THURSDAY, JUNE 11

Jordan Hall of Science, Room 105

1:30 – 1:45 p.m. Opening Remarks
Cindy Parseghian
Ara Parseghian Medical Research Foundation

SESSION I:
Jordan Hall of Science, Room 105

1:45 – 2:15 p.m. NPC1-mediated Cholesterol export from lysosomes
Jian Li, Peter L. Lee, Piyali Saha, Suzanne R. Pfeffer
1Department of Biochemistry, Stanford University School of Medicine

2:15 – 2:45 p.m. Cholesterol trafficking between the endocytic compartment and endoplasmic reticulum at ORP1L-VAP membrane contacts
Nicholas L. Cianciola, Cathleen R. Carlin
1Department of Molecular Biology and Microbiology, Case Western Reserve University

2:45 – 3:05 p.m. CRISPR/Cas9 Technology for the Study and Treatment of NPC Disease
Guosheng Liang
1Associate Professor, Department of Molecular Genetics, University of Texas Southwestern Medical Center
(Poster Talk)

3:05 – 3:20 p.m. Discussion

SESSION II:
Jordan Hall of Science, Room 105

3:35 – 3:55 p.m. Adeno-associated viral gene therapy to treat Niemann-Pick disease, type C1
1National Institutes of Health
(Poster Talk)
3:55 - 4:25 p.m.  Targeting oxLDL to combat NPC1
Tom Houben, Sofie Walenbergh, Tim Hendrikx, Patrick van Gorp, Mike Jeurissen, Jieyi Li, Fons Verheyen, Marion J. Gijbels¹, Christoph J Binder²,³, Ger Koek⁴, Marten Hofker⁵, Dieter Lütjohann⁶, Ronit Shiri-Sverdlov⁷
¹Departments of Molecular Genetics, Molecular Cell Biology and Electron Microscopy, Nutrition and Toxicology Research (NUTRIM) Institute of Maastricht, University of Maastricht, Maastricht, The Netherlands, ²Center for Molecular Medicine (CeMM), Austrian Academy of Sciences, Vienna, Austria, ³Department of Laboratory Medicine, Medical University of Vienna, Vienna, Austria, ⁴Department of Internal Medicine, Division of Gastroenterology and Hepatology, Maastricht University Medical Center (MUMC), Maastricht, The Netherlands, ⁵Department of Pathology and Medical Biology, Molecular Genetics, Medical Biology Section, University of Groningen, University Medical Center Groningen, Groningen, The Netherlands, ⁶Institute of Clinical Chemistry and Clinical Pharmacology, University of Bonn, Germany

4:25 – 4:55 p.m.  Modifying Niemann Pick type C disease
Stephen L. Sturley, Claudia Dall'Armi, Elizabeta Micevska, Gil Di Paulo¹, Karen Reue², Andrew Munkacsi³
¹Columbia University Medical Center, ²UCLA Medical School and ³University of Wellington, New Zealand

4:55 – 5:15 p.m.  Discussion

FRIDAY, JUNE 12

SESSION III:
Jordan Hall of Science, Room 105

8:00 – 8:30 a.m.  Design and Synthesis of Small Molecule Agents for the Study and Treatment of Niemann-Pick Type C Disease
Olaf Wiest, Paul Helquist¹, Edward Holson²
¹University of Notre Dame, ²The Broad Institute of MIT and Harvard

8:30 – 9:00 a.m.  Testing Histone Deacetylase Inhibitors as NPC Therapeutics
Frederick R. Maxfield, Dana Cruz, Shu Mao, Nina Pipalia¹
¹Weill Cornell Medical College, New York, NY

9:00 – 9:30 a.m.  Histone Deacetylase Inhibitors as NPC1 Therapeutics
Daniel Ory, Anita Chacko, Abbey Wolfensen, and Maria Praggastis¹
¹Washington University School of Medicine

9:30 – 10:00 a.m.  HDACi treatment in a murine model of Niemann-Pick Type C disease
Suhail Alam, Michelle Getz, Yana Fedotova, and Kasturi Haldar¹
¹Department of Biological Sciences, University of Notre Dame
SESSION IV: (CLOSED; RESEARCHERS ONLY)
Jordan Hall of Science, Room 105

10:00 – 10:15 a.m.  Discussion

10:30 – 11:00 a.m.  Therapeutic trials for Niemann-Pick Disease, Type C1: Phase 1/2 Studies of HPβCD and vorinostat
Forbes D. Porter1, the TRND Team, and the HDACi Collaborative Group
1The National Institutes of Health

11:00 – 11:30 a.m.  Therapeutic trials for Niemann-Pick Disease, Type C1: Phase 2/3 Studies of HPβCD
Ben Machielse1
1Vtesse

11:30 – 12:00 p.m.  Objective Clinical Efficacy Outcome Measures for Cyclodextrin Treatment in Niemann-Pick Type C (NP-C): A Five-Domain Approach
Elizabeth Berry-Kravis1, 2, 3, Joanne O'Keefe4, Anne Hoffmann1, 5, Amy Winston5, Lisa LaGorio5, Erin Robertson4, Jamie Chin1, Sue Leurgans2, 5

Departments of 1Pediatrics, 2Neurological Sciences, 3Biochemistry, 4Anatomy and Cell Biology, 5Communication Disorders and Sciences, 6Preventive Medicine, Rush University Medical Center

12:00 – 12:15 p.m.  Discussion

SESSION V:
Jordan Hall of Science, Room 105

1:30 – 1:50 p.m.  StARD9 is a Novel Kinesin Required for Motility and Tubulation of Late Endosomes/Lysosomes Containing NPC1
Kevin T. Vaughan, Alexandria Brumfield, Kara L. Huegel, Patricia S. Vaughan, Michelle V. Joyce, Bill Boggess1, Edward H. Hinchcliffe2
1Departments of Biological Sciences and Chemistry and Biochemistry, University of Notre Dame, 2Hormel Institute, University of Minnesota
(Poster Talk)

1:50 – 2:10 p.m.  An Evolutionary Approach to Precision Drug Discovery for Niemann-Pick C
Ethan O. Perlstein, Nina DiPrimio, Tom A. Hartl, Sangeetha Iyer, Tamy Portillo Rodriguez, Alec Ludin1
1Perlstein Lab PBC
(Poster Talk)

2:10 – 2:30 p.m.  Regulation of Cholesterol Homeostasis with Spliceosome Inhibitor GEX1A, A Potentially Novel Lead for Niemann-Pick Type C Disease
Jarred Pickering, Eve Granatosky, D. Cole Stevens, Richard Taylor¹, Nina DiPrimio, Ethan Perlstein²
¹The Warren Family Center for Drug Discovery and Development, Department of Chemistry & Biochemistry, University of Notre Dame, Perlstein Lab PBC²
(Poster Talk)

2:30 – 4:30 p.m.  POSTER SESSION
Jordan Hall Galleria

CRISPR/Cas9 Technology for the Study and Treatment of NPC Disease
Guosheng Liang¹
¹Associate Professor, Department of Molecular Genetics, University of Texas Southwestern Medical Center

Adeno-associated viral gene therapy to treat Niemann-Pick disease, type C1
Randy J. Chandler, Ian M. Williams, Arturo A. Incao, Forbes D. Porter, William J. Pavan, Charles P. Venditti¹
¹National Institutes of Health

STARD9 is a Novel Kinesin Required for Motility and Tubulation of Late Endosomes/Lysosomes Containing NPC1
Kevin T. Vaughan, Alexandria Brumfield, Kara L. Huegel, Patricia S. Vaughan, Michelle V. Joyce, Bill Boggess¹, Edward H. Hinchcliffe²
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Jarred Pickering, Eve Granatosky, D. Cole Stevens, Richard Taylor¹, Nina DiPrimio, Ethan Perlstein²
¹The Warren Family Center for Drug Discovery and Development, Department of Chemistry & Biochemistry, University of Notre Dame, ²Perlstein Lab PBC

The active form of FTY720/FINGOLIMOD is an HDAC inhibitor that increases NPC1 and NPC2 expression
Jason Newton, Nitai C. Hait, Sheldon Milstien, Sarah Spiegel¹
¹Department of Biochemistry and Molecular Biology, VCU School of Medicine

Screening HDAC Inhibitors for Improvement of NPC Cell Cholesterol Accumulation Phenotype
Dana Cruz, Shu Mao, Deepti Gadi, Nina Pipalia¹, Edward Holson², Paul Helquist, Olaf Wiest³, Frederick R. Maxfield¹
The PKC activator, bryostatin 1, improves lipid transport in Niemann Pick C1 cells
Fannie Chen, Jagruti Chaudhari, Yiannis Ioannou1, Sam Kongsamut, Warren Wasiewski2

*Department of Genetics and Genomic Sciences, the Mount Sinai School of Medicine1, and Neurotrope BioScience Inc.2

Intrathecal AAV-mediated gene therapy for feline NPC1 disease
Brittney L. Gurda, Gary Swain, Jessica Bagal, Maria Prociuk, Allison Bradbury, Charles H. Vite1
1University of Pennsylvania, School of Veterinary Medicine

Regulation of Cholesterol Homeostasis with the Polyketide GEX1A, A Potential Lead for Niemann-Pick Type C Disease
Jarred Pickering, Eve Granatosky, D. Cole Stevens, Michael Ahlers, Richard Taylor1, Nina DiPrimio, Ethan Perlstein2
1The Warren Family Center for Drug Discovery and Development, Department of Chemistry and Biochemistry, University of Notre Dame, 2Perlstein Lab PBC

Pharmacokinetic, Biodistribution, and Plasma Protein Binding Properties of 2-Hydroxypropyl-β-Cyclodextrin:Pluronic Polyrotaxanes
Christopher J. Collins, Yawo Mondjinou, Bradley Loren, David H. Thompson1
1Purdue University, Department of Chemistry, Multi-disciplinary Cancer Research Facility

SATURDAY, JUNE 13

SESSION VI:
Jordan Hall of Science, Room 105

8:20 – 8:50 a.m. Mechanism of cholesterol accumulation and cyclodextrin-mediated reversal in NPC disease
Valérie Demais1, Amelie Bartheley, Nicole Ungerer2, Céline Keime3, Martine Perraut, Frank W. Pfrieger2
1Plateforme Imagerie in Vitro, Strasbourg, France, 2Institute of Cellular and Integrative Neurosciences, University of Strasbourg, Strasbourg, France, 3Institut de Génétique et de Biologie Moléculaire et Cellulaire, Illkirch, France

8:50 – 9:20 a.m. Pharmacokinetic, Biodistribution, and Plasma Protein Binding Properties of 2-Hydroxypropyl-β-Cyclodextrin:Pluronic Polyrotaxanes
Bradley Loren, Christopher J. Collins, Yawo Mondjinou, David H. Thompson1
1Purdue University, Department of Chemistry, Multi-disciplinary Cancer Research Facility, Bindley Bioscience Center
9:20 – 9:50 a.m. Neuronal dysfunction in Niemann Pick C1 disease: impact of early developmental defects and efficacy of hydroxypropyl-betacyclodextrin in correcting the phenotype
Sonia Canterini, Giampiero Palladino, Jessica Dragotto, Paola Caporali, Francesco Bruno, Georgia Abate1, Stefano Loizzo2, Robert P. Erickson3, Maria Teresa Fiorenza1
1Department of Psychology, Section of Neuroscience and “Daniel Bovet” Neurobiology Research Center, Sapienza University of Rome, Italy, 2Department of Therapeutic Research and Medicines Evaluation, ISS, Rome, Italy, 3Department of Pediatrics, University of Arizona

9:50 – 10:10 a.m. Use of mitoprotective compounds as a therapeutic strategy to promote survival of Niemann-Pick type C1 neurons
John Steele, Paulina Ordonez, Larry Goldstein1
1University of California San Diego

10:10 – 10:25 a.m. Discussion

SESSION VII:
Jordan Hall of Science, Room 105

10:45 – 11:15 a.m. The active form of FTY720/FINGOLIMOD is an HDAC inhibitor that increases NPC1 and NPC2 expression
Jason Newton, Nitai C. Hait, Sheldon Milstien, Sarah Spiegel1
1Department of Biochemistry and Molecular Biology, VCU School of Medicine

11:15 – 11:45 a.m. Development of a Newborn Screen for NPC1 Disease Based on a Novel Blood-Based Disease Biomarker
Xuntian Jiang, Rohini Sidhu, David E. Scherrer1, Laurel Mydock2, Douglas Covey, Fong-Fu Hsu3, Nicole M. Yanjanin, Forbes D. Porter4, Dennis J. Dietzen5, Elizabeth Berry-Kravis7, Joseph J. Orsini6, Jean E. Schaffer, Daniel S. Ory1
1Diabetic Cardiovascular Disease Center, 2Department of Developmental Biology, 3Department of Internal Medicine, 4Department of Pediatrics, Washington University School of Medicine, 5Program in Developmental Endocrinology and Genetics, Eunice Kennedy Shriver National Institute of Child Health and Human Development, NIH, DHHS, 6New York State Department of Health, 7Rush University Medical Center

11:45 – 12:15 p.m. NPC Disease: new targets, new tissues, new technology
Karen S. Pawlowski, Nancy S. Gonzalez, Jeffrey G. McDonald, and Joyce J. Repa1
1UT Southwestern Medical Center

12:15 – 12:30 p.m. Discussion

12:30 – 12:45 p.m. Closing comments