

WORLD Symposium 2009

Wednesday, February 18, 2009		
BASIC AND BENCH RESEARCH DAY		
Session 1		Co-Chairs: Steven Walkley, Dwight Koerberl
8:00	Chester B. Whitley University of Minnesota, Minneapolis, MN	Welcome
		Introduction of LDN Awardee
8:15	Elizabeth F. Neufeld UCLA, Los Angeles, CA	New findings in the brain of the MPS III B mouse model
8:45	Steven U. Walkley Albert Einstein College of Medicine, Bronx, NY	The Greater Lysosomal System and Lysosomal Disease
9:00	Ralph Nixon Nathan Kline Institute Orangeburg, NY	Autophagy Dysfunction in Alzheimer's Disease and Other Late-Onset Neurodegenerative Diseases
9:20	Andrew Lieberman University of Michigan, MI	Autophagy in the Pathogenesis of Niemann-Pick C Disease
9:40	Break	
10:00	Andrea Ballabio Fondazione Telethon, Italy	Impairment of Autophagy in Lysosomal Storage Diseases
10:20	Silvia Vergarajauregui National Institutes of Health, Bethesda, MD	Autophagic Dysfunction in Mucopolysaccharidosis Type IV Patients
10:35	Matthew Micsonyi Albert Einstein College of Medicine, Bronx, NY	P62/Sequestosome 1 Accumulates in the CNS of Lysosomal Diseases
10:50	Mark Sands Washington University School of Medicine, St. Louis, MO	Metabolic and Clinical Adaptations to Interrupted Lysosomal Recycling
11:05	Charles Roe Baylor University Medical Center, Dallas, TX	IGF-1 Resistance is a Reversible Feature of Late-Onset Acid Maltase Deficiency (AMD)
11:25	Discussion	
11:45	Session concludes	
	Lunch Break	<i>LDN Steering Committee, Advisory Committee, & U54 Research Investigators' Lunch (12:00-1:00 PM) INVITATION ONLY</i>
Session 2	Lysosomal Protein Function and Disease Pathogenesis	Co-Chairs: Beverly Davidson, Patricia Manos
1:15	Rodney Infante University of Texas Southwestern Medical Center, Dallas, TX	NPC2 Facilitates Bidirectional Transfer of Cholesterol Between NPC1 and Lipid Bilayers, A Potential Step in Cholesterol
1:35	Edward Schuchman Mount Sinai School of Medicine, New York, NY	Construction & Characterization of Mutation-Specific Mouse Models for Types A and B Niemann-Pick Disease
1:50	Beverly Davidson University of Iowa, Iowa City, IA	Enzyme Replacement in Animal Models of the Late Infantile Form of Batten Disease
2:05	Sandra Hofmann University of Texas Southwestern Medical Center, Dallas, TX	Metabolic Profiling of Infantile Neuronal Ceroid Lipofuscinosis
2:20	Angela Schulz University Medical Center Hamburg, Hamburg, Germany	Retention of Lysosomal Protein CLN5 in the Endoplasmic Reticulum Causes Neuronal Ceroid Lipofuscinosis in Asian Sibship
2:35	Amelia Ahern-Rindell University of Portland, Portland, OR	Analysis of Beta-Galactosidase in an Ovine Model of GM1-Gangliosidosis
2:50	Discussion	
3:10	Break	
3:25	Mia Horowitz Ramat Aviv, Israel	Interaction Between Parkin and Glucocerebrosidase: A Possible Link
3:40	Xiaoyang Qi	Role of Saposin C in Multivesicular Body

	University of Cincinnati, Cincinnati, OH	Bioformation and Neuropathogenesis
3:55	Kirill Kiselyov University of Pittsburgh, Pittsburgh, PA	Membrane Traffic and Cell Death in Mucopolipidosis Type IV
4:10	Participant Interactive Session	Using ARS technology for participant feedback, discussion, and educational measurement
4:25	Discussion	
4:40	LDN Research Proposals	Co-Chair: John Barranger, Elsa Shapiro
	Elsa G. Shapiro University of Minnesota, Minneapolis, MN	Neuropsychological Outcomes in Hurler Syndrome: 7-20 Years Post-Hematopoietic Stem Cell Transplantation
	Agnes Chen, MD UCLA Medical Center, Torrance, CA	Intrathecal Enzyme Replacement Therapy for Cognitive Decline in MPS I
	Lynda Polgreen University of Minnesota Minneapolis, MN	High Prevalence of Low Bone Mineral Density in Children with Hurler Syndrome after Hematopoietic Stem Cell
	Michael Potegal University of Minnesota, Minneapolis, MN	Behavioral Phenotypes in MPS III
	Raphael Schiffman Institute of Metabolic Disease, Dallas, TX	Mucopolipidosis Type IV is a Both a Developmental Brain Disease and a
	Sara Cathey Greenwood Genetic Center, N. Charleston, SC	Collective Strength in Rare Diseases: Longitudinal Studies of the Glycoproteinoses
5:30	Poster Session opens	
6:30	Poster Session closes	
	TRANSLATIONAL RESEARCH DAY	
Thursday, February 19, 2009		
Session 3	Translational Research I	Co-Chairs: Gary Murray, Robert Steiner
8:00	Gary Murray National Institute of Neurological Disorders and Stroke, NIH, Bethesda, MD	Introduction of the LDN Awardee
8:10	William Sly Saint Louis University School of Medicine, St. Louis, MO	Going the Extra Mile: Strategies to Deliver Enzyme to Resistant Sites Including Delivery Across the Blood-brain Barrier for Neuronal
8:30	Hong Du Cincinnati Children's Hospital Research Foundation, Cincinnati, OH	Wolman Disease/Cholesteryl Ester Storage Disease: Efficacy of Plant-Produced Human Lysosomal Acid Lipase in Mice
8:45	Barry Byrne University of Florida, Gainesville, FL	Treatment Strategies for Pompe Disease: Lessons from Preclinical and Clinical
9:00	Bruce Bunnell Tulane University School of Medicine, New Orleans, LA	Adipose Stem Cell-mediated Therapy for Krabbe's Disease
9:20	Participant Interactive Session	Using ARS technology for participant feedback, discussion, and educational measurement
9:35	Discussion	
9:50	Break	
10:00	Jeffrey Medin Ontario Cancer Institute, University Health Network, Toronto, Canada	Outcomes of Testing Lentivector-Mediated Gene Therapy for Farber Disease in Non-Human Primates
10:15	R. Scott McIvor University of Minnesota, Minneapolis, MN	Ex Vivo Lentiviral Gene Transfer Targeting Hematopoietic Stem Cells for Therapy of Mucopolysaccharidosis Type II, Hunter
10:30	Stephanos Kyrkanides Stony Brook University, NY	The Trigeminal Retrograde Transfer Pathway in Lysosomal Storage Disease Gene Therapy
10:45	David Bedwell University of Alabama, Birmingham, AL	Suppression of a Nonsense Mutation in a Mouse Model of Hurler Syndrome
11:00	Perry Hackett University of Minnesota Minneapolis, MN	Gene Therapy of MPS I Mice Using the Sleeping Beauty Transposon System
11:15	Krystof Bankiewicz	Translational Studies of AAV2-hASM Gene Transfer for Niemann-Pick Disease

	University of California, San Francisco	
11:30	Discussion	
11:45	Session Concludes	
	Lunch Break	<i>Council of Patient Advocates (COPA) Lunch (12:00-1:00 PM) INVITATION ONLY</i>
Session 4	Translational Research II	Co-Chairs: Greg Grabowski, TBD
1:15	Hans Aerts Academic Medical Center, Amsterdam, Netherlands	Biomarkers in Lysosomal Storage Diseases
1:35	Don Mahuran The Hospital For Sick Children, Toronto, ON, Canada	The Effects of Altering Calcium Homeostasis on Mutant Enzyme Activity in Chronic Tay- Sachs and Gaucher Patient Cells Treated
1:50	Kenneth Valenzano Amicus Therapeutics, Inc. Cranbury, NJ	A Combination rhGAA-Small Molecule Therapeutic Approach for the Treatment of Pompe Disease
2:05	Brigitte Rigat The Hospital for Sick Children, Toronto, ON, Canada	Identification of Pharmacological Chaperones for Feline and Human GM1 Gangliosidosis
2:20	Aryan Namboodiri Uniformed Services University, Bethesda, MD	Canavan Disease Treatment Using Glyceryltriacetate
2:35	Synthia Mellon University of California, San Francisco, CA	Neurosteroid Treatment of Lysosomal Storage Disorders
2:50	Cristin Davidson Albert Einstein College of Medicine, Bronx, NY	Chronic Cyclodextrin Administration Ameliorates Clinical Symptoms and Storage Accumulation in Niemann-Pick Type C1 Mice
3:05	Discussion	
3:20	Break	
3:30	Jeffrey Kelly Scripps Research Institute La Jolla, CA	Proteostasis
3:50	Calogera Simonaro Mount Sinai School of Medicine, New York, NY	Novel Biomarkers and Therapies for the Mucopolysaccharidoses
4:05	Melita Dvorak-Ewell BioMarin Pharmaceutical Inc., Novato, CA	Human Primary Chondrocytes, a Relevant Model of Mucopolysaccharidosis IVA,
4:20	Discussion	
4:30	LDN Research Proposals	Co-Chair: Joseph Muenzer, Elsa Shapiro
	Forbes Porter National Institutes of Health Bethesda, MD	Longitudinal Study of Cognition in Subjects with Niemann-Pick Disease Type C
	TBD	Phase I Trial of Pyrimethamine to Treat LOTS
	Kendra Bjoraker University of Minnesota, Minneapolis, MN	A Natural History Study of Hexosaminidase Deficiency
	Chester B. Whitley University of Minnesota Minneapolis, MN	Gene Therapy for Tay-Sachs Disease
5:30	Poster Session opens	
6:30	Poster Session closes	
7:00	Banquet	
Friday, February 20, 2009	CLINICAL RESEARCH DAY	
Session 5	Clinical Research I	Co-Chairs: Roscoe Brady, William Wilcox
8:00	Roscoe O. Brady	Introduction

	National Institute of Neurological Disorders and Stroke, NIH, Bethesda, MD	
8:05	Marc Patterson Mayo Clinic, Rochester, MN	Small Molecule Therapies for Neurologic Manifestations of Lysosomal Storage
8:25	Ellen Sidransky MGB, NHGRI, Bethesda, MD	Gaucher Disease and Parkinsonism
8:40	Neal Weinreb University Research Foundation for Lysosomal Storage Disorders, Coral Springs, FL	A Validated Disease Severity Scoring System for Gaucher Disease Type 1
8:55	Judith Peterschmitt Genzyme Corporation, Cambridge, MA	Genz-112638, an Investigational Oral Treatment for Gaucher Disease Type 1:
9:10	Einat Almon Protalix Biotherapeutics, Carmiel, Israel	Novel Enzyme Replacement Therapy for Gaucher Disease: On Going Phase III Clinical
9:25	Discussion	
9:40	Break	
9:55	Paul Orchard University of Minnesota, Minneapolis, MN	Combined Intrathecal Iduronidase, Intravenous Iduronidase and Transplantation as Therapy for Hurler Syndrome
10:15	Lawrence Charnas University of Minnesota, Minneapolis, MN	Comparison of Concordance Ratios of Symptom Onset in Metachromatic Leukodystrophy (MLD) Sib Pairs Suggests
10:30	Elizabeth Braunlin University of Minnesota, Minneapolis, MN	Cardiac Risk Stratification for Individuals with MPS IH-S: Where We Are Today
10:45	Gerald F. Cox Genzyme Corporation, Cambridge, MA	Genotype Frequencies in the MPS I Registry
11:00	Joseph Muenzer University of North Carolina, Chapel Hill NC	Clinical Phenotype of North American Patients with Hunter Syndrome: Data From HOS: The Hunter Outcome Survey
11:15		Musculoskeletal health in MPS II: Pediatric Outcomes Data Collection Instrument (PODCI) demonstrates functional improvements in patients on ERT
	Klane White Seattle Children's Hospital, Seattle, WA	
11:30	Discussion	
11:45	Session Concludes	
	Lunch Break	Council of Industry Professionals (COIP) Lunch Meeting (12:00-1:00 PM) INVITATION ONLY
Session 6	Clinical Research II	Co-Chairs: Greg Pastores, Elsa Shapiro
1:15	Ed Wraith Royal Manchester Children's Hospital, Manchester, UK	Disease Stability in Patients with Niemann-Pick Disease Type C Treated with Miglustat
1:30	Joe T.R. Clarke Hospital for Sick Children, Toronto, ON, Canada	Substrate Reduction Therapy with Miglustat in Juvenile GM2-gangliosidosis
1:45	Alfried Kohlschuetter University of Hamburg Germany	Metachromatic Leukodystrophy: A Scoring System for Brain MR Observations
2:00	Patricia Duffner Hunter James Kelly Research Institute, Buffalo, NY	Outcomes of Children Transplanted for Krabbe Disease
2:15	Maria L. Escolar University of North Carolina - Chapel Hill, NC	DTI with Quantitative tractography as a Marker of Disease Progression in Newborns
2:30	Discussion	
2:45	Break	
3:00	Laura E Case	Clinical Signs and Symptoms of Pompe

	Duke University Medical Center, Durham, NC	Disease in 120 Infantile Onset and 373 Late Onset Patients: A Report from the Pompe
3:15	Bruno Bembi University Hospital Santa Maria della Misericordia Udine, Italy	Enzyme Replacement Therapy with Alglucosidase Alfa in Juvenile-Adult Glycogenosis Type 2 Patients.
3:30	Victor De Jesus Centers for Disease Control and Prevention, Atlanta, GA	Results from a Pilot Quality Control Program for Lysosomal Storage Disorder Newborn Screening
3:45	Dolan Sondhi Weill Cornell Medical College, New York, NY	AAV-mediated Gene Therapy for the CNS Manifestations of the Lysosomal Storage Disorders
4:00	Participant Interactive Session	Using ARS technology for participant feedback, discussion, and educational measurement
4:15	Discussion	
4:30	LDN Research Proposals	Co-Chair: Chester Whitley, Elsa Shapiro
	Greg Grabowski Cincinnati Children's Hospital Research Foundation, Cincinnati, OH	Longitudinal Natural History Studies of Wolman and Cholesteryl Ester Storage Diseases
	Dwight Koeberl Duke University Medical Center, Durham, NC	CRIM Responses in Pompe Disease
	Marsha Browning Massachusetts General Hospital and Harvard Medical School, Boston, MA	High Throughput Detection and Characterization of Fabry Disease in At-Risk Cardiovascular, Renal, and Neurology Populations
	Michael Mauer University of Minnesota, Minneapolis, MN	Podocyte injury and GL-3 Accumulation are Progressive in Fabry Disease
	Nancy Lyon University of Chicago, Chicago, IL	Health, Developmental, and Functional Outcome Surveillance in Preschool Children
	William Wilcox Cedars-Sinai Medical Center, Los Angeles, CA	Pulmonary Disease and Exercise Tolerance in Boys with Fabry Disease
5:30	Chester B. Whitley	Concluding Remarks