Wednesday
February 10

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 II  
Co-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**  
University of Illinois, Chicago.  
Chicago, IL, US  
Axonopathy in a Mouse Model Of Krabbe Disease

2:45  **Nina Raben**  
NIAMS, National Institutes of Health  
Bethesda, MD, USA  
Suppression of Autophagy as a Therapeutic Approach to Pompe Disease

3:00  **Break and Exhibits**

3:15  **Elizabeth J. White**  
McMaster University  
Hamilton, ON, Canada  
Immune Cell Phenotypes and Cytokine Response in a Mouse Model of Sialidase Deficiency

3:30  **Kostantin Dobrenis**  
Albert Einstein College of Medicine  
of Yeshiva University  
Bronx, NY, USA  
A 34-Amino Acid Peptide Derived from Tetanus Toxin for Neuronal Targeting of Lysosomal Proteins

3:45  **Sandrine Vitry**  
Institut Pasteur  
Paris, Ile de France, France  
Abnormal Vacuoles Distinct from Lysosomes in a Mouse Model of Mucopolysaccharidosis Type IIIB

4:00  **Dao Pan**  
Cincinnati Children's Hospital Medical Center  
Cincinnati, OH, U. S. A.  
Reprogramming HSC-derived Erythroid Cells for Lysosomal Enzyme Production Leads to Visceral and CNS Cross-correction in Mice with MPS Type I

4:15  **Sunita Biswass**  
Harvard Medical School,  
Massachusetts General Hospital  
BOSTON, MA, USA  
A Chemical Genetic Approach to Identifying Therapeutic Targets for NCL

4:30  **Grace Colletti**  
University of Pittsburgh  
Pittsburgh, Pennsylvania, USA  
TRPML1 Downregulation is Associated With Changes in Lysosomal Enzyme Levels

4:45  **Susan Cotman**  
Massachusetts General Hospital  
Boston, MA, USA  
Distinct Features of Disease Phenotypes in Two Genetic Models of NCL

5:00  **Christiane Auray-Blais**  
CHUS-Université de Sherbrooke  
Sherbrooke, Quebec, Canada  
How Useful is Urinary Lyso-Gb3 as a Biomarker for Fabry Disease?
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**

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**Thursday  
February 11**

**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 (prGCD) - Taliglucerase alfa

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 Ramaswarmi  
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 Fabry Outcome Survey

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 Chol
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 for Up to 2 Years

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

8:00  R Rodney Howell
Miller School of Medicine, University of 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 Salveson
Mount Sinai Medical Center/Columbia Universitly, 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

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  

11:15 Raphael Schiffman  
Baylor Research Institute  
Dallas, TX, USA  
The Natural History of Mucolipidosis 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 Reports  
Co-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