2014 NATIONAL UREA CYCLE DISORDERS FOUNDATION ANNUAL FAMILY CONFERENCE

Sample of Invited Speaker/Moderator Bios:

Brendan Lee, M.D., Ph.D.
Principal Investigator, Urea Cycle Disorders Consortium (UCDC)
Investigator, Howard Hughes Medical Institute
Robert and Janice McNair Endowed Chair in Molecular and Human Genetics
Professor, Department of Human & Medical Genetics
Baylor College of Medicine, Houston, Texas

Dr. Lee is a leading UCD researcher and authority on clinical management of urea cycle disorders. Dr. Lee's research in molecular genetics and urea cycle disorders has been published in leading medical journals. He has received numerous honors and awards, including recognition as one of the nation's top physician-scientists via his appointment as an Investigator to the prestigious Howard Hughes Medical Institute, serving as one of an elite group of 12 researchers to improve the translation of basic science discoveries into enhanced treatments for patients. He serves as a member of numerous academic and research study committees, including National Institutes of Health Gene Therapy and Inborn Errors, Graduate Programs in Genetics, Cell and Molecular Biology at Baylor, and as Director of Medical Students Research Track at Baylor. Dr. Lee is a collaborator in the Urea Cycle Disorders Consortium as primary investigator (PI) for several important UCD research projects at the Baylor site. His pioneering work on the role of nitric oxide in the urea cycle has made a significant contribution to general science and human health, published in Nature Medicine in 2012. Dr. Lee is dedicated to expanding the understanding of the pathophysiology of UCDs and development of new treatments. He has participated in and led both investigator-initiated and industry-sponsored interventional studies including the design of Phase II and III studies of a novel ammonia scavenger glyceryl-triphenylbutyrate in urea cycle patients, and combinatorial phenylbutyrate/arginine treatment in patients with argininosuccinic aciduria, He serves on many professional boards, and is an invaluable member of the NUCDF Medical Advisory Board.

Mark L. Batshaw, MD
Principal Investigator, Urea Cycle Disorders Consortium (UCDC)
Executive Vice President, Chief Academic Officer, Physician-in-Chief
Director, Children's Research Institute
Children's National Medical Center, Washington DC
Professor and Chairman, Pediatrics
Associate Dean, Academic Affairs
George Washington University School of Medicine and Health Sciences

Dr. Batshaw is Executive Vice President, Physician-in-Chief, Chief Academic Officer and Director of CNMC's Children's Research Institute, as well as Chair of Academic Medicine and Professor and Chairman of Pediatrics at George Washington University School of Medicine and Health Sciences. Dr. Batshaw has spent more than 25 years as a developmental pediatrician dedicated to providing the best care for children with disabilities. He has authored articles and books on children with disabilities and is a pioneer in the field of urea cycle disorders. As a developmental pediatrician, the focus of his work is to improve cognitive outcome in these devastating disorders. His pioneering research with urea cycle enzyme deficiencies led to his role as co-developer of the first treatment for the disorders, sodium benzoate, followed by sodium phenylacetate. He co-directs the Urea Cycle Disorders Research Consortium with co-principal investigators Mendel Tuchman, MD, Marshall Summar, MD and Cynthia Le Mons (National Urea Cycle Disorders Foundation). The UCD Consortium

is a National Institutes of Health (NIH) funded research collaborative that is conducting a longitudinal study of urea cycle disorders and clinical trials for novel treatments at 15 international academic and clinical research centers. Dr. Batshaw has held innumerable posts and advisory positions within academic, medical and family communities and has served as a member of the NUCDF Medical Advisory Board for over two decades.

Andrea Gropman, M.D.
Principal Investigator, Urea Cycle Disorders Research Consortium (UCDC)
Chief, Division of Neurodevelopmental Disabilities and Neurogenics
Principal Investigator, Children's Research Institute
Children's National Medical Center, Washington DC

Dr. Gropman is a rare combination – a child neurologist and clinical geneticist. After completing her residency training in Pediatrics at Johns Hopkins Hospital, she completed fellowships in neurology and Child Neurology at George Washington University and the Children's National Medical Center, and clinical genetics at the National Human Genome Research Institute. This was followed by a position as a senior staff fellow in the Intramural Research Program at the National Institutes of Neurological Disorders and Stroke (NINDS) at the National Institutes of Health (NIH) in Bethesda, MD, where she was funded by a Howard Hughes Medical Institute physician postdoctoral award. During this time, she was actively involved in basic research, in the study of genetic pathways involved in neuronal migration disorders, and participated as coinvestigator and/or consultant for a number of clinical protocols in the NIH clinical center with focus on neurologic phenotypes in neurometabolic and genetic disorders. She directed the neurogenetics clinic at the Children's National Medical Center. In August of 2003, she joined the pediatrics and neurology faculty at Georgetown University. In August of 2006, she joined the neurology department of Children's National Medical Center. Her research interest focuses on establishing biomarkers of neurological injury in patients with inborn errors of metabolism using specialized neuroimaging modalities at CMFI and in collaboration with researchers at the California Institute of Technology and the Kennedy Krieger Institute in Baltimore, MD. Her work as an investigator for the Urea Cycle Disorders Consortium has resulted in pioneering research studies using these modalities in OTC deficiency. She hopes to use these biomarkers to better characterize and understand the mechanisms of neurologic injury in urea cycle disorders and to accelerate the development of neurotherapeutics to protect against or prevent brain injury in UCD. Dr. Gropman is a committed advocate, serving on the advisory boards of several rare disease organizations, including our NUCDF Medical Advisory Board.

Peter J. McGuire, M.S., M.D.
Genetic Disease Research Branch
Natoinal Human Genome Research Institute
National Institutes of Health, Bethesda, Maryland

Dr. is a Physician-Scientist at the National Human Genome Research Institute (NHGRI) in Bethesda and Principal Investigator for the NIH Urea Cycle Disorders Nutrition and Immunity Study. Dr. McGuire received his medical degree with honors from the Royal College of Surgeons in Ireland. He completed a combined Pediatrics/Medical Genetics Residency and Biochemical Genetics Fellowship at Mount Sinai Medical Center. After completing residency, he stayed on at Mount Sinai as an Assistant Professor in the Department of Genetics and Genomic Sciences and as Attending Physician in the Program for Inherited Metabolic Disorders. As a recipient of the 2008 Genzyme/American College of Medical Genetics Fellowship in Biochemical Genetics and Children's Health Research Center Scholar (2008-2010), Dr. McGuire's research focused on the areas of oxidative stress and inflammation in inborn errors of metabolism, and urea cycle disorders and respiratory viral infection. In 2011, Dr. McGuire

was accepted into the Physician-Scientist Development Program at NHGRI for his proposal on the relationship between urea cycle disorders, nutrition and immunity. This proposal formed the basis for the NIH UNI Study (Urea cycle Disorders, Nutrition and Immunity), as well as translational work on urea cycle disorders performed in his laboratory. He is currently working on a mouse model that allows him to study immune system function in inborn errors of metabolism.

Debra Geary-Hook, M.S., R.D., M.P.H.
Metabolic Dietician
Children's Hospital of Los Angeles, California

Ms. Hook served for many years as metabolic dietician at Loma Linda Children's Hospital in San Bernardino County, CA and metabolic dietitian and clinic coordinator for the UC Irvine Metabolic Team Center in Irvine, California. She is currently metabolic dietician at Miller Children's Hospital in Long Beach, CA and the Department of Gastroenterology at Children's Hospital Los Angeles. She received Bachelor's degrees from Cal State University Chico in Nutrition and Social Psychology, and holds Master's degrees in nutrition and health education from Loma Linda University. Ms. Hook is also certified in nutrition support (CNSD), pediatric nutrition (CSP), health education (CHES) and lactation education (CLE), and has lectured on nutrition and health education at Cal Poly Pomona. Ms. Hook has the distinction of being named Region Five Nutrition Educator of the Year by the American Dietetic Association. She has many years of experience managing children with urea cycle disorders. She is always a favorite of our families when she speaks at NUCDF conferences, is a contributing author to our newsletters, and will be leading new research studies on the role of nutrition in outcomes for urea cycle disorders. Ms. Hook serves on the Education and Technology committees of Genetic Metabolic Dieticians International, and is a valuable member of our NUCDF Medical Advisory Board. "Deb" generously devotes volunteer time to assist our UCD families and their nutritionists with difficult management issues.

Susan Waisbren, Ph.D.
Associate Professor of Psychology, Harvard Medical School
Psychologist, Metabolic Service,
Children's Hospital, Boston, Massachusetts

Dr. Waisbren is the lead psychologist for the Urea Cycle Disorders Consortium, and psychologist at the Clinic for Inborn Errors of Metabolism and Phenylketonuria (PKU) in the Developmental Evaluation Clinic and the Division of Genetics at the Children's Hospital, Boston. Dr. Waisbren received her Ph.D. in Clinical Psychology from the University of California, Berkeley. Dr. Waisbren directs a research program on the behavioral and developmental aspects of phenylketonuria and other inborn errors of metabolism. She has been a leader in developing national and international networks to address the needs of youngsters and their families with inborn errors of metabolism. Dr. Waisbren has authored and coauthored many articles in publications such as the Journal of the American Medical Association (JAMA), Molecular Genetics and Metabolism, American Family Physician, and the New England Journal of Medicine. She is also a member of numerous professional organizations, including the American Psychological Association and the Society for Inherited Metabolic Disorders. In addition, Dr. Waisbren is the founding director of the New England Consortium of Metabolic Programs and is on the Board of the New England Connection for PKU and Allied Disorders. She recently organized the Genetics and Metabolism Psychology Network, which will bring psychologists interested in metabolic and genetic disorders together, with the goal of better describing the psychological aspects of these conditions, establishing guidelines for a core assessment battery, and developing educational tools for families and teachers.

Sandesh Chakravarthy Sreenath Nagamani, MBBS, M.D.
Assistant Professor, Department of Molecular and Human Genetics
Baylor College of Medicine, Houston, Texas

Dr. Nagamani is board-certified in both internal medicine and medical and clinical genetics. He received his MD from the University of Mysore in Karnataka, India, and completed internships and residencies at Gandhi Medical College in India, the Cleveland Clinic in OH, and at Baylor. Dr. Nagamani is a favorite of UCD families being followed in the Baylor Genetics Clinic. He was the recipient of the 2011 National Urea Disorders Foundation (NUCDF) Fellowship Award to continue pioneering research on nitric oxide (NO) in ASA deficiency started in Dr. Lee's lab by two previous NUCDF Fellowship Awardees in 2009 and 2010, Dr. Ayelet Erez and Dr. Oleg Shchelochkov. Dr. Nagamani's landmark work with his colleagues was recently published in the prestigious scientific journal, Nature Genetics, reporting the critical implications of the study on general science and human health. He was recently awarded the Doris Duke Clinical Scientist Development Award for his work pertaining to the nitric oxide in ASA clinical trial.

Renata Gallagher, M.D., Ph.D.
Principal Investigator, Urea Cycle Disorders Research Consortium
Assistant Professor, Pediatrics, Department of Clinical and Medical Genetics
University of Colorado School of Medicine
Children's Hospital Colorado, Aurora, Colorado

Dr. Gallagher specializes in clinical care of patients with urea cycle disorders and other inborn errors of metabolism at Children's Hospital Colorado. She has been at Children's Colorado since 2006. She trained in Genetics and Biochemical Genetics at Stanford Medical Center in California, and trained in Pediatrics in Seattle at the University of Washington. Dr. Gallagher is the local site Principal Investigator for the Urea Cycle Disorders Longitudinal Study at Children's Colorado, one of the studies of the Urea Cycle Disorders Consortium. As a result of her work with the Consortium she has been able to pursue focused research in urea cycle defects. She is currently investigating liver abnormalities in urea cycle disorders.

James Bartley, M.D., Ph.D.
Medical Director, California Children's Services, Metabolic Specialty Care Center, Miller Children's Hospital, Long Beach CA

Dr. Bartley's educational journey led him from Kansas State University at Emporia, to the Department of Biochemistry at the University of Iowa, to the University of Kansas School of Medicine, to an NIH traineeship at the University of California San Francisco, after which he went to the Division of Medical Genetics, Department of Pediatrics, University of Iowa School of Medicine. Dr. Bartley moved to California in 1988, where he has served as Associate Clinical Professor of Pediatrics and Medical Genetics at Loma Linda University Children's Hospital, UC Irvine Metabolic Team Center and Director of the Metabolic Center at Miller Children's Hospital in Long Beach. Dr. Bartley provides care for metabolic and genetic patients at Loma Linda University Children's Hospital, Miller Children's Hospital in Long Beach, and Children's Hospital of Los Angeles. He has been a tireless advocate for children with metabolic disorders in California and throughout the US, being involved in newborn screening, medical foods and formula legislation, and serving on state and regional genetics collaboratives.