

## 7<sup>th</sup> JNCL Young Investigator Symposium – Scientific Agenda

Date & time of virtual meeting: Sunday, 5<sup>th</sup> & Monday, 6<sup>th</sup> December, 2021, 15:00 – 20:00 (CET).

Virtual attendance via Zoom link:

<https://us02web.zoom.us/j/84050872882?pwd=NEh5UzdVQU RuUnpKZjZzWWtYUFp1Zz09>

Meeting-ID: 840 5087 2882

Code: 531068

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## Sunday, 5<sup>th</sup> December 2021 – Part 1

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### Introduction

Chair: **Herman van der Putten** (NCL Foundation)

15:00 – 15:05 **Welcome:** NCL Foundation (GER)

15:05 – 15:35 **Keynote: Angela Schulz** (University Hospital Eppendorf - UKE, GER)

*"CLN3 Clinical Aspects"*

15:35 – 16:05 **Keynote: Susan Cotman** (Massachusetts General Hospital, USA)

*"CLN3 biology"*

16:05 – 16:35 **Keynote: An Ngoc Dang Do** (Eunice Kennedy Shriver National Institute of Child Health and Human Development, USA)

*"CLN3 Biomarkers"*

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16:35 – 16:50 **BIOBREAK**

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### Session 1: Cell Models

Chair: **Sukanya Arcot Kannabiran** (UKE)

16:50 – 17:05 **Daniela Wünkhaus** (Evotec SE, Translational Biology, Hamburg, GER)

*"TRPML1 activation ameliorates lysosomal phenotypes in CLN3 deficient retinal pigment epithelial cells"*

17:05 – 17:20 **Jimin Han** (University of Rochester, Cell Biology of Disease, US)

*"Investigating the retinal pigment epithelium dysfunction as it relates to vision loss in CLN3-Batten"*

17:20 – 17:35 **Sueanne Chear** (University of Tasmania, Wicking Dementia Research and Education CentreHobart, AUS)

*"Using isogenic stem cells to model disease mechanisms of Juvenile Batten disease"*

17:35 – 17:50 **Sukanya Arcot Kannabiran** (UKE, Institute of Biochemistry and Molecular Cell Biology, Hamburg, GER)

*"Elucidating the role of TPCs and TRPMLs in Lysosomal Storage Disorders"*

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17:50 – 18:00 **BIOBREAK**

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Chair: **Elisabeth Butz** (Ludwig-Maximilians-Universität)

18:00 – 18:15 **Llinos Honeybun** (Cardiff University, BioScience, Cardiff, UK)

*"Investigating new medicines to treat CLN3 disease"*

18:15 – 18:30 **Emily Relton** (University of Surrey, Guildford, UK)

*"Translational defects and perturbed stress granule dynamics are associated with loss of CLN3, a batten disease gene"*

18:30 – 18:45 **Masood Ahmad Wani** (University Medical Center, Johannes Gutenberg University of Mainz, Institute of Pathophysiologie, Mainz, GER)

*"Deficits in the intrinsic membrane properties of Cln3 $\Delta^{ex7/8}$  neurons"*

18:45 – 19:00 **Tobias Raisch** (Max Planck Institute of Molecular Physiology, Department of Structural Biochemistry, Dortmund, GER)

*"Sample optimization for structural characterization of CLN3"*

19:00 – 19:15 **Elisabeth Butz** (Ludwig-Maximilians-Universität, Anatomische Anstalt – Lehrstuhl 1, München, GER)

*"Early pathological signs of microglial vulnerability in CLN3 disease"*

19:15 – 19:30 Short Introductions: **Aswini Krishnan, Uche Medoh** and **Kwamina Nyame** (Stanford University, Stanford, CA, USA)

19:30 – 20:00 **General Discussion**

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## Monday, 6<sup>th</sup> December 2021 – Part 2

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### Introduction

Chair: **Frank Stehr** (NCL Foundation)

15:00 – 15:30 **Keynote: Alessandro Ori** (Leibniz Institute on Aging - Fritz Lipmann Institute, Jena, GER)

*"The Aging Brain Proteome"*

15:30 – 16:00 **Keynote: Craig Benson** (Beyond Batten Disease Foundation, USA)

*"Patient Advocacy: Leading Rare Disease Therapeutic Development"*

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16:00 – 16:15 **BIOBREAK**

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## Session 2: Clinical Aspects

Chair: **Hannah Leppert** (Sanford Research).

16:15 – 16:30 **Tufikameni Brima** (University of Rochester School of Medicine and Dentistry, Department of Neuroscience, New York, USA)

*" Event-related potential study assessing the integrity of auditory sensory memory in CLN3 "*

16:45 – 17:00 **Vicki Swier** (Sanford Research, Pediatrics and Rare Diseases, Sioux Falls, USA)

*" Longitudinal characterization of the CLN3 $\Delta$ 7/8 miniswine model shows robust cellular pathology and behavioral deficits "*

16:30 – 16:45 **Hannah Leppert** (Sanford Research, Pediatrics and Rare Diseases, Sioux Falls, USA)

*" Serum biomarker discovery in the Cln3 $\Delta$ ex7/8 minipig "*

## Session 3: Mutant Transcripts

Chair: **Jessica Centa** (Rosalind Franklin University of Medicine and Science)

17:00 – 17:15 **Haoyu Zhang** (University College London, GOS Institute of Child Health, London, UK)

*" Investigating Functionality of Mutant Transcripts of CLN3 "*

17:15 – 17:30 **Christopher Minnis** (University College London, Genetics and Genomic Medicine at the Institute of Child Health (GOSH), London, UK)

*" Are pathogenic transcripts cell-type specific in Juvenile CLN3 disease? "*

17:30 – 17:45 **Matthew Stratton** (Rosalind Franklin University of Medicine and Science, Cell Biology & Anatomy, North Chicago, USA)

*" Spatiotemporal analysis of CLN3 expression in a large animal model of Batten disease "*

17:45 – 18:00 **Jessica Centa** (Rosalind Franklin University of Medicine and Science, Center for Genetic Diseases, North Chicago, USA)

*" Splice-switching antisense oligonucleotides for the treatment of CLN3 Batten disease "*

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18:00 – 18:15 **BIOBREAK**

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## Session 4: Other NCL-Forms, Overlaps with other NDs

Chair: **Seda Yasa** (Harvard Medical School).

18:15 – 18:30 **Rachel Kline** (University of Edinburgh, Functional Genes and Development, Edinburgh, UK)

*" Identifying molecular overlaps between Motor Neuron Diseases and the NCLs. "*

18:30 – 18:45 **Marcel Klein** (UKE, Centre for Molecular- and Neurobiology, Hamburg, GER)

*" CLN3 and Pen2 associate in the autophagy-lysosome system "*

18:45 – 19:00 **Marina Garcia-Macia** (BSAL/USAL, Institute of Functional Biology and Genomics, Salamanca, ES)

*" Aberrant upregulation of glycolysis mediates CLN7 neuronal ceroid lipofuscinosis "*

19:00 – 19:15 **Mitchell Rechtzigel** (BSAL/USAL, Sanford Research, Pediatrics and Rare Diseases, Sioux Falls, USA)

*" Functional and pathway analyses of transmembrane NCLs reveal novel neuron-specific roles of CLN3, CLN6, and CLN8"*

19:15 – 19:30 **Seda Yasa** (Harvard Medical School, Department of Neurology, Boston, USA)

*" CLN3 and CLN5 are regulators of RABA7A function"*

19:30 – 20:00 **General Discussion**

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