



UNIVERSITY OF ROCHESTER BATTEN CENTER (URBC)

Newsletter Winter 2016

University of Rochester Medical Center

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URBC Updates: Inside This Issue

- ❖ Members of the URBC attended an international meeting in Hamburg, Germany focused on the DEM-CHILD Batten disease registry.
- ❖ Members of the URBC have participated in “Ask an Expert” Facebook chats through the Batten Disease Support and Research Association (BDSRA).
- ❖ We are recruiting participants for the Batten Disease Natural History study and the Function and Quality of Life in juvenile Batten Disease (JNCL) study.
- ❖ The final participant completed the JUMP Phase II clinical trial.
- ❖ We received funding from the BDSRA to investigate endpoints for Phase III clinical trials in JNCL.
- ❖ URBC students have been busy carrying out research related to Batten disease.
- ❖ There have been big changes to Rochester’s Golisano Children’s Hospital, home of the URBC

Participation International Batten Meeting

Drs. Jonathan Mink, Erika Augustine, and Heather Adams attended the DEM-Child Registry Workshop meeting on November 20 – 22, 2015. The DEM-CHILD Consortium is an international group of clinical and research sites throughout Europe, and one site in India, that collaborate on sharing expertise and information on all forms of Batten disease. At its heart is the DEM-CHILD registry that has created a network of information on Batten disease genes, biomarkers, and clinical features. The DEM-CHILD Consortium members convened for their annual NCL Registry Workshop meeting in Hamburg, Germany. Members of the URBC were invited to attend the meeting and present our work on JNCL. We also participated in a satellite session to discuss building United States-based partnerships with DEM-CHILD. To learn more about DEM-Child, please visit the Consortium website at: <http://www.dem-child.eu/index.php/aims-and-objectives.html>

“Ask an Expert” BDSRA Facebook Chats

The BDSRA invited the URBC to participate in two “Ask an Expert” Facebook chats. Families were given the opportunity to ask experts from the URBC questions related to Batten disease. Becky Hetteberg, from the BDSRA moderated these chats.

In May, Neurologist Dr. Erika Augustine answered a range of questions about symptoms, like vision loss and seizures, and questions about symptom progression. She talked about the importance of developing an action plan when children have symptoms that can be serious and happen over and over (e.g., seizures). Action plans help parents, caregivers, and teachers know what to look for and what to do when a symptom happens. Dr. Augustine also suggested that families contact the BDSRA and visit www.clinicaltrials.gov for information about current clinical trials.

In October, Child Psychologist Dr. Heather Adams and Pediatric Nurse Practitioner Amy Vierhile led the “Managing Meltdowns and Tantrums” Facebook chat. They explained that challenging behaviors can be caused by dementia. Behavior issues often arise when children are asked to do something that is too difficult for them to do consistently or at all. With Batten disease, this is tricky because children may be both gaining some skills and losing others. It can be more helpful to interpret some behavior as resulting from a loss of a skill (e.g., lacking the skill to wait one’s turn), rather than misbehavior. Strategies like redirection can help children get “unstuck” when they over-focus, or persevere, on one thing.



Ask an Expert continues on next page



Function and Quality of Life in JNCL Research Study

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 Sara Defendorf (Study Coordinator) or Dr. Erika Augustine (Principal Investigator) at (585) 273-3810 or by email at Batten@urmc.rochester.edu.

Ask an Expert (continued)

They discussed aggressive behaviors that kids with Batten disease may have, and ways to manage such behavior using behavioral modification and medication. Because transitions can be very difficult, they also suggested planning very gradual transitions scripted out with the whole team. Importantly, they explained the benefit of having a behavioral specialist do a functional behavioral assessment (FBA). An FBA uses techniques to understand what is happening before, during, and after a certain behavior occurs. The behavioral specialist can help you develop strategies to change the behavior. A great resource for learning more about FBAs is available at the Center for Effective Collaboration and Practice: <http://cecp.air.org/fba>.

You can view the full Facebook chat on the BDSRA Facebook group. Please reach out to the BDSRA or contact the URBC with questions. We look forward to the next "Ask an Expert"



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



Back row (L-R): Jonathan Mink, Heather Adams; Middle row: Travis Amengual, Sara Defendorf, Erika Augustine, Frederick Marshall; Front row: Alyssa Thatcher, Amy Morris, Amy Vierhile

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



Endpoints for Clinical Trials

Dr. Heather Adams recently received funding from the BDSRA to begin a new study focused on identifying endpoints for a future Phase III clinical trial in Juvenile Batten disease (JNCL). A clinical trial endpoint is a measure that tells researchers whether or not a treatment or intervention being studied is beneficial. Endpoints should be sensitive and responsive to clinical trial interventions. For example, a clinical trial designed to investigate the effectiveness of a medication to treat hypertension might use lowered blood pressure as an endpoint, as this measure helps to determine whether or not the medication was effective.

Determining endpoints for clinical trials in JNCL is complicated, as there are a range of symptoms that vary across individuals with JNCL. At a meeting hosted by the URBC in December 2013, experts in Batten disease and parents from around the world agreed that there is an immediate need to develop valid endpoint measures for Phase III clinical trials in JNCL. Dr. Adams is now developing plans to collaborate with parents of affected children and with Batten experts to determine appropriate endpoints. Keep your eyes open for more information related to this study!

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/>

JUMP Update: Final Visit

All study visits have now been completed for the Juvenile NCL Mycophenolate Phase II trial (JUMP) and we anticipate reporting results in 2016. We sincerely appreciate the time that families devoted to the trial and to advancing Batten disease knowledge.

Participate in a Research Study!

We love to hear from you! Please contact us with your questions and comments.

Phone: (585) 275-4762

E-mail: batten@urmc.rochester.edu

Web:

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

 Facebook:

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

Recent Publications

Ragbeer SN, Augustine EF, Mink JW, Thatcher AR, Vierhile AE, Adams HR. Remote assessment of cognitive function in juvenile neuronal ceroid lipofuscinosis (Batten disease): A pilot study of feasibility and reliability. J Child Neurol. Sep 2, 2015. [epub ahead of print]

Summary: Telemedicine (video conferencing) was used to conduct neuropsychological assessments of children with JNCL. The investigators evaluated two different approaches to telemedicine (video conferencing) for remote neuropsychological assessment: having an expert clinician conduct testing remotely, or having the expert clinician supervise a 'local' assessment by someone else. Results of the study suggest that these approaches are feasible and that similar results are obtained by local vs. remote assessment.

Augustine EF, Adams HR, Beck CA, Vierhile A, Kwon J, Rothberg PG, Marshall F, Block R, Dolan J, Mink JW, on behalf of the Batten Study Group. Clinical Features of Epilepsy in Juvenile Neuronal Ceroid Lipofuscinosis (Batten Disease). Dev Med Child Neurol. 2015;57(4):336-371.

Summary: Seizures are reported as a common symptom across various forms of Batten disease, but little is known about the specific seizure experience of children with juvenile neuronal ceroid lipofuscinosis. In this study, we evaluated the seizure history of over 80 children with genetically confirmed CLN3 disease, using questions from the UBDRS. Over 85% of children in this study had experienced at least one seizure but seizures were typically infrequent and could be managed well for most children with one or two medications. There was no difference in seizure severity based on sex or genotype.



URBC Student Spotlight

Several URBC students developed and carried out research studies related to Batten disease. Here are updates on recent student projects.

Travis Amengual: Travis Amengual, a UR Medical Student, developed an electronic survey to learn more about how people impacted by rare disease (including Batten disease) make decisions about participating in clinical research. Almost 1000 people responded to his survey. He is currently analyzing data and hopes to present results at a future conference.

Elizabeth Cozart: UR Medical Student Elizabeth Cozart is currently analyzing data and preparing a manuscript based on her research at the 2014 BDSRA conference. She developed a survey to evaluate the marital well-being and personal support experiences of parents, and the services that children with Batten disease receive. We look forward to sharing the final results with the Batten community.

Shayne Ragbeer, PhD: Dr. Ragbeer participated in multiple BDSRA conferences while a UR Clinical Psychology student. In a pilot study, she and Dr. Adams found evidence that telemedicine (video conferencing) may be a feasible and reliable way to conduct neuropsychological testing with children who have JNCL. This method of testing may make participation in future URBC research easier for families who are unable to travel to the research site. A manuscript about this pilot study titled, *Remote Assessment of Cognitive Function in Juvenile Neuronal Ceroid Lipofuscinosis (Batten disease) – a Pilot Study of Feasibility and Reliability*, was published in September 2015 in the Journal of Child Neurology.

New Golisano Children's Hospital

The University of Rochester Medical Center started building a new children's hospital in September of 2012, and children and their families began moving into the new 245,000 square foot facility this summer. The hospital



includes 52 private rooms and 68 neonatal ICU beds to help children and their families feel more at home. It has a Play Deck, Teen Room, and Toddler Playroom to give kids a place to be kids. It also includes advanced imaging, including the only PET/MRI in a children's hospital.



Golisano Children's Hospital provides care to over 85,000 children and their families each year. Patients travel from the 17-county Finger Lakes region and beyond to meet with pediatric specialists from more than 40 specialty areas. We also welcome children and their families from around the country to the UR Batten Center.

The missions of Golisano Children's Hospital are "to provide comprehensive care for children and their families, educate future pediatricians and pediatric specialists, and perform progressive research that will lead to the medical breakthroughs of tomorrow."