

Wednesday 28th March

- 17.00-18.00 **Opening Lecture – Welcome to NCL2012**
Jon Cooper, Sara Mole, Ruth Williams, Andrea West
- 18.00-19.00 Opening Reception
- 19.00 Evening Meal

Thursday 29th March

- 9.00-12.00 **Session 1 Genetics & Biology of the NCLs**
Chairs: Sara Mole (UCL), Thomas Braulke (Hamburg)
- 9.00-9.10 Introduction by session chairs
- 9.10-9.30 Batten disease: A case of retrograde endosome-Golgi protein transport gone bad? *Jeff Gerst (Weizmann Institute)*
- 9.30-9.50 Identification of potential biomarkers and modifier genes affecting the clinical course of CLN3 Disease.
Georgia Makrypidi (Hamburg)
- 9.50-10.00 Brain endothelial cells deficient in CLN3 display abnormal phenotypes associated with alterations in membrane microdomains. *Luis Tecedor (University of Iowa)*
- 10.00-10.20 CLN3 interacts with motor protein complexes and modifies location of late endosomal/lysosomal compartments.
Kristiina Uusi-Rauva (Helsinki)
- 10.20-10.30 Defining the role of mRNA degradation in Batten disease
Jake Miller (Sanford Research)
- 10.30-10.50 *Coffee Break*
- 10.50-11.10 A novel CRMP2/CLN6/KLC4 signaling complexes and its role in vLINCL. *Jill Weimer (Sanford Research)*
- 11.10-11.20 Next generation sequencing identifies the disease causing mutation for NCL in South Hampshire sheep.
Imke Tammen (Sydney)
- 11.20-11.35 Proteolytic cleavage of the lysosomal membrane protein CLN7 is associated with CLN7 disease. *Pieter Steenhuis (Hamburg)*
- 11.35-11.50 Mutations in DNAJC5, encoding cysteine-string protein alpha (CSP α), cause autosomal dominant adult neuronal ceroid lipofuscinosis. *Stan Knoch (Prague)*
- 11.50-12.00 A recurrent mutation in DNAJC5 causes autosomal dominant Kufs Disease. *Maxime Cadieux-Dion (Montreal)*

12.00-13.00	<i>Poster Session</i>
13.00-14.00	<i>Lunch</i>
14.00-17.00	Session 2 Disease Mechanisms Chairs: David Pearce (Sanford Research), David Palmer (Lincoln, NZ)
14.00-14.10	Introduction by session chairs
14.10-14.25	Mutant glia impair the health of neurons in Juvenile NCL <i>Lotta Parviainen (KCL)</i>
14.25-14.35	Effects of CLN3 loss on inflammasome activation in microglia <i>Tammy Kielian (University of Nebraska)</i>
14.35-14.50	Delayed pathological changes in the thalamocortical system of immunodeficient <i>Ppt1</i> null mutant mice. <i>Thomas Kühl (KCL)</i>
14.50-15.05	Synaptic dysfunction in motor nerve terminals of knock-out mice lacking Cysteine String Protein-alpha, a protein involved in autosomal-dominant adult-onset neuronal ceroid lipofuscinosis. <i>Rafael Fernández-Chacón (Sevilla)</i>
15.05-15.15	Synaptic failure may initiate the neuronal degeneration in cathepsin D deficiency. <i>Maciej Lalowski (Helsinki)</i>
15.15-15.25	Early synaptic abnormalities in multiple models of NCL Megan O'Hare (KCL)
15.25-15.45	<i>Coffee</i>
15.45-16.00	Evidence for altered neurogenesis in mouse and sheep models of NCL – an attempt of self-repair? <i>Sybille Dihanich (KCL/UCL)</i>
16.00-16.15	<i>In vivo</i> intercellular correction in ovine CLN6. <i>Lucy Barry (Lincoln, NZ)</i>
16.15-16.30	Spatial Proteomics Identify a Novel Drug Target in JNCL <i>Mika Ruonala (Frankfurt)</i>
16.30-16.40	Early-stage neurologic and non-neurologic abnormalities in <i>Cln3^{Aex7/8}</i> mice precede overt neurodegeneration Sue Cotman (MGH Boston)
16.40-16.50	Autophagy dysfunction in NCL disease <i>Matt Micsenyi (AECOM, New York)</i>
16.50-17.00	Shared pathological themes between the NCLs and other LSDs <i>Sarah Pressey (KCL/UCL)</i>
17.00-18.00	<i>Poster Session</i>
17.15-17.40	Science Summary Sessions for Parents
18.30	Evening Meal (Followed by 'Pub Social')

Friday 30th March

- 9.00-11.00 **Session 3 Links to Other Diseases**
Chairs: Fran Platt (Oxford), Steve Walkley (AECOM, New York)
- 9.00-9.10 Introduction by session chairs
- 9.10-9.35 Modulating Cellular Clearance in Lysosomal Storage Diseases.
Andrea Ballabio (TIGEM, Naples & Baylor COM)
- 9.35-10.00 Role of the endo-lysosomal system in neurodegenerative diseases
Ralph Nixon (NYU Langone Medical Center/Nathan Kline Institute)
- 10.00-10.20 *Coffee*
- 10.20-10.45 Lysosomal Ca²⁺ homeostasis: role in pathogenesis of lysosomal storage diseases. *Emyr Lloyd Evans (Cardiff)*
- 10.45-10.10 Astrocytes as neuronal energy providers: putative therapeutic targets in the NCLs? *Luc Pellerin (Lausanne)*
- 11.10-12.00 **Session 4 Recent Research Findings**
Chairs: Anu Jalanko (Helsinki), Hannah Mitchison (UCL)
- 12.00-12.50 *Poster Session*
- 13.00 prompt **Conference Photograph**
- 13.10-14.00 *Lunch*
- 14.00-17.00 **Session 5 New Clinical Perspectives**
Chairs: Angela Schulz (Hamburg), Jon Mink (Rochester)
- 14.00-14.10 Introduction by session chairs
- 14.10-14.30 Sex Differences in Clinical Progression and Quality of Life in Juvenile Neuronal Ceroid Lipofuscinosis. *Heather Adams (University of Rochester)*
- 14.30-14.50 Multidimensional Clinical Assessment Tool for late infantile CLN2 Disease. *Ruth Williams (Evelina Children's Hospital, London)*
- 14.50-15.10 Quantitative Brain Volumetric Analysis in Neuronal Ceroid Lipofuscinoses: A tool to precisely monitor disease progression
Angela Schulz (Hamburg)
- 15.10-15.20 Psychopathology in CLN3 disease: Correlation with disease progression and quality of life. *Angela Schulz (Hamburg)*
- 15.20-15.50 *Coffee*
- 15.50-16.10 Sinus node dysfunction in juvenile neuronal ceroid lipofuscinosis
John Østergaard (Aarhus University Hospital)
- 16.10-16.30 Lifelong learning for individuals with Batten disease.
Bengt Elmerskog (Tambartun National Resource Centre)

- 16.30-16.40 Mycophenolate mofetil for the treatment of Juvenile Ceroid Lipofuscinosis. *Amy Vierhile (University of Rochester)*
- 16.40-16.50 Biomarker discovery in Batten disease. *Chun-hung Chan (Sanford Research)*
- 16.50-17.00 Magnetic resonance volumetrics, diffusion tensor imaging and spectroscopy as biomarkers to assess efficacy of gene therapy in a canine model for LINCL. *Fred Wininger (University of Missouri)*
- 17.00-18.00 *Poster Session*
- 17.15-17.40 Science Summary Sessions for Parents
- 18.30 Evening Meal together with Parents Organisation Conference

Saturday 31st March

- 9.00-12.00 **Session 6 Experimental Therapies**
Chairs: Beverley Davidson (Iowa), Mark Sands (Wash U)
- 9.00-9.10 Introduction by session chairs
- 9.10-9.30 The synergistic effects of CNS-directed gene therapy and bone marrow transplantation for infantile neuronal ceroid lipofuscinosis. *Mark Sands (Wash U)*
- 9.30-9.50 A small molecule anti-inflammatory enhances the therapeutic effects of AAV-mediated CNS-directed gene therapy for infantile neuronal ceroid lipofuscinosis. *Charles Shyng (Wash U)*
- 9.50-10.00 Intravenous high-dose enzyme replacement therapy with recombinant palmitoyl-protein thioesterase reduces brain and visceral lysosomal storage in a mouse model of infantile neuronal ceroid lipofuscinosis *Sandy Hofmann (UT SouthWestern)*
- 10.00-10.10 Nonclinical development of recombinant human Tripeptidyl-peptidase 1 (rhTPP1) enzyme replacement therapy (ERT) for Late Infantile Neuronal Ceroid Lipofuscinosis (LINCL). *Brian Vuilleminot (BioMarin)*
- 10.10-10.30 Treatment with recombinant human tripeptidyl peptidase-1 (rhTPP1) delays onset of neurologic signs in a canine model of late infantile neuronal ceroid lipofuscinosis (LINCL). *Christine Sibigtroth (University of Missouri)*
- 10.30-10.50 *Coffee Break*
- 10.50-11.10 AAV-TPP1 Transduction of Brain Ependyma in TPP1-null Dogs Results in Widespread CNS Distribution of TPP1 Enzyme and Improves NCL Disease Phenotypes. *Beverley Davidson (Iowa)*
- 11.10-11.30 Gene delivery to the Perinatal Brain. *Andrew Wong (KCL)*

11.30-11.50	Global CNS gene delivery platform in non-human primates utilizing self-complementary AAV9 vectors. <i>Steven Gray (UNC)</i>
11.50-12.00	Post transplantation fate of human neural stem cells in a mouse model of late infantile NCL. <i>Helen Brooks (KCL)</i>
12.00-13.00	<i>Poster Session</i>
13.00-14.00	<i>Lunch</i>
14.00-17.00	Session 7 Joint Session with Parents Organisations Meeting Session Chairs: Andrea West (BDFa), Sara Mole (UCL) Jon Cooper (KCL), Ruth Williams (Evelina Children's Hospital, London)
14.00-14.10	Introduction by session chairs
14.10-14.40	Summary of Scientific Sessions by respective Chairs
14.40-14.45	Parent Group Presentation
14.45-15.00	Prizes and thanks. Announcement of next NCL congress.
15.00-15.15	<i>Coffee</i>
15.15-16.30	'Market Place'
16.00-17.00	<i>Poster Session</i>
18.30	Conference 'Banquet' for Scientists and Parents