the Johns Hopkins Bloomberg School of Public Health, a Doctor of Medicine (MD) from the University of New Mexico School of Medicine, and a Bachelor of Arts (BA) from Sarah Lawrence College.

Julia Skapik, MD, MPH, FAMIA (she/her)

Julia Skapik, MD, MPH, FAMIA is the Chief Medical Information Officer at the National Association of Community Health Centers (NACHC) and a board-certified Internist and Clinical Informaticist. She came to NACHC after serving as the Chief Health Information Officer for Cognitive Medical Systems and after serving as a Senior Medical Informatics Officer at the Office of the National Coordinator for Health Information Technology, leading quality measure harmonization, interoperability standards and EHR certification programs. Dr. Skapik is also an ongoing leader in health information technology (HIT) interoperability, governance and clinical content as the Board Chair of the HL7 International Board of Directors. In her role at NACHC, Dr. Skapik is focused on broad HIT stakeholder coordination and engagement, common data definitions and measure harmonization, and HIT-enabled clinical quality improvement, care coordination, emerging health center technologies and patient engagement.

Session 5 Speakers

Dominic Smith, MSA (she/her)

Dominic Smith, MSA**,** attended both the University of Tennessee in Knoxville and Central Michigan University. Prior to joining the Michigan Department of Health and Human Services (MDHHS) she worked as a Crime Victim Advocate, and later as Community Health Educator and Deputy Public Information Officer. She joined MDHHS in 2011 as a Project Manager/Health Educator in the Division of Environmental Health leading implementation of the Climate & Health Adaptation Program. She later transitioned to the Lifecourse Epidemiology and Genomics Division becoming the Hemoglobinopathy Program Coordinator and subsequently landed her current position as the Public Health Genomics Section Manager. In this role, she oversees the administration of multiple programs, including the Michigan BioTrust for Health program.

Eric Hendricks, JD (he/him)

Eric Hendricks, JD is a public health attorney and manager of the Public Health and Legal Services section at the Michigan Department of Health and Human Services. He has served as a public health attorney for five years, prior to which he was a law clerk for a U.S. Magistrate Judge. He supports a full range of public health programs, including communicable disease, chronic disease, and environmental health. He is a graduate of the University of Michigan Law School and has lectured on public health law at the U-M School of Public Health.

Jerry Menikoff, MD, JD, MPP (he/him)

Before this year joining the faculty of the National University of Singapore Centre for Biomedical Ethics, Jerry Menikoff, MD, JD, MPP was for 14 years the director of the Office of Human Research Protections, the main U.S. regulatory agency protecting research subjects in federally funded research. He oversaw the multi-year process of successfully revising the "Common Rule," with many of the changes reflecting proposals he promoted for years (in particular, improving consent forms, making consent forms public, and requiring a single IRB for review of multi-centered research). Prior to this, he led the intramural research protections program at the National Institutes of Health. And according to ChatGPT: "The man knows his stuff. He's like the Gandalf of healthcare, waving his ethical staff and saying, 'Thou shall not pass...without informed consent!'"

Biosketches of Committee Members

Jewel Mullen, MD, MPA, MPH (she/her)

Committee Chair

Jewel Mullen, MD, MPA, MPH is Associate Dean for Health Equity and Associate Professor of Population Health and Internal Medicine at the University of Texas at Austin Dell Medical School where she leads strategies to embed health equity as an operating principle across research, education, and practice. Her previous roles in state government and as the Principal Deputy Assistant Secretary for Health at the Department of Health and Human Services focused on assuring equitable access to medical and public health services for all people. During her tenure as Commissioner of the Connecticut Department of Public Health, her department built and opened a new public health laboratory and deliberated adding new conditions to its newborn screening panel. Dr. Mullen also collaborated with the Association of Public Health Laboratories to increase overall state lab efficiency. In addition to serving on NASEM's Board on Population Health and Public Health Practice, she is a current member of the Morbidity and Mortality Weekly Report Editorial Board and the National Vaccine Advisory Committee. Her two most recent NASEM study committees addressed A Fairer and More Equitable Cost-Effective and Transparent System of Donor Organ Procurement, Allocation, and Distribution, and Equitable Allocation of Vaccine for the Novel Coronavirus. Dr. Mullen received her bachelor's and Master of Public Health degrees from Yale University where she also completed a post-doctoral fellowship in psychosocial epidemiology, her MD from the Mount Sinai School of Medicine, a Master in Public Administration from Harvard's Kennedy School of Government, and a certificate in bioethics from Georgetown's Kennedy Institute of Ethics.

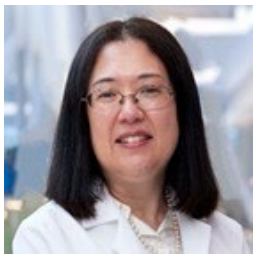
Don Bailey, PhD, MEd (he/him)

Don Bailey, PhD, MEd is a Distinguished Fellow in the Genomics and Translational Research Center at RTI International. Before joining RTI in 2006, he was on the faculty of the University of North Carolina at Chapel Hill (UNC-CH) for 27 years, where he was a W. R. Kenan, Jr. Distinguished Professor and, for 14 years, Director of the Frank Porter Graham Child Development Institute. Dr. Bailey's research addresses early identification and early intervention for children with disabilities, as well as family adaptation to disability. He has an extensive record of publications, with more than 250 peer-reviewed articles, chapters, and books on topics related to newborn screening (NBS), early intervention, disability, and family support. Currently, his work focuses on the future of NBS, family outcomes of NBS, and policy considerations when determining net benefit of screening. He is a Senior Advisor for the Early Check program, a research study offering free additional screening tests to newborns in North Carolina. Early Check has multiple sources of support, including direct funding from the Leona M. and Henry B. Helmsley Charitable Trust, JDRF International (through a collaboration with Janssen Pharmaceuticals), Travere Therapeutics, and Orchard Therapeutics. Early Check also receives donated sequencing services from GeneDx and in-kind contributions from Illumina. Dr. Bailey's work has previously been funded by companies including Asuragen, Janssen Pharmaceuticals, Orchard Therapeutics, Sarepta Therapeutics, Illumina, and Shionogi. Additionally, he has received travel support to present research findings from Travere Pharmaceuticals,

PerkinElmer, and Janssen Pharmaceuticals. Dr. Bailey has presented research findings to organizations including the Association of Public Health Laboratories and EveryLife Foundation. From 2011 to 2017, he served as a member of the Department of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children. He holds a BA in Psychology (Davidson College), an MEd in Early Childhood Special Education (UNC-CH), and a PhD in Early Childhood Special Education (University of Washington).

Mei Baker, MD, FACMG (she/her)

Mei Baker, MD, FACMG is a professor in the Department of Pediatrics, and Director of 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 20 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 improve ongoing screening tests. She is one of the leading scientists who made Wisconsin the first state in the nation and the world to implement universal NBS for severe combined immunodeficiency (SCID) in 2008. She has developed and implemented cystic fibrosis NBS using next-generation sequencing technology in the Wisconsin NBS program. She also implemented the NBS for spinal muscular atrophy in Wisconsin with the unique approach of incorporating additional SMN2 copy number assessment. Dr. Baker recently completed a research project supported by Ultragenyx to help develop a screening assay for Angelman Syndrome. Dr. Baker's contribution to science has been widely recognized, as evidenced by receiving the Harry Hannon Laboratory Improvement Award in Newborn Screening from the Association of Public Health Laboratories (APHL) in 2014, and Everyday Life Saver in Newborn Screening Award from the APHL in 2022. Dr. Baker served as a member of the Department of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children from 2016 to 2021. She currently serves on the scientific advisory boards for the Global Foundation for Peroxisomal Disorders and the Foundation for Angelman Syndrome Therapeutics.



Wendy Chung, MD, PhD (she/her)

Wendy Chung, MD, PhD (NAM) is a clinical and molecular geneticist and the Chair of the Department of Pediatrics at Boston Children's Hospital and Harvard Medical School. Dr. Chung leads the GUARDIAN study piloting genome sequencing as a platform for newborn screening, which is funded by private philanthropy, in addition to sequencing services from GeneDx, in-kind contributions from Illumina, and funds for participant recruitment from Sanofi. She has also led previous pilot studies of newborn screening for SMA. Dr. Chung directs NIH-funded research programs in human genetics of pulmonary hypertension, breast cancer, obesity, diabetes, autism, congenital anomalies including congenital diaphragmatic hernia and congenital heart disease. She is a national leader in the ethical, legal, and social implications of

genomics. She was the recipient of the Rare Impact Award from the National Organization of Rare Disorders and is a member of the National Academy of Medicine and the American Academy of Physicians. She serves as a member on the Board of Directors for Prime Medicine, which is developing platform technologies for potential gene therapy applications. Dr. Chung received her B.A. in biochemistry from Cornell University, her M.D. from Cornell University Medical College, and her Ph.D. from The Rockefeller University in genetics.

Titilope Fasipe, MD, PhD (she/her)

Titilope Fasipe, MD, PhD is Co-Director of the Sickle Cell and Thalassemia Program at Texas Children's Hospital and Assistant Professor of Pediatrics in Hematology/Oncology at Baylor College of Medicine in Houston, Texas. She is involved in community and policy efforts aimed at improving health outcomes in sickle cell disease. Further, she has the unique perspective of relating to and understanding the need for education, community awareness, support, and medical care as she is a pediatric hematologist as well as an individual with sickle cell disease. Dr. Fasipe has been repeatedly appointed to advisory committees of the Texas Department of State Health Services and currently serves on their Newborn Screening Advisory Committee and chairs the Sickle Cell Task Force. Her professional memberships include: the American Academy of Pediatrics, the Heartland-Southwest Sickle Cell Disease Network, the American Society of Hematology, the American Society of Pediatric Hematology/Oncology, and the Global Action Network for Sickle Cell and Other Inherited Blood Disorders. Dr. Fasipe has consulted as a sickle cell disease educator for several pharmaceutical and therapeutic companies, including Forma Therapeutics, Novartis, Global Blood Therapeutics, Bluebird Bio, and Emmaus Medical. Dr. Fasipe received her Bachelor of Science from the University of Texas at Arlington and graduated from the combined MD-PhD program at the University of Texas Medical Branch. She then completed her pediatric residency at Cincinnati Children's Hospital Medical Center and her pediatric hematology/oncology fellowship at Baylor College of Medicine and Texas Children's Hospital.

Faith Fletcher, PhD, MA (she/her)

Faith Fletcher, PhD, MA is an Assistant Professor in the Center for Medical Ethics and Health Policy at Baylor College of Medicine. She is also a senior advisor to The Hastings Center and a Hastings Center fellow. Her research program examines the social and structural barriers to scientific research and healthcare engagement facing traditionally marginalized populations and is grounded in methodological and theoretical approaches from the fields of public health, bioethics, and behavioral science. Her K01 Award funded through the National Human Genomic Research Institute uses a stakeholder engagement approach to develop ethical practices and guidelines for engaging Deep South residents in genomics research. Dr. Fletcher was recently named to the Greenwall Faculty Scholars Program in Bioethics Class of 2026. In 2017, Dr. Fletcher was named one of the National Minority Quality Forum's 40 under 40 Leaders in Health for her commitment to advancing health equity. She recently received the BCM Women of Excellence Award for her outstanding contributions and accomplishments in advancing health and healthcare equity as a field leader in alignment with the mission of the college. Dr. Fletcher received her BS

in biology from Tuskegee University with concentrations in bioethics and philosophy, her MA in bioethics, humanities, and society from Michigan State University, and her PhD in health promotion, education and behavior from the University of South Carolina. Dr. Fletcher also completed a National Cancer Institute R25T-funded Postdoctoral Fellowship in the Department of Behavioral Science at The University of Texas MD Anderson Cancer Center.

Meghan Halley, PhD, MPH (she/her)

Meghan Halley, PhD, MPH is a Senior Research Scholar in the Center for Biomedical Ethics at Stanford University. A medical anthropologist by training, her research focuses on ethical and policy issues arising through the introduction of new genomic technologies for diagnosis and treatment of rare diseases. Her current projects include examining ethical issues related to sustainability and governance of patient data and relationships when large clinical genomic studies transition to new models of funding; ethnographic work exploring how diverse stakeholders perceive value in the use of genome sequencing for diagnosis of rare diseases; and the development of new measures for assessing patient-centered outcomes in pediatric rare diseases. In 2021, she was awarded a career development grant from the National Human Genome Research Institute focused on the ethics and economics of genomic sequencing in rare disease. Dr. Halley received her doctorate in anthropology and her Master of Public Health from Case Western Reserve University in Cleveland, OH. The parent of a child with a rare disease, she serves as President of the Board of Directors for the Undiagnosed Diseases Network Foundation, a nonprofit with the mission of improving access to diagnosis, research, and care for individuals with undiagnosed and ultra-rare diseases and has written on the parental experience navigating complex medical decisions.

Amanda Dawn Ingram, RN (she/her)

Amanda Dawn Ingram, RN is the Director of the Pediatric Case Management and Newborn Screening Follow-up Program for the Tennessee Department of Health. Before becoming director, Ms. Ingram served as a Case Manager and the Case Management Coordinator for the program for 1 year each. Ms. Ingram has been a registered nurse for 23 years and worked as a Neonatal Intensive Care Nurse for 13 years before coming to the Department of Health. As a neonatal nurse, Ms. Ingram had experiences with receiving newborn screening results, requests for repeat testing, and collection of newborn screening specimens. She is a member of the Association of Public Health Laboratories (APHL) and serves on three APHL workgroups to collaborate with other programs to continue newborn screening awareness and improvement. She was selected to participate on the Krabbe Review Technical Expert Panel that informs the Department of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children. Ms. Ingram has co-authored and contributed to multiple articles in the newborn screening field and was a contributor to the recently updated Clinical and Laboratory Standards Institute Guidelines for Newborn Screening Follow-up and Education.

José A. Pagán, PhD (he/him)

José A. Pagán, PhD (NAM) is Professor and Chair of the Department of Public Health Policy and Management at the School of Global Public Health, New York University. He is also Chair of the Board of Directors of NYC Health + Hospitals, the largest municipal healthcare system in the United States. He formerly served as Chair of the National Advisory Committee of the Robert Wood Johnson Foundation's Health Policy Research Scholars and was a member of the Board of Directors of the Interdisciplinary Association for Population Health Science and the American Society of Health Economists. He is a member of the National Academy of Medicine. He has led research, implementation, and evaluation projects on the redesign of health care delivery and payment systems. His areas of focus include population health management, health care payment and delivery system reform, and the social determinants of health. Dr. Pagán received his PhD in economics from the University of New Mexico.

Jochen Profit, MD, MPH (he/him)

Jochen Profit, MD, MPH is the Wendy J. Tomlin-Hess Endowed Professor of Pediatrics at Stanford University and the Chief Quality Officer of the California Perinatal Quality Care Collaborative (CPQCC). Dr. Profit's research focuses on optimizing the quality of neonatal-perinatal health care delivery, with an emphasis on enhancing organizational effectiveness. He is also interested in the use of information technology to support families, care professionals, and policy makers in their efforts to provide optimal care to sick infants. For over a decade, his research has received continuous funding from the National Institutes of Health and other federal, foundation, and intramural sources. Dr. Profit has served on various national scientific and professional organizations, including for the National Institutes of Health. Recently, he served as a member on the NASEM consensus study committee on Research Issues in the Assessment of Birth Settings. He graduated from the University of Freiburg Medical School in Germany and completed his Neonatology and Health Services Research training at Harvard.

Scott Shone, PhD, HCLD (ABB)

Scott Shone, PhD, HCLD (ABB) is the Director of the North Carolina State Laboratory of Public Health. He received his doctorate in Molecular Microbiology and Immunology from The John Hopkins Bloomberg School of Public Health and is a board-certified high-complexity clinical laboratory director. Dr. Shone spent 9 years managing the New Jersey Newborn Screening (NBS) Laboratory. As a senior research public health analyst at RTI International, he focused on NBS pilot studies such as Early Check as the Clinical Laboratory Improvement Amendments (CLIA) Director. Currently, he leads a team of over 230 staff for the delivery of clinical and environmental laboratory services in North Carolina. Dr. Shone received the Jean Dussault Medal for Young Investigators from the International Society for Neonatal Screening and, recently, the Governor's Award for Excellence from the State of North Carolina. Dr. Shone served

as a voting member on the Department of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children from 2017-2022. Currently, he is the Association of State and Territorial Health Officials (ASTHO) organizational representative to this federal advisory committee. He has consulted for the Association of Public Health Laboratories (APHL) to develop contingency plans for NBS programs during states of emergency. He is the APHL organizational representative to the NASEM Forum on Medical and Public Health Preparedness for Disasters and Emergencies, a member of the editorial board for the International Journal of Neonatal Screening, a member of the APHL NBS Committee, and an elected member of the APHL Board of Directors.

Kayte Spector-Bagdady, JD, MBe (she/her)

Kayte Spector-Bagdady, JD, MBe is a health law and bioethics faculty member at the University of Michigan Medical School and interim co-director of the U-M Center for Bioethics and Social Sciences in Medicine, which won the 2022 American Society for Bioethics & Humanities Cornerstone Award. Her research goal is to improve the governance of research with health data and specimens to increase equitable access to scientific advances. She is the PI of two NIH grants studying how and why geneticists select datasets for their research and on hospitals sharing patient data with commercial entities and has been Co-I on an additional $140M in funding. She was also Chair and lead author of the American Heart Association's "Principles for Health Information Collecting, Sharing, and Use." Professor Spector was an Associate Director for President Obama's Presidential Commission for the Study of Bioethical Issues (2010-15) and is a former practicing drug and device attorney. She received her JD and MBe from the University of Pennsylvania.

Beth Tarini, MD, MS, MBA (she/her)

Beth Tarini, MD, MS, MBA is the Associate Director of the Center for Translational Research at Children's National Hospital. She previously served as the Division Director of General Pediatrics at the University of Iowa. She is a formally trained health services researcher (graduate of the Robert Wood Johnson Clinical Scholars Program) who conducts health services research that focuses on optimizing the delivery of genetic services to children and their families, particularly through newborn screening. Her research has been funded by the National Institutes of Health, HRSA, the Robert Wood Johnson Foundation and the Cystic Fibrosis Foundation. Dr. Tarini is a former President of the Society for Pediatric Research, a fellow of the American Academy of Pediatrics, and a member of the Genomics & Society Working Group of the National Human Genome Research Institute. She has been appointed to and led federal, national, and state committees that provide policy recommendations for genetics services and newborn screening, including the Department of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children. Dr. Tarini received her MD from Albert Einstein College of Medicine and her BA from Harvard University. She has an MS in Health Services from University of Washington and an MBA from George Washington University.

Krystal Tsosie (Diné, Navajo Nation), PhD, MPH, MA

Krystal Tsosie (Diné/Navajo Nation), PhD, MPH, MA is an Assistant Professor at Arizona State University in the School of Life Sciences. She co-founded the first US Indigenous-led biobank, a 501c3 nonprofit research institution called the Native BioData Consortium. Dr. Tsosie's research centers on ethical engagement with Indigenous communities in precision health and genomic medicine. Her areas include genetic epidemiology, bioethics, public health, and community research approaches. She previously patented a combined targeted ultrasound imaging and chemotherapeutic drug delivery device for treating early metastases in cancer. Dr. Tsosie is currently on the Board of Directors for the American Society of Human Genetics, and on the ethics committee of the American Society for Cell and Gene Therapies. Her background includes a Master of Arts in Bioethics for studying genetic controversies in Indigenous communities, a Master of Public Health in Genetic Epidemiology for studying gene variation related to hypertension and uterine fibroids, and a PhD in Genomics and Health Disparities. She recently served on the NASEM consensus study committee, Creating a Framework for Emerging Science, Technology, and Innovation in Health and Medicine and on the NASEM planning committee, Engaging Scientists in Central Asia on Data Governance Principles for Life Science Data. Dr. Tsosie accepted a one-time speaker honorarium from Regeneron for serving as a guest speaker at the DRIFT Symposium in 2022 to inform the company how to improve their interactions with Indigenous communities. For year 2024, Dr. Tsosie serves as a paid consultant and member of the Clinical Advisory Board for Cache DNA, a company that developed technology that encapsulates and stores nucleic acids without refrigeration to make DNA and RNA isolation more equitable.

Article I. Preventing Discrimination, Harassment, and Bullying Expectations for Participants in NASEM Activities

The National Academies of Sciences, Engineering, and Medicine (NASEM) are committed to the principles of diversity, integrity, civility, and respect in all of our activities. We look to you to be a partner in this commitment by helping us to maintain a professional and cordial environment. All forms of discrimination, harassment, and bullying are prohibited in any NASEM activity. This commitment applies to all participants in all settings and locations in which NASEM work and activities are conducted, including committee meetings, workshops, conferences, and other work and social functions where employees, volunteers, sponsors, vendors, or guests are present.

Discrimination is prejudicial treatment of individuals or groups of people based on their race, ethnicity, color, national origin, sex, sexual orientation, gender identity, age, religion, disability, veteran status, or any other characteristic protected by applicable laws.

Sexual harassment is unwelcome sexual advances, requests for sexual favors, and other verbal or physical conduct of a sexual nature that creates an intimidating, hostile, or offensive environment.

Other types of harassment include any verbal or physical conduct directed at individuals or groups of people because of their race, ethnicity, color, national origin, sex, sexual orientation, gender identity, age, religion, disability, veteran status, or any other characteristic protected by applicable laws, that creates an intimidating, hostile, or offensive environment.

Bullying is unwelcome, aggressive behavior involving the use of influence, threat, intimidation, or coercion to dominate others in the professional environment.

Section 1.01 REPORTING AND RESOLUTION

Any violation of this policy should be reported. If you experience or witness discrimination, harassment, or bullying, you are encouraged to make your unease or disapproval known to the individual, if you are comfortable doing so. You are also urged to report any incident by:

- Filing a complaint with the Office of Human Resources at 202-334-3400, or
- Reporting the incident to an employee involved in the activity in which the member or volunteer is participating, who will then file a complaint with the Office of Human Resources.

Complaints should be filed as soon as possible after an incident. To ensure the prompt and thorough investigation of the complaint, the complainant should provide as much information as is possible, such as names, dates, locations, and steps taken. The Office of Human Resources will investigate the alleged violation in consultation with the Office of the General Counsel.

If an investigation results in a finding that an individual has committed a violation, NASEM will take the actions necessary to protect those involved in its activities from any future discrimination, harassment, or bullying, including in appropriate circumstances the removal of an individual from current NASEM activities and a ban on participation in future activities.

Section 1.02 CONFIDENTIALITY

Information contained in a complaint is kept confidential, and information is revealed only on a need-to-know basis. NASEM will not retaliate or tolerate retaliation against anyone who makes a good faith report of discrimination, harassment, or bullying.

Article II. Diversity, Equity, and Inclusion Statement and Guiding Principles

We, the National Academies of Sciences, Engineering, and Medicine (the National Academies), value diversity among our staff, members, volunteers, partners, vendors, and audiences. We recognize that talent is broadly distributed in society and that many perspectives enhance the quality of our work and drive innovation and impact.

We pledge to cultivate a workplace culture and climate that promotes inclusion, belonging, accessibility, and anti-racism; upholds equity; and values the participation of all who are engaged in advancing our mission.[1] By embracing the values of diversity, equity, and inclusion in our programs, institutional policies and practices, and products, we will be able to better advise the nation on the most complex issues facing society and the world.

Guiding Principles:

The following diversity, equity, and inclusion principles guide our work at the National Academies:

1. Integrate diverse perspectives and experiences into our programs, institutional policies and practices, and products.
2. Foster a culture of inclusion where all staff, members, and volunteers have full access to participation and feel welcomed, respected, valued, and a sense of belonging.
3. Approach scientific endeavors with a consideration of diversity, equity, and inclusion frameworks.
4. Cultivate mutually beneficial diverse partnerships and collaborations with a variety of communities, including, but not limited to, marginalized and underrepresented communities.

Our institutional strategy for putting these values and principles into practice are outlined in the National Academies DEI Action Plan, a comprehensive five-year plan that charts a path toward achieving our diversity, equity, and inclusion goals. The DEI Action Plan is one of many ways that we commit to systems of accountability and transparency to uphold these principles and allow for continuous learning and improvement.

[1] The National Academies' mission is to provide independent, trustworthy advice and facilitate solutions to complex challenges by mobilizing expertise, practice, and knowledge in science, engineering, and medicine.

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