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Siobhán Addie, PhD, The New York Academy of Sciences
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Siobhan Addie received her Bachelor of Science degree at the State University of New York at Binghamton in Cell and Molecular Biology, and her PhD in Cell Biology and Molecular Physiology from the University of Pittsburgh. Her thesis research examined the role of DNA repair in cancer and aging. Following a postdoctoral research position in genetics at Rockefeller University, she joined the New York Academy of Sciences as a Program Manager in Life Sciences. The mission of the New York Academy of Sciences is three-fold and involves advancing scientific research and knowledge, promoting scientific literacy, and supporting the resolution of global challenges with science-based solutions. Dr. Addie is part of the Conferences Department at the Academy, and is responsible for planning and overseeing scientific symposia that focus on a diverse array of topics within the Life Sciences.

Ravi Amaravadi, MD, University of Pennsylvania
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Ravi Amaravadi is the co- Leader of the Cancer Therapeutics Program of the Abramson Cancer Center, University of Pennsylvania. He is a physician-scientist with expertise in autophagy, conducting early phase clinical trials, and clinically treating patients with advanced melanoma. He was one of the first to demonstrate that autophagy is a targetable mechanism of resistance in cancer therapy using animal models. He has translated this finding into 9 phase I/II clinical trials testing hydroxychloroquine (HCQ) as a possible anticancer agent in multiple cancers. The first 6 clinical trials were published and represent the first deliberate attempt to modulate autophagy therapeutically in any disease. His laboratory published the first evidence that autophagy is elevated in melanoma patient tumors and predicts poor survival. His work on the role of autophagy in melanoma has identified ER stress associated autophagy as a resistance mechanism to BRAF inhibitors. Working with collaborators in chemistry, his lab has designed, synthesized and characterized a novel autophagy inhibitor Lys05, which is more potent than HCQ in animal models. Dr. Amaravadi is also a recognized melanoma medical oncologist and phase I trialist who has served as the site PI or co-investigator on melanoma clinical trials involving BRAF inhibitors, and first in human phase I clinical trials of novel cancer agents.

Norma Andrews, PhD, University of Maryland
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Norma Andrews obtained her PhD degree at the University of São Paulo, Brazil in 1983, followed by postdoctoral training at New York University in the group of Dr. Victor Nussenzweig. In 1990 she joined Yale University as Assistant Professor, and was promoted to tenure in 1996 and to Full Professor in 1999. At Yale she held appointments in the Department of Cell Biology and in the Section of Microbial Pathogenesis. In 2009 she moved to the University of Maryland as Chair of the Department of Cell Biology and Molecular Genetics. Her laboratory studies the cell biology of host-pathogen interactions, and the mechanism of plasma membrane repair. Her laboratory was the first to show that conventional lysosomes in non-specialized cells respond to elevations in cytosolic free calcium by fusing with the plasma membrane, and secreting their contents (Rodriguez et al. *J. Cell Biol.* 1997; Jaiswal, et al. *J. Cell Biol.* 159: 625-635, 2002). The Andrews laboratory also demonstrated that calcium-regulated lysosomal exocytosis promotes the resealing of wounded cells (Reddy et al. *Cell* 106: 157-169, 2001). This process was shown to involve extracellular release of the lysosomal enzyme acid sphingomyelinase, ceramide generation, and a fast form of endocytosis that removes plasma membrane lesions (Idone et al. *J. Cell Biol.* 180:905-914, 2008; Tam et al. *J. Cell Biol.* 189:1027-38, 2010; Corrotte et al. *Traffic* 13:483-494, 2012; Corrotte et al. *eLife* 2013:2:e00926, 2013).

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William E. Balch, PhD, The Scripps Research Institute, La Jolla
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William Balch has over 25 years of experience in the analysis of trafficking through endomembrane compartments using a broad range of molecular, structural, biochemical and bioinformatic approaches. The Balch laboratory is expert on the role of protein folding in human biology through an emergent paradigm he has referred to as 'proteostasis' (*Science* (2008) 319:916; *Science* (2010) 329:766); *Nat Rev Mol Cell Biol* (2013), 14, 237); Roth et al. (2014) *PLoS Biol.* In press) that is now driving the field. Proteostasis pathways include many chaperone and degradation components and requisite signaling pathways that deal with misfolded proteins, including all transmembrane and soluble proteins responsible for human disease. The Balch laboratory has considerable expertise in the analysis of protein folding, trafficking and function using human lung and liver tissue culture models and has access to a wide variety of technologies to follow wild-type and mutant protein synthesis, stability, trafficking. Dr. Balch makes extensive use of advanced live-animal imaging technology to monitor proteostasis health in cell, tissue and whole animal models, state-of-the-art mass spectrometry technologies to monitor protein-protein interactions in health and disease, and analysis of protein function and bioinformatics to mine pathway relationships responsible for disease. The Balch laboratory has authored over 210 papers with a recent focus on understanding the impact of misfolding of the cystic fibrosis transmembrane conductance regulator (CFTR) and alpha-1-antitrypsin (A1AT) on human physiology responsible for chronic obstructive pulmonary disease (COPD) in inherited disease (A1AT deficiency (AATD)) and in response to cigarette smoke.

Andrea Ballabio, MD, Telethon Institute of Genetics and Medicine (TIGEM)
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Andrea Ballabio is the founder and director of the Telethon Institute of Genetics and Medicine (TIGEM) in Naples, Italy. He is also Professor of Medical Genetics at the Faculty of Medicine of the University of Naples "Federico II" and Visiting Professor at both Baylor College of Medicine in Houston, Texas, and at the University of Oxford, UK. Prof. Ballabio's research interests are the elucidation of the biological mechanisms underlying genetic diseases and the development of innovative therapeutic approaches. Prof. Ballabio's team identified numerous genes whose mutations cause human inherited diseases, leading to the discovery of their pathogenetic mechanisms. Among them are: the XIST gene, which is the major player in X-inactivation, the genes for Kallmann syndrome (KAL) Ocular albinism (OA1), spastic paraplegia (SPG7), Opitz/GBB syndrome (MID1) gene, Multiple sulfatase deficiency (SUMF1), and a master transcriptional regulator of lysosomal biogenesis and autophagy (TFEB). Prof. Ballabio's current research focuses on lysosomal biology and the mechanisms underlying lysosomal storage disorders and common neurodegenerative diseases. He has published over 280 papers in international scientific journals. Prof. Ballabio was the President of the European Society of Human Genetics and Council member of the European Molecular Biology Organization (EMBO). He is a recipient of an Advanced Investigator Grant of the European Research Council (ERC). He has received numerous national and international awards for research and culture, among which the 2007 Award of the European Society of Human Genetics. In 2007 he was received the "Knighthood of the Italian Republic" by the President of Italy.

Marco Baptista, PhD, The Michael J. Fox Foundation for Parkinson's Research
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Marco Baptista earned an undergraduate degree in Psychology from the University of Toronto and a PhD in Neuroscience from McMaster University, Canada. After completing his postdoctoral research at The Scripps Research Institute in La Jolla, Marco spent over 5 years in the pharmaceutical industry leading a preclinical Parkinson's program. Marco brings drug discovery knowledge and expertise to The Michael J. Fox Foundation for Parkinson's Research that helps drive the funding of translational research.

Ann Barbier, MD, PhD, Shire
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Ann Barbier obtained her MD in 1989 and a PhD in pharmacology in 1995. She has worked at various pharmaceutical companies, including Johnson & Johnson and Sanofi-Aventis, prior to joining Shire Human Genetic Therapies in 2007, where she is the Global Clinical Lead for the Elaprase, intrathecal treatment for Hunter Syndrome and intrathecal treatment for Sanfilippo A Syndrome programs. Her experience with orphan diseases also includes hereditary angioedema and Huntington's Disease.

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Judith Blanz, PhD, University of Kiel
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Judith Blanz received her PhD in 2005 from the laboratory of Prof. Dr. T. Jentsch at the Center for Molecular Neurobiology (ZMNH) in Hamburg, Germany where she studied the neuronal function of the chloride channel ClC-2. Since 2006, she has been working in the group of Prof. Dr. P. Saftig at the University of Kiel as a group leader. Dr. Blanz is interested in lysosomal function and associated neurodegenerative diseases such as lysosomal storage disorders (LSD). Within recent years, the main focus of her research has been the i) development of a preclinical enzyme replacement therapy for the LSD α-mannosidosis using a recombinant human enzyme as well as ii) studying the function of the lysosomal integral membrane protein LIMP-2 as a trafficking receptor for the sphingolipidase β-glucocrebrosidase. Recent studies suggest an important role of LIMP-2 in α-synuclein metabolism which will be the scope of her future research.

Patricia Boya, PhD, Centro de Investigaciones Biológicas (CSIC)
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Patricia Boya graduated in Biological Sciences in 1994 and obtained her PhD in 2000 at the University of Navarra, Pamplona, Spain. She was a post-doctoral fellow at CNRS in Villejuif, France (2001-2004) and University of Cambridge, U.K. (2005). Then she obtained a competitive Ramón y Cajal researcher contract at the Centro de Investigaciones Biológicas (CSIC), Madrid, where she currently holds a senior research scientist position. Her lab uses cellular and animal models to understand the physiological roles of lysosomes and autophagy and its implications during disease. They have demonstrated that autophagy plays a cytoprotective role in animal models of Parkinson Disease and in models of axonal damage. Her lab has several projects with pharmaceutical companies to screen for new drugs that modulate autophagy with the aim to find new treatments for cancer, neurodegenerative diseases and other pathological conditions.

Melanie Brickman Stynes, PhD, MSc, The New York Academy of Sciences
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Melanie Brickman Stynes serves as the Director of Life Sciences Conferences at the New York Academy of Sciences. Dr. Brickman Stynes has nearly 15 years of experience in public health, primarily as a researcher focused on the juncture of health, demography, policy, and geography. Prior to joining the Academy, Dr. Brickman Stynes was Associate Director of the Institute on Science for Global Policy (ISGP). Additionally, Dr. Brickman Stynes spent nearly a decade as a Research Associate for the Center for International Earth Science Information Network (CIESIN) of Columbia University, where she worked on a range of projects related to health, disease, poverty, urbanization, and population issues. She also taught as an adjunct Professor at Baruch College's School of Public Affairs for five years. She received her Ph.D. in medical geography from University College London (UCL) and her M.Sc. in medical demography from the London School of Hygiene and Tropical Medicine (LSHTM).

Seng H. Cheng, PhD, Genzyme, a Sanofi Company
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Seng Cheng is Head of Research and Early Development of the Rare Diseases Division at Genzyme, a Sanofi Company. Dr. Cheng received his BSc, and PhD degrees in Biochemistry from the University of London, U.K. He trained as a postdoctoral fellow at the National Institute for Medical Research in London, U.K., in the field of tumor biology. He was a Staff Scientist at Integrated Genetics Inc., and later joined Genzyme Corporation to work on several discovery projects including the structure and function of the cystic fibrosis transmembrane conductance regulator. As Group Vice President of Genetic Diseases Science at Genzyme, he also managed the development of novel gene delivery systems as well as translational research in genetic diseases, a number of which transitioned to clinical testing. Areas of focus included inherited metabolic, muscle, lung and neurodegenerative diseases. He has co-authored 245 research articles and reviews, and is a named co-inventor on 50 issued patents in the area of biotechnology. In his current position, he is responsible for directing the translational research and early clinical development activities in rare genetic diseases.

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Sean Clark, PhD, Amicus Therapeutics
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Sean Clark is Director of Biochemistry and Cell Biology at Amicus Therapeutics. He received his BS in Molecular, Cell, and Developmental Biology and his PhD in Molecular Biology at UCLA. His postdoctoral studies at Princeton University focused on the study of actin-related proteins on mitotic spindle orientation and brought to bear tools from molecular biology, biochemistry, cell biology, and yeast genetics. Dr. Clark joined Amicus Therapeutics in 2005 to lead the Gaucher-Parkinson's program. He has had a number of successful collaborations with both Gaucher and Parkinson's experts in academia and managed multiple studies at external CROs. His work has included the investigation of the biochemical mechanism connecting Gaucher and PD, the identification of new small molecules targeting GCase, and their efficacy in Gaucher and Parkinson's models. Dr. Clark has also investigated the origin

Jonathan Cooper, PhD, King's College London
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Jonathan Cooper founded the Pediatric Storage Disorders Laboratory (PSDL) when he was appointed at the Institute of Psychiatry, Psychology & Neuroscience, King's College London in 2000. Prior to this he trained in Anatomy and Cell Biology (BSc, Sheffield), and Neuroanatomy (PhD, Bristol), and spent time as a postdoctoral researcher at the University of Cambridge, the Max-Planck-Institute for Psychiatry (Martinsried, Germany), and in California at the University of California, San Francisco and Stanford University. The PSDL has described many of the important neuropathological features of the Neuronal Ceroid Lipofuscinoses, defining vulnerable neuron populations and the relationship of storage material accumulation, glial activation and synaptic pathology to this neuron loss. A major focus of the lab is in defining the role of glia in NCL pathogenesis. The lab also continues to be involved in a range of pre-clinical studies assessing the efficacy of a range of experimental therapies, and collaborates widely with colleagues around the world. He also works closely with the affected families.

Susan L. Cotman, PhD, Massachusetts General Hospital, Center for Human Genetic Research
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Susan L. Cotman, PhD, is an Assistant Professor of Neurology at Harvard Medical School and Assistant Professor in Neuroscience at Massachusetts General Hospital. Dr. Cotman's research program specializes in developing genetic, cell biological and biochemical tools for investigation of neuronal ceroid lipofuscinosis (NCL/ Batten disease). Current goals include delineating the earliest events that result from the NCL genetic defects to gain insight into the functions of the NCL proteins, identifying pharmacologic and genetic NCL disease modifiers that will lead to NCL therapies, and the identification of new NCL genes, which will improve our understanding of the NCL disease pathways and NCL diagnosis.

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Biography not available at timre of printing.

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Christina H. Eng, PhD, is a Principal Scientist in the Oncology Research Unit at Pfizer. She obtained dual undergraduate degrees in Biology and Chemistry at the Massachusetts Institute of Technology and received her doctorate at Columbia University. Her research interest in autophagy stemmed from work as a postdoctoral fellow at Wyeth Research (currently Pfizer), where she identified a byproduct of glutamine metabolism that modulates autophagy. Current research in her group is focused on examining diverse aspects of cancer metabolism to identify targetable metabolic liabilities in tumor cells, and probing the role of ubiquitin-related enzymes in cancer biology.

Tony Futerman, PhD, Weizmann Institute of Science
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Tony Futerman is the The Joseph Meyerhoff Professor of Biochemistry and head of the Nella and Leon Benoziyo Center for Neurological Diseases at the Weizmann Institute of Science in Rehovot, Israel. He runs a research laboratory of about 20 people that focuses on the cell biology and biochemistry of sphingolipids, a major lipid class in cells, and the roles that they play in health and disease. A major research area concerns understanding the pathological mechanisms at play in neuronopathic forms of Gaucher disease. Dr. Futerman was a member of the Editorial Board of the *Journal of Biological Chemistry* from 2000-2012, was the chair of the 2006 Gordon Conference on Glycolipid and Sphingolipid Biology and was the chair of the first Gordon Conference on Lysosomal Diseases held in 2011.

Brooke Grindlinger, PhD, The New York Academy of Sciences
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Brooke Grindlinger serves as the Executive Director of Scientific Programs at the New York Academy of Sciences, providing strategic development and oversight of the Academy's international portfolio of scientific workshops, conferences, symposia, and related multimedia publications across the broad spectrum of Life Sciences, Physical Sciences, Computer Science, and Engineering. Through this platform – and via strategic alliances with external organizations, foundations, and individuals – the Academy convenes leading international scientists from academia, industry, and government sectors in focused efforts to catalyze advances in science, medicine, engineering, and innovation for the benefit of society. Dr. Grindlinger also serves as a member of the board of the Sackler Institute for Nutrition Science at the New York Academy of Sciences, established in 2010 in partnership with The Mortimer D. Sackler Foundation, Inc., to advance nutrition science research.

Dr. Grindlinger has more than 12 years of experience in scientific research, academic publishing, and science communication. Prior to joining the Academy, she served for 8 years as Science Editor for *The Journal of Clinical Investigation*, managing the review and publication of state-of-the-art basic and clinical biomedical research across the continuum of human physiology and disease, in addition to authoring journal News features, Editorials, Book Reviews, and Press Releases. Dr. Grindlinger received her Bachelor of Science (*First Class Honours*) degree and PhD in molecular and microbial biosciences from the University of Sydney, Australia, studying the pathogenesis of the tuberculosis-causing organism *Mycobacterium tuberculosis* and ways in which to boost the efficacy of the tuberculosis vaccine. For this postgraduate work, Dr. Grindlinger was the recipient of an Australian Postgraduate Award. Dr. Grindlinger also regularly conducts local, national, and international workshops on science communication skills and alternative science careers for early career scientists.

ATTENDEE BIOGRAPHIES

Warren Hirst, PhD, Pfizer Inc
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Warren Hirst, PhD, is an associate research fellow in the Pfizer Neuroscience Research Unit and leads a group focusing on novel therapeutic strategies for Parkinson's disease. This includes research into understanding the role of LRRK2, GBA and protein misfolding in Parkinson's disease. Dr. Hirst leads the programs to identify LRRK2 kinase inhibitors and GBA activators / stabilizers. He was previously in the Wyeth Neurodegeneration Research Department, where he led the molecular pharmacology group and worked for GlaxoSmithKline, UK, before this. Dr. Hirst received his BSc and PhD from Imperial College, London. He has published over 50 peer-reviewed articles.

Michaël Hocquemiller, PhD, Lysogene
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Michaël Hocquemiller, PhD, is the head of the Scientific Reseach Unit at LYSOGENE. He is in charge of all scientific intelligence of the company, he keeps track of biobank samples collected during clinical trials and is responsible for preclinical studies and those related to biomarkers and immune response research. He is also in charge of internal and external scientific communication and he is editor of the website and social networks. Prior to joining LYSOGENE, Michaël combined a twofold experience in preclinical and clinical research that allowed him to have a strong knowledge of lysosomal storage diseases and gene transfer. From 2004 to 2009, Michaël worked as researcher at "Retrovirus et Transfert Génétique" Unit of the Pasteur Institute (INSERM U622). His research focused on neural stem cells reprogramming by gene transfer and on Sanfilippo syndrome neurophysiopathology and his correction by gene therapy in animal models. Michaël has a PhD in neuroscience from University Paris Descartes (Paris V) and a Master in genetics from University Pierre et Marie Curie (Paris VI). He is also certified Clinical Research Associate (SupSanté).

Samantha J. Hutten, PhD, Michael J. Fox Foundation for Parkinson's Research
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Samantha Hutten, after graduating cum laude with a degree in Neuroscience and Behavior from Vassar College in 2005, worked at a small biotechnology company, where she performed behavioral characterization and drug testing in mouse models of Huntington's disease. After that, she obtained her PhD in Biomedical Sciences at Albert Einstein College of Medicine, where she trained in the laboratory of Dr. Ana Maria Cuervo, studying autophagy and neurodegeneration (with a special focus on Parkinson's Disease). As part of her thesis work, she identified the mechanism by which the protein most commonly associated with Parkinson's disease (LRRK2) impairs chaperone-mediated autophagy. Samantha's work was published in *Nature Neuroscience* and was featured on the NIH Director's Blog. She is thrilled to be Associate Director of Research Programs at the Michael J. Fox Foundation for Parkinson's Research, where she manages biomarkers grants in the foundation's portfolio. Her focus is in new assay development for the identification, optimization, and validation of Parkinson's disease biomarkers. She oversees the process of soliciting investigator proposals, peer review, awarding and contracting funds, troubleshooting, and assessing projects as they go forward.

Marja Jäättelä, MD, PhD, Danish Cancer Society Research Center
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Marja Jäättelä is head of the Cell Death and Metabolism research unit at the Danish Cancer Society Research Center in Copenhagen and professor in Cancer Biology at the University of Copenhagen. She received her MD and PhD degrees from the University of Helsinki and performed postdoctoral training at the Memorial Sloan-Kettering Cancer Center in New York and at the University of Michigan in Ann Arbor. The major goal of her present research is to enlighten the mechanisms by which cancer cells escape cell death induced by cancer treatment and to find new ways to kill them. Her research focuses on exploring the cancer-associated changes in the composition (proteins and lipids), trafficking and function of lysosomes and autophagosomes, cancer-relevant signaling pathways responsible for cross-communication between transformation, invasion, cell death and survival as well as mechanisms underlying resistance to chemotherapy.

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Danielle Kerkovich, PhD, Beyond Batten Disease Foundation
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Danielle Kerkovich, PhD, has served as the Principal Scientist of Beyond Batten Disease Foundation for the past 4 years, where she has created a strategic plan to treat and cure juvenile Batten disease [https://beyondbatten.org/research/our-strategy/]. To date, the plan has yielded a dozen published discoveries in the top 10% of 13,000 science and technology journals cited over 500 times and growing, has leveraged into twice the amount of funding originally invested, has initiated 30 partnerships between academic researchers, pharmaceutical scientists, and others, has resulted in the development of multiple animal models of disease, and has resulted in the first large-scale, genetically diverse, human platform for drug discovery in juvenile Batten or any pediatric neurodegenerative disease. Danielle received her PhD and MS degrees in Biomedical Sciences from the Albert Einstein College of Medicine of Yeshiva University with a focus in Developmental Neuroscience. She has extensive experience working with federally funded research through the Department of Veterans Affairs as the Assistant Director for Rehabilitation Research and Spinal Cord Injury/Central Nervous System Disorders Portfolio Manager of the Veterans Administration 157-hospital system. At the start of current U.S. military conflicts through 2007, she identified current gaps in healthcare for soldiers returning from Iraq and Afghanistan, designed over 50 scientific review panels, authored congressional briefing books, and proposed unique funding mechanisms between the VA, the Department of Defense, and others. In addition, she has worked as an advisor to medical research foundations in pediatric cancer, cerebral palsy and burn injury. Dr. Kerkovich has been a peer reviewer for the National Institutes of Health, National Science Foundation, the National Institute on Disability Rehabilitation Research, and Defense Advanced Research Projects Agency.

Thomas Kirkegaard Jensen, PhD, Orphazyme
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Thomas Kirkegaard Jensen is currently the Chief Scientific Officer at Orphazyme ApS, a biotech company developing Heat Shock Protein based therapies for rare, genetic diseases. He has held this position since March 2010. Prior to this, he held positions as CEO of Orphazyme and post-doctoral fellow at the Danish Cancer Society working with Heat shock proteins and lysosomal involvement in cancer and genetic diseases. He is the main founder of Orphazyme, having discovered and patented the founding scientific basis of Orphazyme which was published in *Nature* 2010 (Kirkegaard et al., *Nature* (463), pp549-53). During his training in Medical Biology and his PhD, he became intrigued by the role of the Heat shock response in health and disease. Thus he has spent the past decade focusing on understanding the physiological function and interplay of two key metabolic systems from a molecular pathology perspective: the Heat shock response and lysosomal metabolism with a main interest in the interplay between these systems in various disease states. Besides his position in Orphazyme, he serves on the advisory board of Rare Disease Report (**www.raredr.com**) and is co-founder and vice-chairman of Rare Disease Council Denmark (**www.orphan.dk**), a non-profit organization striving to improve the accessibility of drugs to patients with rare diseases.

Mary Beth Kiser, Beyond Batten Disease Foundation
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Mary Beth Kiser joined BBDF in December 2012. She brings over 20 years of professional experience to the foundation including fundraising and business development, board and volunteer management, and program development. She had been with the Texas Land Title Association since 2003 where she was the Senior Vice President and Chief Operating Officer responsible for the day-to-day operations, including the association's programs, products and services. Mary Beth currently serves on the Board of the Texas Society of Association Executives and earned her Certified Association Executive credentials in 2009. Mary Beth is a graduate of the University of Texas and has been a community volunteer for over 25 years having worked with and led many central Austin organizations and fundraising events.

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Rainer Kuhn, PhD, Evotec AG
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Rainer Kuhn is EVP Neuroscience at Evotec AG (Hamburg, Germany). He received his PhD in 1989 at the Institute of Genetics in Düsseldorf (Germany) studying molecular mechanisms of germ cell development in *Drosophila melanogaster*. Following postdoctoral training in molecular neurobiology at the Salk Institute in La Jolla he joined Ciba-Geigy/Novartis Neuroscience in 1992. He initiated research on metabotropic glutamate receptors, and identified with his team the first allosteric mGluR compounds and the mGluR5 antagonist AFQ056 (Mavoglurant). Over the years he served as Project leader, Unit Head and Executive Director for Neurodegeneration and Neuroregeneration, where he managed large research efforts in psychiatric indications, Alzheimer's disease, Parkinson's disease, multiple sclerosis, Huntington's disease, spinal cord injury, biomarkers and novel stem cell-based neuronal assay systems.

In 2012, he co-founded the biotech startup Promidis (Rome, Italy) focusing on biomarker and therapeutics discovery for Huntington's disease. Since 2013, he serves on the board of the German NCL foundation (neuronal ceroid lipofuscinosis, Batten disease) in Hamburg (Germany). He has published more than 100 peer-reviewed papers.

Salvatore La Rosa, PhD, Children's Tumor Foundation
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Salvatore La Rosa is Senior Director of Research and Development at the Children's Tumor Foundation a non-profit 501(c)(3) medical foundation, dedicated to promote and support research and the development of treatments and cures for the different types of neurofibromatosis or NF. He is responsible for Managing of the Foundation's Drug Discovery activities, providing scientific and knowledgeable review of discovery, preclinical and early development programs in the field of NF. He is also responsible for the development and management of novel partnerships and initiatives with academic research groups and biotech/pharma companies. Among his specific responsibilities is the NFTC preclinical consortium for NF1, and the management of 'Synodos for NF2': a 8 academic centers/12 Principal Investigator initiative. Previous to joining the Foundation, Dr. La Rosa worked as Group Leader of a Medicinal Chemistry unit at Siena Biotech (Italy) serving also as Project Leader for multiple projects in the area of Huntington's Disease and Oncology. He also worked for CRO companies as Senior Medicinal Chemist with Nikem Research (Milan – Italy) and Evotec OAI (Abingdon, UK) developing early hit and lead compounds into clinical candidates. He holds a PhD in Medicinal chemistry from the University of Strathclyde in Glasgow (UK) and a Master of Science in Chemistry from the University of Messina (Italy).

Peter Lansbury, PhD, Lysosomal Therapeutics Inc.
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Peter Lansbury, PhD, is the Chief Scientific Officer at Lysosomal Therapeutics Inc. Peter Lansbury manages the company's research activities. He also retains his position as a professor of neurology at Harvard Medical School. Previously, Peter was Founder and Chief Scientific Officer of Link Medicine until it was acquired by Astra Zeneca in 2012. Peter served as a Director of the Morris K. Udall Research Center of Excellence in Parkinson's disease at Brigham and Women's Hospital. Peter received his doctorate from Harvard University, where he worked for Nobel laureate E. J. Corey.

Jonathan H. LeBowitz, PhD, Biomarin Pharmaceuticals Inc.
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Jonathan H. LeBowitz is a Staff Scientist and Director of Cell and Molecular Biology at BioMarin Pharmaceuticals. Formerly, he was the Executive Vice President and Chief Scientific Officer at ZyStor Therapeutics, Inc. where he was an inventor of a novel peptide-based lysosomal targeting technology called GILT, short for Glycosylation Independent Lysosomal Targeting, and led the development of a candidate Pompe ERT incorporating this technology which now being evaluated in a Phase 3 clinical trial. He is an accomplished investigator with over 35 years of research experience in the biochemistry, molecular, and cell biology of organisms ranging from microbes to humans and an inventor on 14 US patent. Prior to joining ZyStor, Dr. LeBowitz was a tenured Associate Professor of Biochemistry at Purdue University. Dr. LeBowitz was a post-doctoral fellow at Harvard Medical School and MIT and received a PhD in Biochemistry from The Johns Hopkins University.

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Geoffrey Ling, MD, PhD, Defense Advanced Research Projects Agency (DARPA)
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Geoffrey Ling is the Director of the Biological Technologies Office at the Defense Advanced Research Projects Agency (DARPA) and attending neuro critical care physician at Johns Hopkins Hospital. He retired from the US Army in 2012 after serving as a military intensive care physician with multiple deployments to Iraq and Afghanistan. He formerly served in the Science Division at the White House Office of Science and Technology Policy. He received his PhD in pharmacology from Cornell University, and MD from Georgetown University. Dr. Ling is board certified in both Neurology and Neuro Critical Care.

Peter Lobel, PhD, Center for Advanced Biotechnology and Robert Wood Johnson Medical School, Rutgers University
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Peter Lobel is Associate Director of the Center for Advanced Biotechnology and Medicine (CABM) and Professor of Biochemistry and Molecular Biology at Rutgers-Robert Wood Johnson Medical School (RWJMS). He also serves as Director of the RWJMS-Rutgers University Biological Mass Spectrometry Facility. Lobel's laboratory is best known for the development of proteomic approaches to characterize the lysosome and to investigate the role of lysosomal proteins in human disease. This research allowed them to discover disease genes involved in two fatal hereditary childhood neurodegenerative disorders, late infantile neuronal ceroid lipofuscinosis and Niemann Pick type C2 disease. An immediate benefit from this work was the development of methods to definitively diagnose and screen for these diseases. The Lobel laboratory has since made key contributions towards understanding the underlying biology and developing therapies for lysosomal diseases.Lobel received his PhD in Biochemistry from Columbia University. He conducted postdoctoral studies with Dr. Stuart Kornfeld at Washington University School of Medicine in St Louis and was recruited to CABM and RWJMS in 1988. He was named a Searle Scholar and received the 2014 Edward J. Ill Outstanding Medical Research Scientist Award for Basic Biomedical Research.

Shuyan Lu, MS, Pfizer Inc
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Shuyan Lu is a Principal Scientist with the Investigative Toxicology Group at Pfizer La Jolla. Her research efforts have been focusing on how physicochemical properties contribute to compounds' lysosomal accumulation and lysosomal function disruption. This effort led to the mechanism identification of multiple organ toxicities (*e.g.* cardiotoxicity and retinal toxicity). Her current projects aim to understand how lysosomal dysfunction contributes to signaling modulation and various pathological processes.

Frederick R. Maxfield, PhD, Weill Cornell Medical College
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Fred Maxfield is the Vladimir Horowitz and Wanda Toscanini Professor and Chair of Biochemistry at Weill Cornell Medical College. For over 30 years his laboratory has used biophysical methods to carry out quantitative analyses of cell functions. Much of their work involves quantitative fluorescence imaging. He has studied lipoprotein endocytosis and trafficking, and his laboratory developed novel methods to image well-behaved fluorescent sterols and other fluorescent lipids in cells. A major project in the laboratory is analyzing the mechanisms of intracellular sterol and lipid transport. They have used quantitative imaging to develop a very effective method to screen compounds that would be effective in treating Niemann Pick C cells and screened over 40,000 compounds using this screen. Among the validated hits were HDAC inhibitors, including SAHA/Vorinostat – a drug approved by the FDA for treatment of some cancers.

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Matthew Mealiffe, MD, is Medical Director, Clinical Sciences at BioMarin Pharmaceutical Inc. where he works in the group focused on lysosomal storage disorders with involvement in clinical trials focused on Mucopolysaccharidosis Type IVA and Pompe Disease. After receiving his MD from the Yale University School of Medicine, he trained in Internal Medicine and Medical Genetics at the University of Washington. There he pursued research related to furthering understanding of the genetic basis of familial Hodgkin's lymphoma funded by a Doris Duke Clinical Scientist Development Award and saw patients as an attending physician with a practice primarily focused on Clinical Cancer Genetics and Adult Medical Genetics.

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Kalpana Merchant established a life sciences business that provides consultancy services for drug discovery and development and associated technologies in April 2014. In this capacity, she is an Advisor to the Michael J Fox Foundation for Parkinson's Research and a member of their Executive Scientific Advisory Board, a member of the Scientific Advisory Board for Lysosomal Therapeutics, Inc., and an advisor to Third Rock Ventures and other venture or start-up companies. Prior to establishing TransThera Consulting, Kalpana worked in the US pharmaceutical industry in drug discovery research for nearly 21 years as an individual contributor as well as in leadership and management roles. Most recently she was at Eli Lilly and Company as the Chief Scientific Officer for Tailored Therapeutics where she was accountable for scientific and business strategies to deliver tailored therapies with associated biomarkers and companion diagnostics for the neuroscience portfolio – from discovery through Phase III. The neuroscience tailoring strategy leveraged the infrastructure of expertise, technologies, and approaches she established as the Chief Scientific Officer of Translational Science, which supported neuroscience, oncology, metabolic disorders and musculoskeletal therapeutic areas at Eli Lilly. Kalpana received her PhD in neuropharmacology from the University of Utah. Following a postdoctoral research fellowship at University of Washington, she was appointed as an Assistant Professor of Psychiatry at the same institution. She was recruited to Lilly in 2003 from a position of Senior Research Advisor and Fellow at Pharmacia Corp., where she had contributed to neuroscience drug discovery research for about 10 years. Kalpana is engaged in the wider scientific community via her service on NIH Study Sections, Workshops and Advisory Panels, scientific advisory board for the Michael J Fox Foundation for Parkinson's Research as well as membership in several national and international professional societies.

Matthew C. Micsenyi, PhD, Biogen Idec Inc.
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Matthew Micsenyi is a Research Scientist in the Neurology Research Group at Biogen Idec in Cambridge, MA. His current work focuses on developing therapeutic strategies for neurodegenerative proteinopathies. He received his PhD and postdoctoral training from the Albert Einstein College of Medicine in the laboratory of Steve Walkley, working in the field of lysosomal diseases. His research interests are in lysosomal and neurodegenerative disease pathogenesis, with an emphasis on defining autophagy-lysosomal system dynamics and protein aggregation in these disorders.

ATTENDEE BIOGRAPHIES

Myriam Mirza, PhD, Beyond Batten Disease Foundation
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Myriam Mirza currently works as a Consultant Fellow with Beyond Batten Disease Foundation and as a Healthcare Consultant specializing in medical devices and pharmaceuticals with Junicon Healthcare Consultancy Firm. She earned her Bachelors and Masters in Chemical Biology and Biochemistry from McGill University in Montreal, Canada. She then travelled to Regensburg, Germany where she learned German and completed her PhD in Biology at the Institute of Human Genetics in the University Hospital Regensburg. Myriam has focused on inherited childhood diseases since the beginning of her scientific carrier, first working on Cystic Fibrosis and then on Neuronal Ceroid Lipofuscinoses (NCL). Her PhD on CLN6 helped establish that microglia cells in the mammalian retina and brain are indeed responsible for accelerating neuronal cell death, and this damage cycle can be reduced by natural anti-inflammatories found in every day foods. Her goal as a Healthcare Consultant is to create a better information exchange platform where governments, academics, societies and the pharmaceutical industry can collaborate to accelerate research in rare diseases.

Joseph Nabhan, PhD, Pfizer Inc
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Joseph Nabhan is a senior scientist in the rare disease research unit at Pfizer. His training has primarily focused on studies that relate to the ubiquitin proteasome and autophagy pathways. He has studied a proteasome associated deubiquitinase and examined pathways of endosomal sorting that are modulated by ubiquitination, including receptor trafficking and membrane budding. Other studies have focused on modulation of autophagy and autophagic flux as a potential avenue for chemotherapy. At Pfizer, he was involved in a project aimed at developing pharmacological chaperones for glucocerebrosidase as therapy for Gaucher type I disease. Dr. Nabhan has a long-standing interest in lysosomal biology and its role in cellular homeostasis.

Ralph A. Nixon, MD, PhD, New York University Langone Medical Center; Nathan S. Kline Institute
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Ralph Nixon received his PhD from Harvard University, MD from University of Vermont, and training in medicine and psychiatry at Massachusetts General Hospital. He is a Fellow of the American College of Neuropsychopharmacology. Dr. Nixon's research was the first to establish the importance of proteases and defective proteolytic systems in the pathogenesis of Alzheimer's disease and has identified new therapeutic approaches for the disease. A major focus of his research is on the pathogenic importance of endosomal-lysosomal dysfunction in neurodegenerative diseases, which revealed presenilins as essential for lysosome function and presenilin mutations, the most common cause of early onset Alzheimer's disease, as key accelerants of the disease through lysosomal mechanisms. He has published over 250 scientific papers and is the holder of nine issued and pending patents. He currently serves as Immediate Past Chairman of the Medical and Scientific Advisory Council (MSAC) of the national Alzheimer's Association and is also a member of the Association's National Board of Directors. He also serves on the Governor's Commission on Alzheimer's disease for New York State. Dr. Nixon's awards include the Leadership and Excellence in Alzheimer Research, MERIT, and Academic Career Leadership Awards from the National Institutes of Health, and the Zenith and Temple Discovery Awards from the Alzheimer's Association.

Rebecca Oberman, PhD, The ML4 Foundation
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Rebecca Oberman is the first executive director of the Mucolipidosis Type IV (ML4) Foundation. Since she began in 2012, she has worked to facilitate greater communication among the MLIV scientific and patient communities, as well as to build collaborations between those communities and our partners at the NIH and FDA. During the past two years, she has organized two research conferences focused on MLIV science (New York, 2013; Atlanta 2014), with vital participation from the scientific and patient communities. Her work encompasses patient support and communication, fundraising for research, outreach to interested new investigators, maintaining support for current research programs, registry development and enrollment, and more.

ATTENDEE BIOGRAPHIES

Elizabeth Ottinger, PhD, National Institutes of Health (NIH); National Center for Advancing Translational Sciences (NCATS)
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Elizabeth Ottinger is a Senior Project Manager for the Therapeutics for Rare and Neglected Diseases (TRND) program within the National Center for Advancing Translational Sciences (NCATS) at the National Institutes of Health (NIH). In this position, she is responsible for leading projects from pre-clinical studies to IND filing, building team collaborations/partnerships, and developing processes and project management infrastructure within TRND. She currently manages four TRND projects, Niemann Pick Type C1 Disease, Cryptococcal Meningitis, Creatine Transporter Deficiency, and LEOPARD Syndrome. Her most recent position before joining TRND was in the Department of Vaccine Research at Merck Research Laboratories, as part of the antigen lead identification and validation group, working on the development of a broad range of bacterial and anti-viral vaccines. Prior to joining Merck, she was at the University of Pennsylvania developing biochemical and cell based assays for high throughput screening of targets for Spinal Muscular Atrophy (SMA). Dr. Ottinger holds a PhD from the University of Minnesota in Organic Chemistry, and completed her post-doctoral work at the Joslin Diabetes Center, Harvard Medical School.

Frances M. Platt, PhD, University of Oxford
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Frances Platt obtained her PhD from the University of Bath, UK, and was a post-doctoral fellow at Washington University Medical School in St. Louis, USA. She was a Lister Institute Senior Research Fellow and is currently Professor of Biochemistry and Pharmacology at the University of Oxford. Her main research interests include the biology and pathobiology of glycosphingolipids. Her research led to the development of miglustat for the treatment of glycosphingolipid storage diseases. Prof. Platt was awarded the Alan Gordon Memorial Award and the Horst Bickel Award for advances in metabolic disease therapy. She was elected a fellow of the Academy of Medical Sciences in 2011.

Forbes D. Porter, MD, PhD, National Institutes of Health (NIH); The *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD)
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Forbes Porter is a Senior Investigator and Program Head in the intramural research program of the Developmental Endocrinology and Genetics Program (PDEGEN) of the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD). Dr. Porter also serves as the Clinical Director for NICHD. Dr. Porter received both his MD and PhD degrees from Washington University in St. Louis, and trained in both Pediatrics and Clinical Genetics at St. Louis Children's Hospital. He then moved to the NIH as a postdoctoral fellow in 1993, and started his own research group in 1996. His research program studies basic, translational, and clinical aspects of genetic disorders with impaired cholesterol homeostasis. These include Smith-Lemli-Opitz syndrome and Niemann-Pick Disease, type C. The goal of this research program is to understand pathophysiogical processes underlying these disorders, develop therapeutic interventions, and implement therapeutic trials. Dr. Porter works closely with patient advocate groups. He is on the scientific/medical advisory boards of both the RSH/Smith-Lemli-Opitz Foundation and the National Niemann-Pick Disease Foundation.

ATTENDEE BIOGRAPHIES

Alexey Pshezhetsky, PhD, University of Montreal, CHU Sainte-Justine University Hospital
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Alexey Pshezhetsky graduated and obtained PhD degree in chemistry from Moscow State University. In 1989-1992 as a researcher in Moscow State University and Moscow Institute of Medical and Biological Chemistry he studied genetic diseases of children caused by the inherited deficiencies of lysosomal enzymes. In 1993 he joined Departments of Pediatrics and Biochemistry, University of Montreal, where he was subsequently promoted to professor. Since 1998 Dr. Pshezhetsky has been a scientific director of the medical genetics diagnostic laboratory and a director of a Laboratory of lysosomal biology at Ste-Justine Hospital Research Center and an adjunct professor at the Department of Anatomy and Cell Biology, McGill University. He has received many career awards including a National Investigator Award, an Award of Excellence in Pediatric Research from the Foundation for the Research in Children's Disorders and Champion of Genetics award from Genes for Cure Foundation. His current research interests include the molecular basis of lysosomal and other inherited metabolic disorders, glycobiology, proteomics and functional genomics of the cell.

Rosa Puertollano, PhD, National Institutes of Health (NIH); National Heart, Lung, and Blood Institute (NHLBI)
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Rosa Puertollano received a BS and a MS in Biochemistry and Molecular Genetics from the Universidad Autonoma de Madrid, Spain, in 1994, and a PhD in Biochemistry and Molecular Biology from the Consejo Superior de Investigaciones Cientificas (CSIC), Spain, in 1999. Following her graduation she undertook a postdoctoral fellowship at the National Institute of Child Health and Human Development (NICHD), National Institutes of Health (NIH) under the supervision of Dr. Juan Bonifacino. In 2004, Dr. Puertollano joined the National Heart, Lung, and Blood Institute (NHLBI, NIH) as a Principal Investigator where she is the head of the Protein Trafficking and Organelle Biology Section

Marco Sardiello, PhD, Baylor College of Medicine
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Marco Sardiello was trained in Genetics and Molecular Evolution at the University of Bari (Italy), where he identified a gene network that regulates mitochondrial biogenesis and energy metabolism in Drosophila. Subsequent post-doctoral training at the Telethon Institute of Genetics and Medicine (Italy) was focused on the analysis of large cohorts of gene families and lysosomal function and culminated in the identification of a gene network that regulates lysosomal biogenesis and function in mammals, and of its master regulator, transcription factor EB (TFEB). In 2010 he established a laboratory at the Baylor College of Medicine – Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital in Houston (Texas). His lab is currently using genetics, proteomics, cell biology and systems biology approaches to study how the cell regulates its metabolic programs and how dysfunctions in these programs lead to neurological disease. In particular, his lab is focusing on three lines of research: (1) Use of TFEB-mediated lysosomal enhancement to counteract disease progression in Batten disease and other lysosomal storage disorders; (2) Study of the molecular pathogenesis of the Neuronal Ceroid Lipofuscinoses; (3) Characterization of metabolic gene networks regulated by transcription factors or microRNAs that control the basal metabolism of the cell. The ultimate goal of these research programs is to translate knowledge of key regulatory pathways into therapeutic approaches for neurodegenerative disorders.

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Biography not available at time of printing.

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ATTENDEE BIOGRAPHIES

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James A. Shayman, MD is professor of internal medicine and pharmacology at the University of Michigan. He received his undergraduate degree from Cornell University and medical degree from Washington University, St. Louis. He trained in internal medicine and nephrology at Barnes Hospital and received post-doctoral training in pharmacology also at Washington University. His primary research interests have focused on the biochemistry, cellular biology, and pharmacology of glycosphingolipid storage diseases including Gaucher and Fabry disease. His group has targeted glucosylceramide synthase resulting in the development of eliglustat tartrate for Gaucher disease type 1. The Shayman group has recently focused on the design and development of CNS active glycolipid synthesis inhibitors. Additional work includes the discovery and characterization of group XV phospholipase A2, a novel lysosomal lipid hydrolase.

Diana Shineman, PhD, Alzheimer's Drug Discovery Foundation
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Diana Shineman, PhD, is the Director for Scientific Affairs at the Alzheimer's Drug Discovery Foundation, where she develops and manages the Foundation's drug discovery and development grant programs and strategic initiatives. Combining scientific and business expertise, the ADDF manages its research funding portfolio to balance risk, stage of development, and drug target mechanism of action, ensuring that grants meet key milestones before securing follow-on funding. As a measure of success, projects funded by the ADDF have gone on to garner nearly $2 billion in follow-on funding. The ADDF also works strategically with foundations, government and industry partners to tackle unmet needs in the field. As an example of such an initiative, Dr. Shineman led an interdisciplinary effort to standardize animal model study design to improve research efficiency and translatability. Diana joined the ADDF in 2008. She earned a PhD in Cell and Molecular Biology from the University of Pennsylvania working in the Center for Neurodegenerative Disease Research led by Drs. Virginia Lee and John Trojanowski. She also worked as an Editorial Intern for the Journal of Clinical Investigation and was an active member of the Penn Biotechnology Group. Diana received a BA in Biology with a Nutrition concentration from Cornell University, where she was named a Howard Hughes Undergraduate Research Scholar. In addition to maintaining various professional memberships, Diana has also authored numerous articles and peer-reviewed publications.

Katherine B. Sims, MD, Massachusetts General Hospital; Harvard Medical School
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Katherine Sims is Director of the MGH Developmental Neurogenetics and Mitochondrial Disorders Clinic as well as the MGH NCL Disorders Clinic – a BDSRA designated Center of Excellence. These clinics serve as a local, regional and national/international resource for the diagnostic evaluation of genetic neurodegenerative disorders for both pediatric and adult patients and their families. Her principal clinical and research interests have been directed to heritable neurogenetic disorders. She is an active member of the MGH-CHGR Joint Program in the NCL Disorders dedicated to diagnosis, clinical care and translational research studies for patients of all ages with NCL. Dr. Sims established the MGH NCL Clinical Database and BioRepository over 15 years ago. Patient characterization and biosample collection continues as a major effort aimed at characterizing the phenotypes of the NCL disorders and to establish a patient sample resource that will be useful to researchers in the development of model systems for these disorders. Dr. Sims is the founding Director of the MGH Neurogenetics DNA Diagnostic Lab. This molecular DNA service lab was started in 1994 to provide clinical DNA testing services in rare neurogenetic disorders and to accelerate translation of neuromolecular data from MGH/Partners research labs to the clinical arena. Testing is now done for >30 neurodegenerative disorders including all of the NCL disorder genes. This lab serves as national and international resource and as a teaching site for both the Harvard Molecular Genetics and Molecular Pathology fellowship programs.

ATTENDEE BIOGRAPHIES

William S. Sly, MD, Saint Louis University School of Medicine
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William S. Sly, MD, directed the Division of Medical Genetics at Washington University for 20 years, after which he chaired the Department of Biochemistry and Molecular Biology at Saint Louis University for 26 years, ending in 2010. He currently holds the James B. and Joan C. Peters Endowed Chair in Biochemistry and Molecular Biology. His group described the first patient with MPS VII (Sly syndrome) and worked with collaborators at The Jackson Laboratory to characterize the mouse model of this disease. He also headed studies that identified the mannose-6 phosphate and mannose receptors that target enzymes to lysosomes, which provided the rationale for enzyme replacement therapy in Gaucher's disease and other lysosomal storage diseases. For this work, he was inducted into the National Academy of Sciences in 1989. He has also received awards from the National (US) and International MPS Societies. Current work involves ERT for MPS VII and, specifically, brain-directed ERT.

John R. Swart, PhD, Exemplar Genetics
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John R. Swart is the president of Exemplar Genetics, a biomedical research company focused on the development of models of human diseases such as cystic fibrosis, muscular dystrophy, heart disease and cancer. The company is in its sixth full year of business, has seen its revenues grow each year, and has a strong pipeline of models. Exemplar has been able to establish itself as the global commercial leader in this field through its patented technologies, established infrastructure, and proven results. The company currently employs over 20 people and is rapidly growing. Prior to starting with Exemplar, Dr. Swart worked for twelve years for the global animal health business of Boehringer Ingelheim. While with Boehringer, Dr. Swart served in several capacities including leading a team in Research and Development that was focused on new product development and life cycle management, held the position of Executive Director of Operations for a site that produced animal health vaccines, served as Regulatory Affairs Liaison for USDA and FDA inspections, provided oversight for a GMP level Quality Assurance program, and worked with Sales and Marketing for Key Opinion Leader development. He also participated in Boehringer's International Management Development Program leading a team that developed recommendations for improved business planning within the company. Dr. Swart received his BA for Northwestern College of Iowa in 1990 and his PhD from the University of Nebraska in biochemistry in 1994. Dr. Swart serves on the Animal Biotechnology Policy Committee of the Biotechnology Industry Organization and has extensive experience in business planning, product development, regulatory acceptance, and market expansion.

P. Herman van der Putten, PhD, National Contest for Life (NCL) Foundation
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Herman van der Putten is scientific officer of the National Contest for Life (NCL) Foundation in Hamburg, that facilitates and funds targeted research to fight Batten disease. Between 1986 and 2012 he worked in the Pharmaceutical Industry for Ciba-Geigy and Novartis in Basel, Switzerland. From 2004-2012 he was Executive Director in Neuroscience Discovery (Novartis) heading research units in neuropsychiatry and neurodegeneration. He was responsible for innovation, the design and the implementation of scientific strategies in these areas focusing on target discovery, target validation, academic collaborations, preclinical drug development and translational aspects for proof-of-concept testing in human. He also headed is own laboratory and served as the nervous system discipline expert in molecular cell biology and animal model councils. Some of his most recent discovery projects concerned molecular pathways and targets in Huntington disease and Parkinson's disease. He is a co-author of over 100 publications, received EMBO and ZWO postdoctoral fellowships, and worked at the University of Geneva and the Salk Institute in San Diego. In 1981, he received his PhD from the Radbaud University in Nijmegen, The Netherlands.

ATTENDEE BIOGRAPHIES

Steven U. Walkley, DVM, PhD, Albert Einstein College of Medicine
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Steven Walkley is Professor of Neuroscience, Pathology and Neurology and Director of the Rose F. Kennedy Intellectual and Developmental Disabilities Research Center at the Albert Einstein College of Medicine (New York, USA). His research interests began during his early training in Comparative Medicine and Neuroscience and involved animal models of GM1 and GM2 gangliosidosis (Sandhoff disease). His laboratory today is focused on defining the pathogenesis of numerous endosomallysosomal system disorders including Niemann-Pick types A and C, the gangliosidoses, Farber, MLIV, MPS IIIA and the NCIs, as well as the recently discovered endosomal disease, Christianson syndrome. Of particular focus are disease-induced changes in endocytic, autophagic and salvage pathways and their impact on dendritic and axonal integrity and the neuronal connectome. Dr. Walkley's lab has also been in the forefront of therapy development for lysosomal disorders, including the first and presently only approved therapy for Niemann-Pick type C disease. Dr. Walkley was a co-founder of the newly developed Gordon Research Conference on Lysosomal Disease and is an active member of the scientific advisory boards for numerous lysosomal disease organizations.

Qingjun Wang, PhD, University of Kentucky
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Qingjun Wang obtained her undergraduate degree with physics major from University of Science & Technology of China in 1996. She pursued her graduate study on photosynthesis under Dr. John Whitmarsh at the University of Illinois at Urbana-Champaign. After obtaining a PhD in 2002, she joined Dr. Brian Chait's Laboratory of Mass Spectrometry & Gaseous Ion Chemistry at The Rockefeller University, where she began to utilize an integrated mouse genetic-proteomic approach to elucidate the molecular mechanism of autophagy. She identified several novel autophagy protein-protein interactions in mouse brains, and demonstrated that these interactions are important for autophagy regulation. Moreover, using a neuroexcitotoxicity mouse model, she showed unequivocating autophagy induction in neurons exposed to excitotoxic insults; and using a neuron-specific autophagy-deficient mouse model, she demonstrated that autophagy plays a cell-autonomous and indispensable protective role in the healthy brain. In 2009, Qingjun established her independent laboratory at the University of Kentucky to investigate the molecular mechanism of mammalian autophagy, and the roles of autophagy in health and disease. Her lab recently made exciting discovery that autophagy is active in platelets and is essential for hemostasis. Two years ago, her lab began investigating the connections among autophagy, CLN3, and JNCL.