

NCL2012 Scientific Programme Overview

Wednesday 28th March

14.00-19.00	Registration
17.00-18.00	Welcome and Opening Lectures
18.00	<i>Opening Reception followed by Evening Meal</i>

Thursday 29th March

09.00-12.00	Session 1 Genetics & Biology of the NCLs Chairs: Sara Mole (UCL), Thomas Brulke (Hamburg)
12.00-13.00	<i>Poster Session 1</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)
17.00-18.00	<i>Poster Session 2</i>
18.00-18.30	Science Summary Sessions for Parents
18.45	<i>Evening Meal followed by 'Pub Social'</i>

Friday 30th March

08.30-10.20	Session 3 Links to Other Diseases Chairs: Fran Platt (Oxford), Steve Walkley (AECOM, New York)
10.20-10.40	<i>Break</i>
10.40-12.00	Session 4 Recent Research Findings Chairs: Anu Jalanko (Helsinki), Hannah Mitchison (UCL)
12.00-12.50	<i>Poster Session 3</i>
13.00 prompt	<i>Conference Photograph</i>
13.10-14.00	<i>Lunch</i>
14.00-17.00	Session 5 New Clinical Perspectives Chairs: Angela Schulz (Hamburg), Jon Mink (Rochester)
17.00-18.00	<i>Poster Session 4</i>
18.00-18.30	Science Summary Sessions for Parents
18.45	<i>Evening Meal together with Patients Organisation Conference</i>

Saturday 31st March

09.00-12.00	Session 6 Experimental Therapies Chairs: Beverley Davidson (Iowa), Mark Sands (Wash U)
12.00-13.00	<i>Poster Session 5</i>
13.00-14.00	<i>Lunch</i>
14.00-17.00	Session 7 Joint Session with Patients Organisations Meeting Session Chairs: Andrea West (BDFa), Sara Mole (UCL), Jon Cooper (KCL), Ruth Williams (Evelina Children's Hospital)
15.15-16.30	<i>'Market Place'</i>
16.30-17.30	<i>Poster Session 6</i>
18.30	<i>Conference 'Banquet' for Scientists and Parents</i>

NCL2012 Scientific Programme

All talks to be held in the main auditorium, Windsor Building.

Wednesday 28th March

- 14.00-19.00 **Registration**
- 17.00-18.00 **Welcome and Opening Lectures**
Jon Cooper, Sara Mole, Ruth Williams, Andrea West
Perspectives on NCL disease. *Ruth Williams (Evelina Children's Hospital)*
Perspectives on NCL genetics. *Sara Mole (UCL)*
Perspectives on disease mechanisms and therapies. *Jon Cooper (KCL)*
- 18.00-19.00 **Opening Reception** (Picture Gallery, Founder's Building)
- 19.00 **Evening Meal** (Founder's Dining Room, Founder's Building)

Thursday 29th March

- 09.00-12.00 **Session 1 Genetics & Biology of the NCLs**
Chairs: Sara Mole (UCL), Thomas Braulke (Hamburg)
- 09.00-09.10 Introduction by session chairs
- 09.10-09.30 O1 Batten disease: A case of retrograde endosome-Golgi protein transport gone bad? *Jeff Gerst (Weizmann Institute)*
- 09.30-09.50 O2 Identification of potential biomarkers and modifier genes affecting the clinical course of CLN3 Disease. *Georgia Makrypidi (Hamburg)*
- 09.50-10.00 O3 Brain endothelial cells deficient in CLN3 display abnormal phenotypes associated with alterations in membrane microdomains. *Luis Tecedor (University of Iowa)*
- 10.00-10.20 O4 CLN3 interacts with motor protein complexes and modifies location of late endosomal/lysosomal compartments. *Kristiina Uusi-Rauva (Helsinki)*
- 10.20-10.30 O5 Defining the role of mRNA degradation in Batten disease. *Jake Miller (Sanford Research)*
- 10.30-10.50 **Break**
- 10.50-11.10 O6 A novel CRMP2/CLN6/KLC4 signaling complex and its role in vLINCL. *Jill Weimer (Sanford Research)*
- 11.10-11.20 O7 Next generation sequencing identifies the disease causing mutation for NCL in South Hampshire sheep. *Imke Tammen (Sydney)*
- 11.20-11.35 O8 Proteolytic cleavage of the lysosomal membrane protein CLN7 is associated with CLN7 disease. *Pieter Steenhuis (Hamburg)*

11.35-11.50	O9	Mutations in DNAJC5, encoding cysteine-string protein alpha (CSP α), cause autosomal dominant adult neuronal ceroid lipofuscinosis. <i>Stan Knoch (Prague)</i>
11.50-12.00	O10	A recurrent mutation in DNAJC5 causes autosomal dominant Kufs Disease. <i>Maxime Cadieux-Dion (Montreal)</i>
12.00-13.00		Poster Session 1
13.00-14.00		Lunch (Windsor Building)
14.00-17.00		Session 2 Disease Mechanisms Chairs: David Pearce (Sanford Research), David Palmer (Lincoln)
14.00-14.10		Introduction by session chairs
14.10-14.25	O11	Mutant glia impair the health of neurons in Juvenile NCL. <i>Lotta Parviainen (KCL)</i>
14.25-14.35	O12	Effects of CLN3 loss on inflammasome activation in microglia. <i>Tammy Kielian (University of Nebraska)</i>
14.35-14.50	O13	Delayed pathological changes in the thalamocortical system of immunodeficient <i>Ppt1</i> null mutant mice. <i>Thomas Köhl (KCL)</i>
14.50-15.05	O14	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	O15	Synaptic failure may initiate the neuronal degeneration in cathepsin D deficiency. <i>Maciej Lalowski (Helsinki)</i>
15.15-15.25	O16	Early synaptic abnormalities in multiple models of NCL. <i>Megan O'Hare (KCL)</i>
15.25-15.45		Break
15.45-16.00	O17	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	O18	<i>In vivo</i> intercellular correction in ovine CLN6. <i>Lucy Barry (Lincoln)</i>
16.15-16.30	O19	Spatial proteomics identify a novel drug target in JNCL. <i>Mika Ruonala (Frankfurt)</i>
16.30-16.40	O20	Early-stage neurologic and non-neurologic abnormalities in <i>Cln3^{Δex7/8}</i> mice precede overt neurodegeneration. <i>Sue Cotman (MGH Boston)</i>
16.40-16.50	O21	Autophagy dysfunction in NCL. <i>Matt Micsenyi (AECOM, New York)</i>
16.50-17.00	O22	Shared pathological themes between the NCLs and other LSDs. <i>Sarah Pressey (KCL/UCL)</i>
17.00-18.00		Poster Session 2
18.00-18.30		Science Summary Sessions for Parents (Room 1-04)
18.45		Evening Meal (HUB Dining Hall) followed by 'Pub Social' (Medicine Bar)

Friday 30th March

- 08.30-10.20 **Session 3 Links to Other Diseases**
Chairs: Fran Platt (Oxford), Steve Walkley (AECOM, New York)
- 08.30-08.40 Introduction by session chairs
- 08.40-09.05 O23 Modulation of cellular clearance in lysosomal storage diseases.
Andrea Ballabio (TIGEM, Naples & Baylor COM)
- 09.05-09.30 O24 Autophagy failure in Alzheimer's Disease and related diseases.
Ralph Nixon (NYU Langone Medical Center/Nathan Kline Institute)
- 09.30-09.55 O25 Lysosomal Ca²⁺ homeostasis: role in pathogenesis of lysosomal storage diseases. *Emyr Lloyd Evans (Cardiff)*
- 9.55-10.20 O26 Astrocytes as neuronal energy providers: putative therapeutic targets in the NCLs? *Luc Pellerin (Lausanne)*
- 10.20-10.40 **Break**
- 10.40-12.00 **Session 4 Recent Research Findings**
Chairs: Anu Jalanko (Helsinki), Hannah Mitchison (UCL)
- 10.40-10.45 Introduction by session chairs
- 10.45-10.55 O27 A novel genetic link between neuronal ceroid lipofuscinosis and the ubiquitin-proteasome system. *John Staropoli (Mass Gen Hospital)*
- 10.55-11.05 O28 Mutations in the gene encoding Cathepsin F are a cause of type B Kufs disease. *Katherine Smith (Walter and Eliza Hall Institute of Medical Research)*
- 11.05-11.15 O29 Homozygous mutations in progranulin can cause adult onset recessive NCL. *Katherine Smith (Walter and Eliza Hall Institute of Medical Research)*
- 11.15-11.25 O30 Exome sequencing reveals ATP13A2 mutations underlying juvenile NCL. *Jose Bras (UCL)*
- 11.25-11.35 O31 Clinico-pathological features of Kufs disease due to *CLN6* mutation. *Samuel Berkovic (Melbourne)*
- 11.35-11.45 O32 Age-dependent therapeutic effect of memantine in a mouse model of juvenile Batten disease. *David Pearce (Sanford Research)*
- 11.45-12.00 Discussion of the implications of recent genetic findings
- 12.00-12.50 **Poster Session 3**
- 13.00 prompt **Conference Photograph**
- 13.10-14.00 **Lunch** (Windsor Building)
- 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	O33	Sex Differences in clinical progression and quality of life in juvenile neuronal ceroid lipofuscinosis. <i>Heather Adams (U. of Rochester)</i>
14.30-14.50	O34	Multidimensional Clinical Assessment Tool for late infantile CLN2 Disease. <i>Ruth Williams (Evelina Children's Hospital, London)</i>
14.50-15.10	O35	Quantitative Brain Volumetric Analysis in Neuronal Ceroid Lipofuscinoses: A tool to precisely monitor disease progression. <i>Angela Schulz (Hamburg)</i>
15.10-15.20	O36	Psychopathology in CLN3 disease: Correlation with disease progression and quality of life. <i>Angela Schulz (Hamburg)</i>
15.20-15.50		Break
15.50-16.10	O37	Sinus node dysfunction in juvenile neuronal ceroid lipofuscinosis. <i>John Østergaard (Aarhus University Hospital)</i>
16.10-16.30	O38	Lifelong learning for individuals with Batten disease. <i>Bengt Elmerskog (Tambartun National Resource Centre)</i>
16.30-16.40	O39	Mycophenolate mofetil for the treatment of Juvenile Ceroid Lipofuscinosis. <i>Erika Augustine (University of Rochester)</i>
16.40-16.50	O40	Biomarker discovery in Batten disease. <i>Chun-hung Chan (Sanford Research)</i>
16.50-17.00	O41	Magnetic resonance volumetrics, diffusion tensor imaging and spectroscopy as biomarkers to assess efficacy of gene therapy in a canine model for LINCL. <i>Fred Wininger (University of Missouri)</i>
17.00-18.00		Poster Session 4
18.00-18.30		Science Summary Sessions for Parents (Room 1-04)
18.45		Evening Meal together with Patients Organisation Conference (Founder's Dining Room, Founder's Building)

Saturday 31st March

09.00-12.00		Session 6 Experimental Therapies Chairs: Beverly Davidson (Iowa), Mark Sands (Wash U)
09.00-09.10		Introduction by session chairs
09.10-09.30	O42	The synergistic effects of CNS-directed gene therapy and bone marrow transplantation for infantile neuronal ceroid lipofuscinosis. <i>Mark Sands (Wash U)</i>
09.30-09.50	O43	A small molecule anti-inflammatory enhances the therapeutic effects of AAV-mediated CNS-directed gene therapy for infantile neuronal ceroid lipofuscinosis. <i>Charles Shyng (Wash U)</i>
09.50-10.00	O44	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. <i>Sandy Hofmann (UT SouthWestern)</i>

10.00-10.10	O45	Treatment with recombinant human tripeptidyl peptidase-1 (rhTPP1) delays onset of neurologic signs in a canine model of late infantile neuronal ceroid lipofuscinosis (LINCL). <i>Christine Sibigtroth (University of Missouri)</i>
10.10-10.30	O46	Nonclinical development of recombinant human Tripeptidyl peptidase 1 (rhTPP1) enzyme replacement therapy (ERT) for Late Infantile Neuronal Ceroid Lipofuscinosis (LINCL). <i>Brian Vuilleminot (BioMarin)</i>
10.30-10.50		Break
10.50-11.10	O47	AAV-TPP1 transduction of brain ependyma in TPP1-null dogs results in widespread CNS distribution of TPP1 enzyme and improves NCL disease phenotypes. <i>Beverly Davidson (Iowa)</i>
11.10-11.30	O48	Gene delivery to the perinatal brain. <i>Andrew Wong (KCL)</i>
11.30-11.50	O49	Global CNS gene delivery platform in non-human primates utilizing self-complementary AAV9 vectors. <i>Steven Gray (UNC)</i>
11.50-12.00	O50	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		Poster Session 5
13.00-14.00		Lunch (Windsor Building)
14.00-17.00		Session 7 Joint Session with Patients Organisations Meeting Session Chairs: Andrea West (BDFa), Sara Mole (UCL), Jon Cooper (KCL), Ruth Williams (Evelina Children's Hospital)
14.00-14.10		Introduction by session chairs
14.10-14.40		Summary of Scientific Sessions by respective Chairs
14.40-14.45		Patient Group Presentation. <i>Lance Johnston & Irena Newcombe</i>
14.45-15.00		Prizes and thanks. Announcement of next NCL congress.
15.00-15.15		Break, with refreshments available during the Market Place
15.15-16.30		'Market Place'
16.30-17.30		Poster Session 6
18.30		Conference 'Banquet' for Scientists and Families (Founder's Dining Room, Founder's Building)

Microphone carriers

Welcome Session - Sophia kleine Holthaus, Davide Marotta
Session 1 Genetics and Biology - Sophia kleine Holthaus, Francesco Pezzini
Session 2 Disease Mechanisms - Davide Marotta, Katja Kanninen
Session 3 Links to Other Diseases - Christine Sibigtroth, Janos Groh
Session 4 Recent Research Findings - Heidi Larkin, Pieter Steenhuis
Session 5 New Clinical Perspectives - Mervi Kuronen, Matthew Micsenyi
Session 6 Experimental Therapies - Niv Dobzinski, Uma Chandrachud
Session 7 Joint Session - Helen Brooks, Yewande Pearce

NCL2012 Satellite Programme Overview

At various times around or during NCL2012 there will be additional sessions focused on specific topics of interest for particular groups of conference attendees. Some are designed for clinicians or scientists, or particular patient organisations or groups of parents. Many relate specifically to the different work packages of DEM-CHILD, an EU funded collaborative grant, and are thus relevant only to those who are part of this effort. Others are open to those with expertise or interest in the respective area. Some occur in parallel to the main programme. The person responsible is indicated in italics, and additional details for some can be found in the following pages.

Breakfast packs will be provided for those starting at 07.30.

All meetings will be held in the main conference building. If the room for each event is not indicated in the programme below, it will be indicated at the conference information desk prior to its start.

Wednesday 28th March

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| 10.00-12.30 | First NCL Mini-Master class: 'Motor disorders in CLN3 disease (Room 0-04) <i>Ruth Williams (further details provided on page 19)</i> |
| 10.00-12.00 | JNCL PhD Student Meeting (Room 1-02) <i>Frank Stehr/Danielle Kerkovich</i> |
| 12.00-16.30 | First Meeting of the Batten Disease International Alliance (BDIA) (Room 1-05) <i>Tony Heffernan/Andrea West</i> |
| 14.00-16.30 | DEM-CHILD WP03 meeting (Epidemiology / Natural history study) + Outcomes and Registry (Room 0-04) <i>Angela Schulz</i> |

Thursday 29th March

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|-------------|--|
| 07.30-08.30 | NCL Clinical Outcomes Conference (Sept 2012) Planning Session (Room 1-02) <i>Heather Adams</i> |
| 10.30-16.00 | BDFA Training day for Professionals (Room 1-05) <i>Andrea West/Ruth Williams (further details provided on page 19)</i> |
| 17.00-18.00 | DEM-CHILD WP01 meeting (Diagnostic gene chip) (Room 0-04) <i>Angela Schulz</i> |

Friday 30th March

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|-------------|---|
| 07.30-08.30 | DEM-CHILD WP08 (Teaching and dissemination) (Room 0-04) <i>Prathiba Singhi/Angela Schulz</i> |
| 17.00-18.00 | DEM-CHILD WP07 meeting (Identification of new NCL genes) (Room 0-04) <i>Sara Mole</i> |
| 17.30-18.00 | International Rare NCL Gene Consortium joins DEM-CHILD WP07 at 17.45 (Room 0-04) <i>Sara Mole</i> |

Saturday 31st March

- 07.30-08.30 DEM-CHILD WP06 (Innovative therapies for NCLs caused by mutations in transmembrane proteins) (Room 0-04) *Sander Smith*
- 16.00-17.00 BDFA UK Professional Networking Event (Room 0-04) *Andrea West*
- 17.00-18.00 DEM-CHILD WP02 meeting (Automated enzyme testing) (Room 1-04) *Herbert Korall and Susanne Kerlin*
- 17.00-18.00 Combined DEM-CHILD WP04 and WP05 meetings (CLN1 and CLN3 biomarkers) (Auditorium) *Thomas Braulke and Anu Jalanko*

Sunday 1st April

- 09.30-15.30 DEM-CHILD Steering group meeting (Moore building) *Angela Schulz*

NCL2012 Satellite Meetings Details

1st International NCL Mini-Master Class: Movement disorders in CLN3 disease

Wednesday 28th March, Room 0.04, 10.00-12.30

Introduction and welcome

Overview of treatment strategies for Parkinsonian Movement disorders in Childhood

Dr Jean-Pierre Lin and Dr Jonathan Mink

Cases and videos – participants

Questions for discussion

- How should movement disorder in CLN3 disease be described?
- What neuronal pathways are likely to be involved?
- Disease mechanisms?
- Optimum treatment strategies?
- Which drugs? Timing of treatment? How should we monitor treatment?

Conclusions and summing up

BDFA Training Day for Professionals caring for children and teenagers with Juvenile Batten disease

Thursday 29th March Room 1.05, 10.30-16.00

This course is suitable for teachers, nurses, therapists, social workers and all professionals involved in the care of children and teenagers with Juvenile Batten disease.

10.30-10.40	Introductions and welcome
10.40-11.00	Introduction to Juvenile onset Batten disease (CLN3 disease) <i>Ruth Williams</i>
11.00-11.45	Educational issues – Overview then Q&A session <i>Barbara Cole</i>
11.45-12.00	Break
12.00-12.50	What are seizures and how are they treated? – Overview, demonstration of emergency seizure management, Q&A session <i>Ruth Williams</i>
12.50-13.00	How the BDFA can help <i>BDFA Family Support Officer</i>
13.00-14.00	Lunch
14.00-15.00	Talking to the children and other family members <i>Melinda Edwards</i>
15.00-15.15	Break
15.15-16.00	Preparation for the future – maintaining skills and well-being <i>Sarah Kenrick and team</i>
16.00	Thanks and close of meeting

Patient Organisations Programme

Families have a dedicated room in the Windsor Conference Centre throughout the conference in **Room 0-05**.

Friday 30th March

10.00	Welcome (Room 1-04) <i>Pauline Docherty and Heather Band</i>
10.10-10.30	Update on first official meeting of BDIA (Room 1-04) <i>Tony Heffernan</i>
10.30	Break for coffee
10.45-12.00	Discussion groups (Room 1-02/1-03)
12.00-12.50	Poster Session 3
13.00 prompt	Conference Photograph
13.10-14.00	Lunch (Windsor Building)
14.00-17.00	Networking (Room 1-02/1-03) OR Session 5 New Clinical Perspectives (Auditorium)
17.00-18.00	Poster Session 4
18.00-18.30	Science Summary Sessions for Parents (Room 1-04)
18.45	Evening Meal together with Scientific Conference (Founder's Dining Room, Founder's Building)

Saturday 31st March

09.00-12.00	Session 6 Experimental Therapies (Auditorium) Chairs: Beverly Davidson (Iowa), Mark Sands (Wash U)
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14.10-14.40		Summary of Scientific Sessions by respective Chairs
14.40-14.45		Patient Organisation Group Presentation <i>Irena Newcombe and Lance Johnston</i>
14.45-15.00		Prizes and thanks. Announcement of next NCL congress.
15.00-15.15		Break, with refreshments available throughout the Market Place
15.15-16.30		'Market Place' (Rooms 1-02 to 1-05)
16.30-17.30		Poster Session 6
18.30		Conference 'Banquet' for Scientists and Families (Founder's Dining Room, Founder's Building)

Science Summary Sessions 1 & 2: Genetics and Biology; Disease Mechanisms

Chair Lucy Barry, Mariana Vieira, Kim Wager, Megan O'Hare, Andrew Wong, Thomas Kuhle, Lotta Parviainen, Matthew Micsenyi, Mervi Kuronen, Heather Band, Irena Newcombe

Science Summary Sessions 3, 4 & 5 Links to other diseases; Recent Research Findings; New Clinical perspectives:

Chair Richard Tuxworth, Sarah Pressey, Sybille Dihanich, Katherine R Smith, Katja Kanninen, Nicole Neverman, Charles Shyng, Nadia Mitchell, John Staropoli, Heather Adams, Heather Band, Irena Newcombe