Speaker Bios

EVERYLIFE FOUNDATION FOR RARE DISEASES | Scientific Workshop

Ramona Belfiore-Oshan, PhD

Dr. Ramona Belfiore-Oshan, PhD is the Executive Director of the Duchenne Regulatory Science Consortium (D-RSC) at the Critical Path Institute (C-Path). In this role, she provides overall leadership, administrative, and scientific oversight of the Consortium, which focuses on advancing drug development tools for Duchenne muscular dystrophy (Duchenne) and related neuromuscular diseases.

Dr. Belfiore-Oshan earned her bachelor's degree in biological science, master's degree in molecular biology, and PhD in neuroscience from the University of Catania in Italy. Her doctoral research centered on neurodegenerative diseases, including amyotrophic lateral sclerosis (ALS) and Alzheimer's disease. She furthered her research during a postdoctoral position in neurology at the Icahn School of Medicine at Mount Sinai, where she also honed skills in science communication, grant writing, and scientific project management.

Joining C-Path in 2021 as the Associate Director for D-RSC, Dr. Belfiore-Oshan played a pivotal role in supporting the implementation of modeling and simulation work for multiple regulatory submissions. Her efforts contributed to the completion of the development of the DMD Clinical Trial Simulator, a tool designed to enhance the design of clinical trials for Duchenne therapies.

In July 2023, she was appointed as the Executive Director of D-RSC, where she continues to lead initiatives aimed at accelerating therapy development to address the urgent unmet medical needs in Duchenne and other dystrophinopathies.

Dr. Belfiore-Oshan's leadership extends to global collaborative efforts to advance solutions for Duchenne and other dystrophies. She has expressed enthusiasm for working with scientists and patient advocates around the world to promote discussions and mission-driven solutions in the field. Her dedication to advancing research and regulatory science in neuromuscular diseases underscores her commitment to improving outcomes for patients affected by these conditions

Elizabeth Berry-Kravis, MD, PhD

Elizabeth Berry-Kravis MD, PhD is a Professor of Pediatrics, Neurological Sciences and Anatomy/Cell Biology at Rush University Medical Center in Chicago. She established the Fragile X Clinic and Research Program in 1991, through which she has provided care to over 800 patients with fragile X syndrome (FXS). She has studied medical issues, epilepsy and psychopharmacology in FXS, and has been a leader in translational research in FXS for 20 years, including development of clinical outcome measures and biomarkers, natural history studies, newborn screening, and particularly clinical trials of new targeted treatments in FXS, for which she has been PI or Co-PI of 27 trials, both industry and investigator sponsored. Her laboratory studies the cellular role of fragile X messenger ribonucleoprotein (FMRP), relationship between FMRP and clinical function, and optimization of genetic testing methods. More recently she has expanded clinical and translational work to other neurodevelopmental disorders in addition to FXS, including autism spectrum disorders and single gene models of ASD, including Phelan McDermid syndrome, Rett syndrome, and Angelman syndrome.

She also is working on translational research in rare neurogenetic disorders including Niemann-Pick type C, Battens disease, pantothenate kinase-associated neurodegeneration, and creatine transporter deficiency, as well as N-of-1 trials of ASOs and gene therapy for early onset epileptic encephalopathies and other neurogenetic conditions (including but not limited to PACS1, SCN2A, TUBB4A, H3F3A, CLN1). She has received the NFXF Jarrett Cole Clinical Award, FRAXA Champion Award, NFXF William and Enid Rosen Research Award, March of Dimes Jonas Salk Research Award, American Academy of Neurology Sidney Carter Award in Child Neurology, John Merck Fund Sparkplug Award, the FRAXA Ingenuity Award, the FAST Innovation Award, the inaugural Martha Bridge Denckla Award from the Child Neurology Society for work in cognitive disorders of children, and the CureSHANK 2025 PMS Investigator of the Year Award.

Terry Jo Bichell, PhD, MPH

Terry Jo Vetters Bichell, PhD, MPH worked primarily as a public health nurse-midwife until her youngest child, Lou, was diagnosed with Angelman syndrome in 2000. She quickly switched focus to move bench research into the first clinical trials for Angelman syndrome and to help design natural history studies. Dr. Bichell earned a PhD in neuroscience from Vanderbilt University in 2016, studying gene-environment interactions in Huntington's disease rodent models. She was the founding director of the A-BOM Alliance from 2016-2018. In 2019, Dr. Bichell launched COMBINEDBrain, a pre-competitive consortium of patient advocacy organizations, working to identify outcome measures and biomarkers for rare genetic neurodevelopmental disorders. She was a member of the National Academy of Sciences Committee on Processes to Evaluate the Safety and Efficacy of Drugs for Rare Diseases or Conditions in the United States and the European Union. She is the Vice Chair of the Tennessee Rare Disease Advisory Council and teaches a course in Translational Neuroscience at Vanderbilt University. Most recently, Dr. Bichell joined the IRDiRC Patient Advocacy Constituent Committee (PACC) representing COMBINEDBrain. As a parent, Dr. Bichell has been alongside her son Lou many times as he has participated in clinical trials in the search for a treatment for Angelman syndrome.

Michelle Campbell, PhD, MS

Michelle Campbell, PhD, MS is the Associate Director for Stakeholder Engagement and Clinical Outcomes in the Office of Neuroscience, Office of New Drugs (OND) in FDA's Center for Drug Evaluation and Research. Dr. Campbell joined the FDA in 2014 and previously was a reviewer on the Clinical Outcome Assessments (COA) Staff and Scientific Coordinator of the COA Qualification Program in OND. Dr. Campbell's focus is in patient-focused drug development and the use of patient experience data in the regulatory setting. Prior to joining FDA, Dr. Campbell spent more than 10 years conducting research in the academic-clinical setting, including five years in a neurology and developmental medicine department. Dr. Campbell earned her BA in Biology from the College of Notre Dame, her MS in Health Science from Towson University, and her PhD in Pharmaceutical Health Services Research from the University of Maryland School of Pharmacy.

Jenny Carwile, ScD, MPH

Jenny Carwile, ScD, MPH is a Global Medical Director at Sanofi. She leads real world evidence studies on Gaucher disease and ASMD, including the ICGG Gaucher Registry study supporting the recent FDA approval of Cerezyme for Gaucher disease type 3. She lives in coastal Maine and enjoys traveling with her husband and three young children.

Kent Christopherson, PhD

Kent specializes in the most complex corner of biopharma, advanced therapies for rare and complex diseases. The therapies are scientifically demanding, the diseases are complicated, and the path to commercialization requires a caliber of Medical Affairs leadership that is genuinely difficult to find.

He earned a PhD in Biochemistry and Molecular Biology with a minor in Medical and Molecular Genetics, followed by a Postdoctoral Research Fellowship in Experimental Hematology/Oncology and Stem Cell Transplantation at Indiana University School of Medicine. Following an academic career focused on cell and gene therapy, Kent transitioned to industry. His clinical and scientific experience now spans more than 25 years and includes a track record of six global product launches in complex genetic diseases, at Orchard Therapeutics (Libmeldy, Lenmeldy), Ultragenyx (Mepsevii), and Biogen (Spinraza, Elocate, Alprolix).

Kent brings deep fluency across the full Advanced Therapy Medicinal Products (ATMP) platform spectrum, including lentiviral ex vivo gene therapy, AAV in vivo gene therapy, cell therapy, RNAi/ASO molecular therapies, enzyme replacement therapy, and other biologics. That technical breadth is paired with a recognized subject matter authority in rare genetic diseases, neurodegenerative & neurometabolic disorders, immunology & immune deficiencies, and benign & malignant hematology.

As Head of Global Medical Affairs at Orchard Therapeutics, Kent leads global medical strategy across both commercial and clinical-stage programs, while providing medical support for early-stage translational research. His mission is to accelerate innovation and ensure that breakthrough science reaches the patients who need it.

Kathleen Davies, JD, MS

Kathleen Davies, JD, MS is a Senior Policy Advisor to the Center Director for FDA's Center for Drug Evaluation and Research (CDER). She serves in a cross-cutting role across the CDER and FDA, specializing in the modernization of complex regulatory programs and the execution of high-profile public health strategies. In her current role, Ms. Davies has been instrumental in supporting the development and launch of FDA's Rare Disease Innovation Hub (the Hub) and working closely with Amy Comstock Rick, CDER's Associate Director for Rare Disease Strategy and the Director of Strategic Coalitions for FDA's Rare Disease Hub.

With a career spanning 20 years at the agency, she has led multiple initiatives—from launching the agency's first post-market requirement enforcement program to modernizing various aspects of FDA's Advisory Committee Program. Most recently, she served as the lead negotiator for the generic drug reauthorization, successfully securing vital resources to support the dispensing of over 90% of drugs in the U.S. Prior to her role in CDER, she served as a key advisor to three FDA Commissioners across two administrations where she led landmark initiatives, including standing up the Right to Try program, and addressing major public health challenges such as the opioid crisis and COVID-19.

She holds a Juris Doctor with a Health Care Law Certification from the University of Maryland School of Law and a Master of Science in Chemistry from the University of Maryland Baltimore County. She is also a registered patent attorney.

Kate Donigan, PhD, MPhil

Kate Donigan, PhD, MPhil is the Executive Director, Science and Regulatory Policy at Sarepta Therapeutics where she focuses on advancing critical policies that support the development and regulatory approval of innovative genetic medicines for rare diseases. Prior to joining Sarepta in 2022, Kate was a Senior Director of Science and Regulatory at Biotechnology Innovation Organization where she was the regulatory policy lead for cell and gene therapies, diagnostics, and international regulatory policy. Kate also spent seven years at FDA in both CDER and CDRH advising on regulatory policy and developing guidance with a focus on personalized medicine, diagnostics, and rare diseases. She has also worked in the U.S. Senate as a health policy fellow. Kate holds a PhD in Genetics from Yale University and completed postdoctoral training at the National Institute of Child Health and Human Development.

Cartier Esham, PhD

Cartier Esham, PhD is the CEO of Esham Strategies, a strategic consultancy firm in D.C. that works to empower thought leadership, disrupt the status quo and advance innovative health policy solutions. She also represents the Alliance for a Stronger FDA and serves as the organization's Executive Director. Dr. Esham is also a member of the Eshelman Innovation Board.

Dr. Esham has over 20 years of health policy experience. Prior to launching her own firm, she served as the serves as the Chief Scientific Officer at the Biotechnology Innovation Organization (BIO) where she managed and directed BIO's policy development, advocacy, research and educational initiatives for BIO's emerging companies team, the science and regulatory team, the infectious disease team, the industry analysis and research team, and the membership. Dr. Esham has a PhD in Microbiology from the University of Georgia, a master's degree in Marine Biology from the University of North Carolina at Wilmington and a bachelor's degree from the University of Kentucky.

Sorin Fedeles, PhD, MBA

Sorin Fedeles, PhD, MBA is the Executive Director of the Polycystic Kidney Disease Outcomes Consortium (PKDOC) at the Critical Path Institute. With over 15 years of leadership experience in polycystic kidney disease (PKD), his work has focused on basic and translational research advancing, targets, biomarkers, disease modelling, and regulatory strategies to accelerate drug development. He currently leads multi-stakeholder collaborations bridging industry, academia, patient foundations, and regulatory agencies focused on advancing drug development tools for ADPKD, ARPKD, and ADTKD. Dr. Fedeles also holds an adjunct academic appointment (Assistant Professor) at Yale School of Medicine.

Ryan Fischer

Ryan Fischer is the Chief Operating Officer at the Foundation for Angelman Syndrome Therapeutics (FAST), a role he assumed in 2023. In partnership with the President, he leads the development and execution of FAST's strategic vision, overseeing all aspects of the organization's operations, including program development, grants management, stakeholder engagement, and core operational functions. Ryan serves as a key liaison to federal policymakers, regulators, biopharmaceutical companies, fellow foundation leaders, and the broader rare disease advocacy community.

His leadership ensures FAST remains a driving force in advancing potential treatments, promoting patient-focused drug development, and supporting individuals and families affected by Angelman syndrome.

Prior to joining FAST, Ryan spent 18 years with Parent Project Muscular Dystrophy (PPMD), most recently as Chief Advocacy Officer, where he played a critical role in transforming the organization into a globally recognized leader in rare disease advocacy. He was part of the core team that developed and produced the first-ever patient-preference study on caregiver benefit–risk preferences for emerging Duchenne therapies and went on to lead six additional studies, helping advance the integration of patient experience data into regulatory and drug development decision-making.

Ryan also played a key role in advancing major federal legislation during his tenure at PPMD, including the Muscular Dystrophy Community Assistance, Research and Education (MD-CARE) Act Reauthorization (2008), the MD-CARE Act Amendments of 2014, and the Patient-Focused Impact Assessment (PFIA) Act of 2016. PFIA was incorporated into the 21st Century Cures Act and helped establish the Statement of Patient Experience framework now included in regulatory submissions.

Stephanie Fradette, PharmD

Stephanie Fradette, PharmD, is a Senior Vice President and Head of the Neuromuscular Development Unit at Biogen. In her role, she oversees development of therapies for a variety of neuromuscular diseases, including Amyotrophic Lateral Sclerosis (ALS), Spinal Muscular Atrophy (SMA), and Friedreich's Ataxia (FA). Stephanie has spent the last 16+ years in various roles across Research and Development, including Safety, Regulatory, and Clinical Development. Recently, she led the development of the first approved therapy for a genetic form of ALS (SOD1-ALS), which supported significant advancement of neurofilament as a biomarker and optimization of clinical trial design in ALS. Prior to her work in ALS, she supported development of SPINRAZA, the first approved therapy for spinal muscular atrophy (SMA). Steph is also the co-chair of the Accelerating Medicines Partnership (AMP) for ALS, a precompetitive public-private partnership bringing together NIH, CPATH, FDA, academia, patient advocacy groups, and industry to accelerate development of ALS therapies.

Tim Franson, MD

Tim Franson, MD currently serves as a Principal of Faegre Drinker Consulting in the firm's life sciences regulatory consulting practice (2019-present). He spent a majority of his professional career with Eli Lilly and Co. (Lilly Research Laboratories) from 1986- 2008, retiring from the leadership role of Global Vice President for Regulatory Affairs, and was responsible for Lilly's regulatory submissions including over 25 major approvals, as well as regulatory compliance and policy matters. He has experience in all phases of development relating to clinical, regulatory, quality and related disciplines for small molecule, biological & cell/gene therapies. His consulting practice has served over 100 clients involving more than 20 major product approvals.

Dr. Franson has represented industry in policy matters including Congressional testimony for several renewals of the Prescription Drug User Fee Act (PDUFA), notably as co-chair of industry negotiations for PDUFA-3. Dr. Franson was a Steering Committee member for the community developed draft guidance for development of therapies for Duchenne muscular dystrophy and continues to provide rare disease drug development advice for multiple companies and advocacy groups.

He served from 2016-2021 as Chair of the Board of Directors for the Critical Path Institute. He was the President of the U.S. Pharmacopeial Convention from 2010-2015 and served on their Board of Trustees until 2020.

He previously served on the NIH-NCATS Treatment of Rare and Neglected diseases (TRND) review panel. Dr. Franson was Commencement Speaker (2017) for the University of Iowa College of Pharmacy and has published over 50 articles in peer reviewed journals and is board certified in Internal Medicine and Infectious Diseases.

Emily Freilich, MD

Emily Freilich, MD, is a board-certified pediatric neurologist and the Director of the Division of Neurology 1 within the Office of Neuroscience in FDA's Center for Drug Evaluation and Research. Dr. Freilich graduated from Duke University with a Bachelor of Science degree in biology and received her medical degree from Rutgers-New Jersey Medical School. She completed her pediatric residency and child neurology training at Children's National Health System in Washington, D.C. Prior to joining FDA, Dr. Freilich worked at Children's National and the Pediatric Specialists of Virginia, where she was a general child neurologist with special interest in rare pediatric epilepsies, served as co-director of the Tuberous Sclerosis Clinic, and was an Assistant Professor of Pediatrics and Neurology at George Washington University School of Medicine. She joined the Division of Neurology Products at FDA in 2016 as a clinical reviewer working in the areas of epilepsy and migraine and subsequently transitioned to a team leader in the Division of Neurology 1 for review of treatments for neuromuscular and rare neurologic disorders. She subsequently served as Acting Deputy Director in the Division of Neurology 1 prior to her current role.

Heather Gordish-Dressman, PhD, MS

Heather Gordish-Dressman, PhD, MS is a biostatistician and Research Professor of Pediatrics and Genomic and Precision Medicine at the George Washington University School of Medicine and Health Sciences and in the Center for Health Outcomes Research and Delivery Science at Children's National Medical Center. Dr. Gordish-Dressman has over 20 years of experience in the analysis of neuromuscular diseases, ranging from pre-clinical and basic science research to longitudinal natural history studies. She has served as the statistician for Dr. Kanneboyina Nagaraju's Preclinical Murine Testing Facility at CNMC and continues this work as a consulting statistician for AGADA BioSciences. Since 2010, Dr. Gordish-Dressman has been the primary statistician for the Cooperative International Neuromuscular Research Group (CINRG) where she focuses on the analysis of natural history studies and facilitates collaborations with academic and industry partners. Dr. Gordish-Dressman joined the TREAT-NMD TACT as a Core Committee Member in 2018 and is the inaugural Chair of the TREAT-NMD Neuromuscular Advisory Committee (NM-DAC). In addition, she is a co-founder and Board president for TRiNDS, LLC, a CRO specializing in clinical studies in neuromuscular diseases. Dr. Gordish-Dressman has provided statistical support to a large number of collaborators both within her institution and at other institutions around the world. She has co-authored over 200 scientific publications on a wide variety of topics including, Duchenne muscular dystrophy, cerebral palsy, asthma, necrotizing enterocolitis, biomarkers of disease and drug treatment, obesity and metabolic traits, and muscle strength and fitness.

Annie Kennedy

Annie Kennedy, Chief of Mission Officer at the EveryLife Foundation for Rare Diseases, has served within the community for nearly three decades through her roles with Parent Project Muscular Dystrophy, and the Muscular Dystrophy Association. In that time she helped lead legislative efforts around passage and implementation of the MD-CARE Act, the Patient Focused Impact Assessment Act which became the Patient Experience Data provision within the 21st Century Cures Act, engagement with the FDA and Industry around regulatory policy and therapeutic pipelines, led access efforts as the first therapies were approved in Duchenne, and engaged with ICER around the development of the modified framework for the valuation of ultra-rare diseases. Annie's community roles include service on the Board of Directors of Cure SMA, PFDD Works coalition, Patient Driven Values in Healthcare Evaluation Steering Committee, FasterCures Cures for Life initiative, National Health Council's PCORI Valuation Group, Innovation and Value Initiative Patient Advisory Committee, National Duchenne Newborn Screening Pilot Program Steering Committee, Institute for Gene Therapies Patient Advocacy Advisory Council, State Rare Disease Education Initiative National Steering Committee, and as a member of the NIH National Center for Advancing Translational Sciences Advisory Council and the Cures Accelerator Network Advisory Board.

Kimberly Lin, MD

Kimberly Lin, MD is a pediatric cardiologist at the Children's Hospital of Philadelphia, where she serves as Medical Director of the Pediatric Cardiomyopathy Program. Her primary research interest is in the cardiomyopathy that affects individuals with Friedreich Ataxia.

Naomi Lowy, MD

Naomi Lowy, MD is a physician with extensive expertise in overseeing the development of drugs and biologics. With an 18-year career at the U.S. Food and Drug Administration, primarily within the Center for Drug Evaluation and Research, she held multiple leadership roles in the Office of New Drugs, where she played a pivotal role in shaping regulatory science and drug development policies. As Deputy Director in the Division of General Endocrinology, she served as division signatory and thought leader for treatments of rare endocrine diseases affecting the pituitary, adrenal, and parathyroid glands (including acromegaly, Cushing's syndrome, and hypoparathyroidism), growth hormone disorders, and rare bone diseases (including achondroplasia). She now brings her deep regulatory experience to Hyman, Phelps & McNamara, where she advises clients on policy matters, clinical trial design, and strategic solutions for complex regulatory challenges.

Kim Quaintance-Lunn

Kim Quaintance-Lunn serves as Vice President, Head of Regulatory Science and Execution for Alexion Pharmaceuticals. In this role, she is responsible for leading and implementing the regulatory strategies for US and EU and Regulatory Policy and Intelligence. She is also a critical member of the Regulatory Affairs Leadership Team contributing to the development of organizational strategies and focusing on delivering results based on patient's needs and compliance objectives. Kim is a Scientific Advisor to the DIA Board of Directors, and Board Member and Treasurer for the FDA Alumni Association.

Prior to joining Alexion, Kim served as Vice President, Regulatory Affairs Americas, and Distinguished Science Fellow at Bayer; Senior Director, Global Regulatory Policy and Intelligence at Eisai; and Associate Director for Regulatory Affairs in CDER's Office of New Drugs. Kim began her career in the pharmaceutical industry working as a research scientist in drug metabolism at Pfizer. She graduated with a Bachelor of Science in Biochemistry from the University of Illinois.

Nick Manetto

Nick Manetto has more than 20 years' experience working on federal health care policy before both Congress and the executive branch. Specific areas of health policy focus include Medicare and Medicaid, new payment and delivery models, medical research and public health policy, children's health issues, rare disease policy, and patient engagement as part of the drug development process.

Over the past two decades, Nick has worked on most of the major pieces of health care legislation and has helped clients achieve enactment of multiple pieces of legislation into law. He understands client operational needs and goals, how the federal legislative and regulatory processes influence those outcomes, and how clients can maximize their chances of success.

Nick's work has enabled clients to achieve results by working with Congress and multiple executive branch agencies directly. He does this by maintaining strong and productive working relationships with key members of Congress and committees of jurisdiction as well as executive branch officials from political appointees to career civil servants, and by being a trusted and highly responsive resource.

Jewell Martin, MA, MBA, PMP, RAC

Jewell Martin is Director of U.S. R&D and Regulatory Policy at BioMarin Pharmaceutical Inc. In this role, she partners with internal and external stakeholders to identify regulatory opportunities and challenges and to develop strategic regulatory and policy approaches that support innovative drug development.

Prior to joining BioMarin in 2020, Ms. Martin spent more than ten years at the U.S. Food and Drug Administration (FDA), serving in multiple roles across the Center for Drug Evaluation and Research (CDER), including Executive Operations Staff Lead in the Office of New Drugs (OND) and Project Manager in the Office of Pharmaceutical Quality (OPQ). Earlier in her FDA career, she was a biologist in the Center for Veterinary Medicine (CVM), Office of Research. Before joining FDA/CDER, she worked for over five years in clinical trial operations at several institutes within the National Institutes of Health (NIH).

Ms. Martin earned her undergraduate degree and MBA from Howard University in Washington, DC, and a Master of Arts in Medical Sciences from Boston University. She also holds Project Management Professional (PMP) and Regulatory Affairs Certification (RAC) credentials.

Kim McCleary

Kim McCleary has been at the forefront of patient engagement for more than 30 years. Prior to establishing the Kith Collective in 2018 to bolster patient-informed decision-making, Kim served as managing director of FasterCures, a center of the Milken Institute, for five years. For 22 years, she was CEO of a national patient organization, leading patient engagement, research, and public policy for the myalgic encephalomyelitis/chronic fatigue syndrome (ME/CFS) community.

Emily Milligan, MPH

Emily Milligan leads the Barth Syndrome Foundation (BSF), the only organization solely dedicated to Barth syndrome — an ultra-rare, life-threatening mitochondrial disease affecting primarily males from birth.

With a background spanning public health, international relations, and complex R&D management, Emily brings a cross-sector perspective to the challenges of ultra-rare disease therapy development. Before joining BSF, she directed research operations at the nonprofit powerhouse Breakthrough T1D, where she oversaw a $100 million annual research portfolio and launched a $200 million venture fund investing in type 1 diabetes therapeutics — experience that sharpened her understanding of what it takes to move science from promise to patient.

Since joining BSF in 2018, Emily has been an active and credible voice before the FDA and U.S. Congress — advocating for regulatory frameworks and evidentiary standards that account for the realities of ultra-rare disease populations: small sample sizes, limited natural history data, and patient communities that cannot afford to wait. Under her leadership, the FDA approved the first therapy for Barth syndrome in 2025, which also marked the first drug approval for a mitochondrial targeted therapy in the United States. She has deepened BSF's relationships with peer patient advocacy organizations, academic researchers, and industry partners, operating from a conviction she states plainly: advancing the mission of one rare disease community advances the missions of all.

Cara O'Neill, MD, FAAP

Cara O'Neill, MD, is the Chief Science Officer and Co-Founder of the Cure Sanfilippo Foundation. Her daughter, Eliza, bravely lives with Sanfilippo syndrome type A, a rare neurodegenerative metabolic disease. Before shifting her primary focus to research and advocacy, Dr. O'Neill practiced academic and community pediatrics for over a decade, specializing in the care of children with medically complex conditions.

Her passion to advance therapeutic options for children and unique combination of professional and personal experience enables her to bridge gaps between scientists, clinicians, industry, and families. She collaborates extensively with stakeholders across the rare disease community to advance patient-centered research strategies and clinical trial designs, use of surrogate biomarkers, advancement of newborn screening and a wide range of advocacy initiatives. Her contributions include the publication of the first clinical care guidelines for Sanfilippo syndrome, caregiver preference and drug repurposing studies, biomarkers for use in accelerated approval settings and the development of fit-for-purpose clinical outcome measures.

Jennifer Pangoulias

Jennifer has worked in drug development for over 25 years, primarily focused on advancing global development programs for the treatment of rare neurological diseases. Jennifer has experience in working with global regulatory agencies including the US FDA, the European Medicines Agency, and the Pharmaceutical and Medical Device Agency in Japan. She is the co-director of the Angelman Syndrome Outcome Measure and Biomarker Consortium (A-BOM) and has recently taken on the role of Chief Operating Officer for AS² Bio, focused on accelerating drug development for individuals living with Angelman syndrome. She has a niece who lives with Angelman syndrome.

Rosa Sherafat, MD

Rosa Sherafat, MD is a board-certified pediatric endocrinologist and the Deputy Director Office of Clinical Evaluation, in the Office of Therapeutic Products (OTP), in the FDA Center for Biologics Evaluation and Research (CBER). She received her medical degree from Tehran University of Medical Sciences, completed residency in pediatrics at University of Illinois Medical Center, Chicago, IL and fellowship in pediatric endocrinology in Cincinnati Children's Hospital, Cincinnati, OH. She has been an associate professor of pediatrics in the Department of Pediatrics, Medstar Georgetown University Hospital, in Washington, DC (2007-2018).

Dr. Sherafat joined FDA CBER in 2018 and has served as a Clinical reviewer and Team Lead in General Medicine Branch 2 for several cellular, tissue, gene therapy products and medical devices for a variety of neurology, dermatology and pulmonary indications. Dr. Sherafat served as the clinical co-chair and FDA speaker at the 70th Meeting of the Cellular, Tissue, and Gene Therapies (CTGT) Advisory Committee Meeting on Toxicity Risks of AAV Vector-Based Gene Therapy Products (September 2-3, 2021). She participated in the revision of the FDA Guidance for Industry, Long Term Follow Up after Administration of Human Gene Therapy Products (January 2020) and has represented CBER in several public meetings and workshops.

Charlene Son Rigby, MBA

Charlene Son Rigby, MBA is the Chief Executive Officer of Global Genes. She has spent her career building organizations at the intersection of data, technology, and life sciences. Charlene led the merger between Global Genes and RARE-X, a health technology nonprofit. She was previously Chief Business Officer at Fabric Genomics and held executive roles at enterprise software and genomics companies, including Oracle and Doubletwist. She started her career in neuroscience research at Roche. When Charlene's daughter was diagnosed with a rare genetic disease, she co-founded the STXBP1 Foundation. She is committed to finding a cure for her daughter's disorder. Charlene's unplanned connection between her personal life and profession has helped push forward the search for a cure for her daughter and kids like her, and given her work deeper meaning. Charlene is a Termeer Scholar. She holds a BA in Human Biology from Stanford University and an MBA from the Haas School of Business at U.C. Berkeley.

Peter Stein, MD

Peter Stein, MD, is the former Director of CDER's Office of New Drugs (OND). A nationally-recognized leader in pharmaceutical research and development, Dr. Stein joined CDER in 2016 as the OND Deputy Director. Before coming to FDA, he served as Vice President for late stage development, diabetes, and endocrinology at Merck Research Laboratories. He also served as Vice President, head of metabolism development at Janssen. He has more than 30 years of academic, clinical, and industry experience. Presently, Dr. Stein serves as a consultant with NextStage Pharma Consulting, LLC. Dr. Stein holds a bachelor's degree in history from the University of Rochester in New York and a medical degree from University of Pennsylvania. He trained at Yale University and Yale-New Haven Hospital in internal medicine and in endocrinology and metabolism.

Jamie Sullivan, MPH

Jamie Sullivan, MPH, is the Senior Vice President of Policy & Advocacy at the EveryLife Foundation for Rare Diseases, where she leads federal and state policy initiatives to accelerate the development of and access to life-saving treatments for people with rare diseases. With two decades of experience in public health and health policy, Jamie brings deep expertise in legislative strategy, regulatory affairs, and patient engagement. She is a passionate advocate for equitable healthcare and works to ensure that the voices of patients with rare diseases and their families are reflected in policy decisions that impact their lives. Jamie holds a Master's of Public Health from Florida International University and a Bachelor's of Arts in Political Science from the University of Miami.

Marshall Summar, MD

Marshall Summar, MD is the Chief Executive Officer of Uncommon Cures, LLC, a company focused on consolidating rare disease clinical trials and utilizing innovative technologies to reduce costs and timelines. Before co-founding Uncommon Cures in 2022, Dr. Summar led the Rare Disease Institute at Children's National Medical Center, the first clinical home for patients with genetic rare diseases, and the first NORD-designated Rare Disease Center of Excellence.

Dr. Summar's research has resulted in over 180 peer-reviewed publications and more than 100 international patents spanning therapies, software, and devices. His work has contributed to new treatments for sickle cell anemia, organic acidemias, congenital heart disease, and premature birth. A board-certified pediatrician and geneticist, Dr. Summar has held leadership roles with NORD, the Society for Inherited Metabolic Disorders, and numerous advisory boards, earning NORD's Lifetime Achievement Award in 2022.

James Valentine, JD, MHS

James Valentine, JD, MHS is a Director at Hyman, Phelps & McNamara, P.C., where he assists medical product industry and patient advocacy organization clients in a wide range of regulatory matters relating to new drug and biologic development and approval. In this capacity, James has helped secure FDA approvals for many new drugs across a wide range of therapeutic areas and treatment modalities, including ERT, ASO, mRNA, and cell and gene therapy. This work is often in areas where there are no well-defined development pathways, much of which is in the rare disease arena. James provides preapproval development and regulatory strategy, including on the use of novel or repurposed clinical outcome assessments, innovative clinical trial designs and statistical approaches, and leveraging confirmatory evidence to support single study approvals. He has also been involved in a significant proportion of accelerated approvals for non-oncologic treatments, spearheading strategies for novel surrogate biomarkers and intermediate clinical endpoints.

James is also a champion for the patient voice in drug development and review. Before joining the firm in 2014, James worked in the FDA Office of Special Health Issues where he facilitated patient input in benefit-risk decision-making. James helped administer the FDA Patient Representative Program, facilitate stakeholder consultations during the reauthorization of PDUFA and MDUFA, and launch the Patient-Focused Drug Development program. In his current practice, James has helped to plan and moderate roughly 75% of all externally-led Patient-Focused Drug Development meetings and aids patient organizations in planning Patient Listening Sessions and regulatory scientific workshops (eg, Critical Path Innovation Meetings, Science Focused Drug Development). In recognition of his contributions to the rare disease patient community, in 2019 Global Genes named James as a RARE Champion of Hope. James currently sits on the Board of Directors for the EveryLife Foundation for Rare Diseases.

Kristin Van Goor, PhD, MPhil

Kristin Van Goor, PhD, MPhil is US Head, Global Regulatory Policy and Innovation at Takeda where she leads regulatory policy across rare and specialty diseases, with a particular focus on novel development pathways, pediatric evidence generation, and regulatory innovation. In this role, she works closely with product teams, global regulatory colleagues, and external stakeholders to navigate complex regulatory frameworks and advance policies that support timely development and approval of therapies for patients with high unmet need.

Kristin holds a PhD in Genetics from Yale University and completed her postdoctoral training at the National Institutes of Health. She has held a range of positions spanning research, regulatory policy, and industry leadership, bringing a practical, science-based perspective to regulatory and policy challenges. Kristin regularly engages with FDA, patient advocacy organizations, and industry groups on issues affecting rare disease development, orphan incentives, pediatric requirements, and emerging technologies. Kristin is passionate about engaging in cross-sector discussions aimed at strengthening the regulatory ecosystem for rare diseases.

Pei Wang, PhD

Pei Wang, PhD is Senior Vice President of Clinical Development and CMC at Eureka Therapeutics. She has over 17 years of experience advancing T-cell therapies from early research through clinical development.

At Eureka, Dr. Wang directs clinical development and manufacturing strategy across multiple programs. She serves as Principal Investigator for the company's California Institute for Regenerative Medicine (CIRM)-funded ARYA-2 pediatric cancer initiative and has advanced programs that earned multiple FDA designations, including Regenerative Medicine Advanced Therapy, Fast Track, Rare Disease, and Orphan Drug. Her leadership has driven successful IND submissions, process improvements, and strategic collaborations with leading institutions such as Memorial Sloan Kettering Cancer Center, City of Hope, National Cancer Institute, Fred Hutchinson Cancer Center as well as industry partners such as Boehringer Ingelheim and Lyell Immunopharma. Dr. Wang is also a co-inventor on several Eureka patents.

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Lynne Yao, MD

Lynne Yao, MD, is the Director, Division of Pediatric and Maternal Health (DPMH) in the Office of New Drugs, Center for Drug Evaluation and Research, US Food and Drug Administration. Dr. Yao received a BS degree in Biology from Yale University, and an MD degree from the George Washington University School of Medicine. She is board certified in both Pediatrics and Pediatric Nephrology. She has been with the FDA since 2008 and has been DPMH Director since 2012. As DPMH Director, Dr. Yao oversees quality initiatives which promote and necessitate the study of drug and biological products in the pediatric population; and improve collection of data to support the safe use of drugs and biological products in pregnant and lactating individuals. She collaborates with numerous stakeholders both inside and outside of FDA to advance development of safe and effective therapies for children, and pregnant and lactating women.