



UNIVERSITY OF ROCHESTER BATTEN CENTER

Summer 2017 Newsletter

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Dr. Mink Travels to Hamburg, Germany

During the last week of March, 2017, Dr. Jonathan Mink, Director of the University of Rochester Batten Center (URBC) traveled to Hamburg, Germany to work with the neuronal ceroid lipofuscinosis (NCL) research group at the University of Hamburg. He trained Drs. Anegla Schulz, Miriam Nickel, Eva Wibbeler, and Christoph Schwering on the use of the Unified Batten Disease Rating Scale (UBDRS) in children and young adults with CLN3 disease (Juvenile NCL). They saw several patients together and performed parallel evaluations using the UBDRS to establish reliability across raters. Reliability across raters means that the raters agree about the information collected and scored. This is an important step to ensure that the data collected by different raters can be combined and compared from different sites or groups.

One of the highlights of the visit for Dr. Mink was the opportunity to see how children with Batten Disease are evaluated and treated at the University of Hamburg. Our groups share a common philosophy of care and of the importance of natural history research. This visit sets the stage for future collaborations that will enhance opportunities for clinical trials across international borders. It was also an important step toward finalizing an agreement to collaborate on the DEM-CHILD registry, which was developed by Dr. Schulz.



UBDRS Translated to Norwegian

The Unified Batten Disease Rating Scale (UBDRS) has been translated into Norwegian by Dr. Ingrid Helland, a pediatrician in Oslo, Norway. Dr. Helland works in the Department of Clinical Neurosciences for Children at the Oslo University Hospital where she cares for children with Batten Disease.

This is an important step toward adopting a unified approach to comprehensive clinical evaluation of children with all forms of Batten disease. The use of the UBDRS allows for quantitative assessment of symptom severity and impact for Batten disease. For clinical research in rare diseases, international collaboration is important to maximize potential for advances in understanding and treatment development.

Technology & Rare Neurological Disease Symposium (TRNDS)

The University of Rochester hosted a one-day symposium focused on new ways to use technology to accelerate the development of new therapeutics for patients with rare neurological disorders, like Batten disease. The TRNDS Symposium was organized by the Center for Human Experimental Therapeutics at the University of Rochester Medical Center. The conference featured regional and national leaders in rare diseases, health technologies, and clinical trials.

Dr. Erika Augustine, URBC member and Associate Director of the Center for Human Experimental Therapeutics, was Co-Chair of the event. She moderated a panel discussion on *Social Media Tools for Efficient, Effective Recruitment*.

URBC Visits: Child Companion

Volunteer, Wendy Scull recently joined our team as a child companion for children who visit the URBC with their families. She spends time with children if parents are speaking privately with the medical team. Wendy is a retired Teacher of the Visually Impaired and Orientation and Mobility Specialist. She has worked with visually impaired children ranging in age from infancy to age 21 for over 30 years. Wendy also provided services for children with Batten disease during her teaching career.



New Research Opportunity: Batten Disease EEG Study

The URBC is excited to announce a new collaboration with the Cognitive Neurophysiology Lab, a research team led by Dr. John Foxe at the University of Rochester Medical Center. The Cognitive Neurophysiology Lab conducts studies to measure the brain's electrical response to different information (such as sounds). The goal of these studies is to identify biomarkers. Biomarkers are objective, measurable characteristics of biological processes. Biomarkers are important for many reasons. For example, in clinical trials, they can help researchers test the effectiveness of a new treatment. Dr. Foxe's lab uses EEG (see next column) to evaluate whether there are biomarkers for auditory recognition in individuals affected by Batten disease, and if so, how these biomarkers are related to genotype (genetic makeup or "blueprint") and disease progression.



An electroencephalogram (EEG) is a test that measures electrical brain activity. Individuals with Batten disease are invited to participate in this study. In this study, researchers will record the electrical activity of your child's brain through an EEG. To do this, your child will wear a special cap. The cap has small, flat, metal discs called electrodes that will rest against your child's head. The electrodes are connected to wires that measure the small electrical charges caused by brain activity. During the EEG, your child will passively listen to a series of different tones or natural sounds. He/she can listen to or watch movies or do other activities while the sounds are playing. The study itself will take about 90 minutes. After completing the study, you and your child will receive \$14 per hour of your time.



If you are interested in participating in this study, please contact the Cognitive Neurophysiology Lab at (585) 275-1674 to learn more or to schedule an appointment. This research will also take place at the BDSRA meeting.

Natural History Research

We invite families with children who have any form of Batten disease to participate in our ongoing natural history study. We developed the Unified Batten Disease Rating Scale (UBDRS) as a disease-specific evaluation of individuals with JNCL. The UBDRS has two main parts: a physical exam and an interview about medical history and Batten disease symptoms (such as seizures). The UBDRS allows us to track disease progression over time. This has enabled us to describe the natural history of JNCL, including symptoms that lead to disability, and factors that may be related to fewer symptoms or lesser disability. *We are now expanding our research to include all forms of Batten disease.*

The URBC is collaborating with researchers at Massachusetts General Hospital (MGH) to combine the natural history research expertise at URBC with the molecular diagnostic and biorepository expertise at MGH. This will create the opportunity to advance knowledge about the natural history and disease biology of Batten disease.

Study visits take place in Rochester, NY at the URBC and/or at annual BDSRA meetings. Some study activities can also take place remotely (telemedicine). For more information about participating, contact Amy Vierhile at (585) 275-4762 or email us at: batten@urmc.rochester.edu



LINCL: Study of Adaptive Behavior and Family Impact

The URBC is conducting a study focused on learning more about everyday behavior and adaptive skills of children with **Late Infantile Batten disease** and how these behaviors and skills change over time. We also want to learn more about the impact of Batten disease on the family. You may be eligible to be in this study if you are a parent of a child with either a clinical or genetic diagnosis of LINCL.

If you take part in this study, we will ask you to fill out three forms, once every 6 months. The forms will take 45-60 minutes to complete. You can choose to fill these forms out at home, or we can set up an interview with you in person, by phone, or via videoconference.

If you are interested in learning more about the study, please contact Dr. Heather Adams at her private office number: (585) 275-9330.

URBC Contact Registry

Do you want to be informed about future Batten disease research? The URBC Contact Registry asks your permission to keep your contact information on file, so that we can send you updates about the URBC and related research. Individuals with **all forms of Batten disease** and their parents/legal guardians are eligible to participate. Sign up to be in our contact registry by visiting our website: www.rochesterbatten.urmc.edu

Planning Future Clinical Trails

Planning is underway for future clinical trials for CLN3 and CLN1. Representatives from Abeona Therapeutics met with members of the URBC in May 2017 to continue to plan future gene therapy trials.



Participate in a Research Study!

If you are interested in participating in any of our research studies, contact us:

Phone: (585) 275-4762

E-mail: batten@urmc.rochester.edu

Web: www.rochesterbatten.urmc.edu

In-person research visits take place at the URBC or at the annual Batten Disease Support & Research Association (BDSRA) meeting, which will be held at the Wyndham Grand Pittsburgh Downtown on July 6th-9th. We look forward to seeing there!





Student Spotlight

Toby Kashket: Toby is an undergraduate student at the University of Rochester working with Dr. Augustine and Dr. Adams to evaluate quality of life (QoL) and adaptive function in Batten disease. She is reviewing datasets to learn if there are key questions related to quality of life and adaptive function that will help us focus our assessments of children with Batten disease.

Nicola Ross: Nicola is a medical student pursuing Child Neurology. She is working with Dr. Augustine to write a review of the current literature about Infantile Batten Disease. By clarifying what is known about this form of Batten disease, she hopes to identify gaps in knowledge to help guide future research.

Justin Williams: Justin is a 2nd year medical student who has been working with Dr. Mink to develop a staging system for classifying the severity of CLN3 disease (JNCL). This system will provide the opportunity to identify stages of disease progression that can be used to group individuals for enrollment in clinical trials and to track disease progression.

International Batten Disease Conference



A number of URBC members participated at the 15th International conference on Neuronal Ceroid Lipofuscinosis (Batten disease). Researchers and clinicians who study Batten disease convened to discuss advances, challenges, and future directions in Batten disease research. Several members of our team at the URBC attended.

- Dr. Augustine presented results of the recently completed URBC clinical trial for juvenile Batten disease: “Cellcept for Treatment of JNCL.” Details of the trial are posted at www.clinicaltrials.gov (search for ID# NCT01399047). A manuscript is in preparation. We wish to thank the individuals and families who participated in this trial, and the BDSRA for its support of this research.
- Dr. Augustine also co-chaired an interactive session with Margie Frazier, PhD (BDSRA Executive Director): Engaging families in research workshop – Dialogue with parents and researchers.
- Dr. Mink presented a talk, “Rating scales for natural history studies and registries in the NCLs. This session was co-chaired by Dr. Augustine and our colleague in Hamburg, Germany Dr. Angela Schulz.
- Dr. Adams chaired a “Hot Topic in Clinical Research” session: *Neurobehavioral and Psychosocial Function in the NCLs*. This session included three excellent presentations by Bengt Elmerskog: *Experiences on education and adaptations for learnings with juvenile neuronal ceroid lipofuscinosis*; Anne-Grethe Tøssebro: *A study on tactile/body signs or hand signs for students with Batten disease (JNCL)*; and by Andrea West: *The burden of CLN2 disease on families: home-based surveys with caregivers in Germany and the United Kingdom*.

We thank all of the children and families who participate in our research – you make it possible for us to do this work!

We love to hear from you! Please contact us with your questions and comments. With your permission, we may post answers to your question on our Facebook page and in upcoming issues of our newsletter.

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