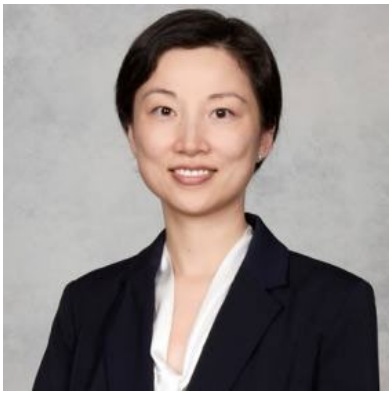
NATIONAL ACADEMIES Sciences Engineering Medicine

Advancing Diagnosis of Rare Diseases: A Webinar
Participant Biographies

Moderators

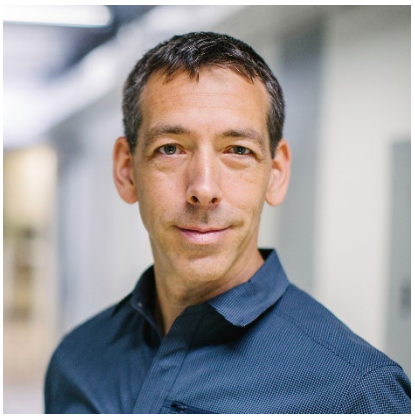
Vanessa Almendro Navarro, Ph.D., MBA, is a life sciences executive who builds at the intersection of innovation, AI-enabled R&D, and commercialization. As Vice President and Head of Science & Technology Innovation at Danaher, she leads enterprise innovation and operating-model modernization across a global portfolio. She designed and scaled the Danaher Beacon distributed R&D program with leading academic partners, launched the Danaher Nexus intrapreneurship pipeline, established the Danaher Antibody Capability Center, and created the Danaher Summits to connect operating companies with top domain expertise. Previously, Vanessa co-founded and led the Brain Tumor Investment Fund; served as Head of Strategy & External Innovation at Eisai and Head of Strategy & Operations at Repertoire Immune Medicines; and held scientific and commercial roles at Vertex. Earlier in her career, she was a research fellow at Dana-Farber Cancer Institute/Harvard Medical School. She holds a Ph.D. in Biochemistry & Molecular Biology (University of Barcelona) and an Executive MBA (MIT). Her expertise spans venture origination, IP strategy, translational development, partnerships and BD, and capability building in diagnostics and therapeutics. She serves on the boards of the Brain Tumor Investment Fund and MIT Sandbox and is a member of the ARM CEO Advisory Council.



Linda N. Geng, M.D., Ph.D., is co-director of the Stanford Long COVID integrated clinical and research program. Dr. Geng is also director of the Stanford Consultative Medicine diagnostic second opinion program. Her primary expertise is in advancing the care of patients with puzzling, complex conditions, and difficult diagnoses. Dr. Geng was part of the 2021 inaugural cohort of NAM Scholars in Diagnostic Excellence program. Dr. Geng has also participated in multiple NASEM workshops including invited speaker this year for workshop to examine overlap in Infection-Associated Chronic Illnesses and workshop to examine the U.S. government definition of Long COVID.

Speakers

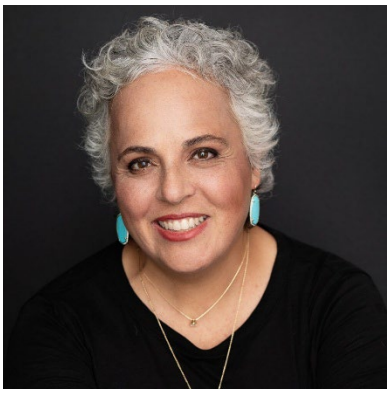
Rebecca Ahrens-Nicklas, M.D., Ph.D., is an Assistant Professor of Pediatrics in the Division of Human Genetics and Metabolism at The Children's Hospital of Philadelphia (CHOP) and the University of Pennsylvania. After completing M.D./Ph.D. training in Physiology and Biophysics, she pursued clinical training in Pediatrics, Clinical Genetics, and Metabolism. She cares for children with rare diseases, with a special interest in neurometabolic disorders. Her research laboratory focuses both on gene discovery and elucidating the pathologic mechanisms underlying rare diseases to guide therapy development. She also partners with advocacy groups to conduct natural history and biomarker studies to promote clinical trial readiness for these rare conditions.



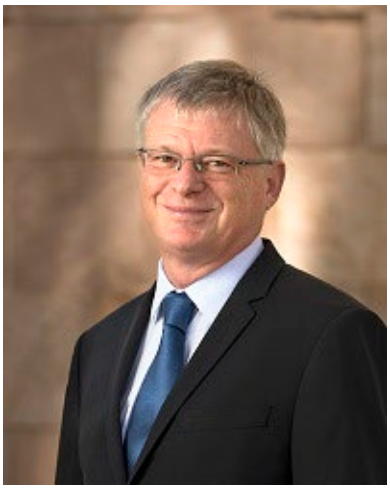
Euan Ashley, BSc, MB ChB, DPhil, FACC, FAHA, FESC, is Chair of the Department of Medicine and Arthur L. Bloomfield Professor of Medicine and Professor of Genetics, of Biomedical Data Science and, by courtesy, of Pathology at Stanford University. He is best known for his contributions to the field of medical genomics and in 2025, he was elected to the National Academy of Medicine. At Stanford, he is the founding director of the Clinical Genomics Program, the Center for Inherited Cardiovascular Disease, and the Catalyst Program for biomedical innovation. Dr. Ashley was awarded the American Heart Association's Medal of Honor for genomics and precision medicine. He was recognized by the Obama Administration for contributions to Personalized Medicine. He has co-founded five biotechnology companies. His book describing the application of genome pioneers to rare diseases, *The Genome Odyssey: Medical Mysteries and the Incredible Quest to Solve Them,* was released in February 2021. Father to three Americans, in his spare time, he continues his quest to understand American football, plays jazz saxophone, pilots small planes, and conducts research on the health benefits of single malt Scotch whisky.

Diana M. Cejas, M.D., MPH, is an Associate Professor of Neurology at the University of North Carolina at Chapel Hill and faculty of the Carolina Institute for Developmental Disabilities. Her clinical work, research, and advocacy focus upon understanding the effects of ableism on health and improving care delivery for youth with neurodevelopmental disabilities. Since surviving cancer and a stroke during her residency, Dr. Cejas has devoted much of her career to patient advocacy and improving communication between healthcare providers and the disability community, particularly young disabled patients of color. She shares her own story and other commentary on health via essays and other nonfiction. She is a 2025 Ucross Foundation Fellow, recipient of the 2024 James Baldwin Fellowship at MacDowell, and 2023 PEN America Emerging Voices Fellowship Finalist. She is working on an essay collection that uses her story as both physician and patient to tell a broader story about dehumanization and discrimination in healthcare.

William A. Gahl, M.D., Ph.D., is a Senior Investigator in the National Human Genome Research Institute and is Head of the Section on Human Biochemical Genetics in the Medical Genetics Branch. He graduated from the Massachusetts Institute of Technology, earned his M.D. and Ph.D. from the University of Wisconsin, and is board-certified in pediatrics, clinical genetics, and biochemical genetics. He elucidated the basic defects in cystinosis and Salla disease, helped bring cysteamine to new drug approval by the FDA as the treatment for cystinosis, published over 700 papers, reviews, book chapters, and editorials, and trained 42 biochemical geneticists. In 2008, he established the NIH Undiagnosed Diseases Program (UDP), which expanded to a national Undiagnosed Diseases Network and a worldwide Undiagnosed Diseases Network International. Dr. Gahl received the Dr. Nathan Davis Award for Outstanding Government Service from the AMA, the Service to America Medal in Science and the Environment, and the EURORDIS Lifetime Achievement Award; he is a member of the National Academy of Medicine.



Maria Kefalas, Ph.D., studied economics at Wellesley College and earned her M.A. and Ph.D. in sociology from the University of Chicago. She worked at the Brookings Institution, held a post-doctoral fellowship at the University of Pennsylvania, and taught at Barnard College (Columbia University) before joining the faculty of Saint Joseph's University in Philadelphia. She is the author of numerous books and articles, and has received grants from the William T. Grant Foundation, the MacArthur Foundation, and the Department of Justice. Her life took an unexpected turn in 2012 when her husband, father, and father-in-law were all diagnosed with cancer. That same year, on the fifth of July, she learned her youngest child, Calliope "Cal", suffered from a fatal degenerative neurological disease called metachromatic leukodystrophy (or MLD). Leukodystrophies are in the same family as Tay-Sachs and Canavan diseases. Cal would lose the ability to walk, talk, and feed herself within months. Cal was not expected to survive beyond the age of six. Dr. Kefalas gained fifty pounds and struggled with depression and grief. Then, a year after her daughter's diagnosis, Cal's nine-year-old brother PJ suggested that the family start selling cupcakes "to raise money and to help kids like Cal." This idea for a bake sale would change everything. She started blogging under the name "The Recovering Supermom" and published essays in Slate, STAT, The Mighty, and The Huffington Post. With her husband, Pat Carr, she is the co-founder of the Calliope Joy Foundation and Cure MLD. Over the next several years, the family would sell 50,000 cupcakes. That money would help establish nation's first Leukodystrophy Center of Excellence at the world-renowned Children's Hospital of Philadelphia. She has become a nationally recognized parent advocate for gene therapy who has spoken at the NIH, NORD, and the FDA. Tragically, her husband, Rutgers University sociologist Pat Carr died from multiple myeloma on April 16, 2020. Cal would beat the odds for over a decade. Cal succumbed to the disease at the age of 12 on March 24th, 2022. She was surrounded by her family, nurses, and doctors at her home. She left this world while her brother PJ held her in his arms. The story of Cal and the cupcakes has been featured on CBS Sunday Morning with Jane Pauley and the Chan Zuckerberg Initiative. Dr. Kefalas's work was funded by the Philadelphia Eagles, and she received the 2018 Rare Impact Award by National Organization of Rare Disorders (NORD). Her latest book Harnessing Grief: One Mother's Quest for Meaning and Miracles was published by Beacon Press. Maria and her family continue to sell cupcakes, and they have raised over a million dollars to help children impacted by leukodystrophies. Libmeldy, a gene therapy to treat Cal's disease, was approved in the EU and UK in 2021. That same year, the state of New York began screening newborns for MLD. And Maria is working tirelessly to ensure that MLD will become one of the five percent of rare diseases with an FDA approved treatment in the near future. Maria has embraced a healthy lifestyle with the help of her family, and she only sells cupcakes these days, but rarely eats them anymore herself. She lives outside of Philadelphia with her children Camille and PJ and a rescued dog named Brody. Camille recently graduated from the College of Wooster with a degree in sociology and religion and has been inspired to work in the health care field because of Cal. PJ is a transfer student at Rutgers University studying biochemistry, he hopes to use his degree in science to join the fight against genetic diseases like leukodystrophy. Maria is a professor at Saint Joseph's University.



Stephen F. Kingsmore, M.B., Ch.B., B.A.O., D.Sc., is President and CEO of Rady Children's Institute for Genomic Medicine at Rady Children's Hospital, San Diego, which is implementing pediatric genomic/precision medicine at unprecedented scale. Previously he was the Dee Lyons/Missouri Endowed Chair in Genomic Medicine at the University of Missouri-Kansas City School of Medicine and director of the Center for Pediatric Genomic Medicine at Children's Mercy Hospital, Kansas City. He has been the president and CEO of the National Center for Genome Resources, Santa Fe, New Mexico, chief operating officer of Molecular Staging Inc., vice president of Research at CuraGen Corporation, founder of GatorGen, and assistant professor at the University of Florida's School of Medicine. Kingsmore received M.B., Ch.B., B.A.O. and D.Sc. degrees from the Queen's University of Belfast. He trained in clinical immunology in Northern Ireland and 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, and received the 2013 Scripps Genomic Medicine award and 2013 ILCHUN prize of the Korean Society for Biochemistry and Molecular Biology. TIME magazine ranked his rapid genome diagnosis among the top 10 medical breakthroughs of 2012. In March of 2015, Kingsmore surpassed his previous record in genetic sequencing by reducing the process to 26 hours, which was recognized in April 2016 by Guinness World Record as the fastest genetic sequencing in the world.

Edward "Ed" Neilan, M.D., Ph.D., is the Chief Medical and Scientific Officer of the National Organization for Rare Disorders (NORD®). He is a physician-scientist and rare disease expert, and he joined NORD in 2021 to lead its medical and research programs. Dr. Neilan seeks to encourage and enable institutions and companies to develop innovative approaches and new treatments to help rare disease patients. He is an expert in clinical trial design and drug development, has contributed data that helped support the FDA and global regulatory approvals of five new rare disease therapies, and has authored or co-authored multiple clinical trial protocols and safety and regulatory reports to global health authorities. Prior to joining NORD, Dr. Neilan worked at Sanofi Genzyme, a major biopharma company, where he led global medical affairs strategy and execution for the rare neurological diseases portfolio and contributed medical expertise to clinical development efforts across multiple programs. Prior to that, he served as the President of the Medical Staff at Boston Children's Hospital. As a staff physician, clinical geneticist, and the Director of Quality Improvement in the metabolism program at Boston Children's, he directly cared for and studied patients with many genetic diseases. After completing BS and MS degrees in Biology at Yale University, Dr. Neilan earned his MD and PhD degrees at Stanford University. He completed residency and fellowship training at Harvard Medical School, where he subsequently served as a faculty member for more than 12 years. He is triple board-certified in pediatrics, clinical genetics,

and clinical biochemical genetics. He is a fellow of both the American Academy of Pediatrics and the American College of Medical Genetics and Genomics.