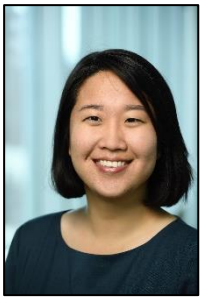
Primary Mitochondrial Diseases:
A Rare Disease Virtual Workshop

Virtual Public Meeting
May 22, 2025; 10AM – 3:30PM (eastern time)

Presenters & Panelists

Anna Choe, MD, MPH
Center for Drug Evaluation and Research, FDA



Anna Choe, MD, MPH is a clinical team leader in the Division of Rare Diseases and Medical Genetics, Office of New Drugs, Center for Drug Evaluation and Research. She holds a BA in Neuroscience from Washington University in St. Louis, an MD from the University of Alabama at Birmingham, and an MPH from Johns Hopkins School of Public Health. Her training encompasses pediatrics and preventive medicine. She specializes in reviewing rare disease programs, with a special interest in real-world evidence and endpoint development. In her leisure time, she enjoys hiking and learning new crafts.

Jason Colquitt
Across Healthcare

Jason Colquitt is the CEO and Founder of Across Healthcare, a technology company empowering rare disease patients, caregivers, researchers, and clinicians through innovative digital health platforms. With over 25 years of experience in healthcare technology, Jason has led the development of patient-centered tools that support data collection, clinical research, and treatment optimization, particularly for underserved and rare disease populations. Under his leadership, the Matrix platform—used by over 110 patient advocacy groups across 280 rare diseases in 137 countries—has become a global resource for multimodal patient data, real-world evidence generation, and insights. Jason is a frequent advisor to government, industry, and academic initiatives, championing patient voice, ethical data use, and interoperability standards. His work supports global collaborations aimed at accelerating drug development and improving health equity across the global rare disease community.

Marni Falk, MD
Children's Hospital of Philadelphia & University of Pennsylvania Perelman School of Medicine

Marni J. Falk, MD, is a physician-scientist who holds a Distinguished Endowed Chair in the Department of Pediatrics and serves as Founder and Executive Director of the Mitochondrial Medicine Frontier Program at The Children's Hospital of Philadelphia (CHOP), as well as Professor in the Division of Human Genetics within the Department of Pediatrics at the University of Pennsylvania (UPENN) Perelman School of Medicine in Philadelphia, Pennsylvania. A board-certified Clinical Geneticist and Pediatrician, Dr. Falk works to support patients with rare disorders, with a focus on primary mitochondrial disease, to improve precision clinical care, diagnostic modalities, precision therapeutics, genomic resources, and community consortium activities. Dr. Falk is PI of an active translational research laboratory program investigating causes and global metabolic consequences of mitochondrial disease, and advancing the preclinical development of targeted therapeutics and novel diagnostic nanosensors through human clinical trial stage, using *C. elegans*, zebrafish, mouse, and human cell models of genetic and/or pharmacologic based respiratory chain dysfunction.

Chad Glasser, PharmD, MPH
Tisento Therapeutics

Chad brings more than 12 years of clinical research experience, having worked on a range of preclinical to late-stage programs spanning infectious disease, oncology, and multiple rare disease therapeutic areas. For the last five years, he has focused on advancing the zagociguat clinical program in MELAS, first at Cyclerion then through the transition to Tisento. Prior to Cyclerion, Chad was a clinical scientist at Acceleron Pharma (acquired by Merck) where he designed clinical studies for multiple rare neuromuscular diseases, and before that, he completed a two-year post-doctoral clinical research fellowship and started his career at Cubist Pharmaceuticals (acquired by Merck).

Magnus Hansson, MD, PhD
Abliva AB, A Member of Pharming Group

Magnus Hansson received his Medical Degree in 2002 and PhD in 2007 from Lund University, Sweden, where he holds an Associate Professorship. He served as a specialist physician in clinical physiology prior to joining Abliva in 2016. At Abliva, he has headed the development of a portfolio of innovative compounds for mitochondrial disease including KL1333 which is currently in a pivotal stage clinical trial, the FALCON study. He has a long-standing commitment to mitochondrial medicine development authoring 60+ scientific publications and 10+ patent families.

Michio Hirano, MD
Columbia University Medical Center

Dr. Michio Hirano is the Lucy G. Moses Professor of Neurology, Chief of the Division of Neuromuscular Medicine, and Director of the H. Houston Merritt Neuromuscular Research Center at Columbia University Medical Center. For over 30 years, Dr. Hirano's translational research focused on mitochondrial disease. His laboratory has identified causative genes for more than a dozen disorders. To understand how some of these mutant genes cause diseases, the laboratory has been studying cell and mouse models. For the autosomal recessive mitochondrial disease thymidine kinase 2 (TK2) deficiency, Dr. Hirano's group has developed a deoxynucleoside therapy in Tk2-deficient mouse and has delivered the treatment to patients under an expanded access program. This therapy is currently under investigation by UCB, which is sponsoring clinical studies for potential drug registration. Dr. Hirano has directed the NIH U54-funded North American Mitochondrial Disease Consortium (NAMDC).

Kerry Jo Lee, MD
Center for Drug Evaluation and Research, FDA

Dr. Kerry Jo Lee is the Associate Director for Rare Diseases and Lead for CDER's Accelerating Rare Disease Cures (ARC) Program in the Office of New Drugs (OND), Center for Drug Evaluation and Research (CDER). In this role she also leads CDER's Rare Diseases Team, a multidisciplinary rare disease programming and policy team that anchors ARC operations and works across CDER to promote their mission to facilitate, support, and accelerate the development of drugs and therapeutic biologics for rare diseases. Dr. Lee is a pediatric gastroenterologist/hepatologist who joined the FDA as a medical officer in 2014. Through her previous roles in CDER/OND, Dr. Lee has served as a lead in the areas of benefit-risk assessment, modernization efforts (including the integrated review for marketing applications), and real-world data/evidence programming in CDER drug review and policy.

Amel Karaa, MD
Massachusetts General Hospital & Harvard Medical School

Dr. Amel Karaa is an internist and clinical geneticist by training and currently the director of the mitochondrial disease program at the Massachusetts General Hospital in Boston where she oversees clinical care, clinical research and trials for mitochondrial disease patients. She is the recipient of the 2013 United Mitochondrial Disease Foundation (UMDF) Fellowship, is the immediate past president of the Mitochondrial medicine Society, Chair of the United Mitochondrial Disease Foundation Scientific and Medical Advisory Board and is part of the scientific and medical advisory boards of several advocacy and private groups. Dr. Karaa is also a founder and a board member of the Mitochondrial Care Network (MCN), a US-wide network overseeing expert centers for mitochondrial medicine and the founder of TREAT MITO, a clinical trial and research consortium for primary mitochondrial diseases.

Naomi Knoble, PhD
Center for Drug Evaluation and Research, FDA

Naomi Knoble, PhD, is a pediatric neuropsychologist and Associate Director specializing in rare disease measurement in clinical trials in the Division of Clinical Outcome Assessment (DCOA), Office of Drug Evaluation Science (ODES), Office of New Drugs (OND), Center for Drug Evaluation Research (CDER), with the US Food and Drug Administration (FDA). Dr. Knoble has a PhD in Counseling Psychology from the University of Oregon and completed clinical training in pediatric neuropsychology at the Oregon Health & Science University and University of Minnesota Medical School. Prior to joining FDA in 2020, Dr. Knoble was a research scientist developing patient-centered outcomes in global clinical trials and post-market evidence generation. Dr. Knoble is the FDA liaison for the Critical Path Institute's Rare Disease Clinical Outcome Assessment Consortium.

Reenie McCarthy, JD
Stealth BioTherapeutics

Reenie McCarthy is the Chief Executive Officer and a member of the Board of Stealth BioTherapeutics. Since 2015, she has led the company as President and CEO, focusing on advancing therapies for diseases involving mitochondrial dysfunction. With over 20 years of experience on the investment team at Morningside, Stealth's principal investor, she has worked extensively with private nonclinical and clinical-stage companies developing drugs across a broad range of therapeutic areas. In addition to her role at Stealth, Reenie serves on the Board of the Biotechnology Innovation Organization (BIO), where she supports companies in advancing science to bring therapies to patients with unmet needs.

Catherine Pilgrim-Grayson, MD, MPH
Center for Drug Evaluation and Research, FDA

Catherine Pilgrim-Grayson is the Director of the Division of Rare Diseases and Medical Genetics at the US FDA, providing scientific, clinical and technical direction on all medical and scientific decisions and judgment in connection with the review and evaluation of drugs in that arena. She advances OND's policies and research agenda for rare diseases, liaising with other FDA offices and other regulatory agencies, industry, professional organizations, academia, and the public. In addition to overseeing several recent approvals for drugs to treat rare inborn errors of metabolism with unmet need, an important recent Division accomplishment is the establishment of the Genetic Metabolic Diseases Advisory Committee, which provides the FDA independent, knowledgeable advice and recommendations on key issues around medical products for genetic metabolic diseases. In prior roles at FDA she was a Clinical Reviewer, acting Clinical Team Leader, acting Deputy Director and the Deputy Director for Safety. Dr. Pilgrim-Grayson is a board-certified obstetrician/gynecologist. She graduated from Swarthmore College with Honors, the University of Pennsylvania Perelman School of Medicine and the Johns Hopkins Bloomberg School of Public Health with Honors. She completed her Gynecology and Obstetrics residency at the Johns Hopkins Medical Institutions, and had a varied clinical career before coming to FDA, in academic medicine at Johns Hopkins, leading the Johns Hopkins Fibroid Center and teaching, conducting research and international public health outreach. Dr.

Pilgrim-Grayson has also previously held a faculty position at Thomas Jefferson University, leading the Obstetrics and Gynecology clinic, and has served as the Chief of the Department of Obstetrics and Gynecology at the University of Maryland St. Joseph Medical Center.

Brian Tseng, MD, PhD
The POLG Foundation

Brian Tseng is a board-certified MD/PhD pediatric neurologist who has served in leading roles as a clinician, academic researcher, and vice-president in global biopharma companies. He has over 18 years experience leading teams of teams to advance pediatric and rare disease medicines, including gene therapies. His past clinical/academic practice has been at Massachusetts General Hospital – Harvard Medical School and previously Colorado Children's Hospital. Currently, Dr. Tseng serves as the Chief Executive Officer of The PolG Foundation (Home - The POLG Foundation), a relatively new non-profit research advocacy organization. In his spare time, he is also a Wilderness Medicine certified Parkmedic with the Rocky Mountain National Park (RMNP) Search and Rescue and volunteers on the Board of Directors for the Estes Valley Fire Protection District.

Yan Wang, PhD
Center for Drug Evaluation and Research, FDA

Yan Wang is a statistical reviewer in the Division of Biometrics IV in the Office of Biostatics at the Center for Drug Evaluation and Research at the U.S. Food and Drug Administration (FDA). She currently provides statistical support for the medical Division of Rare Disease and Medical Genetics, which focuses on drugs and biologics development programs for the prevention and treatment of rare inborn errors of metabolism. Her prior experience at the FDA includes drug development in anti-infective, ophthalmology, and transplant. Prior to joining the FDA in 2005, Dr. Wang was a senior statistician in a pharmaceutical company developing products for the treatment of diabetes. She received her PhD in Biostatistics from the University of University of California at Los Angeles.

Kasey Woleben
Cure Mito Foundation

Kasey Woleben, the cofounder and Executive Director of the Cure Mito Foundation, is motivated by her own experiences to promote research on mitochondrial diseases. After her son was diagnosed with Leigh syndrome in 2014, she has been dedicated to leading the foundation in partnership with researchers and families to create effective treatments.

Philip Yeske, PhD
United Mitochondrial Disease Foundation

Philip Yeske received a BS in Chemistry from Allegheny College in 1985 and a doctorate in Organic Chemistry from Emory University in 1990. Dr. Yeske has been active in the mitochondrial disease community for more than 20 years, first as a parent of an affected child who lost her life to mitochondrial disease at age 1, then later as a Trustee of the United Mitochondrial Disease Foundation (UMDF). In 2013 he joined the staff of UMDF as Science & Alliance Officer, responsible for managing all scientific and business development efforts of the foundation related to improved diagnoses, development of treatments and cures, and optimized patient care.

Sophia Zilber
Lived Experience Perspective

Sophia Zilber is an advocate, data expert, and rare disease leader who blends deep personal commitment with over two decades of professional experience in drug development. With a background in clinical data analysis and statistical programming, she brings both expertise and heart to everything she does. She is especially focused on raising awareness around patient registries and the importance of collecting high-quality data for rare disease research and drug development. Sophia currently serves as Vice President and Patient Registry Director at the Cure Mito Foundation, where she is leading the development of a global Leigh syndrome patient registry to support research, accelerate treatments, and center the voices of families.

Zarazuela Zolkipli-Cunningham, MBChB, MRCP
Children's Hospital of Philadelphia & University of Pennsylvania Perelman School of Medicine

Zarazuela Zolkipli-Cunningham, MBChB, MRCP, is Director of Clinical Research in the Mitochondrial Medicine Frontier Program in the Division of Human Genetics at The Children's Hospital of Philadelphia (CHOP) and Assistant Professor, at the University of Pennsylvania Perelman School of Medicine.

Dr. Zolkipli-Cunningham has established a Mitochondrial Myopathy Clinical Research program to design and validate new quantitative methods of Mitochondrial Myopathy for clinical trials, precision trials, and natural history studies. She is PI of NIH-R01, Industry, and Foundation grants and co-I of a Department of Defense Focused Development Award. As Clinical Research Director of the Mitochondrial Medicine Frontier Program, she is actively engaged in bridging academia-industry collaborations to pursue precision and clinical trials. She is also CHOP-site PI of the Pediatric Neuromuscular Research Network on Spinal Muscular Atrophy natural history studies and outcome measure development, co-leads a ClinGen NICHD U24 Leigh syndrome Mitochondrial Disease consortium gene-disease curation effort, member of the North American Mitochondrial Disease Consortium (NAMDC) committee, Rare Disease Clinical Research Network (RDCRN) Career Enhancement Committee, Scientific and

Medical Advisory Board of the United Mitochondrial Disease Foundation (UMDF), co-leads the UMDF Bench to Bedside Education Seminars and is Scientific Organizer of the annual UMDF Clinical Research Pavilion. The focus of her work is to improve clinical care, validate new outcome measures of Mitochondrial Myopathy for clinical trials, and advance new therapies in Mitochondrial Disease.

Moderator

Susan C. Winckler, RPh, Esq.
Chief Executive Officer, Reagan-Udall Foundation for the Food and Drug Administration

Susan C. Winckler is CEO of the Reagan-Udall Foundation for the Food and Drug Administration. The Foundation is the non-profit organization created by Congress to advance the mission of the FDA. Prior to accepting the Foundation post in May of 2020, Ms. Winckler served as President of Leavitt Partners Solutions. As President and Chief Risk Management Officer for the Leavitt Partners family of businesses, Ms. Winckler advised corporate executives on policy and business matters. As CEO of the Food & Drug Law Institute, she provided attorneys, regulators, industry leaders, and consumers with a neutral forum to address domestic and global issues. As FDA Chief of Staff from 2007-2009, Ms. Winckler managed the Commissioner's office; served as his/her senior staff adviser; analyzed policies; and represented FDA before myriad government and external stakeholders. She simultaneously led FDA's Offices of Legislation, External Relations, Public Affairs, and Executive Secretariat. As APhA Vice President Policy/Communications and Staff Counsel, she served as the association's lead spokesperson and senior liaison to Congress, the executive branch, state associations, and allied groups. Ms. Winckler earned a bachelor's degree from the University of Iowa College of Pharmacy and her juris doctorate *magna cum laude* from Georgetown University Law Center. She is an APhA Fellow, an elected member and Chair of the United States Pharmacopeial Convention (USP) Board of Trustees (2015-2020, 2020-2025), a member of the Purgo Scientific, LLC board, and a member of the Virginia Commonwealth University School of Pharmacy National Advisory Council.