show me the PATH TO A CURE
July 12 – 14 • St. Louis, Missouri

Annual Family Conference 2024 Research Study Opportunities

Dear Families,

We are pleased to present several Batten disease studies for your participation at this year's conference. Included in this packet is more detailed information for each study, and below is a brief review of the studies and which CLN types are being investigated in each study. We invite you to visit the Research Study Registration table for more information.

Study Name	Institution	PI	CLN Variant(s) Included
Batten Natural History; Neuropsychological Testing; Medical History	University of Rochester	Heather Adams & team	All CLN Variants
Cognitive Neurophsyiology Lab	University of Rochester		All CLN Variants
Characterizing Sleep in Batten Disease	University of Rochester	Heather Adams	CLN2, CLN3
Cognitive and Developmental Outcomes Over Time in Batten Disease	Nationwide Children's Hospital	Emily de los Reyes & Jessica Scherr	CLN2
CoRDS: Batten Questionnaire	Sanford Research	Ben Forred	All CLN Variants
Comprehensive Blood and Biomarker Analysis Study	Sanford Research	Jill Weimer	All CLN Variants
BOLD: Basic Observations of Lysosomal Diseases	Washington University	Patricia Dickson	CLN2, CLN3, CLN4, CLN5, CLN6, CLN7, CLN8
Eye Scan Study	Flaum Eye Institute	Ruchira Singh	All CLN Variants

To participate, an appointment must be made **in advance** for each study at the Research Study Registration table. Some researchers will be prepared for appointments on Thursday, should you arrive in time. If you would like to schedule a time before the conference, please email the address listed with the study. Studies that include blood collection will be consenting on Thursday from 1-6:30 PM; Friday from 8:30 AM-6 PM; and Saturday from 9-Noon.

Please note that **BDSRA will not make Research Study appointments**. Appointments must be made with **each** study's team in advance or on-site at the Research Study Registration table. You may participate in as many studies as you wish, noting that each study is individual and will require a separate consent form. Participants may decide which studies are best for their families.

We are grateful to the many researchers who collaborated to make these opportunities happen at the conference. Your participation is vital for advancing research toward treatments and cures. Thank you for your consideration and for partnering with us on the "Path to a Cure."

BDSRA foundation

Dear families,

The University of Rochester Batten Center (URBC) is inviting all families to participate in research during the week of the virtual BDSRA family conference this summer. We have several research opportunities available to individuals with all forms of Batten disease and their families.

Research Study Activities:

- **Batten Natural History**: Using the Unified Batten Disease Rating Scale (UBDRS), we are tracking the natural history of Batten disease. This study has two parts: an interview about medical history and symptoms and a brief physical exam with your child. Your child must be present for the physical exam (5-10 minutes). You may also elect to complete additional questionnaires.
- **Neuropsychological Testing**: If your child has the ability to complete this testing, Dr. Heather Adams will conduct a brief (<30 minutes) evaluation of his/her cognitive abilities such as attention and memory to better understand the cognitive and behavioral functioning of those with Batten disease.
- **Medical History**: We will gather information about your child's current medications, any testing completed within the past year and his or her current health.

The URBC knows that you and your family are busy, and they will do all they can to accommodate your schedules. Please speak with them to learn more! Please contact a team member through email at Batten@urmc.rochester.edu or by phone at (585) 275-4762 to set up an appointment.

If you are interested in taking part in a URBC study but can't do so during the week of the BDSRA conference, there are also opportunities to do so during other times, either by video or in person at the University of Rochester Batten Center in Rochester, NY.

BE A PART OF BATTEN RESEARCH!

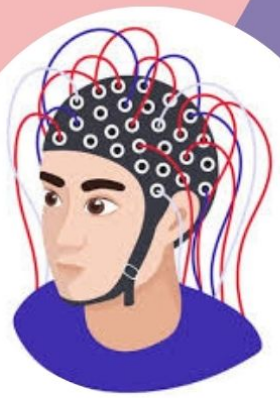
Cognitive Neurophysiology Lab (CNL)

Our Study:

Our EEG studies help us understand how the brain processes information from our senses.

This study hopes to find markers of brain activity that correspond to instances of cognition in response to auditory stimuli.

The goal of this study is to understand more about the symptoms of NCL disorders and how those symptoms change over time.



What will participants do?

Participants will complete an electroencephalogram (EEG). An EEG is designed to evaluate the electrical activity in the brain. It is safe and non-invasive.

Participants will passively listen to tones through headphones.

The experiment should last around 3 hours and we offer participants breaks whenever needed.

Participants will be compensated for their time

Contact Us!

Email: erin_bojanek@urmc.rochester.edu, sile_nimhurchu@urmc.rochester.edu

Or

Call: 315-335-2602

THE FREDERICK J. AND MARION A. SCHINDLER
COGNITIVE NEUROPHYSIOLOGY LABORATORY

RSRB Approval Date: 3/22/2024

University of Rochester Batten Center Research Study

Characterizing Sleep in Batten Disease

This study aims to gather more information about sleep function in individuals with CLN2 and CLN3 Batten Disease. Study activities include:

Affected individuals: provide saliva samples to test melatonin concentration and wear an actigraph (wrist-watch style activity monitor)

Parents/caregivers: complete daily sleep/activity logs, questionnaires about the impact of sleep problems on the well-being of affected individuals and family, and help the affected individual with study participation

We are looking for individuals who…

- Have a confirmed genetic or enzyme-based diagnosis of CLN2 or CLN3 disease and
- Have any symptoms of CLN2 or CLN3 disease and
- Live at home with at least one primary caregiver
- Have not taken oral melatonin (supplement) in the past 2 weeks, or has only taken it occasionally (no more than 3 times per week).

FAQs:

Q: How long does the study last?
A: Study participation is approximately 7-10 days long

Q: Will my affected child wear the actigraph only at night-time?
A: We will ask that your child wear the actigraph continously during day and night-time, as much as they can. At the beginning of the study, we will discuss strategies to help your child get used wearing the actigraph.

Q: How long will you be recruiting people for the study?
A. At this time we do not have an "end date" for recruitment, but we will notify the Batten community if this changes.

Q: We live outside of the United States. Can we still participate?
A: Unfortunately due to some new guidelines for international shipping and for transport of samples, only residents of the USA can participate.

To learn more, contact: BattenSleepStudy@URMC.Rochester.edu or call Study Coordinator Marianna Pereira-Freitas at: (585) 274-0205.

RSRB Approval Date: 3/1/2024

University of Rochester Medical Center
presents a new Batten study led by Heather Adams, Ph.D.

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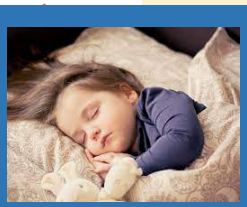

University of Rochester Batten Center

Department of Child Neurology
www.rochesterbatten.urmc.edu
601 Elmwood Avenue, Rochester, NY 14580
(585) 275-2808

Nationwide Children's Hospital is recruiting *children diagnosed with CLN2 disease, a form of Batten Disease,* to participate in a study looking at cognitive and developmental outcomes over time.

Who can take part in this study?

We are recruiting individuals with CLN2 disease to participate in a study to look at cognitive and developmental outcomes over time in patients that are untreated and those that are receiving enzyme replacement therapy. The purpose of the study is to better understand the natural progression of the disease, as well as examine how best to measure developmental outcomes through clinical assessment to inform standard of care. Results from this study will provide a better understanding of CLN2 disease to guide assessment and treatment efforts.

What will happen during the study?

Participants that meet screening criteria will:

- Complete a battery of developmental assessments that include measures of early learning, language, and motor development.
- Caregivers will also complete a series rating forms regarding symptoms and behaviors they see
- Study participation will take1-2 hours to complete for each visit. Families can participate up to 2 times per year.

Compensation

Participants will be reimbursed $50 for their time for each visit completed.
Participation in research is completely voluntary

Contact Information

If you are interested in participating in this study or would like more information please contact Jessi Scherr, PhD at 614-355-7500 ext. 3-8230 or
Jessica.scherr@nationwidechildrens.org

Coordination of Rare Diseases at Sanford

About CoRDS

The Coordination of Rare Diseases at Sanford (CoRDS) is a registry that stores information on individuals affected by a rare disease to help accelerate research.
CoRDS provides a better way for researchers conducting studies or clinical trials to search for individuals who are interested and may be able to participate.

Who can participate in CoRDS?

- A diagnosis of a rare disease
- A person with an uncommon disease with unknown occurrence
- A person who has not yet been diagnosed
- Unaffected carrier

Batten Questionnaire

If you choose to participate in the CoRDS Registry, you will be asked to complete the CoRDS standard questionnaire along with the Batten questionnaire. CoRDS will send an annual reminder to update your information or confirm that it is up to date. You can update your information at any time.

How to Register

Simply scan the barcode below:

Contact Information

- cords@sanfordhealth.org
- (877)-658-9192

SANFORD RESEARCH

WASHINGTON UNIVERSITY IN ST. LOUIS

BASIC OBSERVATIONS OF LYSOSOMAL DISEASES

BOLD RESEARCH STUDY

PI: PATRICIA DICKSON, MD

Researchers at Washington University School of Medicine and St. Louis Children's Hospital are investigating Lysosomal Storage Diseases in order to better understand disease mechanisms. CLN2 Disease is one of the study's primary targets and we are recruiting participants.

We are also interested in other forms of Batten Disease:
CLN1, CLN3, CLN4, CLN5, CLN6, CLN7, & CLN8

We will be investigating the genes that cause neuronal ceroid lipofuscinosis ("Batten disease"), as well as other lysosomal diseases, to better understand how lysosomal dysfunction can cause disease.

To qualify for this study you or your child must be:

- English-speaking
- Have a confirmed diagnosis of CLN1, 2, 3, 4, 5, 6, 7, or 8

Your or your child's participation would involve:

- Collection of medical record data and reporting of health information which allows the study to accurately describe an individual's condition.
- Collection of a blood sample for testing and pluripotent cell development.

Participating in this research study is completely optional. The research team is available and willing to answer any questions.

For more information or if you are interested in participating in the BOLD Research study, please contact:

Kimberly Turner
Clinical Research Coordinator
Department of Pediatrics
Washington University School of Medicine
Phone: 314-747-0791
E-mail: turner.k@wustl.edu

Sanford Research

Batten study led by Dr. Jill Weimer

Comprehensive Blood and Biomarker Analysis

This study aims to collect blood samples from Batten patients (all forms) along with blood samples from healthy subjects to investigate biomarkers or surrogate markers of disease. Furthermore, small divided-out portions of these samples will be stored in a biorepository (a facility for the collection, storage, and management of biological samples) at Sanford Research.

Study activities include:

- ***Affected individuals:*** Provide blood samples to test levels of various substances in blood.
- ***Relatives/caregivers:*** Caregivers of a participating affected individual will complete a questionnaire about the affected individual's diagnosis and symptoms. May provide blood samples to test levels of various substances in blood.

We are looking for individuals who are:

- Genetically diagnosed with any of the 13+ NCL subtypes
- Over 6 months of age
- Unaffected individuals who have a biological child or sibling that has been genetically diagnosed

Contact to learn more:

WeimerLab01@gmail.com

(605)-312-6407

Sanford Research

Pediatrics and Rare Disease - Weimer Lab

2301 E 60th St N Sioux Falls, SD 57106

(605)312-6300

SANFORD RESEARCH

RSRB Approval Date: 06/26/2024

EYE SCAN STUDY!

Conducted by the Flaum Eye Institute

We wanted to let everyone know that representatives from the Flaum Eye Institute in Rochester, NY, who specialize in Battens disease research, are here conducting a research study to help us learn more about the disease. **They will be offering free eye scans of the retina called Ocular Coherence Tomography, or OCT to anyone attending the conference this weekend that either has Battens disease or is a biological parent or sibling of someone with Battens disease,** as long as they meet certain criteria. The scans take less than 10-15 minutes and will be analyzed by scientists and researchers to see if they can better understand what factors contribute to vision loss. If you meet the eligibility criteria, you will be paid $35 for participating. So, if you are a patient or parent or sibling of someone with Battens and interested in participating in this research and willing to have a few scans taken of your eyes, please feel free stop by the Flaum Eye Institute booth this weekend! Oh, and no pupil dilation is required!

University of Rochester representatives from the Flaum Eye Institute in Rochester, NY, who specialize in Batten disease research, are here conducting a study to better understand "how" vision loss occurs in NCLs.

PLEASE STOP BY THE FLAUM EYE INSTITUTE BOOTH (Room 228) FOR MORE INFORMATION & TO RECEIVE FREE OCT & OCT-A IMAGES (retina scans) OF YOUR EYES!

You will be paid $35 if you qualify to participate

If you would like to schedule the imaging study in advance, Please contact Dr. Ruchira Singh at ruchira_singh@urmc.rochester.edu or Nathaniel Foley Foley, at Nathaniel_Foley@URMC.Rochester.edu or 585-259-1701 to set up an appointment.