

University of Rochester Batten Center

Newsletter

Winter 2013/2014

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What is the University of Rochester Batten Center?

The University of Rochester Batten Center (URBC) is a comprehensive Batten disease clinical and research center at the University of Rochester Medical Center in Rochester, New York. The URBC provides support and clinical services, contributes new knowledge, and works to find treatments that will slow, halt, or prevent disease in persons with Batten disease. The URBC is a Batten Center of Excellence of the Batten Disease Support and Research Association (BDSRA). Centers of Excellence are selected based on expertise and services offered related to Batten disease.

Clinical services include...

- Genetic diagnosis of Batten disease
- Clinical consultation for Batten disease
- School and behavioral support consultation
- Education about Batten disease

Research activities include...

- Continued refinement of the Unified Batten Disease Rating Scale (UBDRS), a disease-specific clinical rating scale
- Establishment of clinical outcome measures for JNCL
- Development of clinical trials for JNCL



From left to right: Heather Adams, PhD; Elisabeth de Blieck, MPA, CCRP; Sara Defendorf, BS, CCRP (back); Jonathan Mink, MD, PhD; Frederick Marshall, MD; Amy Vierhile, RN, PNP; Alyssa Thatcher, BS; Paul Rothberg, PhD; Erika Augustine, MD

Clinical Trial - Juvenile NCL Mycophenolate Phase II trial (JUMP)

In 2012, the URBC began a clinical trial for Juvenile Batten disease (JNCL). The trial examines whether mycophenolate mofetil, a drug FDA-approved to suppress the immune system in children with organ transplants, is safe for children with JNCL.

Dr. Erika Augustine has taken over the role of Principal Investigator. Dr. Fred Marshall will remain involved with the study as a Co-Investigator. The change in leadership was a natural transition; Dr. Augustine, a pediatric neurologist who specializes in rare diseases research, has been dedicated to this trial from the beginning.

Enrollment continues as we strive for our goal of 30 participants. Ten children have successfully enrolled and finished participation in the trial. We thank all of the local physicians who have worked with us on the trial.

To qualify for the study, children must have genetically confirmed JNCL and be able to walk at least 10 feet on their own or with a walker. During the study, each child will take study medication for 8 weeks and placebo for 8 weeks. Children will take a 4 week break in the middle of the trial to clear the body of medication. In this double-blind study, the researchers and families will not know when the child is taking placebo or active medication. Children will travel with a parent to Rochester four times over the 22 week study period. For more information, contact Amy Vierhile (585)-275-4762 or Sara Defendorf (585)-273-3810.



MEDICINE of THE HIGHEST ORDER



Workshop on JNCL Clinical Trials Outcomes

On December 6th, 2013, researchers and parents from around the world gathered at the University of Rochester Medical Center to discuss the future of research for Juvenile Batten disease.

There are a number of recent and ongoing clinical trials in Batten Diseases. Most of these are focused on asking if these potential new treatments are *safe*. After safety studies are completed, the next stage of clinical trials research is to learn if these or other interventions are *effective*. In order to answer this question, we will need ways to measure whether or not a treatment actually makes a difference for patients.

Therefore, the URBC hosted a workshop titled, “*Outcome Measures and Infrastructure for Phase III Studies in JNCL*”.

The workshop brought together experts in JNCL, experts from other fields (clinical trials in rare diseases, statistics, etc.), and Batten family representatives. Attendees included representatives from each Batten Center of Excellence. Each expert was asked to bring along a trainee so that we can continue cultivating the next generation of researchers who are focused on Batten Disease. They evaluated existing registries for patients with Juvenile Batten Disease and discussed steps needed to make Phase III trials possible.

The conference was made possible by a grant from the National Institutes of Health, and through the support of the BDSRA, Beyond Batten Foundation, and many collaborators.

We would love to see you at the 2014 BDSRA National Conference



This year's conference, planned by the staff of BDSRA and the Ohio Chapter of BDSRA, will be headquartered at the Columbus, Ohio airport Marriot from July 24th-July 27th. Online conference registration can be accessed at www.bdsra.org For questions, please call Tracy Kirby at 800-448-4570.

**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.

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