Newborn Screening: Current Landscape and Future Directions – Meeting 2

March 26th, 2024 | 9:15am–5:00pm ET

A Virtual 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.

Agenda

Virtual Meeting

TUESDAY, MARCH 26TH, 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 near-term and fundamental needs and challenges reflected in bullets 1 and 4 of the study's statement of task. Future information gathering sessions will continue to explore other issues to be addressed in the study.

Meeting Objectives

- Explore the mission and goals of the NBS system, and what implications these have for future program evolution.
- Discuss the information assessed and processes involved in evaluating conditions for inclusion on the federal Recommended Uniform Screening Panel (RUSP)
- Understand a range of equity, economic, clinical, ethical, and other considerations involved in adding conditions to NBS programs and discuss these implications for the future of the NBS system.

9:15am **Welcome and Opening Remarks**

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

Session I: Reflecting on the Mission of Newborn Screening Programs

- Explore the mission and goals of the NBS system, and what implications these have for future program evolution.
- Discuss the information assessed and processes involved in evaluating conditions for inclusion on the federal Recommended Uniform Screening Panel (RUSP)
- Understand a range of equity, economic, clinical, ethical, and other considerations involved in adding conditions to NBS programs and discuss these implications for the future of the NBS system.

9:20am **Overview**

Meghan Halley (she/her), Stanford University, *Committee Member, Session Moderator*

9:25am **History of and Ethical Justification for NBS Programs**

Aaron Goldenberg (he/him), Case Western Reserve University School of Medicine

9:40am **Reflections on the Mission of NBS Programs**

Michele Wright (she/her), National Organization of African Americans with Cystic Fibrosis (NOAACF)

Terry Wright (he/him), National Organization of African Americans with Cystic Fibrosis (NOAACF)

Stanton Berberich (he/him), State Hygienic Laboratory at the University of Iowa [Retired]

Natalie Ram (she/her), University of Maryland Carey School of Law

10:05am **Moderated Discussion with Speakers and Q&A**

10:25am **BREAK**

Session II: Examining Current Processes for Adding New Conditions

- Provide an overview of the evidence review process for adding conditions to the federal Recommended Uniform Screening Panel (RUSP).
- Examine the strength of evidence and different types of evidence required federally and across different states and territories.
- Reflect on how processes for adding conditions to NBS programs vary state to state.
- Discuss barriers (e.g. limited resources, workforce, cost) to adding new conditions.

10:40am **Session Overview**

Mei Baker (she/her), Wisconsin State Laboratory of Hygiene; *Committee Member; Session Moderator*

10:45am **Overview of the Current Evidence Review Process for the RUSP**

Alex Kemper (he/him), Nationwide Children's Hospital

11:00am **Perspectives on the Process of Adding New Conditions**

Niki Armstrong (she/her), Foundation for Angelman Syndrome Therapeutics

Barb Ballard (she/her), SCID Angels for Life Foundation

M. Christine Dorley (she/her), Tennessee Department of Health

John Thompson (he/him), Washington State Department of Health

11:35am **Moderated Discussion with Speakers and Q&A**

12:00pm **LUNCH**

Session III: Balancing Benefits and Harms When Considering Adding New Conditions

- Discuss additional factors that could be included in the evidence review process when considering adding new conditions to screening programs.
- Discuss potential benefits (e.g., reducing a diagnostic odyssey) and harms (e.g., expanding those who undergo a prognostic odyssey) to individuals and their families from expanding or limiting newborn screening.
- Discuss potential tradeoffs to expanding the list of screened conditions including cost, workforce, resources, and follow-up considerations for neglected conditions.

1:00pm **Session Overview**

José Pagán (he/him), New York University; *Committee Member; Session Moderator*

1:05pm **Potential Benefits and Harms of Screening Newborns for Complex Conditions**

Melissa Wasserstein (she/her), Albert Einstein College of Medicine

1:15pm **Health Economics Considerations for Newborn Screening**

Scott Grosse (he/him), Centers for Disease Control and Prevention

1:25pm **Potential Tradeoffs When Considering Adding New Conditions**

Lainie Ross (she/her), University of Rochester

1:35pm **Moderated Discussion with Speakers and Q&A**

1:55pm **BREAK**

Session IV: Centering Equity in Newborn Screening Programs

- Highlight important equity and engagement considerations.
- Discuss inequities across states in terms of what is screened, timeliness of screening, and resource/workforce availability.
- Discuss inequities in the follow-up care after newborn screening that disproportionately affect those from underserved populations.

2:05pm **Session Overview**

Amanda Ingram (she/her), Tennessee Department of Health; *Committee Member; Session Moderator*

2:10pm **Reflections from Chan Zuckerberg Initiative: Perspectives on the Study Task and the Rare As One Project**

Anne Claiborne (she/her) and Jeanie Kim (she/her), Chan Zuckerberg Initiative

2:20pm **Moderated Discussion with Speaker and Q&A**

2:35pm **Health Disparities Related to Unequal Social Safety Nets**

Heeju Sohn (she/her), Emory University

2:45pm **Considerations for Newborn Screening Among Alaska Native and American Indian Peoples**

Julie Beans (she/her), Southcentral Foundation

2:55pm **Characteristics of Newborn Screening Programs in the U.S.**

Jelili Ojodu (he/him), Association of Public Health Laboratories

3:05pm **Inequities in Follow-Up Care: A Family Physician's Perspective**

Deanna Wathington (she/her), American Public Health Association and REACHUP

3:15pm **Moderated Discussion with Speakers and Q&A**

3:35pm **BREAK**

Session V: Envisioning the Future for Adding New Conditions to Newborn Screening

- Discuss proposals/models for NBS programs moving forward.
- Discuss considerations for scaling review processes to efficiently handle more conditions.
- Reflect how changing the evidence review process/requirements may affect the ethical justification for NBS programs.
- Discuss solutions for sustainability for NBS programs when considering adding new conditions.

3:45pm **Session Overview**

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

3:50pm **Implementation of Consent Models for Newborn Screening: Historical Insights and Future Considerations**

Amy Gaviglio (she/her), Connetics Consulting

4:00pm **Considerations for Newborn Screening Programs Moving Forward**

Michael Watson (he/him), Washington University School of Medicine

11:15am **Moderated Discussion with Speakers and Q&A**

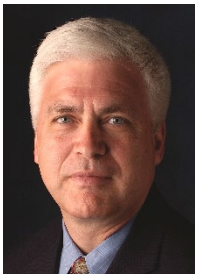
4:55pm **Closing Remarks**

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

4:15pm **ADJOURN**

Biosketches of Speakers

Session 1 Speakers



Stanton Berberich, PhD

Stanton Berberich, PhD served as Program Manager Medical Screening at the State Hygienic Laboratory at The University of Iowa (Iowa's Environmental and Public Health Laboratory) for almost 30 years before retiring in 2023. His work included overseeing the Newborn Screening (NBS) operations, which provided laboratory support not only for the Iowa NBS Program but for the North Dakota, South Dakota, and Alaska NBS Programs as well. Dr. Berberich has served on multiple NBS national committees and workgroups over 3 decades and continues to promote and enable the mission of NBS.

Aaron Goldenberg, PhD

Aaron Goldenberg is a Professor and Vice Chair in the Department of Bioethics at Case Western Reserve University. He is also Director for the Case Western Bioethics Center for Community Health and Genomic Equity. Dr. Goldenberg has a background in bioethics, health behavior/health education, public health ethics, and public health genetics. He has focused his work on the ethical, social, and equity issues associated with newborn screening and the integration of new genomic technologies into research, clinical and public health settings. He is currently Co-Chair for the Newborn Screening Translational Research Network's Bioethics and Legal Workgroup and Co-Chair for the Ethical, Legal, Social and Policy Implications of Newborn Screening Subcommittee for the Association of Public Health Laboratories

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Natalie Ram, JD

Natalie Ram is professor of law at the University of Maryland Francis King Carey School of Law and adjunct faculty with the Berman Institute of Bioethics at Johns Hopkins University. Professor Ram is a leading scholar on the intersection of genetic privacy and the criminal legal system, including work examining whether and how law enforcement access to newborn screening resources is regulated across the United States. Professor Ram was a 2021 Greenwall Faculty Scholar in Bioethics. She is an elected member of the American Law Institute. Professor Ram previously clerked for Associate Justice Stephen G. Breyer, U.S. Supreme Court, and for Judge Guido Calabresi, U.S. Court of Appeals for the Second Circuit. She earned her JD at Yale Law School and AB in public and international affairs at Princeton University.

Michele Wright, PhD

Michele Wright, PhD is the Co-Founder of the National Organization of African Americans with Cystic Fibrosis (NOAACF). She concurrently serves as the Director of Business Development and Outreach for the Society of Motion Picture and Television Engineers. Throughout her career as an innovator, educator, entrepreneur, creative, senior executive, nonprofit leader, mentor, and advocate, Dr. Wright has earned accolades for her profound commitment to highlighting and driving diversity, equity, and inclusion (DEI). Leveraging advanced degrees in both engineering and public policy, Wright had led an array of initiatives, campaigns, and organizations focused on improving visibility, access, and opportunity for underrepresented communities, with a particular emphasis on health equity. As the board chair and senior executive director of NOAACF, Dr. Wright has championed a diverse range of health equity initiatives, including the annual Blacks, Indigenous, and Other Minority Ethnicities with Rare and Genetic Diseases (BIOMERGD) Conference and the Wright Cystic Fibrosis Screening Tool. Using her rare voice within the rare disease space, Wright continues to advocate for "Terry Wright's Law," which will require that all known CF-causing gene variants (also called mutations) be used for newborn screening and diagnostic testing for CF. This will markedly improve equity in newborn screening for BIPOC individuals while benefiting all people with CF who may have delayed or missed diagnosis due to the presence of rare CFTR variants. Dr. Wright has received many honors, including the 2022 USA TODAY Woman of the Year for Arkansas and a 2023 AARP Purpose Prize Fellow —in recognition of her global impact and nationally recognized contributions to diversity, inclusion, and health equity. Through all these achievements, Wright always has believed that "the best is yet to come." She takes this philosophy forward as she continues her work to make a positive impact on children, women, and underrepresented populations worldwide.

Terry Wright

Terry Gene Wright is the Co-founder and President of the National Organization of African Americans with Cystic Fibrosis ("NOAACF"), a 501(c)(3) organization with a mission to connect individuals with CF, help build diverse communities, and, through its national platform, raise CF awareness within the African American community and beyond, and the co-founder of the BIOMERGD (Blacks, Indigenous, and Other Minority Ethnicities with Rare and Genetic Diseases) Annual Conference. Terry is a 2022-2023 AARP Purpose Prize Fellow in recognition of his global impact and nationally recognized contributions to diversity, inclusion, and health equity. In 2022, he became the first African American to serve as CF Ambassador for the Cystic Fibrosis Foundation (CFF) Arkansas Chapter. Additionally, he and his wife, NOAACF Co-Founder, Dr. Michele Wright, became the first persons of color to receive the CFF Arkansas Chapter's annual "Breath of Life Award", CFF's Highest Award bestowed. He is the real-life subject and inspiration behind the film, "54 YEARS LATE": The Terry Wright Story. At the age of 54, Terry was unexpectedly diagnosed with cystic fibrosis. He shares his surreal journey to CF Land in his children's coloring storybook, "Terry's Journey to CF Land," which was listed in the top spot on BookAuthority's "8 Best New Cystic Fibrosis Books To Read In 2021". Today, at the age of 61, Terry wholeheartedly utilizes his deep-rooted passion for gardening, nature, agriculture, horticulture, fitness, nutrition, and health to help individuals from all walks of life to achieve the best in health! He is fully committed to utilizing his expertise to help others achieve a better quality of health and life.

Session 2 Speakers

Niki Armstrong, MS, CGC

Niki Armstrong, MS, CGC is the Vice President of Genetic Services and Education at the Foundation for Angelman Syndrome Therapeutics. Niki is a board certified genetic counselor at FAST, providing counseling to families, developing educational tools on genetics and clinical trials, and directing data strategy related to the Global Angelman Syndrome Registry. She is also charged with supporting the development of newborn screening for AS. Niki previously led PPMD's Duchenne newborn screening efforts, including the RUSP nomination package currently under review. Before moving to advocacy, Niki worked as a clinical genetic counselor in oncology at a community hospital and as a pediatric genetic counselor in an academic children's hospital. Niki is a graduate of the University of Minnesota Genetic Counseling Graduate Program.

Barb Ballard

Barb's only child was born with X-linked Severe Combined Immune Deficiency. Despite his numerous medical complications, in 1997 she developed an internet listserv so that families with SCID children could connect and would no longer have to be isolated while coping with a rare disease. Barb served as a member of the Board of Trustees for the Immune Deficiency Foundation for 18 years where she advocated for SCID patients, developing programs aimed to improve their quality of life including the national campaign for the implementation of SCID Newborn Screening. She was a founding board member for SCID Angels For Life. A Virginia native and a graduate of Virginia Tech with a major in Finance, Barb brings not only the patient and parent perspective, but also a financial and business background. Her diversified career includes both private businesses and non-profits, varying from Chief Accounting Officer for a Washington, DC area homebuilder, to Communications Officer for The Royal Society of the Arts in the US, to owning and managing an automotive service business.

M. Christine Dorley, PhD

M. Christine Dorley, PhD, currently serves as the Newborn Screening (NBS) and Environmental Division Director for the Tennessee Department of Health, Division of Laboratory Services. She has been with the Division of Laboratory Services for almost 30 years serving in different capacities. Dr. Dorley became involved with NBS in 2004, temporarily overseeing NBS during a critical period in which screening for inborn errors of metabolism by Tandem Mass Spectrometry was added to the Tennessee panel. Soon, NBS became her passion, so she permanently moved to the section in 2007. She has been instrumental in migrating the NBS laboratory to a seven-day workweek with a significant decrease in turnaround time for reporting time critical and time sensitive results. Under her leadership, the laboratory has quickly adopted and implemented screening tests to match the core disorders on the Recommended Uniform Screening Panel (RUSP) for the United States. She recently became a member of the Advisory Committee on Heritable Disorders in Newborns and Children and also serves as a member of the RTI Expert Advisory Committee. She is a member of the Association of Public Health Laboratories and has chaired two workgroups: Hemoglobinopathies workgroup and the NBS CONPLAN workgroup. She currently serves on other APHL NBS committees including the NewSteps Steering committee, the QA/QC subcommittee, and the New Disorders Subcommittee.

Alex Kemper, MD, MPH, MS

Alex R. Kemper, M.D., M.P.H., M.S., is the Division Chief of Primary Care Pediatrics at Nationwide Children's Hospital and Professor of Pediatrics at the Ohio State University College of Medicine and Deputy Editor of PEDIATRICS. Dr. Kemper completed his pediatric residency training at Duke University followed by combined fellowship training in health services research and medical informatics with residency training in preventive medicine at the University of North Carolina. His research focuses on preventive services delivery. Dr. Kemper is a former member of the United States Preventive Services Task Force and serves as Chair of the Evidence Review Group for the Advisory Committee on Heritable Disorders in Newborns and Children.

John Thompson, PhD, MPA, MPH

John Thompson received a bachelor's degree from Brigham Young University in molecular biology. He then pursued graduate degrees in public administration and public health genetics at the University of Washington, focusing on newborn screening policy. John has worked for the Washington State Newborn Screening Program since 2003, as a follow-up consultant, the short-term follow-up supervisor and currently as the office director. He oversees all operations for bloodspot screening in the regional NBS laboratory screening babies born in Washington, Hawaii, and Idaho. He had a very rewarding experience as cochair for APHL's NewSTEPs Short-Term Follow-Up workgroup and currently serves on the NewSTEPs Steering Committee.

Session 3 Speakers

Scott Grosse, PhD

Scott Grosse, PhD, is senior health economist in the Office of the Director, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention (CDC), US Department of Health and Human Services, Atlanta, Georgia. He joined CDC in 1996 after completing doctoral studies in economics and public health at the University of Michigan. He conducts health services research, economic evaluations, and policy analyses on childhood-onset rare disorders and neurodevelopmental disorders and associated risk factors and policies such as newborn screening. He serves as federal advisor to the Evidence Review Group that is contracted to prepare evidence reviews to inform recommendations of the Advisory Committee on Heritable Disorders in Newborns and Children for the addition of disorders to the federal Recommended Uniform Screening Panel.

Lainie Ross, MD, PhD

Lainie Friedman Ross, MD, PhD, is the Dean's Professor and Inaugural Chair of the Department of Health Humanities and Bioethics, and the Director of the Paul M Schyve, MD Center for Bioethics at the University of Rochester School of Medicine and Dentistry. She also holds secondary appointments in pediatrics and philosophy at the University of Rochester. Dr. Ross is a graduate of Princeton University (AB from the School of Public and International Affairs), The University of Pennsylvania Perelman School of Medicine (MD) and Yale University (MPhil and PhD in Philosophy). She trained in pediatrics at the Children's Hospital of Philadelphia and the Morgan Stanley Children's Hospital of New York-Presbyterian. She is a Hastings Fellow, a John Simon Guggenheim Memorial Foundation Fellow, and a member of the National Academy of Medicine. Dr Ross' research portfolio focuses on ethical and policy issues in pediatrics, organ transplantation, genetics and rare diseases, and translational research ethics. She has published 5 books and over 300 peer-reviewed articles. She has lectured nationally and internationally about ethical and policy issues related to newborn screening. She was the lead author of the American Academy of Pediatrics/American College of Medical Genetics and Genomics policy statement and technical report on the genetic testing of children (2013). She also served on the Illinois Department of Public Health, Genetic and Metabolic Diseases Advisory Committee, (2009-2016), and the National Institutes of Health Recombinant DNA Advisory Committee "the RAC" (2014-2018).

Melissa Wasserstein, MD

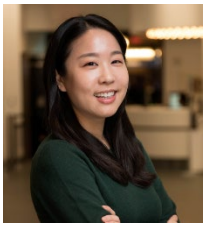
Dr. Melissa Wasserstein is a Professor of Pediatrics and Genetics and a Raizin Distinguished Faculty Scholar in Pediatrics at the Albert Einstein College of Medicine. She is Co-Director of the Rose F. Kennedy Intellectual and Developmental Disability Research Center and the Chief of the Division of Pediatric Genetic Medicine at the Children's Hospital at Montefiore. A board-certified biochemical geneticist and pediatrician, Dr. Wasserstein diagnoses and manages individuals with inborn errors of metabolism. She is an international leader in the diagnosis and management of acid sphingomyelinase deficiency. She directs NIH-funded research activities focused on expanding newborn screening, enhancing its accuracy, and evaluating the practical and ethical implications of expansion through exploration of parental perspectives.

Session 4 Speakers

Chan Zuckerberg Initiative Representatives

Tania Simoncelli, MS is vice president of Science in Society at the Chan Zuckerberg Initiative. In 2019, Simoncelli launched the Rare As One Project, a program that supports patient communities in their quest to accelerate research. Her previous roles include senior advisor to the director and executive director of the Count Me In initiative at the Broad Institute of MIT and Harvard and assistant director for forensic science and biomedical innovation at the White House Office of Science and Technology Policy. She has also worked in the Office of the Commissioner at the Food and Drug Administration and as science advisor to the American Civil Liberties Union. Simoncelli earned a B.A. in Biology and Society from Cornell University and a M.S. in Energy and Resources from the University of California, Berkeley.

Anne Claiborne, MPH, JD is director of Strategic Operations and Policy in the Science in Society program at the Chan Zuckerberg Initiative. In this role she provides leadership for strategic explorations and special projects, including grounding CZI's Science in Society work in equity and understanding the policy landscape in which programs are situated. Claiborne joined CZI from the National Academies of Sciences, Engineering, and Medicine, where she was a Senior Program Officer in the Health and Medicine Division, where her work focused particularly on therapeutics discovery and development, clinical research, and ethics of health science, including emerging biotechnology. She received her JD from Harvard Law School and MPH from the Johns Hopkins Bloomberg School of Public Health.



Jeanie Kim, JD is a Health Science Policy Analyst on the Science in Society team and supports the Rare As One project by helping to better understand the policy landscape and emerging issues around biomedical research, technology, and innovation. Prior to joining CZI, she worked at the Open Society Foundations, where she focused on promoting health equity through legal tools. Earlier in her career, she was a Research Fellow at Yale Law School, where she worked on a collaborative project at the intersection of law, medicine, and public health to advance clinical research integrity and transparency.

Heeju Sohn, PhD

Dr. Heeju Sohn is an Assistant Professor of Sociology at Emory University with graduate training in Demography (Ph.D.) and Information Engineering (M.Eng). Dr. Sohn completed her NIH-funded (K99/R00) postdoctoral training at UCLA's Health Policy and Management and California Center for Population Research. Her research examines how unequal safety-nets exacerbate health and social disadvantages among vulnerable groups in the United States. Her work directly addresses knowledge gaps stemming from limited data by developing new methods to combine and analyze openly available data sources.

Julie A. Beans, MPH

Julie Beans (Yup'ik and Oneida) is a public health researcher who has worked with Southcentral Foundation Research since 2014. She's worked on several community-based research projects that aim to improve health outcomes for the Alaska Native and American Indian community in Alaska through research, while maintaining community protections. Ms. Beans works to bring the voice of community members to the forefront of health research to ensure research directly benefits the community

Jelili Ojodu, MPH

Mr. Ojodu 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 at APHL. He received his MPH in Maternal and Child Health from The George Washington University and a Bachelor of Science degree in Biological Sciences from the University of Maryland, College Park.

Deanna J. Wathington, MD, MPH, FAAFP

Deanna J. Wathington currently serves as President-Elect of the American Public Health Association. She is the clinical director at REACHUP, Inc., an affiliate professor in the College of Public Health (University of South Florida), and the executive director of the Consortium of African American Public Health Programs. She serves on the APHA Executive Board and the ASPPH Workforce Advisory Committee. Previous service includes the Council on Education for Public Health, the National Partnership for Women and Families Listening to Mothers Advisory Group, and the HHS OMH Southeastern Health Equity Council. Dr. Wathington's work and scholarly efforts have centered on health equity, clinical-community linkages, maternal and child health, equitable community development, and expanding diversity within the health professions. She is co-editor of Black Women in Public Health: Strategies to Name, Locate, and Change Systems of Power. Dr. Wathington has received the distinction of Fellow of the American Academy of Family Physicians, an AAMC GIA Award of Excellence, the Dr. Hildrus A. Poindexter Award, and the Florida Outstanding Woman in Public Health Award.

Session 5 Speakers

Amy Gaviglio

Amy Gaviglio is a certified genetic counselor and founder of Connetics Consulting, LLC, which provides public health genetics, genomics, and rare disease services nationwide and internationally. She has worked in the newborn screening and rare disease space for the past 17 years. Amy currently works with the Centers for Disease Control and Prevention's Newborn Screening and Molecular Biology Branch, the Association of Public Health Laboratories (APHL), Expecting Health, RTI International, and several other rare disease organizations. She is co-chair of APHL's New Disorders in Newborn Screening taskforce and is a member of additional national groups including the Rare Disease Diversity Coalition and EveryLife Foundation's Community Congress. She also serves as an Advisor for the Midwest Genetics Network and is a member of the MPS Society's Scientific Advisory Board. Finally, Amy serves as Chair of the NBS Expert Panel for the Clinical and Laboratory Standards Institute and is currently the Chair of Minnesota's Rare Disease Advisory Council.

Michael Watson, PhD, MS, FACMG

Michael S. Watson, MS, PhD, FACMG is an Adjunct Professor of Pediatrics at Washington University School of Medicine. His postdoctoral training was in the Medical Genetics Training Program at Yale University School of Medicine where he was Associate Director of Clinical Cytogenetics 1984-1986. He directed the Clinical and Molecular Cytogenetics Laboratories at Washington University School of Medicine in St. Louis from 1986–2001. He's certified by the American Board of Medical Genetics and Genomics in Clinical Cytogenetics and Genomics and earned a PhD in Medical Genetics. He served on the Board of Directors of the American College of Medical Genetics (ACMG) (1992-1998) (Vice President for Laboratory Affairs). He directed the HRSA-funded project "Newborn Screening: Toward a Uniform Screening Panel and System" and HRSA's National Coordinating Center for Regional Genetics and Newborn Screening Collaboratives and NIH/NICHD's Newborn Screening Translational Research Network (NBSTRN) Coordinating Center (2007-2019) and was a co-P.I. of the ClinGen Resource Project (2013-2019). He remains on its Steering Committee and co-chairs its Partnership Work Group. He was Executive Director of the American College of Medical Genetics and Genomics (ACMG) and the ACMG Foundation for Genetic and Genomic Medicine from 2000 through December 2019.

Biosketches of Committee Members

Jewel Mullen, MD, MPA, MPH

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.

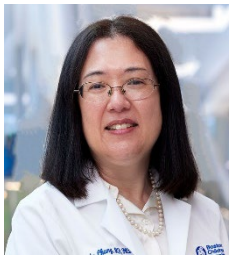
Don Bailey, PhD, MEd

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

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.



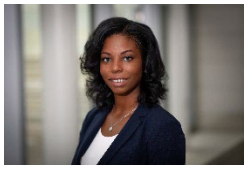
Wendy Chung, MD, PhD (NAM)

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

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

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

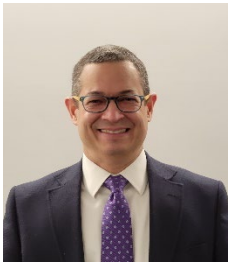
contributions and accomplishments in advancing health and healthcare equity as a field leader in alignment with the mission of the college. Dr. Fletcher received her BS in biology from Tuskegee University with concentrations in bioethics and philosophy, her MA in bioethics, humanities, and society from 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

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

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 (NAM)

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

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

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

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.

Updated June 7, 2018