CYSTINOSIS RESEARCH NETWORK

CYSTINOSIS SYMPOSIUM

A Rare Disease Model for Comprehensive Care

Speaker Bios

Andrea Atherton, MS, CGC

Andrea Atherton is a board-certified genetic counselor who currently works for Amgen as a Regional Medical Director in Ultra-Rare Medicines where her primary focus is centered around scientific education and dissemination of information on the medical management and treatment for Cystinosis. Andrea started her career over 20 years ago as a genetic counselor at Children's Mercy Hospital after graduating from the University of Texas Health Science Center with a master's degree in genetic counseling. While working as a genetic counselor, Andrea was extremely involved in the evaluation, diagnosis and management of pediatric and adult patients with inborn errors of metabolism, lysosomal storage disorders, mitochondrial conditions, neuromuscular disorders, neurogenetic diseases and became a huge advocate for newborn screening at the state and federal level. She served as a member of the Newborn Screening Translational Research Network steering committee and was recently appointed as a board member to RareKC in 2023. Andrea has published dozens of articles on inborn errors of metabolism, lysosomal storage disorders and newborn screening over the years with her most recent article focusing on reproductive health and family planning in Cystinosis.

Rachel Bishop, MD, MPH

Rachel Bishop, M.D., M.P.H., is a board-certified comprehensive ophthalmologist at Johns Hopkins where she serves as medical director of the Wilmer Frederick eye clinic. She is an expert in the ocular management of cystinosis. During her 14-year career at the National Institutes of Health (NIH), Dr. Bishop cared for the many patients with cystinosis who traveled to NIH to participate in pioneering clinical research studies there. Dr. Bishop's medical career began in the U.S. Army, where she served as chief of ophthalmology at Fort Hood, Texas. After transferring to the U.S. Public Health Service, she served as chief of eye consult services at the National Eye Institute (NEI) at NIH. She co-created and ran a state-of-the-art eye clinic in Liberia, West Africa, studying the ocular effects of Ebola. She also served as lead clinical communicator for NEI, providing hundreds of interviews to print, radio, and network media sources.

Separate from her clinical ophthalmology practice, Dr. Bishop is a certified executive coach. She runs a leadership coaching program at Johns Hopkins, and also serves clients from industry, government, and medicine in her private coaching practice.

Dr. Bishop received a BS in Economics from MIT, a medical degree from the University of Pennsylvania, a master of public health from Johns Hopkins, and an executive certificate in leadership coaching from Georgetown University.

Dr. Bishop enjoys trail runs with her dogs, skiing, reading, and hanging around the fire pit with friends. She is married to a toolbelt-wearing writer from the Midwest. Their blended family includes two girls, two boys, and two dogs.

Stephanie Cherqui, PhD

Stephanie Cherqui is Professor in the Department of Pediatrics, Division of Genetics at the University of California San Diego. Her laboratory primarily centers on two research areas: the development of hematopoietic stem cell and gene therapy-based treatments for genetic disorders, and investigating the mechanism behind hematopoietic stem cell-mediated tissue repair. She demonstrated that multisystemic disorders could be rescued by hematopoietic stem cell transplantation even if the protein involved is an intracellular membrane protein like in cystinosis. Her work led to the first-

Speaker Bios

Stephanie Cherqui, PhD, *continued*

in-human HSPC gene therapy clinical trial for cystinosis. She is now applying this strategy to other multisystemic disorders including the neuromuscular degenerative disease, Friedreich's ataxia.

Dr. Cherqui is the Chair of the Cystinosis Stem Cell and Gene Therapy Consortium, and a member of the American Society of Gene and Cell Therapy (ASCGT) Gene and Cell Therapy of Genetic and Metabolic Diseases committee. She is also a member of the Scientific Review Board of the Cystinosis Research Foundation and a Scientific Council member for the Cure Cystinosis International Registry (CCIR). Her research is mainly funded by grants from the National Institute of Health (NIH), California Institute of Regenerative Medicine (CIRM), the Cystinosis Research Foundation, and Friedreich's ataxia Research Alliance (FARA).

Jonathan Dicks, MSN-FNP

Jonathan's professional journey began in the high-pressure environment of the Emergency Department at UCMC, where he honed his skills as a critical care RN for over 4 years. Yet, it was a serendipitous encounter with an attending physician in Emergency Medicine that sparked not only a deep friendship but also a shared vision for redefining fitness.

Transitioning his career path, Jonathan attained his MSN-FNP from the University of Cincinnati, a testament to his unwavering commitment to advancement. In 2020, he co-founded Revive Strength and Wellness, a trailblazing personal training facility in Cincinnati, Ohio, dedicated to innovative fitness solutions.

But beyond his professional achievements, Jonathan's heart lies in advocacy, particularly in the realm of rare diseases. His personal journey as a father to three children Finn, Teddy and Eleanor, who bravely battles the rare condition cystinosis, has ignited his fervor for championing their cause.

From attending his first Cystinosis Research Network family conference in 2019 to representing Ohio at Rare Disease Week on Capitol Hill, Jonathan's advocacy knows no bounds. His dedication led to his election as Vice President of Development for CRN, and his subsequent election as President, guiding the organization with passion and purpose.

Amidst his advocacy endeavors, Jonathan finds solace in the arts, drawing from his background as a classically trained thespian. While medicine and fitness remain integral parts of his life, it's his advocacy work that truly defines his purpose, driving him to create positive change for those in need.

Maya Doyle, MSW, PhD, LCSW

Maya Doyle (MSW, PhD) is a social worker, educator, and rare disease researcher-advocate who lives in New York and teaches in the department of Social Work/School of Health Sciences and the Frank Netter School of Medicine at Quinnipiac University in Connecticut. She has been the coordinator for the kidney camp program at Frost Valley YMCA for more than 20 years. Maya is currently an Interprofessional Education fellow at QU and mentors the annual Rare Disease Day events at Quinnipiac. She completed her doctoral work with a study on emerging adulthood and healthcare transitioning for young people with cystinosis and has published articles and chapters related to the experience of illness, transition to adult care, and genetics. She has been a speaker at many patient/family advocacy meetings, created educational resources for patients and caregivers, and hosted transitioning workshops for teens and their parents. Maya is a professional advisor to the Cystinosis Research Network and a mentor to the Adult Leadership Advisory Board.

Speaker Bios

James Drakakis, DO

Dr. James Drakakis, DO, is a distinguished nephrologist and Clinical Assistant Professor in the Department of Medicine at NYU Langone Hospital – Long Island. His journey in medicine began with a Bachelor of Arts in Biology and Psychological and Brain Sciences from The Johns Hopkins University in 2003, followed by a Doctor of Osteopathic Medicine degree from Lake Erie College in 2007. He completed his residency in Categorical Internal Medicine and fellowship in Nephrology at NYU Langone Hospital – Long Island, where he has remained a dedicated faculty member.

Throughout his career, Dr. Drakakis has been recognized for his exceptional contributions to medicine and education. He has received numerous accolades, including the Internal Medicine Attending of the Year award in 2015 and the Fellow of the Year award in 2012. His commitment to excellence was evident early in his career, as he earned the Categorical Internal Medicine Resident of the Year award consecutively from 2008 to 2010. Dr. Drakakis is board certified in both Internal Medicine and Nephrology and holds an active physician license in New York State since 2010. His professional affiliations include the American Society of Nephrology, where he has been an active member since 2010. Dr. Drakakis's research interests are reflected in his numerous publications and presentations on nephrology and internal medicine topics. He has contributed to studies on acute tubular necrosis, hyponatremia, and renal transplant complications, among others. His research work has been published in reputable journals such as the American Journal of Medical Sciences and Kidney International Reports. Additionally, he has participated in various clinical trials aimed at improving treatment outcomes for patients with chronic kidney disease and other nephrological conditions.

Bill Gahl, MD, PhD

Dr. William A. Gahl graduated from the Massachusetts Institute of Technology and earned his M.D. and Ph.D. from the University of Wisconsin. He served as pediatric resident and chief resident at the University of Wisconsin hospitals and completed clinical genetics and clinical biochemical genetics fellowships at the NIH. Dr. Gahl elucidated the basic defects in cystinosis and Salla disease and helped bring cysteamine to new drug approval by the Food and Drug Administration as the treatment for cystinosis. He has published over 650 papers, reviews, book chapters, and editorials, trained 42 biochemical geneticists and cultivated international experts in Hermansky-Pudlak syndrome, alkaptonuria, Oculocerebrorenal Syndrome of Lowe, Menkes disease, Congenital Disorders of Glycosylation, Griscelli Syndrome, Gray Platelet Syndrome, Joubert Syndrome, polycystic kidney disease and other ciliopathies, Hutchinson-Gilford Progeria, GNE myopathy, oculocutaneous albinism, sialuria, and free sialic acid storage disorders. His group identified the genes responsible for Hartnup disease, Gray Platelet Syndrome, two types of renal Fanconi syndrome, 3-methylglutaconic aciduria type III, a new neutrophil defect, and many other disorders. In 2008, he established the NIH Undiagnosed Diseases Program (UDP), which has made more than 350 rare disease diagnoses and discovered 30 new genetic diseases. Dr. Gahl expanded the UDP to a national Undiagnosed Diseases Network and a worldwide Undiagnosed Diseases Network International. He established American Board of Medical Specialties certification for medical biochemical genetics. 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, the EURORDIS Lifetime Achievement Award, and numerous other awards. In 2019, he was elected to the National Academy of Medicine.

Ashley Gefen, MD, MS

Ashley Gefen, MD, MS, is a pediatric nephrologist at Phoenix Children's Hospital specializing in genetic kidney disease, tubular disorders and kidney stone disease.

After earning her medical degree at Washington University School of Medicine in St. Louis, Dr. Gefen completed her pediatrics residency at New York University Hospitals/Hassenfeld Children's Hospital/Bellevue Hospital in New York. She

Speaker Bios

Ashley Gefen, MD, MS, *continued*

completed a pediatric nephrology fellowship at Northwell Health/Cohen Children's Medical Center in Queens, New York, and earned a master's degree in clinical research at Drexel University College of Medicine in Philadelphia.

Dr. Gefen is certified by the American Board of Pediatrics. She's a member of numerous professional organizations, including the American Society of Pediatric Nephrology and the National Kidney Foundation. Dr. Gefen also co-chairs the tubular/genetic/congenital kidney disease committee of the Pediatric Nephrology Research Consortium.

Cybele Ghossein, MD

Cybele Ghossein, MD is a Professor of Medicine in the Division of Nephrology and the Vice Chair of Academic and Faculty Affairs in the Department of Medicine at Northwestern University. She has devoted her career to the care of patients with kidney disease with a special interest in young adults transitioning from pediatric to adult kidney care. She previously was the Associate Chief of Clinical Operations for the Division of Nephrology and in that role, had an interest in the clinical operations and programing of the division. During her tenure she established a chronic kidney disease clinic as well as a pediatric to adult transition program for young adults with kidney disease. In her role as Vice Chair in the Department of Medicine, she has worked to implement a variety of professional development programs including a peer faculty coaching program, a faculty development lecture series and a Women in Medicine yearly conference.

Ladan Golestaneh, MD, MS

Dr. Golestaneh holds various administrative positions within the Division of Nephrology and Department of Medicine including service as Medical Director of two large hemodialysis centers and director of the largest home dialysis program in the Bronx. Her research interests include acute kidney injury (AKI) and the cardiorenal and hepatorenal syndromes. She has also published on the healthcare disparities affecting patients with kidney disease. Her recent research interests include care delivery models for end-stage kidney disease patients, a project in which she collaborates with the Montefiore CMO. She is lead PI for an NIH funded study examining the effects of peer mentorship in patients receiving hemodialysis.

Paul Goodyer, MD

Dr. Goodyer is the Professor of Pediatrics and Human Genetics at McGill University and a Pediatric Nephrologist at the Montreal Children's Hospital, where he directs the Hereditary Renal Disease Clinic. He provides care for children with a wide variety of rare genetic kidney diseases, including Cystinosis. We discovered that this disease is comparatively common in Quebec because of a specific CTNSW138X nonsense mutation transferred to the French Canadian community from Ireland 2-3 hundred years ago. His young cystinosis patients participated in the North American trial of cysteamine sponsored by the National Institutes of Health and have been followed during the transition to adulthood.

Paul Goodyer, MD, *continued*

For many years he has directed a research lab focused on the molecular basis of hereditary renal disease and the development of novel treatments for these conditions. His lab has made significant contributions to the role of PAX2 mutations in renal hypoplasia, the function of WT1 in hereditary Wilms tumor, the genetics of cystinuria, nephrogenic diabetes insipidus, Dent disease, X-linked hypophosphatemic rickets and cystinosis. They examined the WNT signaling pathway in the pathogenesis of ADPKD cysts during embryonic life.

More recently, his lab showed that a new generation of non-toxic aminoglycosides (developed by Eloxx Pharmaceuticals) permits translational readthrough of the CTNSW138X nonsense mutation common among French Canadian cystinosis patients. Based on experimental data, they initiated a clinical trial of an Eloxx compound for the treatment of cystinosis patients with the CTNSW138X mutation (NCT04069260). Although the clinical trial was brought to a halt by the COVID pandemic, our preliminary data suggested that ELX-02 was safe and probably effective in lowering leukocyte cystine at doses above 1mg/kg.

Although unfortunate for the clinical trial, they think the COVID pandemic came with a silver lining. Following discussions with Moderna scientists, they were funded by the Cystinosis Research Foundation to investigate whether the mRNA/lipid nanoparticle technology used to generate COVID vaccines could be adapted for the treatment of cystinosis.

John M. Greally, MD, PhD

John Greally is a medical doctor, originally trained in pediatrics who went on the specialise in Genetics. He sees patients at Montefiore Medical Center in the Bronx, New York, focusing on the use of advanced genomic technologies to reveal the causes of rare diseases in his patients. He is Co-Director of the NORD-recognised New York Center for Rare Diseases, a Montefiore initiative recognised in 2023 as a Center of Excellence by the National Organization for Rare Diseases (NORD).

He is also a PhD scientist with expertise in genomics. His research program aims to understand how to use genomic information in diagnosing and managing patients with genetic conditions. He is an expert in epigenetics, how cells use the 97% of DNA that is not genes to cause cells to change how they function, and how DNA sequence changes in this under-explored majority of the genome can cause diseases. His group also studies how to use digital technologies to improve clinicians' ability to examine patients with rare diseases.

His lab research uses stem cell systems and computational biology to study human diseases. Working in the Bronx, his work includes a focus on health equity in genomics, specifically the delivery of medical genomic services to minoritised communities without exploitation, and without distracting from the non-genetic causes of health disparities.

Dr. Greally is Professor of Genetics and Pediatrics, and the Founding Director of the Center for Epigenomics at the Albert Einstein College of Medicine, with an Affiliate Membership at the New York Genome Center. He received his honours degree in Medicine from the National University of Ireland (NUI) in Galway, Ireland. He trained in pediatrics at the Children's Hospital of Pittsburgh and Clinical Genetics at Yale University.

He received his higher degrees in medicine (D.Med.) and science (Ph.D.) from the University of Galway in Ireland, and has been awarded a Fellowship of the American College of Medical Genetics (FACMG). He has over 200 peer-reviewed publications and is the recipient of numerous National Institutes of Health and other grant awards.

He is CEO and Founder of Latent Genomics Inc., a startup allowing genomic information to be used easily in patient care by all clinicians. His book entitled Epigenetics: History, Molecules and Diseases is due to be published in 2025.

Jack Greeley

Jack is 24 years old. He was born in Portland and moved to Chicago at one year old. He was diagnosed with Cystinosis shortly after moving to Chicago. He has not had a kidney transplant yet. Considering the fact that he has Cystinosis,

Speaker Bios

Jack Greeley, *continued*

his health has been good as of late. His biggest issues have been with his bones and specifically with his legs, ankles, and feet. He has had two major leg surgeries and wore considerable leg braces until college. He currently still wears inserts that aren't visible when wearing shoes. The past two years he has taken up hobbies including lifting weights, fitness, and nutrition as a major focus on health.

Larry Greenbaum, MD, PhD, FAAP

Larry Greenbaum, MD, PhD is Division Director of Pediatric Nephrology and the Bernard Marcus Professor of Pediatric Nephrology at the Emory School of Medicine and Children's Healthcare of Atlanta. He received his MD and PhD degrees from the Yale School of Medicine, and completed a residency in Pediatrics and a fellowship in Pediatric Nephrology at the UCLA School of Medicine. Dr. Greenbaum, an NIH funded investigator, conducts clinical research in a variety of areas in pediatric nephrology, including cystinosis, chronic kidney disease, and nephrotic syndrome. He co-edited the textbooks Practical Strategies in Pediatric Diagnosis and Therapy, Clinical Pediatric Nephrology and Pediatric Kidney Disease. Dr. Greenbaum has served as President of the American Society of Pediatric Nephrology and as the chair of the Executive Committee of the American Academy of Pediatrics Section on Nephrology. He is currently on the steering committee of the Pediatric Nephrology Research Consortium. He enjoys caring for patients with cystinosis and sees children and adults with cystinosis from throughout the United States.

Shannon Henderson, RN

Shannon Henderson is a 35-year-old wife, proud aunt, loving daughter, Sunday school teacher, adventure seeker, devoted friend, and she happens to have Cystinosis. In 2022, after years of being the patient in the hospital bed, she was inspired to become a registered nurse and work alongside professionals she's admired her entire life. She is thankful for the perseverance, compassion, attitude, grit, and outlook on life that has been strengthened by her experience with this chronic illness. Above all else, she cherishes her faith and relationships. This too has largely been influenced by a life with Cystinosis. She looks forward to connecting with new friends and old at this Symposium.

Frederick Kaskel, MD, PhD, FAAP, FASN

Dr. Kaskel is a translational investigator with expertise in basic renal physiology and research in glomerular disease and chronic kidney failure in children. He has considerable experience in leading national and international scientific activities and was a PI on the NIDDK Focal Segmental Glomerulosclerosis Clinical Trial, President of the 15th Scientific Congress of the International Pediatric Nephrology Association and former President of the American Society of Pediatric Nephrology. He served on the FDA Cardiovascular Renal Advisory Panel and currently on the Pediatric Advisory Panel, the Scientific Steering Committee of Nephcure and the Standing Renal Committee of the National Qualify Forum, and Chair of the Clinical Translational Science Awards Consortium Child Health Oversight Committee, and PI on a T32 Training Grant on Developmental Nephrology for the past 15 years as the former Chief of the Division of Pediatric Nephrology at Albert Einstein College of Medicine, Children's Hospital at Montefiore. As a faculty member of the Global Health Department at Einstein and have mentored medical undergraduates

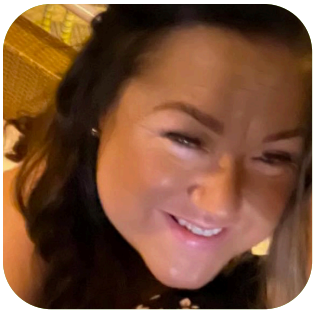
Frederick Kaskel, MD, PhD, FAAP, FASN, *continued*

in Peru, Nigeria, and Viet Nam as well as being a Visiting Professor at the Children's Hospital of Fudan University in Shanghai, the Children's Hospitals in Ho Chi Minh City, and Hanoi, the Philippine Pediatric Nephrology Association in Cebu, and lead our ISN Renal Sister Center with pediatric nephrology colleagues in Port Hartcourt, Nigeria. Dr. Kaskel has considerable experience in administering projects, leading multidisciplinary teams of investigators, and mentoring high school, university and medical undergraduates, and residents/fellows along their various career pathways, and implementing effective communication regarding time-lines for research development, data analyses, submission of new scientific information for presentation and publication, and maintaining budgetary adherence to funded investigations. As an Emeritus Chief of Nephrology, he is actively involved in these mentoring activities as well as organizing the historical perspectives in Pediatric Nephrology, the American Society of Pediatric Nephrology and the International Pediatric Nephrology Association.

Lesli King

Lesli King is the Director of Marketing at Leadiant Biosciences, Inc., where she has been an integral part of the team since 2001. With a career dedicated to healthcare, particularly rare diseases, Lesli brings extensive expertise in patient advocacy, market access, commercial operations, marketing, public relations, and product launches.

Lesli's leadership is marked by strategic vision, innovative thinking, and meaningful relationships, driving the company's mission to bringing novel therapies to market that transform the lives of those affected by rare diseases. For over 23 years, Lesli has been deeply involved with the cystinosis community, and is committed to making a difference in the lives of those affected by cystinosis.



Laura Krummenacker

Laura Krummenacker was diagnosed with Cystinosis 35 years ago. She has been a part of Dr. William Gahl's protocol at the NIH since she was three years old and is part of the first generation on two of the first FDA approved medications Cystagon and Cystaran. Laura was also under the care of Dr. Frederick Kaskel, her pediatric nephrologist and doctor who diagnosed her and was the lead in her care for 25 years, guiding her transition into the adult world of nephrology. She is now followed by Dr. James Drakakis and has developed a strong self-advocating relationship with him. Laura received her Associate Degree in Early Childhood Development and has worked in after school child care programs and was a receptionist for a large real estate company and a busy hair salon. While Laura has overcome many obstacles since she was diagnosed 35 years ago, none was more devastating than a diagnosis of breast cancer almost seven years ago. Laura faced that unthinkable diagnosis with courage and strength and was consistently surrounded by the love of her family and friends. She had to make some very tough choices and held herself with the same strength and courage that she still maintains. A testament to her tenacity and the great outcome she had was due to the phenomenal medical group of specialists who communicated constantly and the fact that after surgery and chemotherapy treatment she was able to maintain perfect kidney function and will soon be celebrating 25 years post-transplant. Laura continues to handle every "bump in the road" with grace, humor and a true fighting spirit! She continues to be an inspiration to all of her family and friends, and her biggest cheerleader is her mom!

Marybeth Krummenacker

Marybeth Krummenacker graduated from State University of New York at Farmingdale with an Associate Degree in business and for many years worked in the health insurance industry. For the last 27 years she has been the Assistant to the Public Information Officer of her local township. Marybeth has been a volunteer for various community organizations

Speaker Bios

Marybeth Krummenacker, *continued*

and has said that her 15 years of PTA experience is where she learned the “art” of advocating. Marybeth completed six years as a Board Member of NORD, serving on the Executive, Advocacy and Membership Committees. She is also one of the founding members of the Cystinosis Research Network and is serving a second term as the Vice President of Education and Awareness. Marybeth’s daughter, Laura was diagnosed with Cystinosis 35 years ago and this firsthand knowledge has allowed her to share the real stories of exactly what it means to live with and manage a rare disease.

France Lebel, MSc

Senior Director, Medical Affairs Canada & Metabolic North America, and Lead, Market Access Canada. Recordati Rare Diseases Inc.

France graduated from Université Laval with a Master of Science, Experimental Medicine Genetics Epidemiology in 1992. It was the start of her passion for rare genetic disorders working on Tyrosinemia Type-1. Her career in the pharmaceutical industry spans over three decades working for Jansen, Pharmacia, BMS with different roles in commercial affairs, R&D and the last 20 years in medical affairs at Actelion, InterMune, Eisai and more recently Recordati Rare Diseases Inc. In her current role, France is medical counsel to Recordati Rare Diseases Marketing, Regulatory, and Legal functions. She coordinates and directs medical activities, including the MSL team, Medical Information, Medical Communications, medical review of promotional materials, Health Outcomes Research, and medical support of Commercial training activities.

Rare diseases have been at the center of most of her roles, and disease awareness, medical education and communication are her focus and passion. At Recordati Rare Diseases we are “Focused on The Few”.

Chelsea Meschke, LMSW

Chelsea recieved her bachelors in Social Work in 2010, after years working in Lansing with the homeless youth population, she set out to obtain her master’s degree in Social Work from Grand Valley State University. From there, she has spent my career working with the elderly in Hospice and care management of those with multiple chronic illness. Meschke started with the Cystinosis Research Network in 2023 on the Board of Directors. Her son, Jaxon was diagnosed with Cystinosis in 2018 and her son Myles in 2021. Her passion is to educate, advocate, and support all those that are affected by chronic illness and need a voice to help them be heard.

Sophie Molholm, PhD

Dr. Sophie Molholm is a Cognitive Neuroscientist at the Albert Einstein College of Medicine, with appointments in the departments of Pediatrics, Neuroscience and Psychiatry and Behavioral Sciences. She uses multimodal brain imaging, psychophysics, and standardized cognitive and clinical assessments to probe the brain processes underlying perception and cognition in neurodevelopmental and neuropsychiatric conditions such as autism and schizophrenia as well as in rare genetic disorders including cystinosis, 22q11.2 deletion syndrome and Rett syndrome. Her research is directed at gaining mechanistic understanding of the neurobiological processes underlying these conditions and developing functional biomarkers. In addition to running her lab, she serves as Director of the NIH

Sophie Molholm, PhD, *continued*

funded Rose F. Kennedy Intellectual and Developmental Research Center. Her research has been continuously funded by federal and foundation grants for the past 20 years.

Christina Morris

Christina is 35 years old. She currently lives in Charleston, SC with her boyfriend and two dogs both named Lola (they both had dogs named Lola when they met). She works as a nanny and is currently finishing up her psychology degree. In her free time, Christina enjoys going to concerts, attending art classes, and drinking coffee. She is a member of The Adult Leadership Advisory Board (ALAB) under the Cystinosis Research Network and helps advocate for those living with Cystinosis.

Kristina Sevel, MSN, RN

Kristina Sevel is a registered nurse and Vice President of Research for the Cystinosis Research Network. With a passion for advancing medical knowledge and improving patient outcomes, Kristina's work focuses on driving research initiatives to combat cystinosis. As a devoted parent of a five-year-old with cystinosis, Kristina brings a personal perspective to her advocacy and research efforts, striving to make a difference in the lives of individuals and families affected by this condition.

Reza Seyedsadjadi, MD

Reza Sadjadi is an assistant professor of neurology and neuromuscular neurologist with research interests in disease outcome measures, biomarkers and clinical trial readiness in neuromuscular diseases and neuromuscular complications of rare degenerative hereditary processes. He has been involved in developing and validating multiple clinician and patient reported outcome measures using modern psychometrics and item response theory models. He is leading a series of clinical trial readiness studies of distal myopathy and dysphagia in nephropathic cystinosis; clinical, neurophysiological and pathological characterization of myopathy and dysphagia in adults with nephropathic cystinosis; evaluation for inherent muscle resilience and regenerative capacity.

Melinda S. Sharkey, MD

Melinda S. Sharkey, MD, is an Attending Orthopedic Surgeon at Montefiore. She specializes in the operative and nonoperative treatment of general pediatric and adolescent musculoskeletal problems, including traumatic injuries, as well as congenital and developmental disorders. Her practice particularly focuses on the surgical treatment of bone deformities, foot deformities, limb length discrepancies, and metabolic bone diseases.

Dr. Sharkey received her Bachelor of Arts in Biochemistry in 1999 at Wellesley College. In 2004, she received her Doctor of Medicine at the University of Chicago Pritzker School of Medicine. Dr. Sharkey completed an internship in General Surgery and a residency in

Speaker Bios

Melinda S. Sharkey, MD, *continued*

Orthopedic Surgery at the University of California San Francisco from 2004 to 2009. She then completed a fellowship in Pediatric Orthopedic Surgery at the Children's Hospital of Philadelphia.

Herberth Sigler

Herberth R. Sigler is a civil engineer who graduated from The Bolivian Catholic University with a Bachelor's Degree. He moved to New York City and earned a Master's in Civil Engineering from The City College of New York. He is a few weeks away from earning a post-graduate certificate in Project Management from Harvard University. He has over 20 years of professional experience, including 12 years in the commercial electrical construction industry in NYC. Herberth is a board member of Cystinosis Research Network (CRN) and the IRB at the NIH. He firmly believes that caregivers in the cystinosis community will go beyond the extra mile to find proper treatment for their loved ones. His 15-year-old daughter, Martina, has Cystinosis, and Herberth and his wife, Jessica, are her caregivers. They live in Queens, New York.

Adam C. Stein, MD

Dr. Stein specializes in the evaluation and treatment of a variety of gastrointestinal and nutritional disorders that involve the small bowel, including Crohn's disease, intestinal failure (such as short bowel syndrome), celiac disease, enteropathy, malabsorption, cystic fibrosis, cystinosis, obscure gastrointestinal bleeding, and functional bowel disorders. Dr. Stein is also the Director of Nutrition Support, leading a multidisciplinary team that manages nutrition through the vein (parenteral nutrition, or "TPN") as well as tube feeding.

Jess Thoene, MD

Dr. Jess Thoene is currently an Active Emeritus Professor of Pediatrics in the Division of Pediatric Genetics, Metabolism & Genomic Medicine in the Department of Pediatrics at the University of Michigan in Ann Arbor. He was Director of the Pediatric Clinical Research Center at the University of Michigan and is a past Joseph P. Kennedy, Jr. Foundation Fellow. He serves as a consultant to the pharmaceutical industry and was a member of the Board of Directors of Copley Pharmaceuticals, Inc. in Boston. He is past Chairman of the Board of Directors of The National Organization for Rare Disorders,and was Chairman of the National Commission on Orphan Diseases. He has authored more than 100 articles in the peer-reviewed literature on inborn errors of metabolism, published 3 medical reference texts, holds six U.S. patents, with 1 pending, and is certified in Pediatrics and Clinical Biochemical Genetics. He is a member of the American Society of Clinical Investigation, the American Society of Human Genetics, the American Pediatric Society, the Society for Pediatric Research, the Society for Inherited Metabolic Disorders (Past Counselor), and the Mid-west Society for Pediatric Research (Past-President). From 2000- 2006 he was Director of the Hayward Genetics Center at Tulane University School of Medicine and Karen Gore Professor of Pediatrics at that institution. He currently follows cystinosis patients at the University of Michigan Biochemical Genetics Clinic and pursues clinical research.

Speaker Bios

Eileen Wall

Eileen Wall is the Senior Manager of Medical Affairs at Leadiant Biosciences, Inc. She holds a master's degree in public health in epidemiology from the University of Massachusetts and received her Bachelor of Science in Nursing from Wayne State University, Detroit.

She has 15 years of Medical Affairs experience in the biopharmaceutical industry, primarily in rare diseases.

Melissa P. Wasserstein, MD

Chief, Division of Genetics, Children's Hospital at Montefiore
Professor of Pediatrics, Albert Einstein College of Medicine

Joshua Zaritsky, MD, PhD

Dr. Joshua Zaritsky is a Pediatric Nephrologist who has been in practice for over 15 years. He received his Medical and PhD Degree from Stanford University School of Medicine and did his fellowship in Pediatric Nephrology at UCLA. He is currently a pediatric nephrologist at Phoenix Children's hospital in Phoenix, Arizona. He has a history of basic and clinical research in iron metabolism. Dr. Zaritsky has recently focused on the treatment of rare renal diseases such as XLH, HPP, FSGS, cystinosis, cystinuria and BBS.

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