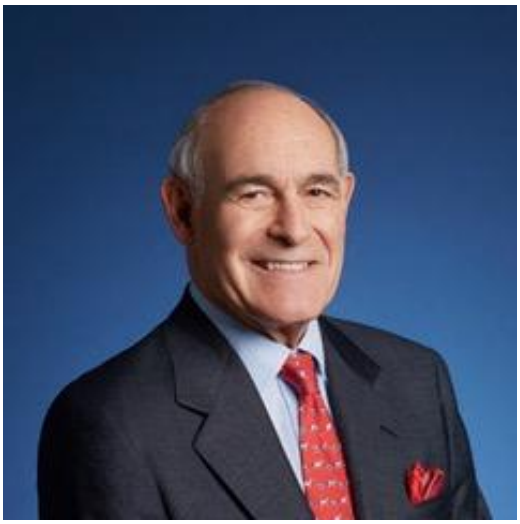
ISMRD'S 2025 INTERNATIONAL FORUM SPEAKERS

Robert J Desnick, M.D., Ph.D., D.Sc. (Hon)



Robert J Desnick is Dean for Genetics and Genomic Medicine Emeritus and Professor and Chairman Emeritus of the Department of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai, Mount Sinai Health System, New York, NY, USA. In 1977, he joined the faculty as the Arthur J and Nellie Z. Cohen Professor of Pediatrics and Genetics, and Chief of Medical and Molecular Genetics. From 1993–2011, he was the Inaugural Chairman of Sinai's Department of Genetics and Genomic Sciences. In 2011, he was appointed Dean for Genetics and Genomic Medicine. (2021-2021)

Dr Desnick's research interests include genomics, pharmacogenetics, personalized medicine and the delineation and treatment of lysosomal storage diseases (LSDs) and the inherited porphyrias. His basic, translational, and clinical research have led to the development of enzyme replacement therapy (ERT) for Fabry disease (agalsidase beta, EMA/FDA approved 2001/2003) and Nieman-Pick disease A/B (olipidase alfa, FDA/EMA 2021), oral pharmacologic chaperone therapy for Fabry disease (Migalastat, FDA/EMA 2017) and RNA interference therapy for the four Acute hepatic Porphyrias (FDA/EMA 2019/2020). He co-founded Amicus Therapeutics (NASDAQ: FOLD) and served as the SAC Chair for Synageva Biopharma (NASDAQ:GEVA) during the development of ERT for Lysosomal Acid Lipase Deficiency (Seblipase alfa, FDA/EMA 2015) and served as the SAB Chair of Kiniska Pharmaceuticals (NASDAQ: KNSA)

He has published >620 research articles, >265 chapters in text books, and 10 edited books. He is board-certified in Clinical, Biochemical, and Molecular Genetics and is a Fellow of the American Academy of Pediatrics. He is an elected member of numerous medical/scientific societies including the Society of Pediatric Research, American Pediatric Society, American Society for Clinical Investigation and American Association of Physicians. He is a past Director of the American Board of Medical Genetics and Genomics (ABMGG), a Founding Diplomat of the American College of Genetics (ACMG) and a member of the ACMG Foundation. He was Chair of the Association of American Medical Colleges (AAMC) and recipient of the AAMC Distinguished Service Award (2010). He received the NORD Rare Impact Award (2017). a Lifetime Achievement Award from the University of Minnesota (UoM, 2018) and the UoM Distinguished Alumni Award (2019). He is an elected Fellow of the American Academy for the Advancement of Science, member of the National Academies of Sciences, Engineering and Medicine, and of the National Academy of Inventors).

Patricia Dickson, MD

Dr. Patricia I. Dickson is a distinguished geneticist and pediatrician known for her expertise in medical genetics, dysmorphology, and inborn errors of metabolism.

She currently serves as the Centennial Professor of Pediatrics and Genetics and Division Chief of Genetics and Genomic Medicine at Washington University School of Medicine in St. Louis.

Education & Training

- Undergraduate: Classics degree from the University of Chicago (1995)
- Medical School: MD from Columbia University, New York (1999)
- Residency: Pediatrics at Harbor-UCLA, Chief Resident (2002–2003)
- Fellowship: Medical Genetics at UCLA/Harbor/Cedars-Sinai Research & Clinical Focus

Dr. Dickson is renowned for her work on mucopolysaccharidosis (MPS), a group of rare genetic disorders caused by enzyme deficiencies. Her research includes:

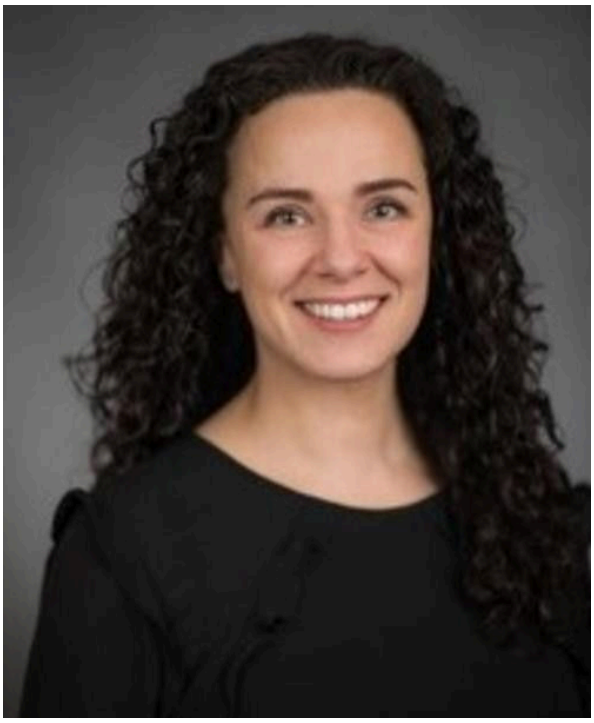
- Intrathecal enzyme replacement therapy for MPS I
- Clinical trials targeting spinal cord compression in Hurler-Scheie and Scheie syndromes

She's passionate about improving care for patients with rare genetic conditions and has helped transform treatment approaches through recombinant enzyme therapies.

Honors & Recognition

- Elected to the American Society for Clinical Investigation in 2021, affirming her role as a physician-scientist
- Helped Washington University/BJC Healthcare earn designation as a NORD Rare Disease Center of Excellence

Julie Eisengart, PhD, LP



Dr. Julie Eisengart is an Associate Professor in the Department of Pediatrics and the Director of the Neurodevelopmental Program in Rare Disease. As a pediatric neuropsychologist, she specializes in rare neurodegenerative disorders of childhood as well as a range of complex medical conditions, and she works with patients from infancy to young adulthood.

Her clinical interests include assessing the strengths and needs of children affected by complex medical diagnoses, throughout their medical journeys, to optimize whole-child, whole-family care and support.

As a clinical supervisor and mentor, she values her role in training future psychologists and supporting their development into independent, balanced professionals.

Dr. Eisengart's research focuses on the changing relationships between brain structure, biochemical abnormalities, and brain function in rare disease, with the goal of predicting outcomes and improving supportive planning.

She has been heavily involved in examining outcomes of early diagnosis, newborn screening, and/or novel therapies for rare disease.

Her research extends to defining and measuring aspects of disease that are under-represented in the clinical and research communities but are important and meaningful to patients and their families, such as neurobehavioral symptoms and the caregiver's lived experience.

Leigh Ellen Fremuth, Phd

Dr. Leigh Ellen Fremuth is a postdoctoral researcher specializing in neuroimmunology and lysosomal biology.

Her academic journey includes training at St. Jude Children's Research Hospital, where she contributed to groundbreaking work on lysosomal storage diseases and their links to neurodegeneration and cancer.

Currently, Dr. Fremuth is a postdoctoral fellow in the Eyo Lab at the University of Virginia's Department of Neuroscience. She joined the lab in April 2025 and holds a Fellow position on the prestigious Cardiovascular Training Grant through the Robert M. Berne Cardiovascular Research Center.

Her current research focuses on microglial function and brain-immune interactions, contributing to the lab's broader mission of understanding neurovascular and neuroimmune mechanisms.

She has co-authored publications on AAV-mediated gene therapy for sialidosis and galactosialidosis, rare lysosomal disorders, showcasing her expertise in translational approaches to neurodegenerative diseases.

Dr. Leigh Ellen Fremuth earned her Ph.D. in Biomedical Sciences with a focus on Neuroscience from the University of Tennessee Health Science Center in May 2022. Her doctoral research, under the mentorship of Dr. Alessandra d'Azzo, explored the immunomodulatory roles of the lysosomal sialidase Neuraminidase 1 (NEU1)—a key enzyme implicated in neurodegenerative diseases like sialidosis and Alzheimer's disease.

Jeanine Jarnes, PharmD, BCOP, BCPS

Jeanine Jarnes, PharmD, BCOP, BCPS, is an Assistant Professor in the University of Minnesota Department of Pediatrics and is an adjunct assistant professor in the College of Pharmacy, University of Minnesota.

Jeanine is a board certified Pharmacotherapy Specialist, as well as having a board certification in oncology pharmacy. Jeanine works as a pharmacotherapy provider for patients with inherited metabolic diseases at the University of Minnesota Specialty Clinics.

She also serves as a Clinical Pharmacogeneticist for the Lysosomal Disease Network and as a clinical researcher for patients with lysosomal diseases. Dr. Jarnes's research includes projects in the following areas: natural history of gangliosidosis diseases, such as Tay-Sachs disease, Sandhoff disease, and GM1-gangliosidosis, using a substrate reducing therapy protocol for treatment of childhood gangliosidosis diseases, classifying and managing infusion reactions to intravenous enzyme replacement therapies for lysosomal diseases, and pharmacokinetics and pharmacodynamics of ERT and of small molecule therapies.

Research Summary/Interests:

- Longitudinal study of the gangliosidosis diseases and development of new treatment therapies.
- Pharmacogenetics, pharmacodynamics, and pharmacokinetics of enzyme replacement therapies and small molecule therapies for lysosomal diseases and phenylketonuria (PKU).
- Combination therapies for lysosomal diseases, including Tay-Sachs disease, Gaucher disease, and MPS diseases.
- Optimizing management of acute, delayed, and biphasic infusion reactions to intravenous enzyme replacement therapy for lysosomal diseases.
- Pharmaceutical care impact on clinical outcomes in lysosomal diseases and phenylketonuria (PKU).
- Lysosomotropic agents and their impact on lysosomal diseases.

Virginia Kimonis, M.D., M.R.C.P.

Dr Kimonis is currently a Clinician Scientist and tenured Professor in the Division of Genetics and Genomic Medicine, Department of Pediatrics, with appointments in Neurology and Pathology, University of California Irvine, and Children's Hospital, Orange County. Dr. Kimonis received her medical degree from Southampton Medical School, United Kingdom and trained in pediatrics and general practice in the UK before moving to the USA. She completed a residency in pediatrics at Massachusetts General Hospital, Boston and fellowship training in Clinical and Biochemical Genetics at the National Institutes of Health, Johns Hopkins and Washington D.C. Children's Hospital. She is board certified in Pediatrics, in addition to Clinical Genetics and Biochemical Genetics. She previously served as the Chief of Genetics at Southern Illinois University School of Medicine. She worked at Boston Children's Hospital/Harvard Medical School before joining UC Irvine in 2006 serving as the Chief of the Division of Genetic Medicine and Genomic Medicine until 2012.

Dr. Kimonis specializes in the diagnosis and management of neuromuscular, neurodegenerative, lysosomal storage diseases and other complex rare disorders. She leads an active clinical and basic research programs that focus on inherited muscle disorders, lysosomal storage and mitochondrial diseases.

Dr. Kimonis discovered an important disease: multisystem proteinopathy associated with mutations in the VCP gene. Her research is funded by the NIH, MDA, FDA, VoLo, AMDA, Sanofi, Chiesi, Idorsia, and foundations. Dr. Kimonis's goal is to establish a premier clinical, and translational research program for rare genetic diseases.

Dr. Kimonis established a Lysosomal Disease Center of excellence at UCI. Recognizing the lack of effective treatments, she directs a comprehensive research program focused on beta-mannosidosis, and assisted the families who created The Lost Enzyme Project.

The lab's objectives include conducting natural history studies to document the clinical progression and variability of the disease, utilizing patient cell lines and the MANBA -/-

mouse model to better understand the disease mechanisms, and for translational studies. This initiative, in collaboration with the families, and JCR Pharmaceuticals, is dedicated to developing β-mannosidase, the enzyme deficient in affected patients as an

Virginia Kimonis, M.D., M.R.C.P. continued

enzyme replacement therapy. JCR's patented J-Brain Cargo® enables the enzyme to cross the blood-brain barrier-a critical advancement for treating neurological symptoms. These studies are very important in advancing IND enabling studies for treatment of patients with β-mannosidase.

Troy Lund, MD, PhD, FAAP

Troy Lund, MD, PhD, FAAP, is a pediatric blood and marrow transplant physician who cares for children with adrenoleukodystrophy, leukemia and lymphoma. He sees patients at the University of Minnesota Masonic Children's Hospital Pediatric Specialty Care Journey Clinic. Dr. Lund's research focuses on improving the outcomes for children with rare and ultra-rare diseases undergoing blood and marrow transplantation in both the clinical and laboratory settings. He is also a Professor in the Department of Pediatrics and a faculty member in the Division of Pediatric Blood and Marrow Transplantation & Cellular Therapy.