ISMRD'S 2025 INTERNATIONAL FORUM SPEAKERS

Angela Martin Rios M.D., MSC

Dr. Angela Martin Rios received her medical degree in 2005, and a master's degree in Human Genetics in 2016 from the National University of Colombia, School of Medicine. She moved to Irvine in 2022 to complete a Lysosomal Storage Diseases research fellowship at the Kimonis Laboratory until June 2025.

Her research interests in lysosomal storage disorders had led to participate in publications on Sanfilippo disease, Pompe disease and Fabry disease, and her current work on a potential treatment for Beta-Mannosidosis under the mentoring of Dr. Virginia Kimonis.

She is a recipient of the Travel Award winner from the Society for Inherited Metabolic Disorders in 2024, and the Trainee Awards for Excellence in Human Genetics Research in 2023 from the American Society of Human Genetics ASHG, for her work on antisense oligonucleotides treatment in Pompe disease.

In July 2025, Dr. Martin started her medical residency in Pediatrics and Medical Genetics at the University of Iowa Stead Family Children's Hospital.

Emeritus Professor Marc C. Patterson, MD, FRACP, FAAN, FANA

Marc Patterson, MD, FRACP, FAAN, FANA, was born in Australia, where he was educated, and graduated with first class honors in medicine from the University of Queensland in 1981.

He trained in neurology and child neurology at the University of Queensland (1982-1988), in child neurology at Mayo Clinic (1988-1990) and neurometabolic disease at the Developmental and Metabolic Neurology Branch, NINDS/NIH, under the direction of Roscoe Brady, MD (1990-1992). Dr. Patterson is a fellow of the Royal Australasian College of Physicians and is certified by the ABPN in Neurology with Special Qualification in Child Neurology, and in Neurodevelopmental Disabilities.

He served as Professor of Neurology, Pediatrics and Medical Genetics at Mayo Clinic through his retirement in December 2024. During his tenure at Mayo, he was appointed as Director of the Child Neurology Training Program (2008-2016), and Chair of the Division of Child and Adolescent Neurology (2008-2017). Dr Patterson had previously served as Professor and Director of Pediatric Neurology and Director of the Child Neurology Training Program at Columbia University in New York (2001-2007).

Following his retirement from practice, in January 2025, Dr Patterson joined IntraBio, Inc., as US Chief Medical Officer, continuing his focus on developing safe and effective therapies for neurodevelopmental and neurodegenerative disorders. Dr Patterson served as Editor for the Journal of Inherited Metabolic Disease and JIMD Reports, Editor-in Chief of the Journal of Child Neurology and Section Editor (Pediatric Neurology) for Up-To-Date through his retirement from practice. He has previously served on the editorial board of Neurology and the oversight committee for Annals of Neurology.

Dr Patterson served as an examiner and committee member for the ABPN for more than 20 years and was Vice-Chair of the Neurodevelopmental Disabilities MOC committee through December 2024. He has previously served on the RITE examination committee of the American Academy of Neurology (AAN), as program director for educational courses at multiple AAN annual meetings, and as a counselor and scientific program director for the Child Neurology Society (CNS).

His research and practice have focused on rare diseases in children, including inherited metabolic disorders in general, with special interests in Niemann-Pick disease, type C (NPC), other lysosomal diseases, and congenital disorders of glycosylation, areas in which he has published more than 250 peer-reviewed papers and book chapters. He led the pivotal trial which led to the approval of miglustat for the treatment of Niemann-Pick disease, type C in

Emeritus Professor Marc C. Patterson, MD, FRACP, FAAN, FANA continued

the European Union and more than countries worldwide; he has also participated in the design and execution of clinical studies which

led to the recent FDA approval of arimoclomol and N-acetyl-L-leucine for the treatment of Niemann-Pick disease, type C, in the United States.

Dr Patterson has served on the scientific advisory boards of several rare disease foundations and chaired the scientific advisory board for the international Niemann-Pick disease registry, an entity supported by a consortium of international Niemann-Pick Disease Foundations, and with an initial grant from the European Union. He has received funding support from NIH, industry, and private foundations.

Robert Šáhó, MD

Dr Robert Šáhó is a pediatrician and current Ph.D. student specializing in inherited metabolic disorders. He works as a clinician at the Department of Pediatrics and Inherited Metabolic Disorders in Prague, Czech Republic.

He provides outpatient care at the dedicated Clinic for Lysosomal Storage Diseases and the National Center for Gaucher Disease Treatment, managing patients with Gaucher disease, Fabry disease, Pompe disease, LAL-deficiency, MPS, Alpha-Mannosidosis, and Niemann-Pick type C.

He is actively involved in international clinical research, serving as subinvestigator in multiple clinical trials, including studies on MPS II, PMM2-CDG and LAL deficiency.

Dr. Šáhó is the first-author of an international collaborative study, which presents the most extensive multicenter dataset to date on hematopoietic stem cell transplantation in alpha-mannosidosis since 2010. He is presenting his work at national and international meetings, including for example the WOLRD*Symposium*, International Symposium on MPS and Related Diseases and Alpha-Mannosidosis CEE Expert Meeting. He is also engaged in undergraduate medical education, with active roles in simulation-based teaching and medical school admissions.

Mathias Schmidt, PD, Ph.D.

Mathias Schmidt, PD, Ph.D. has spent over 25 years in the (Bio)pharmaceutical Industry in various positions including research, preclinical and clinical development, business development, global business expansion and executive management.

He has been with JCR Pharmaceuticals since 2020 where he has been serving in multiple different functions including clinical development, executive management, business internationalization, and business development. Mathias also serves as President and CEO of ArmaGen. Previous positions include Takeda Pharmaceuticals, Nycomed, Altana Pharma and ASTA Medica.

He received his master's degree from the University of Stuttgart, a Ph.D. in tumor biology from the University of Freiburg and the venia legendi in Pharmacology from the University of Konstanz where he has been lecturing Disease Biology for many years. Earlier in his career Mathias worked at several cancer hospitals in Germany and the United States.

Mathias is driven by a strong sense of purpose based on the recognition that all human life has equal value and that the genes we are born with are not a choice of lifestyle.

His greatest respect is for the families and caregivers who give their love and compassion to support their children affected with rare genetic diseases.

Richard Steet, Ph.D

Dr. Steet joined Greenwood Genetic Center in August 2018 as the Director of Research and Head of the JC Self Research Institute following twelve years as a professor in the Complex Carbohydrate Research Center at the University of Georgia. His current research program, funded by the NIH and private foundations, is focused on defining disease mechanisms for two different classes of inherited diseases: lysosomal storage disorders and the congenital disorders of glycosylation. This work takes advantage of cell and animal-based models and uses a combination of chemical, molecular, and developmental approaches to unravel the complex pathogenesis of these disorders and explore new ways to treat them.

Dr. Steet's goals for the Research Division are to integrate strengths in basic science research with clinical and translational studies, to enhance partnerships with pharmaceutical companies that can drive therapeutic development, and collaborate with the Clinical and Diagnostic divisions of GGC to enhance our understanding of the genetic basis for birth defects and disabilities.

Dr. Steet is a dedicated advocate of rare disease research and serves on the scientific advisory boards for the National MPS Society and ISMRD.

Professional Appointments

Instructor of Medicine, Washington University in Saint Louis, Department of Medicine 2005-2006
Assistant Professor, Department of Biochemistry and Molecular Biology, Complex Carbohydrate Research Center, University of Georgia, 2006-2012
Associate Professor, Department of Biochemistry and Molecular Biology, Complex Carbohydrate Research Center, University of Georgia, 2012-2017
Professor, Department of Biochemistry and Molecular Biology, Complex Carbohydrate Research Center, University of Georgia, 2017-2018
Director of Research and Head of JC Self Research Institute of Human Genetics, Greenwood Genetic Center, 2018-present

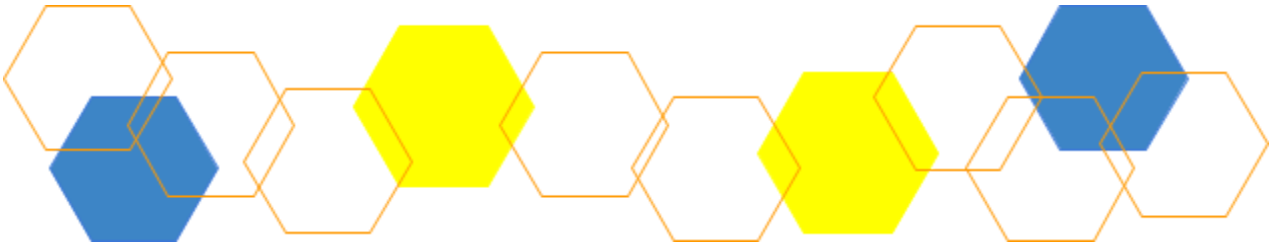
Richard Steet, Ph.D continued

Other Appointments and Professional Service

Adjunct Assistant Professor; Department of Cell Biology, University of Georgia, Athens, GA; 2006-present
Adjunct Professor, Department of Genetics and Biochemistry, Clemson University, Clemson, SC, 2018-present
Treasurer, Society for Glycobiology, 2013-present
Member, Scientific Advisory Board, ISMRD 2012-present
Member, Scientific Advisory Board, MPS Society, 2015-present
Member, Editorial Board, Glycobiology (official journal of the Society for Glycobiology), 2016-present

Awards and Honors

Recipient of the University of Georgia nomination for the Searle Scholar Award competition, 2006
NIH Extramural Loan Repayment Program (LRP) Award in Pediatric Research, 2009



Karolina M Stepien, M.D., Ph.D

Dr. Karolina M. Stepien, M.D., Ph.D. is a distinguished expert in Adult Metabolic Medicine, with a career spanning clinical care, academic research, and international collaboration.

Professional Background

- Consultant in Adult Metabolic Medicine at Salford Royal Hospital, Northern Care Alliance NHS Foundation Trust in Greater Manchester, UK
- Honorary Senior Lecturer at the University of Manchester, Division of Cardiovascular Sciences Formerly worked at the National Centre for Inherited Metabolic Diseases in Dublin

Areas of Expertise

- Inherited Metabolic Diseases (IMD) including lysosomal storage disorders
- Natural history of IMDs and long-term complications including endocrine dysfunction
- Transitioning care from pediatric to adult metabolic services
- Management of pregnancy in IMDs

Academic Contributions

- Author of over 100 publications and book chapters in metabolic medicine
- Principal investigator in multiple academic and commercial studies focused on IMD treatment and long-term outcomes
- Section Editor for Orphanet Journal of Rare Diseases and guest editor for journals such as *Frontiers in Genetics*, *Genes*, and *Frontiers in Cardiovascular Medicine*

International Recognition

- Active in cross-border collaborations and surveys on rare diseases
- Contributor to research on Mucopolysaccharidoses, Alpha-Mannosidosis and Urea Cycle Disorders

Her work bridges clinical excellence with scientific innovation, making her a leading figure in the field of metabolic medicine.

Dr. Chester B. Whitley, PhD, MD.

Dr. Whitley is the Principal Investigator for the Lysosomal Disease Network (LDN), a growing consortium of medical centers collaborating since 2003—and funded by the National Institutes of Health beginning in 2008—to address critical gaps in taking lysosomal disease research from the lab to clinical practice. Chester (Chet) B. Whitley, Ph.D., M.D. is a tenured Professor in the Department of Pediatrics, the Department of Experimental and Clinical Pharmacology, and Director of the Advanced Therapies Program at the University of Minnesota.

Dr Whitley was the Founding Organizer of the 1st International Symposium on Mucopolysaccharidosis and Related Diseases (May 20-22, 1988), University of Minnesota, Minneapolis, USA

Dr. Whitley holds many distinguished posts, including:

- Principal Investigator, Lysosomal Disease Network, www.LysosomalDiseaseNetwork.org
- Professor, Department of Pediatrics and the Department of Experimental and Clinical Pharmacology, University of Minnesota, Minneapolis MN, USA
- Director, Advanced Therapies Program, University of Minnesota, Minneapolis MN, USA
- Director, Gene Therapy and Diagnostics Lab
- Director, Gene Therapy Center, University of Minnesota, Minneapolis MN USA

John H Wolfe, VMD, PhD

John H. Wolfe, VMD, PhD, is Professor of Pathology and Medical Genetics in the Departments of Pathobiology and Pediatrics in the
School of Veterinary Medicine and Perlman School of Medicine; and is Director of the W F Goodman Center for Comparative Medical Genetics at the University of Pennsylvania.

He is also a Stokes Investigator in the Children's Hospital of Philadelphia Research Institute. He investigates gene and stem cell therapies in animal homologues of human ultra-rare monogenic disorders, mostly for the central nervous system in lysosomal storage diseases.

He has been continuously funded by the NIH for nearly four decades; does extensive grant reviewing for the NIH and private foundations; and reviews manuscripts for numerous scientific journals.

The Wolfe Lab investigates vector-mediated gene transfer and stem cell transplantation in animal homologues of pediatric neurodevelopmental genetic diseases. The Wolfe Lab has made significant contributions to testing and developing gene therapy approaches, particularly for intellectual developmental disorders. The lab investigates gene therapy approaches to treating lysosomal storage disorders, which require global brain correction, currently focusing on AAV vectors.

THANK YOU TO OUR ESTEEMED SPEAKERS