University of Rochester Batten Center Newsletter

Summer 2014

What is a Clinical Trial?

Clinical trials are research studies to evaluate whether new potential treatments are safe, well-tolerated, and/or effective. Clinical trials are part of a long research process, which often begins in a laboratory where scientists test new ideas. A clinical trial is conducted according to a carefully designed research plan called a protocol. Any clinical trial or study involving human volunteers must be reviewed, approved, and monitored by an institutional review board (IRB). An IRB is made up of researchers, physicians, and members of the community. The role of an IRB is to make sure studies are ethical and to protect the rights and welfare of the participants.

Why participate in a trial or research study?

Participating in a trial or study is a way to contribute to medical knowledge that may help others. Furthermore, participation can sometimes provide access to new potential treatments before they become available to the public. Only with clinical research can we gauge the safety and effectiveness of potential new treatments. The groundbreaking advancements, both past and present have been made possible by the participation of volunteers.

For more information on clinical trials along with their risks and benefits, you can visit www.clinicaltrials.gov and http://www.nih.gov/health/clinicaltrials/whyparticipate.htm

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.

Thirteen children have participated 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. **We continue to enroll new participants.** 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 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). The UBDRS is used in two ways – as part of a clinical evaluation at the URBC and as a research tool.

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. Since 2002, 124 children and young adults with Batten disease have taken part in this study, and many return each year! Participation each year is extremely valuable because it helps 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

UNIVERSITY of ROCHESTER MEDICAL CENTER

MEDICINE of THE HIGHEST ORDER

From left to right: Heather Adams, PhD; Elisabeth de Blieck, MPA, CCRC; 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

Workshop on JNCL Clinical Trials Outcomes

On December 6-7th, 2013, the URBC hosted a conference, "Outcome Measures and Infrastructure for Phase III Studies in Batten Disease (JNCL)". Researchers from England, the USA, Germany, Italy, and Denmark, and parents and advocacy groups from England and the USA met to discuss clinical research issues in JNCL. The conference was made possible by a grant from the National Institutes of Health.

The conference participants convened to discuss challenges and opportunities for Phase III clinical trials. Phase III trials focus on clinical efficacy, or in other words, testing whether experimental treatments are effective at treating disease.

There is much preparation to do for future trials:

- study outreach, since the disease is so rare
- set standards to measure if treatments are effective
- identify potential targets of treatment (i.e., specific symptoms of the disease)
- identify and expand patient registries
- build/strengthen international collaborations
- identify potential funding opportunities

The URBC will be preparing meeting proceedings to be published in a peer-reviewed scientific journal. A website is also in development to help continue the work that was started at the conference: http://www.urmc.rochester.edu/neurology/batten-disease-center/batten-disease-conference/

URBC Members Attend WORLD Meeting

Dr. Jonathan Mink and Sara Defendorf attended the 10th annual WORLD Symposium, which is organized and run by the Lysosomal Disease Network (LDN). The WORLD Symposium is an annual meeting where researchers and clinicians from around the world convene to discuss research related to lysosomal storage diseases like Batten disease.

Dr. Mink presented data related to movement disorders in children with JNCL and Late Infantile NCL (LINCL). Although both groups had high rates of movement disorders, the main type of movement disorder exhibited by children with JNCL and LINCL differed. Most children with JNCL had symptoms of parkinsonism, or slow movement with muscle stiffness and problems with balance, which worsened with age. Children with LINCL were more likely to have myoclonus, or brief, involuntary twitching of muscles. Sara Defendorf presented a poster about the challenges of carrying out a clinical trial for a rare disease.

We thank all of the 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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