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

<table>
<thead>
<tr>
<th>TIME</th>
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<tbody>
<tr>
<td>11:00–1:00 PM</td>
<td><strong>Registration</strong></td>
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<td>(Sonoran Room Foyer; West)</td>
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<td>1:00–1:10 PM</td>
<td><strong>Welcome &amp; Introduction</strong></td>
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<td>Sean Kassen &amp; Andy Lieberman</td>
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<td></td>
<td>(Sonoran Room)</td>
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<td>1:10–2:00 PM</td>
<td><strong>Keynote Presentation: Phyllis Hanson (40+10)</strong></td>
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<tr>
<td>Session #1</td>
<td><strong>Lysosomal Biology and Cholesterol Trafficking</strong></td>
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<td>(Sonoran Room)</td>
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<td></td>
<td>Andy Lieberman: session leader</td>
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<td>2:00–2:30 PM</td>
<td><strong>Sandra Maday (20+10)</strong></td>
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<td><em>Lysosomal pathways in synaptic signaling between neurons and astrocytes</em></td>
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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 Pfeffer (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

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<tr>
<th>TIME</th>
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<tbody>
<tr>
<td>7:00-8:00 AM</td>
<td>Breakfast</td>
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**Session #2** *Cholesterol trafficking and signaling*  
(Sonoran Room) Eamonn Dickson: session leader

<table>
<thead>
<tr>
<th>TIME</th>
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| 8:00-8:30 AM | Emily Eden *(20+10)*  
*The importance of lysosomal lipids in inter-organelle communication.* |
| 8:30-9:00 AM | Arun Radhakrishnan *(20+10)*  
*Role of NPC1 in microbial infection* |
| 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)*  
*Structural Insights into Lysosomal Transporters* |
| 9:40-9:55 AM | Daniel Wustner *(10+5)*  
*Role of Niemann Pick type C2 protein in cellular cholesterol efflux and inflammation* |
| 9:55-10:10 AM | Break *(15)* |
Session #3  Cholesterol trafficking and signaling
(Sonoran Room)  HY Yang: session leader

10:10–10:40 AM  Fred Maxfield (20+10)
Testing chaperone modulation in NPC1 mutant cell lines

10:40–11:10 AM  Judith Storch (20+10)
Understanding the mechanisms and utility of using lysobisphosphatidic acid (LBPA)/
bismonoacylglycerophosphate (BMP) as an NPC disease therapeutic.

11:10–11:30 AM  Eamonn Dickson (15+5)
NPC1-dependent remodeling of ion channels at membrane contact sites contribute to
neurodegeneration

11:30 – 11:50 AM  Rajat Rohatgi (15+5)
Lipid trafficking pathways in Niemann–Pick Type C disease

11:50 – 12:10 PM  Chris Patzke (15+5)
Platform to Analyze Mutations Causing Niemann Pick Type C Disease in Human Neurons

12:10 – 1:00PM  Lunch

Session #4  Biomarkers and Characterization
(Sonoran Room)  Elizabeth Berry-Kravis: session leader

1:00 – 1:15 PM  Frank Pfreiger (10+5)
Progressive pathologic changes in the retinal pigment epithelium of NPC1-deficient mice
1:15 – 1:30 PM  Wenping Li (10+5)
Progranulin alterations in the Niemann-Pick type C1 11061T mouse model

1:30 – 1:50 PM  Jorge L. Rodriguez-Gil (15+5)
Molecular and Cellular Basis of NPC1 Lung Disease

1:50 – 2:10 PM  Denny Porter (15+5)
Identification and Characterization of Serum Protein Biomarkers in Niemann-Pick disease, type C1

2:10 – 2:30 PM  Stephanie Cologna (15+5)

2:30 – 2:40 PM  Critical Path Institute (10)

2:40 – 5:00 PM  Afternoon Break

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

6:30 – 7:30 PM  Dinner
(Murphy Room)
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<td>8:00-8:30 AM</td>
<td><strong>Session #5</strong> Therapeutic Approaches &amp; Modeling</td>
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<td><strong>(Sonoran Room)</strong> Stephani Cologna: session leader</td>
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| 8:30 - 8:50 AM | Elsa Rodrigues *(20+10)*  
*Unveiling CYP46A1 as a Therapeutic Target in Niemann–Pick Type C Disease: Promising Advances and Potential Challenges* |
| 8:50 - 9:10 AM | Mark Schultz *(15+5)*  
*Exploring mRNA therapeutics in Niemann–Pick type C1 disease* |
| 9:10 - 9:30 AM | Kevin Vaughan *(15+5)*  
*Overcoming Cholesterol Transport Defects in Niemann Pick Type C Disease.* |
| 9:30 - 9:50 AM | Marco Segatto *(15+5)*  
*Modulation of the neurotrophin receptor p75 reduces cholesterol accumulation in Niemann–Pick type C cells* |
| 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(Monoacylglycerol)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  Valentina Pallotina (10+5)

A possible new therapeutic drug target for Niemann–Pick type C1 disease: the BET proteins

11:15 – 11:30 AM  Ben Calvarey (10+5)

Understanding the Spatial Covariance of NPC1 Fold Design through Machine Learning to Address Clinical Disease

11:30–12:30 PM  Lunch

Session #7  Pharmaceutical & Clinical Data
(Sonoran Room) Denny Porter: session leader

12:30–1:00 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

1:05-1:25 PM  Solomon Mbua (15+5)
*The International Niemann-Pick Disease Registry – An update and progress in North America*

1:25-1:45 PM  Ross Fredenberg (15+5)
*Update on NPC1 Stabilizer Drug Development Program*

1:05-1:25 PM  Melissa Wasserstien (15+5)
*AZ-3102, a novel brain-penetrant small molecule for the treatment of Niemann-Pick disease Type C*

1:25-1:45 PM  Scenic Biotech (15+5)
*Discovery of a novel Niemann Pick type C (NPC) disease modifier and development towards its clinical application*

1:25-1:45 PM  Intrabio (15+5)
*Evaluation of the safety and efficacy of N-acetyl-L-leucine for NPC*

1:45-5:00 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

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