

# University of Rochester Batten Center

## Newsletter

### Spring 2013

#### 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 neuronal ceroid lipofuscinosis.

#### 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 development of the Unified Batten Disease Rating Scale (UBDRS), a disease-specific clinical rating scale.
- Collaboration with other investigators.
- Development of clinical trials for JNCL.

#### Clinical Trial - Juvenile NCL Mycophenolate Phase II Clinical 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.

The trial will enroll 30 individuals with JNCL. 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.

#### Natural History Research

We developed the Unified Batten Disease Rating Scale (UBDRS) to provide a consistent and disease-specific approach to evaluating children with JNCL. The UBDRS has several parts: a physical exam, review of the child's medical history, and questions about the various symptoms of Batten Disease (such as seizures). The UBDRS is used in two ways – as part of a clinical evaluation at the URBC, and as a research tool.

Because the UBDRS is always completed the same way, we are able to consistently 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 less disability. Since 2002, 120 children and young adults with Batten disease have already taken part in this study, and many return each year! Repeat evaluations are extremely valuable. They help us understand how the disease changes over time.

We invite families to participate in this study. Study visits take place in Rochester, NY at the URBC and/or at annual Batten Disease Support and Research Association meetings. For more information about participating in our natural history study, contact Amy Vierhile at (585) 275-4762 or email us at: [batten@urmc.rochester.edu](mailto:batten@urmc.rochester.edu)



1<sup>st</sup> row: Amy Vierhile, Nicole Newhouse, Heather Adams; 2<sup>nd</sup> row: Fred Marshall, Jon Mink, Paul Rothberg, Jennifer Kwon, Erika Augustine, Lisa de Bleeck



## Workshop on JNCL Clinical Trials Outcomes

There are a number of recent and ongoing clinical trials in Batten Disease. 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 a way to measure whether or not an intervention actually makes a meaningful difference in disease course.

Therefore, the URBC is currently planning to host a workshop titled, “*Outcome Measures and Infrastructure for Phase III Studies in JNCL*” (date t.b.d.). The workshop will bring together experts in the clinical features of JNCL, experts from other fields (clinical trials in rare diseases, statistics, etc.), and Batten family representatives. Each expert will be asked to bring along a trainee so that we can continue cultivating the next generation of researchers who are focused on Batten Disease. Our goals are to a) identify possible outcome measures for future clinical trials, b) develop a roadmap for further research to test and refine outcome measures, and c) strengthen research collaborations to support outcome measure development.



The University of Rochester Batten Center is a member of the Lysosomal Disease Network which is part of the National Institutes of Health (NIH) Rare Diseases Clinical Research Network (RDCRN), supported through collaboration between the NIH Office of Rare Diseases Research (ORDR) at the National Center for Advancing Translational Science (NCATS), the National Institute of Neurological Disorders and Stroke (NINDS) and National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK).

## Who Are We?

**Jonathan W. Mink, MD PhD** is medical director of the URBC. He is Professor of Neurology and Pediatrics and Chief of the Child Neurology Division at the University of Rochester Medical Center (URMC). He is Principal Investigator (PI) of the natural history study and Co-PI of the JUMP study.

**Frederick J. Marshall, MD** is Associate Professor of Neurology and Director of the Memory Care Program at URMC. He is PI of the JUMP study.

**Erika F. Augustine, MD** is Assistant Professor of Neurology and Pediatrics. She is Co-PI of the JUMP study.

**Jennifer M. Kwon, MD** is Associate Professor of Neurology and Pediatrics at URMC. She is the medical monitor for the JUMP study.

**Heather Adams, PhD** is Assistant Professor of Neurology and Pediatrics. She is a neuropsychologist and the PI of the Neurobehavioral Outcomes in Batten Disease study.

**Paul G. Rothberg, PhD** is Professor of Pathology and Laboratory Medicine and is Director of the Section of Molecular Diagnosis at URMC. He conducts genetic testing.

**Elisabeth de Blieck, MPA CCRC** is Program Manager at the Clinical Trials Coordination Center at URMC.

**Amy Vierhile, RN PNP** is a Pediatric Nurse Practitioner in Child Neurology at URMC. She is a study coordinator for the natural history study and the JUMP study.

**Sara Defendorf, BS, CCRP** is the clinical site monitor for the JUMP study.

**Alyssa Thatcher, BS**, is a study coordinator for the Neurobehavioral Outcomes in Batten Disease study and the natural history study.

**We thank all of the children and families who participate in our research –  
we would be unable to do this work without you!**

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

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