

**Ara Parseghian Medical Research Fund at Notre Dame
Michael, Marcia, and Christa Parseghian Scientific Conference for
Niemann-Pick Type C Research**

**Conference Agenda
June 8 – 11, 2024**

Day 1

Saturday, June 8, 2024

TIME	EVENT
11:00-1:00 PM	Registration (Sonoran Room Foyer; West)
1:00-1:10 PM	Welcome & Introduction Sean Kassen & Andy Lieberman (Sonoran Room)
1:10-2:00 PM	Keynote Presentation: Phyllis Hanson (40+10) <i>Beyond Repair: ESCRTs Enhance Membrane Resilience</i>
Session #1 (Sonoran Room)	<i>Lysosomal Biology and Cholesterol Trafficking</i> Andy Lieberman: session leader
2:00-2:30 PM	Sandra Maday (20+10) <i>Lysosomal pathways in synaptic signaling between neurons and astrocytes</i>

- 2:30-3:00 PM Roberto Zoncu (20+10)
Regulation of Lysosomal Membrane Homeostasis in Niemann-Pick type C
- 3:00-3:15 PM Break (15)
- 3:15-3:45 PM Yvette Wong (20+10)
Regulation and function of lysosomal inter-organelle contact sites in health and disease
- 3:45-4:15 PM Suzanne Pfefer (20+10)
Loss of neuroprotection in the striatum in NPC and other neurodegenerative diseases
- 4:15 - 4:35 PM Mike Henne (15+5)
SNX-RGS proteins as lipid transporters that influence lysosome phosphatidic acid (PA) and LBPA metabolism in Niemann Pick disorder
- 4:35 - 4:55 PM Albert Lu (15+5)
Uncovering the role of SNX13 in an NPC1 bypass cholesterol egress pathway
- 4:55 - 5:10 PM Hijai-Regina Shin (10+5)
Lysosome at the center of lipid sensing and growth regulation
- 5:15 - 8:00 PM** *Reception & dinner*

Day 2
Sunday, June 9, 2024

TIME	EVENT
7:00-8:00 AM	Breakfast
Session #2 (Sonoran Room)	<i>Cholesterol trafficking and signaling</i> Eamonn Dickson: session leader
8:00-8:30 AM	Emily Eden (20+10) <i>The importance of lysosomal lipids in inter-organelle communication.</i>
8:30-9:00 AM	Arun Radhakrishnan (20+10) <i>Role of NPC1 in microbial infection</i>
9:00-9:20 AM	HY Yang (15 + 5) Phosphatidylserine regulates plasma membrane repair through tetraspanin-enriched macrodomains
9:20-9:40 AM	Xiaochun Li (15+5) <i>Structural Insights into Lysosomal Transporters</i>
9:40-9:55 AM	Daniel Wustner (10+5) <i>Role of Niemann Pick type C2 protein in cellular cholesterol elux and inflammation</i>
9:55-10:10 AM	Break (15)

Session #3 *Cholesterol trafficking and signaling*

(Sonoran Room) **HY Yang: session leader**

10:10-10:40 AM Judith Storch (20+10)
Understanding the mechanisms and utility of using lysobisphosphatidic acid (LBPA)/bismonoacylglycerophosphate (BMP) as an NPC disease therapeutic.

10:40-11:00 AM Eamonn Dickson (15+5)
NPC1-dependent remodeling of ion channels at membrane contact sites contribute to neurodegeneration

11:00 - 11:20 AM *Rajat Rohatgi (15+5)*
Lipid trafficking pathways in Niemann-Pick Type C disease

11:20 - 11:40 PM *Chris Patzke (15+5)*
Platform to Analyze Mutations Causing Niemann Pick Type C Disease in Human Neurons

11:40 - 11:55 AM Valentina Pallottini **(10+5)**
A possible new therapeutic drug target for Niemann-Pick type C1 disease: the BET proteins

11:55 – 12:45 PM Lunch

Session #4 **Biomarkers and Characterization**

(Sonoran Room) **Elizabeth Berry-Kravis: session leader**

12:45 - 1:00 PM Frank Pfrieger **(10+5)**
Progressive pathologic changes in the retinal pigment epithelium of NPC1-deficient mice

1:00 - 1:15 PM Wenping Li **(10+5)**
*Progranulin alterations in the Niemann-Pick type C1
I1061T mouse model*

1:15 - 1:35 PM Jorge L. Rodriguez-Gil **(15+5)**
Molecular and Cellular Basis of NPC1 Lung Disease

1:35-1:55 PM Denny Porter **(15+5)**
*Identification and Characterization of Serum Protein
Biomarkers in Niemann-Pick disease, type C1*

1:55-2:15 PM Stephanie Cologna **(15+5)**
The growing biomarker landscape in NPC1

2:15-2:25 PM Critical Path Institute **(10)**

2:25-5:00 PM Afternoon Break

5:00-6:30 PM Poster Session
(Terrace Level) *Foyer*

6:30-7:30 PM Dinner
(Murphy Room)

Day 3
Monday, June 10, 2024

TIME	EVENT
7:00-8:00 AM	Breakfast
Session #5 (Sonoran Room)	Therapeutic Approaches & Modeling Stephani Cologna: session leader
8:00-8:30 AM	Elsa Rodrigues (20+10) <i>Unveiling CYP46A1 as a Therapeutic Target in Niemann-Pick Type C Disease: Promising Advances and Potential Challenges</i>
8:30 - 8:50 AM	Mark Schultz (15+5) Exploring mRNA therapeutics in Niemann-Pick type C1 disease
8:50 - 9:10 AM	Andy Lieberman (15+5) <i>Model systems for the identification of Niemann-Pick C1 proteostatic therapies</i>
9:10 - 9:30 AM	Kevin Vaughan (15+5) <i>Overcoming Cholesterol Transport Defects in Niemann Pick Type C Disease.</i>
9:30 - 9:50 AM	Marco Segatto (15+5) <i>Modulation of the neurotrophin receptor p75 reduces cholesterol accumulation in Niemann-Pick type C cells</i>
9:50 - 10:05 AM	Coffee Break

Session #6 Therapeutic Approaches & Modeling

(Sonoran Room) Suzanne Pfeffer: session leader

- 10:05 - 10:25 AM Charles Vite (15+5)
The feline NPC1 colony at the University of Florida – new collaborations.
- 10:25 - 10:45 AM Jian Xiong (15+5)
Modulating Bis(Monoacylglycero)Phosphate for enhanced lysosomal function -- A novel therapeutic strategy for Niemann Pick C1?
- 10:45 - 11:00 AM Thomas Grewal (10+5)
Targeting TBC1D15-mediated Rab7 inactivation to overcome cholesterol accumulation in NPC1 disease
- 11:00 - 11:15 AM Ben Calvarey (10+5)
Understanding the Spatial Covariance of NPC1 Fold Design through Machine Learning to Address Clinical Disease
- 11:15-11:35 PM Solomon Mbua (15+5)
The International Niemann-Pick Disease Registry – An update and progress in North America
- 11:35-12:25 PM Lunch

Session #7 Pharmaceutical & Clinical Data

(Sonoran Room) Denny Porter: session leader

- 12:25-12:55 PM Elizabeth Berry-Kravis (20+10)
Long-term Open Label Treatment of Neurological Symptoms in

Niemann-Pick Type C1 with Intrathecal ADRABETADEX Through Expanded Access

- 12:55-1:25 PM Fred Maxfield (20+10)
Testing chaperone modulation in NPC1 mutant cell lines
- 1:25-1:55 PM Ross Fredenberg (15+5)
Update on NPC1 Stabilizer Drug Development Program
- 1:55-2:15 PM Melissa Wasserstein (15+5)
A multi-tiered approach to screening newborns for complex disorders
- 2:15-2:35 PM Scenic Biotech (15+5)
Discovery of a novel Niemann Pick type C (NPC) disease modifier and development towards its clinical application
- 2:35-2:55 PM Intrabio (15+5)
Evaluation of the safety and efficacy of N-acetyl-L-leucine for NPC
- 2:55-5:00 PM Afternoon Break
- 5:00-6:30 PM Poster Session
(Terrace Level) *Foyer*
- 6:30-8:00 PM Reception and Dinner
(Murphy Room) *Special remarks from NPC family*

Day 4

Tuesday, June 11, 2024

TIME

EVENT

7:30-9:00 AM

Breakfast