

WORLD Symposium 2008 Program

Subject to change

Wednesday, February 13, 2008

Session 1

Wednesday Morning

BASIC AND BENCH RESEARCH

Pathophysiology of Lysosomal Diseases

**Steven Walkley,
Don Mahuran,
Co-Chairs**

- 8:30 Chester Whitley** Welcome
University of Minnesota, Minneapolis, MN
- 8:45 Edwin Kolodny** 1 Keynote Address:
New York University School of Medicine, New York, NY "Reflections On The Current State of Diagnosis For LSDs"
- 9:00 Susan Slaugenhaupt** 2 A murine model for mucopolidosis type IV
Massachusetts General Hospital/Harvard Medical School, Boston, MA
- 9:15 Tyler Mark Pierson** 3 Novel mutations in juvenile Sandhoff disease
Neurogenetics Branch, NINDS/NIH, Bethesda MD presenting as motor neuron disease
- 9:30 David Priestman** 4 Glycosphingolipid profiling of tissues from mouse
Department of Pharmacology, University of Oxford, Oxford UK models of human lysosomal storage disorders.
- 9:45 Discussion**
- 10:00 Break**
- 10:15 Tim Edmunds** 5 Biochemical characterization of the N370S
Genzyme Corporation, Framingham, MA glucocerebrosidase mutant: Implications for chaperone therapy
- 10:30 Kathleen Hruska** 6 Functional evaluation of predicted regulatory
MGB, NHGRI, NIH, Bethesda, MD sequences at the GBA locus
- 10:45 Richard Steet** 7 Altered chondrocyte differentiation in
University of Georgia, Athens, GA mucopolidosis II zebrafish
- 11:00 Margaret McGovern** 8 Skeletal manifestations in Niemann-Pick disease
Stony Brook University, Stony Brook, NY due to acid sphingomyelinase deficiency

WORLD Symposium 2008 Program, cont.

- 11:15 Edward Schuchman** 9 Mouse models for types A and B Niemann-Pick disease: Insights into pathogenesis and treatment
Mount Sinai School of Medicine, New York, NY
- 11:30 David Pearce** 10 Moving toward therapies for JCNL
University of Rochester, Rochester, NY
- 11:45 Discussion**
- 12:00 Lunch on your own**

Session 2 Wednesday Afternoon

Future Therapeutic Approaches

**David Pearce,
Edward Schuchman,
Co-Chairs**

- 1:00 Dwight Koeberl** 11 Muscle-targeted gene therapy in glycogen storage disease type II with adeno-associated virus serotypes 7,8, and 9
Duke University Medical Center, Durham, NC
- 1:15 Perry Hackett** 12 Comparison of gene expression in vivo from Sleeping Beauty transposons after hydrodynamic delivery or condensation by polyethylenimine and cationic lipids
University of Minnesota, Minneapolis, MN
- 1:30 Don Mahuran** 13 Identification of Ambroxol as a potential enzyme enhancement-agent for Gaucher disease
Hospital For Sick Children, Toronto, ON, Canada
- 1:45 Ozlem Goker-Alpan** 14 The contribution of lysosomal pathways to the pathogenesis of common neurodegenerative disorders: Glucocerebrosidase and the synucleinopathies
MGB, NHGRI, NIH, Bethesda, MD
- 2:00 Mia Horowitz** 15 The possible implication of lowering cholesterol level in Gaucher disease patients
Tel Aviv University, Ramat Aviv, Israel
- 2:15 Catherine Kielar** 16 Reactive and synaptic changes precede neuronal loss in mouse models of NCL
Institute of Psychiatry, King's College London, UK
- 2:30 Jonathan Cooper** 17 Immunosuppression as a novel therapeutic target in Batten disease
Institute of Psychiatry, King's College London, UK
- 2:45 Discussion**
- 3:00 Break**

WORLD Symposium 2008 Program, cont.

3:15	Walter Low 18 <i>University of Minnesota, Minneapolis, MN</i>	Targeting neural stem cells in the mammalian brain with intraventricular injections of lenti, AAV5, or Sleeping Beauty vectors
3:30	Charles Pontikis 19 <i>King's College London, UK</i>	In situ perfusion in a mouse model of Sanfilippo syndrome
3:45	Mark Haskins 20 <i>University of Pennsylvania School of Veterinary Medicine, Philadelphia, PA</i>	Intrathecal enzyme therapy in mucopolysaccharidosis I cats reduces storage throughout the brain
4:00	R. Scott McIvor 21 <i>University of Minnesota, Minneapolis, MN</i>	Gene therapy of mucopolysaccharidosis type I: Intraventricular administration of adeno-associated virus vector transducing the human alpha-L-iduronidase gene in a murine model of the disease
4:15	Edward Ginns 22 <i>University of Massachusetts Medical School, Shrewsbury, MA</i>	Development of a novel ingestible macrophage-targeted gene therapy for Gaucher disease
4:30	<i>Poster Session</i>	
6:00	<i>Poster Session Adjourns</i>	

Thursday, February 14, 2008

Session 3 Thursday Morning

Research Projects

Chester Whitley, Chair

8:30	<i>Introduction and Overview</i>
8:45	<i>LDN Research Projects</i>
9:30	<i>LDN Research Cores</i>
10:00	<i>Discussion</i>
10:30	<i>Break</i>

Measurement of Disease Progression Panel

Edward Giannini,
Elsa Shapiro,
Co - Chairs

10:45	Edward Giannini 23 <i>Cincinnati Childrens Hospital, Cincinnati, OH</i>	Development of a disease severity scoring system for Anderson-Fabry disease
--------------	---	---

WORLD Symposium 2008 Program, cont.

10:55	Nicole Yanjanin 24 <i>NICHD, NIH, DHHS, Bethesda, MD</i>	Disease progression in Niemann-Pick disease, type C: Longitudinal
11:05	Edward Giannini 25 <i>Cincinnati Childrens Hospital, Cincinnati, OH</i>	Development of a disease severity scoring system for patients with Pompe disease
11:15	Jennifer Kwon 26 <i>University of Rochester Medical Center, Rochester, NY</i>	Quantifying the rate of neurologic decline in Batten disease in clinical trials
11:25	Ari Zimran 27 <i>Shaare Zedek Medical Center, Jerusalem, Israel</i>	Development of a disease severity scoring system for type I Gaucher disease
11:35	<i>Discussion</i>	
11:50	<i>Lunch on your own</i>	

Session 4 Thursday Afternoon

Newborn Screening Panel

**Rodney Howell,
John Barranger,
Co - Chairs**

1:00	Rodney Howell <i>Special Assistant to the Director, NICHD, NIH</i>	Introduction
1:10	Michele Caggana 28 <i>NYSDOH/Wadsworth Center, Albany, NY</i>	Newborn screening for Krabbe disease in New York state: Experience from the first year
1:20	Joan Keutzer 29 <i>Genzyme Corporation, Framingham, MA</i>	An update on newborn screening for Pompe disease
1:30	Olaf Bodamer <i>University Children's Hospital, Vienna, Austria</i>	Newborn Screening in Austria
1:40	Christiane Auray-Blais 30 <i>Universiti de Sherbrooke, Sherbrooke, Quebec, Canada</i>	Mass urinary screening for Fabry disease: Is it feasible?
1:50	Victor De Jesus 31 <i>Centers for Disease Control and Prevention</i>	Recent developments on lysosomal storage disorder activities at the centers for disease control and prevention
2:00	<i>Discussion</i>	
2:15	<i>Break</i>	

WORLD Symposium 2008 Program, cont.

Clinical Outcomes		William Wilcox, Ellen Sidransky, Co-Chairs
2:30	Elsa Shapiro 32 <i>University of Minnesota, Minneapolis, MN</i>	Neuropsychological function and neuroimaging in severe and attenuated mucopolysaccharidosis
2:45	Patricia Dickson 33 <i>LA Biomed at Harbor-UCLA, Torrance, CA</i>	Initial experience with intrathecal recombinant human alpha-L-iduronidase for spinal cord compression in two mucopolysaccharidosis I patients
3:00	<i>Break</i>	
3:15	Jeanine Utz 34 <i>University of Minnesota, Minneapolis, MN</i>	Delayed infusion reactions associated with enzyme replacement therapies for lysosomal storage disorders.
3:30	Maria Escolar 35 <i>University of North Carolina, Chapel Hill, NC</i>	Long-term developmental follow-up of babies treated for infantile Krabbe disease with unrelated cord blood transplantation
3:45	Lynda Polgreen 36 <i>University of Minnesota, Minneapolis, MN</i>	Growth hormone therapy improves growth in Hurler syndrome with minimal effect on scoliosis, kyphosis, or genu valgum.
4:00	Christine Eng 37 <i>Department of Molecular and Human Genetics, Houston, TX</i>	Characterization of symptom onset and clinical events in patients with Fabry disease: findings from the Fabry Registry
4:15	<i>Discussion</i>	
4:30	<i>Poster Session</i>	
6:00	<i>Poster Session closed</i>	
6:30	<i>Banquet</i>	
7:30	<i>Banquet and Program conclude</i>	

WORLD Symposium 2008 Program, cont.

Friday, February 15, 2008

Session 5
Friday Morning

CLINICAL TRIALS Spectrum of Clinical Outcomes

Robert Steiner,
Danilo Tagle, Co-Chairs

8:30	Presentation of 2007 LDN Award	
8:45	Roscoe Brady <i>Scientist Emeritus, NINDS, NIH, Bethesda, MD</i>	Keynote Address, 2008 LDN Awardee for Innovation and Accomplishment
9:15	Hans Andersson 38 <i>Tulane University Medical School, New Orleans, LA</i>	Clinical outcome following 8-year enzyme replacement therapy in 884 children with type I Gaucher disease (GD1)
9:30	David Lockhart 39 <i>Amicus Therapeutics, Cranbury, NJ</i>	Pharmacological chaperone treatment for lysosomal storage disorders
9:45	Jakub Tolar 40 <i>University of Minnesota, Minneapolis, MN</i>	Antioxidant neuroprotection with hematopoietic cell transplantation in cerebral adrenoleukodystrophy
10:00	<i>Discussion</i>	
10:15	<i>Break</i>	
10:30	William Wilcox 41 <i>Cedars-Sinai Medical Center, Los Angeles, CA</i>	Natural history of Fabry disease: Progression of the nephropathy in a large series of affected males and heterozygous females
10:45	Beth Thurberg 42 <i>Genzyme Corporation, Framingham, MA</i>	Pathology of Fabry nephropathy: Renal fibrosis may begin in adolescence
11:00	Elizabeth Jacklin 43 <i>Royal Manchester Childrens Hospital, UK</i>	The natural history of Niemann-Pick disease type C
11:15	Neal Weinreb 44 <i>University Research Foundation for Lysosomal Storage Diseases, Coral Springs, FL</i>	Effect of enzyme replacement therapy with imiglucerase (Cerezyme.) every 4 weeks in patients with type I Gaucher disease
11:30	<i>Discussion</i>	
11:45	<i>Lunch on your own</i>	

WORLD Symposium 2008 Program, cont.

Session 6 Friday Afternoon

Gaucher Disease: Focus for Innovation

Joseph Muenzer,
Gregory Grabowski,
Co-chairs

1:00	Gregory Grabowski 45 <i>University of Cincinnati College of Medicine, Cincinnati, OH</i>	Dose-response relationships for enzyme replacement therapy with imiglucerase / alglucerase in patients with Gaucher disease type I
1:15	Ellen Sidransky 46 <i>MGB, NCHRI, NIH, Bethesda, MD</i>	The association between mutant glucocerebrosidase and Parkinsonism
1:30	Gregory Pastores 47 <i>NYU School of Medicine, New York, NY</i>	Clinical trials with miglustat in patients with type 1 Gaucher disease (GD1)
1:45	Einat Almon 48 <i>Protalix Biotherapeutics, Carmiel, Israel</i>	Novel enzyme replacement therapy for Gaucher disease: On-going phase III clinical trial with recombinant human glucocerebrosidase expressed in plant cells
2:00	Ari Zimran 49 <i>Shaare Zedek Medical Center, Jerusalem, Israel</i>	36 months on treatment: Open-label phase I/II long-term study of enzyme replacement therapy (ERT) with "gene-activated" human glucocerebrosidase (GA-GCB) in patients with type I Gaucher disease
2:15	Brandon Wustman 50 <i>Amicus Therapeutics, Cranbury, NJ</i>	Pharmacological chaperone therapy for Gaucher disease: Mechanism of action, a survey of responsive mutations and phase I clinical trial results
2:30	Judith Peterschmitt 51 <i>Genzyme Corporation, Cambridge, MA</i>	Preliminary results of a phase II clinical trial of Genz-112638 in patients with type I Gaucher disease
2:45	<i>Discussion</i>	
3:00	<i>Break</i>	

WORLD Symposium 2008 Program, cont.

Longitudinal Studies and Outcomes

Christine Eng,
Ed Wraith, Co-Chairs

3:15	Olaf Bodamer 52 <i>University Children's Hospital, Vienna, Austria</i>	Natural history and treatment of MPS I: The MPS I Registry
3:30	Maurizio Scarpa 53 <i>University of Padova, Padova, Italy</i>	Neurological manifestations of Hunter syndrome: Insights from HOS, The Hunter Outcome Survey
3:45	Michelle Wood 54 <i>Great Ormand Street Hospital, London, UK</i>	Changes in gait pattern as assessed by the GaitRite walkway system in MPS II patients undergoing enzyme replacement therapy.
4:00	Ed Wraith 55 <i>Royal Manchester Children's Hospital, Manchester, UK</i>	Miglustat in Niemann-Pick disease type C (NPC)
4:15	Barry Byrne 56 <i>University of Florida, Gainesville, FL</i>	The Pompe Registry: Centralized data collection to track the natural course of Pompe disease
4:30	Alison Skrinar 57 <i>Genzyme Corporation, Cambridge, MA</i>	Response to enzyme replacement therapy in 18 juvenile and adult patients with advanced Pompe disease
4:45	<i>Discussion</i>	
5:00	<i>Meeting Adjourns</i>	