

Conference Agenda

Wednesday, October 5, 2016

5:00-6:30 Opening Session

- 5:00-5:30 **Welcome: Susan Cotman, Ph.D.**, Chair, NCL 2016 Organizing Committee
Margie Frazier, Ph.D., LISW-S, BDSRA Executive Director, NCL 2106 Sponsoring Organization
- 5:30-6:30 **Keynote Address: James Gusella, Ph.D.** (MGH/Harvard Medical School) The genetic research cycle in human disease
- 6:30-8:00 **Welcome Reception**

Thursday, October 6, 2016

8:30-12:20 Session 1: Genetics and Cell Biology of the NCLs

- 8:30-8:35 **Introduction**-Session Chairs: Marco Sardiello, Ph.D., Jill Weimer, Ph.D.
- 8:35-9:00 **Sara Mole, Ph.D.** (University College London) Perspectives on NCL genetics: genes, mutations and genotype-phenotype correlations (O1)
- 9:00-9:25 **Monkol Lek, Ph.D.**, (Broad Institute of MIT and Harvard) Using genomic technologies for the diagnosis of rare diseases (O2)
- 9:25-9:40 **Murat Bastepe, Ph.D.**, (MGH) Identification of large exonic deletions within genes responsible for neuronal ceroid lipofuscinosis (NCL) (O3)
- 9:40-9:55 **Sreeganga Chandra, Ph.D.**, (Yale University) Identification of PPT1 substrates: Implications for CLN1 (PPT1 and CSPalpha) (O4)
- 9:55-10:20 **Stephane Lefrancois, Ph.D.**, (Centre INRS-Institut Armand-Frappier) CLN3 and CLN5 regulate endosomal sorting (O5)
- 10:20-10:35 **Coffee break**
- 10:35-11:00 **Emyr Lloyd-Evans, Ph.D.**, (Cardiff University) Identifying an ion channel function for CLN3 and a role in regulating lysosomal response to changes in swelling and osmolarity (O6)
- 11:00-11:15 **Favio Pesaola**, (National University of Cordoba) Implication of CLN8 in the spatial distribution of lysosomes (O7)
- 11:15-11:30 **Robert Huber, Ph.D.**, (Trent University) Aberrant migration and adhesion in a Dictyostelium model for juvenile neuronal ceroid lipofuscinosis (O8)
- 11:30-11:55 **Jill Weimer, Ph.D.**, (Sanford Research) CLN6's role in regulation of vesicular trafficking (O9)
- 11:55-12:20 **Marco Sardiello, Ph.D.**, (Baylor College of Medicine) TFEB, lysosomal biogenesis, and neuronal ceroid lipofuscinosis (O10)

12:20-1:30 Poster Session I (Lunch will be provided)

1:30-4:45 Session 2: Disease models and mechanisms

- 1:30-1:40 **Introduction**-Session Chairs: Sara Mole, Ph.D., Tammy Kielian, Ph.D.
- 1:40-2:05 **Michelle Hastings, Ph.D.**, (Rosalind Franklin University of Medicine and Science) Antisense oligonucleotides for the treatment of juvenile neuronal ceroid lipofuscinosis (O11)
- 2:05-2:20 **Ryan Geraets**, (Sanford Research) A new cLINCL mouse model for studying various therapeutic approaches (O12)

- 2:20-2:45 **Jon Cooper, Ph.D.** (UCLA) The nature of glial dysfunction and its impact upon neurons varies between forms of NCL (O13)
- 2:45-3:00 **Claire Russell, Ph.D.**, (Royal Veterinary College University of London) Valproic acid attenuates seizures and extends lifespan of the zebrafish model of CLN2 disease (O14)
- 3:00-3:15 **Coffee break**
- 3:15-3:30 **Rosanna Beraldi, Ph.D.**, (Sanford Research) Generation of a Juvenile Batten disease porcine model (O15)
- 3:30-3:45 **Carolyn Schmidtke, Ph.D.**, (University Medical Center Hamburg-Eppendorf) Transcriptional control of lysosomal components in CLN3 defective cells (O16)
- 3:45-4:00 **Megan Bosch**, (University of Nebraska Medical Center) Abnormal astrocyte glutamate regulation and metabolic dysfunction contribute to neuronal pathology in juvenile neuronal ceroid lipofuscinosis (O17)
- 4:00-4:15 **Colleen Stein, Ph.D.**, (University of Iowa) Use of a transgenic system to study cell-type specific restoration of CLN3 in a JNCL mouse model (O18)
- 4:15-4:30 **Uma Chandrachud, Ph.D.**, (MGH, Harvard Medical School) Targeting Ca²⁺ homeostasis rescues lysosomal phenotypes in neuronal cell models of juvenile NCL (O19)
- 4:30-4:45 **Sophia Kleine-Holthaus, Ph.D.**, (University College London) AAV-mediated transduction of bipolar cells is essential to prolong the photoreceptor survival in Cln6^{ncif} mice. (O20)
- 5:15 Boarding for Boston Duck Boats Tour and Quincy Market Dinner**

Friday, October 7, 2016

8:30-11:35 Session 3: Genome-wide and systems level strategies for NCL

- 8:30-8:40 **Introduction**-Session Chairs: Beverly Davidson, Ph.D., David Sleat, Ph.D.
- 8:40-9:05 **Stephen Haggarty, Ph.D.**, (MGH, Broad Institute of MIT and Harvard) Chemical genomics of neurodegeneration: Patient-specific iPSC models (O21)
- 9:05-9:30 **Carmine Settembre, Ph.D.**, (TIGEM) Physiological role of the lysosome as a signaling organelle (O22)
- 9:30-9:45 **Elisa Tinelli, Ph.D.**, (University College of London) Screening for lead compounds to treat juvenile CLN3 disease (O23)
- 9:45-10:00 Coffee break
- 10:00-10:25 **Ethan Perlstein, Ph.D.**, (Perlstein Lab PBC) Using worms and flies to discover drugs for lysosomal storage disorders (O24)
- 10:25-10:50 **Stephan Storch, Ph.D.**, (University Medical Center Hamburg-Eppendorf) Neurodegeneration and lysosomal pathology in a Cln7 mouse model (O25)
- 10:50-11:05 **Ursula Heins Marroquin**, (University of Luxembourg) Towards small-molecule therapies for juvenile forms of Batten disease: drug screens in yeast and zebrafish models of JNCL (O26)
- 11:05-11:20 **Thomas Wishart, Ph.D.**, (University of Edinburgh) Identifying novel therapeutic targets for the NCLs (O27)
- 11:20-11:35 **Maica Llaverro Hurtado**, (University of Edinburgh) Identifying potential peripherally accessible biomarkers in the NCLs (O28)

11:35-1:05 Poster Session II (Lunch will be provided)

1:05-5:15

Session 4: Recent advances and challenges in translational research

- 1:05-1:15 **Introduction**-Session Chairs: David Pearce, Ph.D., Sandra Hofmann, M.D.
- 1:15-1:40 **Alessandra Biffi, M.D.**, (Dana Farber Cancer Institute, Boston Children's, Harvard Medical School) HSC gene therapy for LSDs: current and new indications
- 1:40-2:05 **Mark Sands, Ph.D.**, (Washington University of St. Louis) A new therapeutic target for infantile neuronal ceroid lipofuscinosis (O29)
- 2:05-2:30 **David Sleat, Ph.D.**, (Rutgers University) Chronic enzyme replacement therapy to the brain of a mouse model of late infantile neuronal ceroid lipofuscinosis (O30)
- 2:30-2:55 **Beverly Davidson, Ph.D.**, (The Children's Hospital of Pennsylvania) Emerging therapies for the NCLs
- 2:55-3:10 **Coffee break**
- 3:10-3:35 **Tammy Kielian, Ph.D.**, (University of Nebraska Medical Center) Self-complementary AAV9 gene delivery partially corrects neuropathology associated with Juvenile Neuronal Ceroid Lipofuscinosis (CLN3) (O31)
- 3:35-4:00 **Ken Hensley, Ph.D.**, (University of Toledo) XN001 promotes autophagy to treat Batten disease (O32)
- 4:00-4:15 **Alejandra Rosenberg, Ph.D.**, (University of North Carolina at Chapel Hill) Preclinical Intrathecal Gene Therapy for Infantile Neuronal Ceroid Lipofuscinosis (O33)
- 4:15-4:30 **Janos Groh, Ph.D.**, (University Hospital Wuerzburg) Immunomodulatory therapy in mouse models of CLN1 and CLN3 disease (O34)
- 4:30-4:45 **Rebecca Whiting, Ph.D.**, (University of Missouri) Retinal function and structure are preserved in a canine model of CLN2 Batten disease after intravitreal implantation of stem cells genetically modified to overproduce TPP1 enzyme (O35)
- 4:45-4:50 **Dave Palmer, Ph.D.**, (Lincoln University) An introduction to 'in vivo monitoring of viral gene injection therapy in ovine Batten disease' (P41)
- 4:50-5:15 **Nadia Mitchell, Ph.D.**, (Lincoln University) Gene transfer can prevent stereotypical disease development in ovine CLN5 and CLN6 models of NCL (O37)

Dinner on own

Saturday, October 8, 2016

8:00-9:15 Late Breaking News and Selected Talks from Posters

- 8:00-8:10 **Introduction**
- 8:10-8:30 **Betsy Ferguson, Ph.D.**, (Oregon National Primate Research Center) A non-human primate model of neuronal ceroid lipofuscinosis (O38)
- 8:30-8:45 Short talk selected from Posters (TBD)
- 8:45-9:00 Short talk selected from Posters (TBD)
- 9:00-9:15 Short talk selected from Posters (TBD)

9:15-10:15 Hot Topic in Clinical Research Workshop: "Neurobehavioral and Psychosocial Function in the NCLs"

- 9:15-9:20 Introduction-Workshop Chair: Heather Adams, Ph.D. (O39)
- 9:20-9:30 **Bengt Elmerskog** (Statped midt, Norway) Experiences on education and adaptations for learners with juvenile neuronal ceroid lipofuscinosis (O40)

- 9:30-9:40 **Anne-Grethe Tøssebro**, (Statped midt, Norway) A study on tactile/body signs or hand signs for students with Batten disease (JNCL) (O41)
- 9:40-9:50 **Andrea West**, (Batten Disease Family Association, UK) The burden of CLN2 disease on families: home-based surveys with caregivers in Germany and the United Kingdom (O42)
- 9:50-10:15 Panel discussion
- 10:15-10:45 **Coffee break**
- 10:45-11:30** **Margie Frazier, Ph.D., LISW-S** (Batten Disease Support and Research Association), **Erika Augustine, M.D.**, (University of Rochester Medical Center), Engaging families in research workshop (O43)
- 11:30-1:00** **Poster Session III (Lunch will be provided)**
- 1:00-2:10** **Session 5: Registries, Biorepositories and Rating Scales**
- 1:00-1:10 **Introduction**-Session Chairs: Erika Augustine, M.D., Angela Schulz, M.D.
- 1:10-1:25 **Jonathan Mink, M.D., Ph.D.**, (University of Rochester) Rating scales for natural history studies and registries in the NCLs (O44)
- 1:25-1:40 **Ines Noher de Halac, Ph.D.**, (National University of Cordoba) Argentina next generation sequencing technology (NGS) in human medicine, and registry of NCL disease in databases focused on genotype-phenotype relationships (O45)
- 1:40-1:55 **Angela Schulz, M.D.**, (University Medical Center Hamburg-Eppendorf) The DEM-CHILD Consortium: an international collaboration aimed at improved diagnosis and natural history data for outcomes-oriented research in the NCLs (O46)
- 1:55-2:10 **Jan Hochstein, M.D.**, (University Medical Center Hamburg-Eppendorf) Longitudinal 8-years brain volumetric analysis in 35 CLN3 patients: Successful development of a sensitive marker to measure clinical outcome (O47)
- 2:10-2:25 **Panel discussion**
- 2:25-2:40 **Coffee break**
- 2:40-4:30** **Session 6: Progress towards therapies: Clinical trial updates**
- 2:40-2:50 **Introduction**-Session Chairs: Ruth Williams, M.D., Jonathan Mink, M.D., Ph.D.
- 2:50-3:15 **Ronald Crystal, M.D.**, (Weill Cornell Medical College) Gene therapy for CLN2 disease
- 3:15-3:40 **Angela Schulz, M.D.**, (University Medical Center Hamburg-Eppendorf) Intracerebroventricular cerliponase alfa (BMN 190) in children with CLN2 disease: Results from a Phase 1/2, open-label, dose-escalation study (O48)
- 3:40-4:05 **Erika Augustine, M.D.**, (University of Rochester) Developing therapies for individuals with CLN3 disease – results from a phase 2, double-blind, crossover study of mycophenolate (O49)
- 4:05-4:30 **Emily de los Reyes, M.D.**, (Nationwide Children’s Hospital) CLN6: The promise of gene therapy (O50)
- 4:30-4:50 **Conference closing and announcement of the 16th International Conference on NCL**
- 7:30-9:30** **Boston Batten Brew Bash!**