

**18<sup>th</sup> National NCL Congress**  
"Retinal degeneration & Role of adaptive and  
innate immune cells in CLN3 disease"

Date of congress: Sunday, 18<sup>th</sup> April 2021 & Monday, 19<sup>th</sup> April 2021

Congress takes place via Webex.

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*We thank our sponsor and partner for the generous support:*



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**Sunday, 18<sup>th</sup> April 2021 (17:00 – 21:00 CET)**

## **Retinal degeneration in CLN3 Batten disease**

17:00 – 17:05      Welcome: **NCL Foundation** (GER)

### **Clinical aspects of retinal degeneration in NCL**

**Chair: Angela Schulz** (University Medical Center Hamburg-Eppendorf, Hamburg, GER)

17:05 – 17:30      **Szilárd Kiss - Progressive retinal degeneration in CLN2 disease**  
(Weill Cornell Medicine, New York-Presbyterian Hospital, USA)

17:30 – 17:55      **Simon Dulz - An ophthalmic rating scale to assess ocular involvement in juvenile CLN3 disease**  
(University Medical Center Hamburg-Eppendorf, Hamburg, GER)

17:55 – 18:20      **Michalis Georgiou - Juvenile Batten Disease (CLN3): Detailed ocular phenotype, novel observations, delayed diagnosis, and masquerades**  
(Moorfields Eye Hospital, London, UK)

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18:20 – 18:25      BIOBREAK

### **Gene Therapy**

**Chair: Beverly Davidson** (The Children's Hospital of Philadelphia, Philadelphia, USA)

18:25 – 18:50      **Dominik Fischer - Gene therapy in the eye: lessons from a first-in-man application**  
(Centre for Ophthalmology, University of Tübingen, GER)

18:50 – 19:15      **Sophia kleine Holthaus - Ocular Gene Therapy for CLN3 disease, a proof-of-concept study**  
(UCL Institute of Ophthalmology, London, UK)

19:15 – 19:40      **Shaomei Wang - Intracranial delivery of AAV-Cln6 partially prevents photoreceptor degeneration and visual deficits in Batten disease mice**  
(Cedars-Sinai Medical Center, Los Angeles, USA)

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19:40 – 19:45      BIOBREAK

### **CLN3 disease retinal models**

**Chair: Jill Weimer** (Sanford Research, Sioux Falls, USA)

19:45 – 20:10      **Qingjun Wang - Effects of CLN3 deficiency on RPE autophagy-lysosomal system and metabolism**  
(University of Kentucky, Lexington, USA)

20:10 – 20:35      **Ruchira Singh - Retinal pigment epithelium dysfunction in CLN3-Batten disease**  
(University of Rochester Medical Center, Rochester, USA)

20:35 – 21:00      **Jill Weimer - Modeling CLN3-Batten Disease in a Pig**  
(Sanford Research, Sioux Falls, USA)

**Monday, 19<sup>th</sup> April 2021 (17:00 – 21:15 CET)**

**Role of adaptive and innate immune cells in Batten disease**

17:00 – 17:10      Welcome: **NCL Foundation** (GER)

**Role and translational aspects of adaptive immune cells**

**Chair: Robert Steinfeld** (University Children`s Hospital, Zuerich, CH)

17:10 – 17:30      **Rudolf Martini - Neuroinflammation as a disease amplifier in NCL: chances for treatment?**  
(University Clinic Würzburg, GER)

17:30 – 17:50      **Kevin Rostásy - Two case reports of children with NCL treated with fingolimod: an off-label approach repurposing an established drug for treatment of MS**  
(Children`s Hospital Datteln, GER)

17:50 – 18:10      **Janos Groh - Neuroinflammation in disease and aging: lessons from rare genetic disorders**  
(University Clinic Würzburg, GER)

18:10 – 18:20      **Robert Steinfeld - Laudatio**  
(University Children`s Hospital, Zuerich, CH)

18:20 – 18:30      **Graeme Bilbe - New SAB member: experience & achievements**  
(DNDI, Neuchâtel, CH)

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18:30 – 18:35      BIOBREAK

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**Role of innate immune cells/microglia**

**Chair: Sabina Tahirovic** (German Center for Neurodegenerative Diseases, Munich, GER)

18:35 – 18:55      **Susan Cotman - Investigation of microglia in CLN3 disease using mouse and human iPSC-derived models**  
(Massachusetts General Hospital, Boston, USA)

18:55 – 19:15      **Sabina Tahirovic - Microglial pathology in NPC and CLN3 disease**  
(German Center for Neurodegenerative Diseases, Munich, GER)

19:15 – 19:35      **Jeremy Burns - Differential accumulation of storage bodies with aging identifies discrete subsets of microglia in the healthy brain**  
(Biogen, Boston, USA)

19:35 – 19:55      **Anja Capell - Cellular mechanism of progranulin deficiency in NCL and FTD**  
(Ludwig-Maximillian`s University, Munich, GER)

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19:55 – 20:00      BIOBREAK

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## **Neurons and Astrocytes**

**Chair: Dirk Isbrandt** (German Center Neurodegenerative Diseases, University Clinics, Cologne, GER)

20:00 – 20:20      **Markus Damme - TMEM106B – a major risk gene for neurodegenerative diseases and aging**  
(University of Kiel, GER)

20:20 – 20:40      **Rebecca Ahrens-Nicklas - Contribution of specific cell types to network pathology in CLN3 disease**  
(The Children's Hospital of Philadelphia, Philadelphia, USA)

20:40 – 20:50      **Final Discussion - Outlook**