SPEAKER BIOGRAPHIES

- **Charles Abrams, M.D., Ph.D.** is professor of Neurology and Physiology & Pharmacology at SUNY Downstate Medical Center. He received M.D and Ph.D. degrees from the Albert Einstein College of Medicine and Clinical training at New York Hospital-Cornell and Johns Hopkins University. His work focuses on the roles of gap junction forming connexin proteins in myelinating glia in health and disease.

- **John Aitchison, Ph.D.** is Professor and Scientific Director at the Center for Infectious Disease Research (formerly Seattle Biomedical Research Institute). He also holds an appointment as Professor and is a founding member of the Institute for Systems Biology. As a student, he studied biochemistry, specializing in biotechnology and genetic engineering, at McMaster University in Ontario, Canada. There, in the laboratory of Dr. Richard Rachubinski, he investigated the molecular mechanisms responsible for sorting proteins to peroxisomes. In the Blobel lab, Dr. Aitchison applied classic cell biology techniques and yeast genetics to the study of protein sorting in cells. He began his career as a principal investigator as an Assistant Professor in the Faculty of Medicine and Dentistry at the University of Alberta. In 2000, he joined the Institute for Systems Biology as a founding faculty member. In Seattle, he began to develop systems cell biology approaches to study peroxisome biogenesis. Aitchison also holds affiliate appointments at the University of Washington, University of Alberta, University of British Columbia and Rockefeller University.

- **Genevieve Bernard, M.D., MSc, FRCPC** is a pediatric neurologist and clinician-researcher at the McGill University Health Center and an assistant professor at McGill University. She did her medical school at Université de Montréal, her residency in Neurology at McGill and her post-doctorate fellowship at Université de Montréal in Neurogenetics and Movement disorders. She is the leader of the Montreal research group on Leukodystrophies and her research studies focus on 4H or POLR3-related leukodystrophies. Her group identified the 3 causative genes and is working on understanding the pathophysiology of the disease to lead to way toward finding therapeutic strategies.

- **Josh Bonkowsky, M.D., Ph.D.** is an Associate Professor in Pediatrics, and the Bray Chair in Pediatric Neurology, at the University of Utah. After growing up in Georgia and then going to college at Harvard University, Dr. Bonkowsky moved out west and did his MD and PhD training at the University of California, San Diego. Dr. Bonkowsky then moved to the mountains to complete his residency and fellowship at the University of Utah. Dr. Bonkowsky's interests are in understanding the normal processes of brain development, and the effects of disease and how to treat those diseases. His research team combines studies at the bench with large-scale clinical database studies in humans.

- **Nancy Braverman, M.D., M.S.** is an Associate Professor in the Department of Human Genetics and the Department of Medical Genetics and Pediatrics. She completed her medical degree at Tulane University in Louisiana and completed a fellowship in Clinical and Biochemical Genetics at the Johns Hopkins Medical Center in Baltimore, MD. Dr. Braverman has been studying peroxisome disorders for 2 decades, identifying and characterizing relevant genes, their mutation spectrum and determining protein functions. Her laboratory has generated mouse models to study peroxisomal disease pathophysiology and has developed a successful high throughput drug screen for these disorders. Dr, Braverman has also recently begun a natural history study for peroxisomal disorders.

- **Andrea DeBarber, Ph.D.** is a Research Assistant Professor in the Oregon Health & Science University (OHSU) School of Medicine. She obtained her Ph.D. in chemistry in 1997 and Postdoctoral Fellowships at the NIH and OHSU, further solidified her knowledge of synthetic and analytical chemistry. In 2004 she was recruited to remain at OHSU as Associate Director of the Bioanalytical Shared Resource Facility (BSR). In this role her responsibilities include development of LC-MS/MS methods for identification and quantification of drugs and metabolites, as well as analysis of endogenous steroids, sterols and lipids. In 2008 she expanded her BSR role to become Director of the Portland State University High Resolution Mass Spectrometry Facility. Dr. DeBarber has also overseen the OHSU Sterol Analysis Diagnostic Laboratory since 2013. Funding from the Friends of Doernbecher Foundation, the United Leukodystrophy Foundation and Retrophin, Inc. have allowed Dr. DeBarber to develop an independent research program, where the focus of her research has been on developing improved diagnostic and screening tests for rare genetic disorders of sterol and bile acid synthesis. Most recently she has been working toward developing LC-MS/MS based newborn screening for cerebrotendinous xanthomatosis (CTX), a rare genetic disorder of bile acid synthesis.

- **Fabrice Dabertrand, Ph.D.** received his Ph.D. in Neurosciences from the University of Bordeaux, France, in 2006. After finishing his first postdoctoral work on the effect of microgravity on cerebral arteries, he joined the Department of Pharmacology of the University of Vermont, US, in 2009, where he is currently Research Assistant Professor. Over the last six years, he developed novel and unique techniques to isolate and study the mouse brain microvasculature under physiologic and pathologic conditions, particularly cerebral small vessel disease (cSVD). His work was published in leading journals (*Circ Res, JCBFM, PNAS*) and in 2012, in partnership with Dr. Anne Joutel (Paris, France) Dr. Dabertrand has applied these techniques to the mouse model of CADASIL, a monogenic form of cSVD, that her group developed. This collaboration is now the core of a transatlantic network, involving 8 laboratories, currently funded by the prestigious Foundation Leducq. Fabrice Dabertrand, who just received the New Investigator Award from the American Physiological Society (Cardiovascular Section), was awarded his own peer-reviewed funding from the United Leukodystrophy Foundation (CADASIL Research Grant).

- **Gabriele Dodt, M.D.** is a Professor at the Interfaculty Institute of Biochemistry, University of Tübingen, Germany. She received her M.D. from the Institute for Physiological Chemistry at the University of Cologne, and completed a Research Fellowship at the Institute for Physiological Chemistry, Ruhr-University Bochum. Additionally, Dr. Dodt also completed a postodoctoral fellowship in the Department of Biological Chemistry at Johns Hopkins University. Dr. Dodt's research group focuses on investigation of the dynamics and turnover of peroxisomes and its implication for peroxisomal disorders, using a broad array of methods in biochemistry, cellular and molecular biology. This includes fluorescence imaging techniques and structural analysis to get insights into the molecular details of peroxisome biogenesis and maintenance.

- **Ralf Erdmann, Ph.D.** is a Full Professor and Chair of the Department of System Biochemistry, Ruhr-University Bochum in Germany. He received his Ph.D. in Biology from Ruhr-University and completed a post-doctoral fellowship at the Rockefeller University of New York. Dr. Erdmann's research interests include the biogenesis of peroxisomes, identification and characterization of peroxins, the molecular basis for peroxisome biogenesis disorders, and the characterization of the peroxisomal protein import machinery.

- **John Fink, M.D.** is a Professor in the University of Michigan Department of Neurology. Following medical school (Medical College of Ohio), Internship (Mayo Clinic), Neurology Residency (University of Virginia), Dr. Fink completed postdoctoral training in Developmental and Metabolic Neurology and Medical Genetics at the National Institutes of Health. Dr. Fink serves as Medical Advisor to the Spastic Paraplegia Foundation. Dr. Fink's clinical and laboratory research investigates the molecular genetic basis of inherited neurologic disease, particularly disorders such as hereditary spastic paraplegia, primary lateral sclerosis, and adrenomyeloneuropathy that are characterized by axon degeneration. This research includes gene identification, biochemical analysis, creation of animal and stem cell models of disease, and clinical investigations of natural history, phenotype analysis, and clinical trials.

- **Yukio Fujiki, Ph.D.** is a Professor at the Medical Institute of Bioregulation, Kyushu University. He completed his Ph.D at Kyushu University and completed a post-doctoral fellowship in the Department of Medicine and Endocrinology at Cornell University Medical College. His research interests include peroxisome biogenesis and homeostasis, protein targeting, and peroxisome biogenesis disorders.

- **Joseph Hacia, Ph.D.** is currently an Associate Professor in the Department of Biochemistry and Molecular Biology at the Keck School of Medicine of the University of Southern California (USC). He earned his Ph.D. in Biology at the California Institute of Technology (Pasadena, CA) and completed his postdoctoral training in Medical Genetics at the National Institutes of Health (Bethesda, MD). Dr. Hacia is also a member of the Institute for Genetic Medicine as well as the Eli and Edythe Broad Center for Regenerative Medicine and Stem Cell Research at USC. He has a long-term interest in the development of improved therapies for peroxisomal disorders. In addition to directing an active research laboratory, Dr. Hacia is a Chair of Medical Education at the USC Keck School of Medicine.

- **Ewald Hettema, Ph.D.** trained at the Academic Medical Centre in Amsterdam under Henk Tabak and Ron Wanders. After obtaining his Ph.D. 1998 he went on to study peroxisome dynamics as a postdoc in Henk Tabak's lab (1998-2000). Subsequently, he moved to Cambridge, UK, to join Hugh Pelham's lab at the MRC-LMB on an HFSP long-term fellowship, where he studied membrane protein trafficking through the endosomal system. From 2004 to 2014, he was funded by Wellcome Trust Fellowships to establish an independent research group to study peroxisome dynamics in model organisms at the University of Sheffield. He currently has a position there as reader in molecular cell biology and continues his research in yeast and fruit fly peroxisome biogenesis.

- **Peter K. Kim, Ph.D.** is a Cell Biologist at the Hospital for Sick Children (Toronto) and an Associate Professor in the Department of Biochemistry at the University of Toronto. His research group studies the molecular mechanism of peroxisomes and mitochondria maintenance in mammalian cells in order to understand various diseases ranging from common diseases such as Parkinson's disease, to rare understudied diseases such as Peroxisome Biogenesis Disorders. Dr. Kim's interest in peroxisome biology first germinated during his Ph.D. studies at McMaster University in Hamilton, Ontario, Canada, where he studied the mechanisms of membrane protein trafficking. Inspired by the development of novel microscopy imaging techniques, he joined the laboratory of Jennifer Lippincott-Schwartz (NIH, Bethesda, USA) to explore the mechanism of peroxisomal protein trafficking. There he developed various tools to investigate the maintenance of peroxisomes in living cells. In 2009, he established his own group at the Hospital for Sick Children in Toronto, Canada, where he is examining the role of peroxisomes and mitochondria in Neurodegenerative Disorders. Outside of the laboratory, Peter enjoys fishing and tennis. His research is supported by the Government of Canada, SickKids Foundation, and by his wife and their three lovely children.

- **Troy Lund, M.D., Ph.D.** is an Assistant Professor in the Department of Pediatrics, Division of Blood and Marrow Transplantation at the University of Minnesota. He completed his Ph.D. in Medical Sciences at the University of South Florida in 1996. In 2002, Dr. Lund received his medical degree from the University of Minnesota and continued there for a Fellowship in Pediatric Hematology, Oncology and Bone Marrow Transplant. Dr. Lund is interested in the use of blood and marrow transplantation (BMT) primarily for patients with leukemia, lymphoma, and rare inherited metabolic disorders, such as Adrenoleukodystrophy (ALD) and Hurler syndrome. His work both in his laboratory and with his patients has created many new approaches to treatment, which will ultimately make transplant safer and more effective. His international work takes him to several countries in Sub-Saharan Africa each year where he sees a variety of patients with a broad spectrum of diseases from tuberculosis to cancer. His research in Africa is focused on the oxidative stress associated with hemolytic anemia. He also supports community-health based projects through his non-profit: the Medicine for Sick Children Foundation.

- **Ann Moser, B.A.** is a research associate in neurology and co-director of the Peroxisomal Diseases Laboratory at Kennedy Krieger Institute and Johns Hopkins University. She received a bachelor's degree in biochemistry in 1961 from Radcliffe College. During the time she was an undergraduate, she was a technician in Dr. Konrad Bloch's laboratory at Harvard University. After working as a technician in laboratories in different hospitals, Moser joined the John F. Kennedy Institute (later Kennedy Krieger Institute) in 1976 as a senior technician. In 1982, she became an assistant in neurology. Since 1991, Moser has been working as a research associate in neurology. She is a co-director of the Peroxisomal Diseases Laboratory in the Hugo W. Moser Research Institute at the Kennedy Krieger Institute.

- **Bwee Tien Poll-The, M.D., Ph.D.** is a Professor of Pediatric Neurology at the University of Amsterdam, Academic Medical Center in the Netherlands. She completed her medical degree (with a specialization in Pediatrics) at the University of Amsterdam and completed fellowships in both Pediatric Neurology and Genetic and Metabolic Disorders at the Hôpital Nêcker-Enfants Malades in Paris, France. In 1988, Dr. Poll-The received her Ph.D. from the University of Amsterdam with her dissertation entitled "Genetic Peroxisomal Disorders". Dr. Poll-The's current research interests include neurometabolic diseases, with a focus on peroxisomal disorders.

- **Richard Rachubinski, Ph.D.** is is currently Distinguished University Professor and Chair of the Department of Cell Biology at the University of Alberta. He received his Ph.D. from McGill University under the supervision of Dr. John Bergeron and did postdoctoral work first at McGill with Dr. Gordon Shore and then at The Rockefeller University with Dr. Paul Lazarow, where he began his studies on peroxisome function and formation. He was faculty in the Department of Biochemistry at McMaster University from 1984 to 1993 and joined the University of Alberta in 1993. He is a Fellow of the Royal Society of Canada, the Canadian Academy of Health Sciences, and the American Association for the Advancement of Science. Dr. Rachubinski's research interests are in protein targeting and organelle biogenesis, particularly of the peroxisome. He has contributed to the identification and characterization of numerous genes whose mutation causes peroxisome biogenesis disorders, and provided a molecular foundation for the understanding and eventual rational treatment of these disorders linked to dysfunction of peroxisome assembly.

- **William B. Rizzo, M.D**. is Professor of Pediatrics at the University of Nebraska Medical Center in Omaha and Director of the Hobart Wiltse Center for the Study of Inherited Metabolic Disorders. He obtained a B.A. from Northwestern University in 1972 and a M.D. from the University of Illinois in 1977. After residency training in Pediatrics at Johns Hopkins University and a Medical Genetics fellowship at the National Institutes of Health (NIH), he joined the Medical College of Virginia in 1983, where he initiated research on adrenoleukodystrophy and later Sjögren-Larsson syndrome. In 2002, he joined the University of Nebraska

Medical Center where he oversees the care of children with inherited metabolic diseases and performs laboratory research in the area of biochemical and molecular genetics.

- **Andrew Simmonds, Ph.D.** has been a member of the Department of Cell Biology at the University of Alberta since 2002. His research group studies macromolecule trafficking and gene regulation related to human disease using the model developmental system *Drosophila melanogaster* (fruit flies). Since 2010, working in collaboration with Dr. Rick Rachubinski, Dr. Simmonds' group established *Drosophila* as an effective model system for determining the tissue specific requirements of peroxisomes. This led to a proof-of-concept paper based on their systematic biochemical, developmental and gene regulation based characterization of a Drosophila *Pex1* mutation. Subsequently, Dr. Simmonds' Drosophila animal model has been added to a national team, encompassing both the basic developmental biology of Drosophila with studies of patients and cultured cell models in a rational and systematic attempt to identify potential therapeutic interventions for both peroxisome biogenesis disorders as well as enzymatic deficiencies.

- **Gary D. Smith, M.D**. is Professor of Ob/Gyn, Physiology, and Urology and Director of the University of Michigan's MStem Cell Laboratory. His work on stem cells spans and links areas including: 1) political and public policy/engagement involving human embryonic stem cells (hESCs); 2) derivation of disease-specific hESCs with national and international distribution; 3) fostering cross-discipline collaborations using disease specific-hESCs to understand molecular mechanisms of congenital birth defects and adult diseases; 4) practical solutions to enhance clinical utility of hESCs in drug testing and therapies. Dr. Smith was a participant in drafting of Michigan Proposal 2008-2, which proposed to amend the Michigan Constitution to remove restrictions on embryonic stem cell research. With passage of Michigan Proposal 2008-2 Dr. Smith established the State's first hESC derivation laboratory in 2009, now called MStem Cell Laboratory. The University of Michigan's MStem Cell Laboratory has become the leading U.S. institution in derivation of disease-specific hESCs with acceptance on the NIH Stem Cell Registry.

- **Steven Jeffrey Steinberg, Ph.D., FACMG** is an Assistant Professor in the McKusick-Nathans Institute of Genetic Medicine at Johns Hopkins University in Baltimore, MD. He earned his Ph.D. in Biochemical Genetics from the University of London for the graduate work he conducted in the Paediatric Research Unit at Guy's Hospital in London, England. In 2005 he completed his training in Clinical Biochemical Genetics and Clinical Molecular Genetics at Johns Hopkins University. Currently he spends most of his time co-directing the Johns Hopkins DNA Diagnostic Lab. Steven's most recent peroxisomal research effort yielded the Pex1-G844D mouse model. He hopes that this model will be used widely as a tool to test therapies for Zellweger spectrum disorders.

- **Marjo van der Knaap, M.D., Ph.D.** was trained in Adult and Pediatric Neurology. She wrote a Ph.D. thesis on MRI and MRS of myelination and white matter disorders in children and young adults (1991). She is currently professor of Child Neurology, VU University Medical Center, Amsterdam and head of the department of Child Neurology. Since 1987, her research has been focused on magnetic resonance of childhood white matter disorders with a special focus on unclassified childhood leukoencephalopathies. She developed MRI pattern recognition to facilitate the diagnostic work-up of white matter disorders and used this tool to define novel disorders. Her research group has identified the mutated genes in several novel disorders, including vanishing white matter. Her subsequent studies are focused on elucidation of disease mechanisms and development of treatment. In 2000, she founded the Center for Childhood White Matter Disorders in Amsterdam, in which diagnostics, patient care and research are integrated.

- **Ronald J.A. Wanders, Ph.D.** is Professor Clinical Enzymology of Inherited Metabolic Diseases and Head of Laboratory Genetic Metabolic Diseases (LGMD) at the Academic Medical Center (AMC) at University of Amsterdam in the Netherlands. He received his Ph.D. degree in Biochemistry (metabolism) at the University of Amsterdam and completed a postdoctoral fellowship at the LGMD at the AMC. Dr. Wanders is an internationally recognized expert on biochemistry and enzymology in relation to metabolism and metabolic disorders. His research focus in the last 5 years has been on biochemical and

pathophysiological aspects of disorders of mitochondrial fatty acid oxidation, peroxisome metabolism & biogenesis and isoprenoid/cholesterol biosynthesis, areas of research for which LGMD has a longstanding international reputation.

- **Michael Wangler, M.D., M.S** is a physician-scientist specializing in childhood genetic disease and merging clinical genomics and *Drosophila* biology. Michael is an Assistant Professor in the Department of Molecular and Human Genetics at Baylor College of Medicine, board-certified in pediatrics and medical genetics. Michael's primary research focus is to understand gene function for better clinical sequencing interpretation. Michael has pursued this in three areas of focus, 1) using human genomics for diagnosis and gene discovery in rare disease, for example, *AHDC1* in syndromic speech delay and *ACTG2* in Berdon syndrome. 2) studying rare disease pathogenesis in cases diagnosed by genomics by employing *Drosophila* studies including *PEX16* and *DNM1L* and 3) establishing a framework for the latest technology in *Drosophila* to be employed in combination with human genomics, an effort which led to an exciting gene discovery, *ANKLE2* in microcephaly identified through *Drosophila* screens. He is currently working on using genomic data from patients and *Drosophila* to understand peroxisomal disorders.

- **Hans Waterham, Ph.D** is AMC Principal Investigator, Associate Professor and certified European Clinical Laboratory Geneticist (EBMG) in the Laboratory Genetic Metabolic Diseases of the Academic Medical Center at the University of Amsterdam, The Netherlands. He gained his MSc and PhD degree in Biological Sciences at the University of Groningen, the Netherlands. His PhD study was on peroxisome biogenesis in yeast. After his PhD, he performed 3 years postdoctoral research in Portland, Oregon, USA and 0.5 year in Nijmegen, the Netherlands after which he joined the Laboratory Genetic Metabolic Diseases in 1998. His current research focuses on Functional Genetics of Metabolic Disorders with emphasis on Inherited Defects of Isoprenoid Biosynthesis and Human Peroxisome Biogenesis Disorders. In addition, he is head of the laboratory's DNA diagnostic unit specialized in clinical genetic testing of inborn errors of metabolism (>70 different disorders), including all peroxisomal disorders. He is (co-) author on >225 peer-reviewed manuscripts.

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