



UNIVERSITY OF ROCHESTER BATTEN CENTER

Summer 2016 Newsletter

University of Rochester Medical Center

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Ask An Expert Facebook Chat

On Monday, May 23rd 2016, Dr. Heather Adams was joined by Dr. Daniel Mruzek, PhD, a child Psychologist and behavior analyst at the University of Rochester, for a recent Batten Disease Support and Research Association (BDSRA) Ask an Expert Facebook Chat, "Helping Your Child/Teen with Batten disease: Functional Behavioral Assessments". They shared information about how functional behavioral assessment (FBA) can be a tool for understanding triggers and perpetuating factors for challenging behaviors. The FBA can be the first step in developing a management plan to help prevent some challenging behaviors from developing, and to help manage them effectively when they do occur. Please see the BDSRA official group Facebook page for the full details from this one-hour session. Also, the website, the "Center for Effective Collaboration and Practice" has information about FBAs: www.cecp.air.org/fba/



Batten Clinical Corner: Seizures 101

A seizure is a type of abnormal electrical brain activity that affects how a person moves or interacts for a brief period of time. Generalized tonic-clonic seizures come to mind for most people when they think of a seizure, but there are many types of seizures.

There are two main categories of seizure: generalized and focal. Generalized seizures begin in both sides of the brain at once. Examples of generalized seizures include: tonic-clonic (jerking movements on both sides of the body with loss of consciousness, formerly known as grand mal), absence (staring spells, formerly known as petit mal), myoclonic (brief jerks), tonic (stiffening spells), and others. Focal seizures start in one specific brain region. Determining the type of seizure can be important for treatment selection.

The first treatment choices are usually based on the seizure type, EEG patterns, interactions with other medications, and side effects. It can take a while to find the right treatment plan for your child, with your child's clinicians. There are many seizure medications and other types of treatments, including specialized diets. More than one treatment may be required. We aim for seizure freedom, with one medicine, and minimal to no side effects. Often, we are not able to completely stop the seizures. Even with treatment, sometimes seizures happen during illnesses, during medication changes or missed doses, during a change in sleep habits, or sometimes for no obvious reason at all. It is important to work closely with your care team to set goals for the balance between seizure control and medication side effects like sedation. It is also important for family friends, and school teachers and staff to know what to do during a seizure. Your provider can help to create an emergency plan.

www.epilepsy.com is a helpful resource for more information.

For more information on seizures in JNCL:

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4610252/>



Meaningful Outcome Measures for JNCL Focus Group Study

Dr. Heather Adams is currently leading a new study focused on developing meaningful and reliable “outcome measures” for future clinical trials in juvenile Batten disease (JNCL). At the present time, there are no known treatments that can change the course of JNCL. When such treatments become available in the future, we will need a way to reliably measure the treatment effect. Some questions that we will need to ask are, “Does the treatment make a difference in the disease course?” “How large of a difference does the treatment cause?” “Does the change in the disease course result in better function, improved quality of life, or other benefits to the person with Batten disease?” “How long will we have to give a treatment before we know that it is making a difference?” “Are there specific symptoms (but not others) that are changed as a result of the treatment?” Outcome measures are important in helping us answer these questions.

Parents of individuals with JNCL are experts in the daily impacts of the disease. Therefore, we are inviting parents to participate in a 90 minute focus group via teleconference, where we will be asking about what is meaningful to individuals with JNCL and their families when considering treatment-related changes. During the focus group, we will ask you and the other participants to discuss critical events and symptoms of JNCL and how they have impacted your child. Parents will be paid \$25 for participating in the focus group. If you are interested in learning more, please contact Alyssa Thatcher (Study Coordinator) or Dr. Heather Adams (Principal Investigator) at (585) 276-5966 or by email at Batten@urmc.rochester.edu

New Collaboration

The URBC is excited to announce a new collaboration with a research team led by Dr. John Foxe at the University of Rochester. Dr. Foxe and his research team conduct studies to identify biomarkers in childhood neurological and neuropsychiatric conditions. They also study how these biomarkers are related to genotype (genetic makeup or “blueprint”). Biomarkers are objective, measurable characteristics of biological processes. They are important in clinical trials because they help to determine if a treatment is effective. Dr. Foxe and his research team will begin collaborating with the URBC to test whether or not biomarkers for auditory recognition and comprehension exist in Batten disease. To do this, electroencephalography (EEG) will be used. An EEG is a test that looks at electrical activity in the brain using small electrodes embedded in a cap that rests on the child’s head. The child can sit and listen to a favorite movie or music while the EEG is being completed. Keep your eyes out for more information about this collaboration and future participation!

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 other 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



LINCL: Study of Adaptive Behavior and Family Impact

The URBC is conducting a new 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? Sign up to be in our contact registry by visiting our website: <http://www.urmc.rochester.edu/neurology/batten-disease-center/>

Participate in a Research Study!

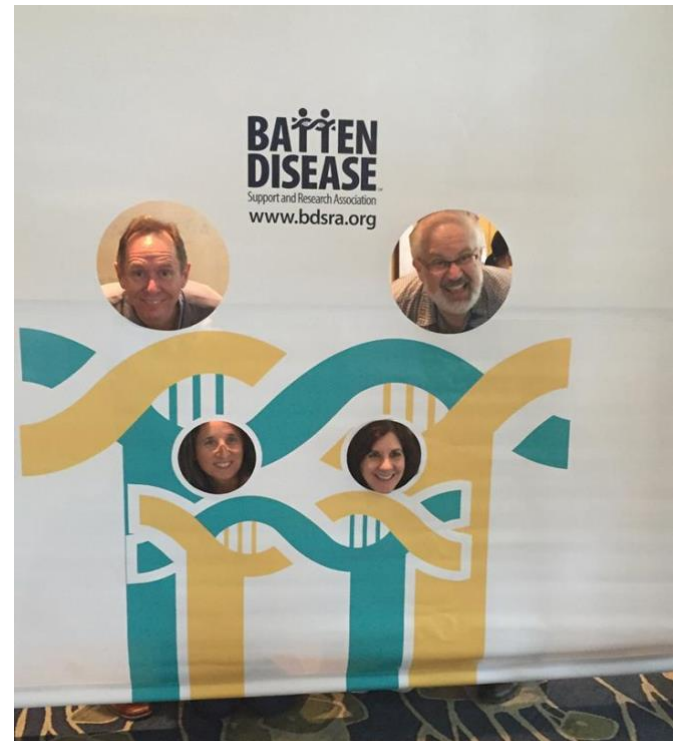
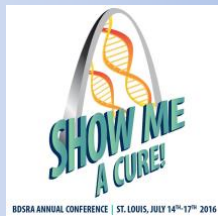
If you are interested in participating in any of our research studies, contact us:

Phone: (585) 275-4762

E-mail: batten@urmc.rochester.edu

Web: <http://www.urmc.rochester.edu/neurology/batten-disease-center/>

In-person research visits can take place at the URBC or at the annual Batten Disease Support & Research Association (BDSRA) meeting headquartered at the Renaissance St. Louis Airport Hotel in St. Louis, MO from July 14th-17th.



Function and Quality of Life in JNCL

The University of Rochester Medical Center is **currently recruiting parents** of individuals with **genetically confirmed (CLN3) JNCL** for a research study. The purpose of this study is to learn how to measure changes in function and quality of life of people with JNCL, using parent questionnaires. The study will last a minimum of 18 months and up to 24 months. It involves completing a series of four online surveys once every six months. You must be a parent of an individual with genetically confirmed JNCL, able to speak English, and have access to a computer, internet, and a current email address. You will be paid with a \$25 gift card for each complete set of questionnaires, up to \$125 for completion of 5 assessments over 24 months. There will be no cost to you to participate.

To learn more about this study, you can click on the following link: <https://redcap.urmc.rochester.edu/redcap/surveys/> and enter the code: **WEJTCH8P**. This link will allow you to enroll in the study if you choose to participate. If you have any questions, please contact Alyssa Thatcher (Study Coordinator) or Dr. Erika Augustine (Principal Investigator) at (585) 276-5966 or by email at Batten@urmc.rochester.edu.



URBC Student Spotlight

Each year, URBC students develop and carry out research studies related to Batten disease. Last summer, Travis Amengual, a UR Medicine student, developed and distributed an electronic survey to learn more about how people who are impacted by rare disease (including Batten disease) make decisions about participating in clinical research. In April, 2016, Travis presented results at the American Academy of Neurology (AAN) annual meeting. He explained that people affected by rare disease often live far from locations where the disease is studied, which makes participation in clinical research difficult. He found that distance, time, and level of convenience are important factors in making a decision about whether or not to participate in research. When recruiting for research studies, it is important that the group of participants is representative of the population being studied because we want to make sure that study results are relevant and transferrable to that population. Findings also underscored the potential for use of technology (e.g., videoconferencing and wearable sensors) to make participation in research easier. People who completed the survey reported a high level of access to and comfort with technology that could allow for tele-research in the future.

International Batten Disease Conference

The 15th International Conference on Neuronal Ceroid Lipofuscinosis (Batten Disease) will take place in Boston, MA on October 5-8, 2016. Researchers and clinicians who study Batten disease will convene to discuss advances, challenges, and future directions in Batten disease research. Several members of our team at the URBC look forward to attending. For more information: <http://nclboston2016.com/>



Travis Amengual at the AAN 2016 annual meeting

**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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 <https://www.facebook.com/URMCBattenCenter>