Tuesday, February 11, 2014

Basic Science I Co-Chairs: Roscoe Brady and Perry Hackett

8:00	**Chester B Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and opening remarks
8:15	**Gregory A Grabowski** Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Keynote address: How did we get here, and where are we going?
8:45	**Kirsten E McKay** Birmingham Women's Hospital Birmingham, United Kingdom	Detection of Niemann-Pick disease type C mutations in infants with liver disease using targeted next generation sequencing
9:00	**Emyr Lloyd-Evans** Cardiff University Cardiff, United Kingdom	Uncovering the mechanism by which miglustat mediates benefit in NPC disease; role of acid sphingomyelinase in NPC disease pathogenesis
9:15	**Mengqiao Wang** National Center for Advancing Translational Sciences, National Institutes of Health Rockville, MD, United States	Delta-tocopherol facilitates generation of induced pluripotent stem cells from fibroblasts with Niemann-Pick disease type C
9:30	**Yoshikatsu Eto** Institute of Neurological Disease Kawasaki City, Japan	Generation and application of iPS cells for lysosomal disorders
9:45	**Jeong-A Lim** NIAMS/NIH Bethesda, MD, United States	Mitochondrial abnormalities in Pompe disease
10:00	**Break & Exhibits**	
10:15	**Paula Rozenfeld** Universidad Nacional de La Plata, LISIN La Plata, Argentina	Osteoclastogenesis functional study of osteoclasts precursors from Gaucher disease patients
10:30	**Rick Hamler** Amicus Therapeutics Cranbury, NJ, United States	Glucosylceramide and glucosylsphingosine quantitation by liquid chromotography-tandem mass spectrometry to enable studies of neuronopathic Gaucher disease
10:45	**Mia Horowitz** Tel Aviv University Ramat Aviv, Israel	The connection between ERAD, UPR, Gaucher disease and Parkinson disease
11:00	**Gheona Altarescu** Shaare Zedek Medical Center Jerusalem, Israel	α-Synuclein polymorphisms in patients with Gaucher disease and Parkinson disease
11:15	**Manoj K Pandey** Division of Human Genetics,	Gaucher disease: glucosylceramide-mediated TLR4-MyD88 induction cause enhanced CXCL-13

	Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	secretion and increased B cell trafficking in a mouse model
11:30	**Lunch Break**	On your own – *or* - COPA (Council of Patient Advocates)

Basic Science II Co-Chairs: Elizabeth Neufeld and Scott McIvor

1:00	**Anatália Labilloy** University of Pittsburgh Pittsburgh, PA, United States	Altered dynamics of a lipid raft associated protein in a kidney model of Fabry disease
1:15	**Xingli Meng** Baylor Research Institute Dallas, TX, United States	Abnormal intracellular calcium handling: a key pathogenic and therapeutic target of the cardiac manifestations in Fabry disease
1:30	**Haiyan Fu** Research Institute at Nationwide Children's Hospital Columbus, OH, United States	Correction of broad molecular impairments in a mouse model of MPS IIIA by systemic rAAVrh74-hSGSH gene delivery
1:45	**Janine Gilkes** University of Florida Gainesville, FL, United States	AAV8 is preferential candidate for neonatal gene transfer in Sanfilippo syndrome type B model
2:00	**Keisuke Kitakaze** The University of Tokushima Tokushima, Japan	Replacement effects of human modified lysosomal β-hexosaminidase B on Tay-Sachs and Sandhoff disease models and Imaging with novel pH-activatable fluorescent probes
2:15	**Shaalee Dworski** University of Toronto Toronto, ON, Canada	Altered MCP-1 and ceramide metabolite levels in serum from Farber mice and Farber patients
2:30	**Raymond Y Wang** CHOC Children's Specialists Orange, CA, United States	Aortic gene expression from the canine model of MPS I identifies upregulation of genes related to antigen presentation and inflammatory cytokines, and downregulation of cellular adhesion and cytoskeletal genes
2:45	**Jan Lukas** University of Rostock Rostock, Germany	Small molecule enhancers for mutant enzymes in lysosomal disorders
3:00	**Break & Exhibits**	
3:15	**Jessica L Fletcher** The University of Sydney Camperdown NSW, Australia	Early molecular changes in canine fucosidosis
3:30	**Brittney Gurda** University of Pennsylvania Philadelphia, PA, United States	Gene therapy for mucopolysaccharidosis VII: Evaluation of intrathecal rAAV vectors in the canine model
3:45	**Silvia Muro**	Pulmonary delivery and effects of recombinant acid