

2010 Scientific Conference on Niemann-Pick Type C Disease

Friday September 24th, 2010

Session 1: Biochemistry of NPC Proteins Chair: Joyce Repa PhD

- 7:00 – 8:00 am **Breakfast**—La Paloma Country Club
- 8:00 – 8:15 **Welcome and Opening Announcements**
- 8:15 – 8:45 **Lina Abi-Mosleh PhD**—Cyclodextrin Overcomes Deficient Lysosome-to-Endoplasmic Reticulum Transport of Cholesterol in Niemann-Pick type C Cells. *Abstract #1*
- 8:45 – 9:15 **Suzanne Pfeffer PhD**—Functional analysis of NPC1 protein. *Abstract #18*
- 9:15 – 9:45 **Massoud Motamed PhD**—Identification of Surface Residues on Niemann-Pick C2 Essential for Hydrophobic Handoff of Cholesterol to NPC1 in Lysosomes. *Abstract #2*
- 9:45 – 10:15 **Coffee Break**
- 10:15 – 10:45 **Judith Storch PhD**—Cyclodextrin uses a NPC2-like mechanism to transport cholesterol in Niemann-Pick C disease. *Abstract #22*
- 10:45 – 11:15 **Kevin Vaughan PhD**—A comparison of ERAD and dynamic membrane models of NPC disease. *Abstract #11*
- 11:15 – 11:45 **Guosheng Liang PhD**—Cholesterol Binding to N-terminal Domain of NPC1 is Essential for NPC1 Function in Mice. *Abstract #15*
- 12:00 – 1:30 pm **Lunch**—La Paloma Country Club

Session 2: Clinical Science Chair: Suzanne Pfeffer

- 1:45 – 2:15 **Charina M. Ramirez PhD**—Pulmonary Disease in the Niemann-Pick Type C Mouse. *Abstract #6*
- 2:15 – 2:45 **Akshay Muralidhar, Medical Student**—Pulmonary Function and Pathology in Treated and Untreated *Npc1*^{-/-} Mice. *Abstract #9*

- 2:45 – 3:15 **Coffee Break**
- 3:15 – 3:45 **Cristin D. Davidson PhD**—Treatment of Niemann-Pick Type C Disease with Cyclodextrin. *Abstract #8*
- 3:45 – 4:15 **John Totenhagen, Graduate Student**—Magnetic Resonance Imaging of Mouse Models of NPC Disease. *Abstract #13*
- 4:15 – 4:45 **Olaf Wiest PhD & Paul Helquist PhD**—Modeling of Rare Diseases: From Networks to Drugs. *Abstract #26*
- 5:30 – 8:00 pm **Cocktails and Dinner**—La Paloma Country Club
-

Saturday September 25th, 2010

Session 3: Cell and Developmental Biology of NPC **Chair: Judy Storch**

- 7:00 – 8:00 am **Breakfast**—La Paloma Country Club
- 8:00 – 8:30 **Ting Yu, PhD Candidate**—Deconstructing Niemann-Pick type C neuropathology using conditional *Npc1* knockout mice. *Abstract #17*
- 8:30 – 9:00 **Fabian Bartz, PhD Student & Heiko Runz PhD**—Characterizing the role of TMEM97 in NPC disease. *Abstract #20*
- 9:00 – 9:30 **Kanagaraj Subramanian PhD**—Trafficking of Human NPC1 Mutants Responsible for NPC Disease. *Abstract #23*
- 9:30 – 10:00 **Coffee Break**
- 10:00 – 10:30 **Andrés Klein PhD**—Temporal and spatial rescue of Niemann Pick type C disease. *Abstract #21*
- 10:30 – 11:00 **H. Rob Yang PhD**—A role for oxysterol binding protein-related protein 5 in endosomal cholesterol trafficking. *Abstract #10*
- 11:00 – 11:30 **Forbes D Porter MD**—Development of surrogate markers for Niemann-Pick Disease, type C. *Abstract #19*
- 11:45 – 1:15 pm **Lunch**—**WESTIN LA PALOMA – MURPHY ROOM**

Session 4: Drug Discovery

Chair: Rob Yang

- 1:30 – 2:00 **Paul Helquist PhD**—Chemistry core: Collaborative drug discovery strategies for Niemann-Pick Type C disease therapeutics. *Abstract #27*
- 2:00 – 2:30 **Fred Maxfield PhD**—Tissue culture models for assessing NPC therapies. *Abstract #16*
- 2:30 – 3:00 **Joyce Repa PhD**—Comparing the Molecular Mechanisms of Novel Therapies for NPC Disease. *Abstract #14*
- 3:00 – 3:30 **Coffee Break**
- 3:30 – 4:00 **Mary M Cloutier PhD**—A rapid neuromuscular and behavioural score for quantitative assessment of disease progression in a murine model of Niemann Pick Type C Disease. *Abstract #7*
- 4:00 – 4:30 **Ron Browne PhD**—Clinical experience with intravenous infusions of hydroxy-propyl-beta-cyclodextrin in identical twin patients with Niemann-Pick Type C disease under individual investigator new drug exemptions. *Abstract #4*
- 4:30 – 5:00 **Charles Vite DVM, PhD**—Intrathecal cyclodextrin therapy of feline Niemann-Pick Type C disease. *Abstract #25*
- 6:00 – 8:00 pm **Cocktails and Dinner—La Paloma Country Club**
-