

***Scientific Conference on Niemann-Pick Type C Disease***

Sponsored and Hosted by:

***Ara Parseghian Medical Research Foundation***

Co-Sponsored by:

***Office of Rare Diseases, NIH***

*The Westin La Paloma Resort & Spa – Tucson, Arizona – June 1-2, 2007*

***Friday, June 1, 2007***

**7:15-8:00 Continental Breakfast - La Paloma Country Club**

**8:15-8:30 Cindy and Michael Parseghian, Welcoming Address - Murphey Room**

***NPC1 Protein and Biochemistry***

**8:30-8:50 Frances Sharom, Ph.D., University of Guelph - “Purification of human NPC1 and characterization of its binding to sterols and lipids”**

**9:00-9:20 Suzanne Pfeffer, Ph.D., Stanford University - “Functional analysis of NPC1 protein”**

**9:30-9:50 Kevin Vaughan, Ph.D., University of Notre Dame - “The Role of Dynein-Driven Motility in the Trafficking of NPC1 Membranes”**

**10:00-10:10 Haruaki Ninomiya, M.D., Tottori University - “Proteosomal degradation of NPC1”**

**10:15-10:30 Coffee break**

***NPC1 Function and Neurobiology***

**10:30-10:40 Jeffrey Krise, Ph.D., University of Kansas - “Evaluating NPC-1’s functional role in both sensing and facilitating the intracellular trafficking of lysosomal stores of amine-containing molecules”**

**10:45-10:55 Hongyuan Yang, Ph.D., University of New South Wales - “Ncr1p, the yeast ortholog of the human NPC1 protein, regulates the exit of sterols from the yeast vacuole”**

**11:00-11:20 Barbara Karten, Ph.D., Dalhousie University - “The role of NPC1 in synaptic and neuronal function”**

**11:30-11:50 Matthew Scott, Ph.D., Stanford University - “In Vivo Studies of NPC Disease: Why do we need new mice?” (Manuel Lopez to present)**

**12:00-1:45 LUNCH – La Paloma Country Club (SAB to meet over lunch in 19<sup>th</sup> Hole)**

### *NPC2 Function*

- 2:00-2:20**     **Peter Lobel, Ph.D., Robert Wood Johnson Medical School** - “Molecular Characterization of Niemann-Pick C2 Disease”
- 2:30-2:50**     **Ann Stock, Ph.D., Robert Wood Johnson Medical School** - “Structural and functional characterization of NPC2”
- 3:00-3:20**     **Judith Storch, Ph.D., Rutgers University** - “The role of NPC2 in lysosomal cholesterol transport”
- 3:30-5:00**     **Poster Session – Finger Rock Room III**
- 6:00-6:45**     **Cocktails (cash bar) – La Paloma Country Club**
- 6:45**            **Buffet Dinner – La Paloma Country Club**

*Saturday, June 2, 2007*

- 7:00-7:45**     **Continental Breakfast – La Paloma Country Club**

### *Therapeutic Strategies for NPC Disease*

- 8:00-8:20**     **Frederick Maxfield, Ph.D., Weill Medical College of Cornell University** - “Chemical Compounds that Reduce Cholesterol Accumulation in NPC Cells”
- 8:30-8:50**     **Stephen Sturley, Ph.D., Columbia University** - “Intervention Points in NPC Disease, Insight from NPC-Protein Interactions”
- 9:00-9:20**     **Daniel Ory, M.D., Washington University** - “Mechanism of neurosteroid protection in a mouse model of NPC Disease”
- 9:30-9:50**     **Charles Vite, D.V.M., Ph.D., University of Pennsylvania** - “Effect of allopregnanolone administration on disease markers in NP-C cats”
- 10:00-10:20**   **Joyce Repa, Ph.D., UT Southwestern** - “Liver X Receptor agonists increase cholesterol loss from the CNS and attenuate microglia inflammatory responses to enhance the survival of *Npc1* - null mice”
- 10:30-10:45**   **Coffee Break**
- 10:45-11:05**   **Steven Walkley, D.V.M., Ph.D., Albert Einstein College of Medicine** - “Combinatorial Approaches to Therapy for NPC Disease”

- 11:15-11:35 Silvana Zanlungo, Ph.D., Pontificia Universidad Catolica de Chile - “Signaling pathways leading to neuronal death in NPC: Finding new therapeutic targets”**
- 11:45-12:00 Discussion/Conclusion**
- 12:15-1:30 Lunch – La Paloma Country Club**
- 12:15-4:00 SAB Meeting over lunch – La Paloma Country Club, 19<sup>th</sup> Hole**
- 1:30-3:00 Poster Session – Finger Rock Room III**
- 3:00-6:00 Open afternoon for conference attendees**
- 6:00-6:45 Cocktails (cash bar) - La Paloma Country Club**
- 6:45 Final Buffet Dinner - La Paloma Country Club**