



UNIVERSITY OF ROCHESTER BATTEN CENTER

Summer 2018 Newsletter

University of Rochester Medical Center

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Batten Days Research Weekend

The University of Rochester Batten Center (URBC) held its first annual Batten Days research weekend on February 15th-17th in Rochester, NY. Families with children who have CLN1 and CLN3 disease travelled from across the country to take part. During the weekend, they participated in research and attended informational presentations on topics related to Batten disease. Many families had a chance to explore Rochester and visit the Strong Museum of Play!

One goal of this event was to continue to gather information about the symptoms and progression of CLN1 and CLN3 disease. Families participated in a range of research activities, including the Batten Natural History study, which involved an interview about medical history and symptoms and a brief physical exam using the Unified Batten Disease Rating Scale (UBDRS). Some individuals participated in neuropsychological testing to evaluate cognitive abilities, like attention and memory as well as behavioral function. Others participated in an electroencephalography (EEG) study, where researchers used a special cap with sensors to measure the electrical activity of the brain in an effort to explore whether there are biomarkers for auditory perception in individuals with Batten disease. Finally, some individuals participated in an eye exam, where investigators took pictures of the eye using special cameras to better understand vision loss experienced by those with CLN1 and CLN3.

Another aim of this event was to train new medical providers on the use of the UBDRS, which is a disease-specific assessment tool designed to measure the symptoms and progression of Batten disease. The UBDRS was developed at the URBC, and it continues to be an important way to consistently evaluate symptom severity and rate of change in both clinical and research settings. Medical providers and Batten disease researchers attended this meeting to learn about how to use the UBDRS to evaluate individuals with CLN1 and CLN3, so that they will be able to use this tool in the future.

New Medical Records Research Study

We are carrying out a new study at the URBC. The goal of this study is to understand more about the symptoms of NCL disorders and how those symptoms change over time. If you have a child or children, living or deceased with any NCL disorder, then you can contribute to this study. For this study, our research team will request healthcare and school records from your child's providers. We will review these records to better understand how Batten disease progresses. Although this study won't benefit your child directly, the information is vital to learn about these diseases. This study will help researchers understand the best ways to test new treatments in the future. If you are interested in participating or if you have questions, please call (585) 275-4762.

Visit from Hamburg Batten Experts

Drs. Angela Schulz and Miriam Nickel from the University of Hamburg Eppendorf made a visit to the URBC in May 2018. Dr. Schulz leads the DEM-CHILD international NCL registry. The groups met to discuss ways to improve our understanding of natural history of the NCLs and to foster future research collaboration.





Magnetic Resonance Imaging (MRI) Research

The Cognitive Neurophysiology Lab at the University of Rochester Medical Center, led by Dr. John Foxe has extended the study of Batten Disease to include Magnetic Resonance Imaging (MRI). MRI is a state of the art tool that lets you see inside any organ of the body including the brain. It is a safe and painless test that uses magnetic fields, radio frequency pulses and a computer to produce detailed pictures. The aim of this addition is to link EEG data (that informs about the electrical activity in the brain) with structural changes that occur over time in the brain of people with Batten disease. This combination will yield a powerful marker of disease progression. Disease markers such as these are important to track the natural course of Batten disease and to help researchers test the effectiveness of new treatments.

Individuals with Batten disease who have already done the EEG study are invited to get their MRI pictures taken. For those who have not yet done either the EEG or MRI portions of this study you are also invited to be a part of this study.

EEG portion of the study: your child will be fitted with a special cap. This cap has small channels where sensors, called electrodes, will be positioned. Electrodes look like small, flat metal discs connected to wires. When placed on the scalp, electrodes can detect small electrical charges resulting from brain activity. Once the cap is fitted, your child will passively listen to a series of different tones or natural sounds.

MRI portion of the study: You will be asked questions to make sure your child doesn't have any internal metal clips from previous surgery or anything else that might cause a problem near a strong magnetic field. To obtain the highest quality MRI results, your child will need to be completely still during the scan. For this reason, we will have mock-scan session before the real MRI scan to make your child familiar with the scanner environment, and will help your child to stay still during the scan. The mock session takes about 10 minutes and the MRI session takes 30 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.



Natural History Research

We invite families with children who have all forms of Batten disease to participate in our ongoing natural history study. We developed the Unified Batten Disease Rating Scale (UBDRS) to provide a consistent and disease-specific approach to evaluating individuals with JNCL. The UBDRS has two main parts: a physical exam and an interview about medical history and symptoms of Batten Disease (such as seizures). Using the UBDRS, we track disease progression over time. This has enabled us to describe the natural history of JNCL, including the symptoms that lead to disability, and potential factors that are 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. For more information about participating, contact Amy Vierhile at (585) 275-4762 or email us at: batten@urmc.rochester.edu



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**.



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 in Nashville, TN on July 19th-22nd. We look forward to seeing there!



Welcome New URBC Team Members!

Tufikameni Brima, PhD is a research assistant professor at the University of Rochester in the department of Neuroscience. She is based in the Cognitive Neurophysiology Laboratory of Dr. Foxe where she uses high-density electrophysiology (EEG) and Magnetic Resonance Imaging (MRI) to investigate neural correlates in rare genetic neurodevelopmental diseases including Batten disease. The aim of her current study is to identify disease markers specific to Batten disease using both EEG and MRI. These biomarkers are important to track disease progression as well as the effect of current and future therapeutic intervention.

Shannon Dean, MD, PhD has recently joined the pediatric neurology division. She is completing a fellowship in pediatric movement disorders and experimental therapeutics.

Nasir Maidanwal, MD, MPH is a research coordinator at the University of Rochester in the Division of Child Neurology. He holds an MD degree from Kabul Medical University, Afghanistan. He earned his first master's degree in public health from the University of Mahidol in Thailand and second master's degree in One Health/biosecurity from Massey University, New Zealand. He has worked with various international organizations, such as W.H.O., MSH, and USAID.



Visiting the University of Rochester Batten Center (URBC)

Families visit the URBC for clinical appointments, to participate in research, or both! When a child visits the URBC, many different activities can take place. Each family is able to choose which activities they wish to take part in. Depending on the number of activities, the visit may be spread out over one or two days. A visit to the URBC often begins with a call to Amy Vierhile, DNP, our Clinical Coordinator. Amy works with parents / caregivers to schedule a date to visit the URBC and to help you make travel arrangements.

Review of Medical Background: Amy Vierhile, DNP, or another coordinator will meet with you to review your child's medical history, path to diagnosis, and other background information.

Unified Batten Disease Rating Scale (UBDRS): One of the neurologists on the team will complete the UBDRS. The UBDRS includes a brief physical exam where we test your child's strength, balance, and coordination, and ask you some questions about your child's behavior, everyday skills, and his or her symptoms of Batten disease.

Neuropsychological Evaluation: If your child is able to complete a brief evaluation of their cognitive skills, they can do so with Dr. Adams, the pediatric neuropsychologist for the URBC. Dr. Adams will spend about 30 minutes with your child to complete tasks of attention, memory, and language.

Questionnaires: We recognize that parents are the true experts on their child! Therefore, we ask a lot of questions to learn as much as possible about the broad impacts of Batten disease on your child and family. We may ask you to complete questionnaires about your child's behavior, everyday skills, sleep patterns, and other aspects of function. We can arrange to complete these with you by phone or mail either before or after your visit to the URBC.

EEG (research activity only): An electroencephalogram (EEG) is a test designed to evaluate the electrical activity in the brain. In this study, researchers measure the electrical activity of your child's brain using an EEG while wearing a special cap with sensors. This 75-90 minute study aims to evaluate whether there are biomarkers for auditory cognition in individuals with Batten disease.

MRI (research activity only): Magnetic Resonance Imaging (MRI) is a state of the art tool that lets you see inside any organ of the body (e.g., the brain) with great clarity. The aim of this study is to link EEG data with structural changes that occur over time in the brain of people with Batten disease. If you choose to have your child participate, your child will get to practice lying still in a pretend MRI scanner, so that he or she can become familiar with the scanner environment. If your child is able to tolerate this practice session, your child will have an MRI, which typically takes about 30 minutes.

Vision Exam: You might be asked to have your child participate in an eye exam by pediatric retinal specialist, Dr. Mina Chung. During this exam, your child will have tests to check how well your child sees, and pictures of your child's eyes will be taken using a special camera. Your child may be shown flashing lights and may be asked to push a button when they see lights. In order to do these tests, eye drops will be given to your child to dilate their eyes.

For more information, visit our website (www.rochesterbatten.urmc.edu) or contact us at by phone (585-275-4762) or email (batten@urmc.rochester.edu).

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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