

**Wednesday
February 10**

BASIC RESEARCH

Session 1 Basic Research I

Co-Chairs:
Roscoe Brady
Beverly Davidson

8:00 Chester Whitley

Welcome
Introduction of the LDN Awardee

8:15 William Sly
Saint Louis University School of
Medicine
Saint Louis, MO, USA

Keynote Address: New Hope for Delivering Enzymes Across
the Blood-Brain Barrier

8:45 Walter Low
University of Minnesota
Minneapolis, MN, USA

Stem Cell Repair of the Central Nervous System

9:15 Andrew Wong
King's College London
London, London, UK

Viral Vector and Neural Stem Cell Therapies for Batten
Disease

9:30 Marcy Weatherspoon
Medtronic, Inc.
Minneapolis, MN, USA

Scalability of an AAV4-Mediated Gene Therapy in Sheep
Following Intracerebroventricular
Administration

9:45 Katherine Ponder
Washington University School of
Medicine
St. Louis, MO, USA

The Role of Cathepsin S in Aortic Disease in MPS I and
MPS VII Mice and Dogs

10:00 Break and Exhibits

10:15 Cristin Davidson
Albert Einstein College of Medicine
Bronx, NY, USA

Cyclodextrin Treatment Not Only Delays But Also Reduces
Established Intraneuronal Storage
in Niemann-Pick Type C Disease

10:30 Michael Tropak
Sick Childrens Hospital
Toronto, ON, CANADA

Identification of Pyrimethamine Derivatives Showing
Improved Enzyme Enhancement Efficacy Towards Mutant
Hex A

- 10:45 Jess Thoene**
University of Michigan
Ann Arbor, MI, USA
Correction of Cystine Storage In Cystinotic Fibroblasts by Recombinant Cystinosin
- 11:00 Nidhi Gupta**
NHGRII
Bethesda, MD, USA
Are Mutations in Limp-2 associated with Myoclonic Epilepsy in Patients with Gaucher Disease?
- 11:15 Don Mahuran**
The Hospital for Sick Children
Toronto, ON, Canada
Demonstration of the In Cellulo Efficacy of Pyrimethamine as a Pharmacological Chaperone for Late Onset Tay-Sachs Disease Using a Fluorescent GM2 Ganglioside Analogue
- 11:30 Richard Steet**
University of Georgia
Athens, GA, USA
Identifying the Pathogenic Mechanisms Associated with ML-II Using Zebrafish and Feline Models
- 11:45 Session Concludes**

Lunch Break

COPA Meeting

Session 2 Basic Research IICo-Chairs:
Steven Walkley
Robert Steiner

- 1:00 David Begley**
Kings College London
London, UK
The Blood-Brain Barrier: A Central Role in the Pathology and Treatment of Neuronopathic Lysosomal Storage Disorders
- 1:30 Beverly Davidson**
University of Iowa
Iowa City, IA, USA
Disease Brain Endothelia Provide Unique Molecular Signatures for CNS-directed Enzyme Therapy
- 2:00 N. Matthew Ellinwood**
Iowa State University
Ames, Iowa, USA
Brain Response to Intrathecal or High Dose Enzyme Replacement Therapy in the MPS I Dog
- 2:15 David Sleat**
Center for Advanced Biotechnology and Medicine and University of Medicine and Dentistry of New
Comparative Proteomics and Lysosomal Disease

- 2:30 Ernesto Bongarzone** Axonopathy in a Mouse Model Of Krabbe Disease
University of Illinois, Chicago.
Chicago, IL, US
- 2:45 Nina Raben** Suppression of Autophagy as a Therapeutic Approach to
NIAMS, National Institutes of Health Pompe Disease
Bethesda, MD, USA
- 3:00 Break and Exhibits**
- 3:15 Elizabeth J. White** Immune Cell Phenotypes and Cytokine Response in a
McMaster University Mouse Model of Sialidase Deficiency
Hamilton, ON, Canada
- 3:30 Kostantin Dobrenis** A 34-Amino Acid Peptide Derived from Tetanus Toxin for
Albert Einstein College of Medicine Neuronal Targeting of Lysosomal Proteins
of Yeshiva University
Bronx, NY, USA
- 3:45 Sandrine Vitry** Abnormal Vacuoles Distinct from Lysosomes in a Mouse
Institut Pasteur Model of
Paris, Ile de France, France Mucopolysaccharidosis Type IIIB
- 4:00 Dao Pan** Reprogramming HSC-derived Erythroid Cells for Lysosomal
Cincinnati Children's Hospital Enzyme Production Leads to
Medical Center Visceral and CNS Cross-correction in Mice with MPS Type I
Cincinnati, OH, U. S. A.
- 4:15 Sunita Biswass** A Chemical Genetic Approach to Identifying Therapeutic
Harvard Medical School, Targets for NCL
Massachusetts General Hospital
BOSTON, MA, USA
- 4:30 Grace Colletti** TRPML1 Downregulation is Associated With Changes in
University of Pittsburgh Lysosomal Enzyme Levels
Pittsburgh, Pennsylvania, USA
- 4:45 Susan Cotman** Distinct Features of Disease Phenotypes in Two Genetic
Massachusetts General Hospital Models of NCL
Boston, MA, USA
- 5:00 Christiane Auray-Blais** How Useful is Urinary Lyso-Gb3 as a Biomarker for Fabry
CHUS-Université de Sherbrooke Disease?
Sherbrooke, Quebec, Canada

- 5:15 Brian Bigger**
University of Manchester
Manchester, Lancashire, UK
The Effect of Long-Term Substrate Reduction Therapy with Genistein in a Mouse Model of MPS IIIB
- 5:30 Forbes Porter**
Washington University
St. Louis, MO, USA
Cholesterol Oxidation Products are Sensitive and Specific Blood-based Biomarkers for Niemann-Pick C1 Disease
- 5:45 Poster Session Opens** *Poster sessions are not accredited by ACCME.*
- 7:00 Poster Session Closes**

**Thursday
February 11**

TRANSLATIONAL RESEARCH

Session 3 Translational Research I

Co-Chairs:
William Wilcox
Christine Eng

- 8:00 Chester Whitley**
Introduction
- 8:00 Anne Pariser**
Food and Drug Administration
Silver Spring, MD, USA
Regulation and Review of Small Clinical Trials
- 8:30 Emil Kakkis**
Kakkis EveryLife Foundation
Novato, CA, USA
Transforming the Development of Treatments for Lysosomal Storage Disorders
- 9:00 Neal Weinreb**
University Research Foundation for
Lysosomal Storage Disorders
Coral Springs, FL, USA
Long-term Data from the ICGG Gaucher Registry: 10 Years of Treatment
- 9:15 M. Judith Peterschmitt**
Genzyme Corporation
Cambridge, Ma, USA
Bone Response to Genz-112638 in a Phase 2 Study in Gaucher Disease Type 1

- 9:30 Ari Zimran**
Shaare Zedek Medical Center
Jerusalem, Jerusalem, Israel
Enzyme Replacement Therapy with velaglucerase alfa Improves Key Clinical Parameters in a Pediatric Subgroup with Type 1 Gaucher Disease.
- 9:45 Juan Ruiz**
Shire Human Genetic Therapies
Cambridge, MA, USA
Antigenic Differences in Patients Receiving Velaglucerase Alfa or Imiglucerase Treatment
- 10:00 Break and Exhibits**
- 10:15 David Aviezer**
Protalix Biotherapeutics
Carmiel, Israel,
Novel Enzyme Replacement Therapy for Gaucher Disease: Phase III Pivotal Clinical Trial with Plant Cell Expressed Recombinant Glucocerebrosidase
- 10:30 David Warnock**
University of Alabama at Birmingham
Birmingham, AL, USA
End Stage Renal Disease in Patients with Fabry Disease: Natural History Data from the Fabry Registry
- 10:45 Uma Ramaswami**
Addenbrooke's University Teaching Hospital
Cambridge, United Kingdom
Two-year Longitudinal Follow-up Showing Safety and Effectiveness of Enzyme Replacement Therapy using Agalsidase Alfa in Children: Data from the
- 11:00 Michael West**
Dalhousie University
Halifax, NS, Canada
A Randomized Controlled Trial of Enzyme Replacement Therapy in Fabry Disease: The Canadian Fabry Disease Initiative at Year Three.
- 11:15 Stephen Waldek**
Salford Royal NHS Foundation Trust
Salford, Manchester, United
A Validated Disease Severity Scoring System for Fabry Disease
- 11:30 Ken Valenzano**
Amicus Therapeutics
Cranbury, NJ, USA
Pharmacological Chaperones Increase ERT-Mediated Substrate Reduction In Mouse Models of Fabry and Pompe Disease
- 11:45 Session Concludes**
- Lunch Break**
COIL Meeting

Session 4 Translational Research II

Co-Chairs:
 Gregory Grabowski
 Elsa Shapiro

- 1:00 Elizabeth Braunlin**
 University of Minnesota
 Minneapolis, MN, USA
 Cardiac Valvular Interstitial Cells in MPS I
- 1:30 Robert Steiner**
 Oregon Health & Science University
 Portland, OR, US
 CNS Transplantation of Purified Human Neural Stem Cells in Infantile and Late-Infantile Neuronal Ceroid Lipofuscinoses: Summary of the Phase I Trial
- 2:00 Jae Choi**
 NIH
 Bethesda, MD, USA
 Alpha-Synuclein Aggregation in Gaucher Patients and Carriers with Synucleinopathies
- 2:15 Mia Horowitz**
 Tel Aviv University
 Ramat Aviv, Israel
 Interaction Between Mutant Glucocerebrosidase And Parkin: Its Possible Implication to the Development Of Parkinson Disease
- 2:30 Sean Clark**
 Amicus Therapeutics
 Cranbury, NJ, USA
 Genetic and Pharmacological Chaperone Modulation of Brain GCase Activity Affects Synuclein Accumulation in Mice
- 2:45 Break and Exhibits**
- 3:00 Derralynn Hughes**
 University College London
 Hampstead, London, UK
 Preliminary Long-Term Safety, Tolerability, and Assessments of Renal Function of Adult Fabry Patients Receiving Treatment with AT1001, a Pharmacological Chaperone
- 3:15 Lawrence Charnas**
 Shire HGT
 Cambridge, MA, USA
 A Re-analysis of Disease Stage Progression in Krabbe Disease (infantile Globoid Cell Leukodystrophy, iGLD)
- 3:30 Alia Ahmed**
 University of Minnesota
 Minneapolis, MN, USA
 Preliminary Data on Quantitative MRI and Neuropsychological Function in the Mild Form of MPS II
- 3:45 Julie Eisengart**
 University of Minnesota
 Minneapolis, MN, USA
 Differences In Language Functioning In Hurler Syndrome Before And After HCT: A Qualitative Comparison Of Treatments And Risk Factors

4:00 Poster Session Opens

Poster sessions are not accredited by ACCME.

6:00 Poster Session Closes

6:00 Reception and Banquet

During the Banquet, only the presentations (on the following page) will be accredited by ACCME.

Session 5 Clinical Care Symposium

IMPROVING CLINICAL OUTCOMES

Co-Chairs:

John Barranger

Marc Patterson

6:30 Joan Keutzer

Genzyme Corporation
Cambridge, MA, USA

Newborn Screening for Lysosomal Diseases

7:00 Chester Whitley

University of Minnesota
Minneapolis, MN, USA

Small Molecules for Treatment of Lysosomal Diseases

7:30 Jeanine Utz

University of Minnesota, Fairview
Pharmacy Services
Minneapolis, MN, USA

Medication Therapy Management for Lysosomal Diseases

8:00 John Crowley

Amicus Therapeutics
Cranbury, NJ, USA

When Drug Research is Personal

8:20

Presentation of Lysosomal Disease Network WORLD
Symposium 2010 Advocate Award

**Friday
February 12**

CLINICAL RESEARCH

Session 6 Newborn Screening

Co-Chairs:
Joan Keutzer
Rodney Howell

- 8:00 R Rodney Howell**
Miller School of Medicine, University of Miami
Miami, FL, USA
Developing an Evidence Review Process for Newborn Screening Decision-Making
- 8:30 Patricia Duffner**
University at Buffalo/Hunter James Kelly Research Institute
Buffalo, New York, USA
Longitudinal/Outcome Studies of Children with Krabbe Disease
- 9:00 Roberta Salvesson**
Mount Sinai Medical Center/Columbia University
New York, NY, USA
Expansion Of Newborn Screening Panels: A Systematic Evaluation of Krabbe Disease Screening in New York State
- 9:15 Hui Zhou**
Centers for Disease Control and Prevention
Atlanta, GA, USA
Update on Laboratory Support at the Centers for Disease Control and Prevention for Newborn Bloodspot Screening to Detect Lysosomal Storage Disorders
- 9:30 Dietrich Matern**
Mayo Clinic College of Medicine
Rochester, MN, USA
First Steps Towards Determination Of The Most Efficient And Effective Newborn Screening (NBS) Approach For LSDs
- 9:45 Trisha Duffey**
University of Washington
Seattle, WA, USA
Newborn Screening For Lysosomal Storage Disorders: Tandem Mass Spectrometry To Quantitate Enzymatic Activity.
- 10:00 Break and Exhibits**

Session 7 LDN NIH-Funded Project Reports

Co-Chairs:
Danilo Tagle
Mary Lou Oster-Granite

- 10:15 Elsa Shapiro**
University of Minnesota
Minneapolis, MN, USA
Longitudinal Studies of Brain Structure and Function in MPS Disorders: A Study of the Lysosomal Disease Network
- 10:30 Agnes Chen**
Los Angeles Biomedical Institute at Harbor-UCLA Medical Center
Torrance, CA, USA
A Study of Intrathecal Enzyme Replacement for Cognitive Decline in Mucopolysaccharidosis I

- 10:45 Lynda Polgreen**
University of Minnesota
Minneapolis, MN, USA
Update on the Longitudinal Study of Bone Disease and the Impact of Growth Hormone Treatment in MPS I, II, and VI.
- 11:00 Michael Potegal**
University of Minnesota Medical School
Minneapolis, MN, USA
Empirical Assessment of Social/Emotional Function in Children with MPS III: Preliminary Observations.
- 11:15 Raphael Schiffman**
Baylor Research Institute
Dallas, TX, USA
The Natural History of Mucopolidosis Type IV
- 11:30 Jonathan Mink**
University of Rochester
Rochester, NY, USA
The UBDRS Predicts Rate of JCNL (CLN3) Disease Progression
- 11:45 Session Concludes**

Lunch Break and Exhibits

LDN Investigators Meeting

Session 8 LDN NIH-Funded Project ReportsCo-Chairs:
Catherine McKeon
Joseph Muenzer

- 1:00 Jeffrey Krischer**
University of South Florida
Tampa, FL, USA
The Rare Diseases Clinical Research Network's (RDCRN) Data Management and Coordination Center
- 1:30 Kyle Rudser**
University of Minnesota
Minneapolis, MN, USA
Statistical Issues In Clinical Trials: Information Growth In Longitudinal Trials
- 2:00 Sara Cathey**
Greenwood Genetic Center
N. Charleston, SC, USA
Longitudinal Studies Of The Glycoproteinoses: An International Update
- 2:15 Ronald G. Crystal**
Joan & Sanford I. Weill Medical
College of Cornell University
New York, NY, USA
Assessment of Neurological Deterioration in Subjects with LINCL

- 2:30 Priya Kishnani**
Duke University Medical Center
Durham, NC, USA
Immunological Aspects of Treatment of Pompe Disease
- 2:45 Marc Patterson**
Mayo Clinic
Rochester, MN, USA
Longitudinal Study of Cognition in Subjects with Niemann-Pick Disease, Type C
- 3:00 Break and Exhibit**
- 3:15 Gregory Grabowski**
Children's Hospital Research
Foundation
Cincinnati, OH, USA
Epidemiology and Natural History of Wolman and
Cholesteryl Ester
Storage Diseases
- 3:30 Marsha Browning**
MGH/Harvard
Boston, MA, USA
Fabry Disease Identification
- 3:45 Michael Mauer**
University of Minnesota
Minneapolis, Minnesota, USA
Natural History and Structural-Functional Relationships in
Fabry Renal Disease
- 4:00 Michael Msall**
University of Chicago
Chicago, IL, USA
Developmental and Functional Surveillance in Preschool
Children with Lysosomal Storage Diseases
- 4:15 William Wilcox**
Cedars-Sinai
Los Angeles, CA, USA
Pulmonary Disease and Exercise Tolerance in Boys with
Fabry Disease
- 4:30 Joe Clarke**
Hospital for Sick Children
Toronto, ON, Canada
Open-Label Phase I/II Clinical Trial of Pyrimethamine for the
Treatment of Chronic GM2 Gangliosidosis
- 4:45 Chester B. Whitley**
University of Minnesota
Minneapolis, MN, USA
A Natural History Study of Hexosaminidase Deficiency
- 5:00 Chester B. Whitley**
Closing remarks

Curriculum and Faculty are subject to change.

As of 1.15.2010