Tuesday, February 11, 2014

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

Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States 8:45	8:00	Chester B Whitley	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, United Kingdom Uncovering the mechanism by which miglustat mediates benefit in NPC disease; role of acid sphingomyelinase 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; role of acid stargeted next generation sequencing 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 Mitochondrial abnormalities in Pompe disease 9:45 Jeong-A Lim NIAMS/NIH Bethesda, MD, United States Mitochondrial abnormalities in Pompe disease 10:20 Paula Rozenfeld Universidad Nacional de La Plata, LISIN La Plata, Argentina Osteoclastogenesis functional study of osteoclasts		University of Minnesota	
Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States		Minneapolis, MN, United States	
Medical Center Cincinnati, OH, United States	8:15	Gregory A Grabowski	Keynote address: How did we get here, and where
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	Cincinnati Children's Hospital	secretion and increased B cell trafficking in a mouse
	Medical Center	model
	Cincinnati, OH, United States	
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

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