

# BDCRC Newsletter – December 2025




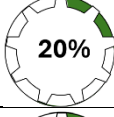

Catch up on the latest updates, opportunities, and upcoming events across the Batten Disease Clinical Research Consortium. Note, this newsletter is best viewed on a desktop, laptop, or tablet.

## Progress in Motion

*Tracking milestones and celebrating momentum across consortium projects.*

### Start-up Item

### Progress

Draft recruitment materials and circulate for feedback		<i>Thank you to the recruitment workgroup; Nadia Moore (KKI), Isabella Garcia (Rush), Joelle Anderson and Colette Free (Sanford), Nubia Fernandez (CHOC), Claire Cinnamon (NYU), Yael Shiloh-Malawsky (UNC), Noah Siedman (BDSRA)</i>
Receive <b>SMART IRB Letter of Acknowledgement</b>		<i>Congratulations, Colorado, NYU, and Rush for ceding reliance this month!</i>
Complete the <a href="#">BDCRC Site Information &amp; Contact Form</a>		<i>Congratulations NICHD for being the first to complete!</i>
Confirm Box access		<i>Congratulations University of Rochester for being the first team to all confirm!</i>
Issue subawards to sites for review		<i>Thank you to Research Administration teams across the BDCRC for their work on the subaward process!</i>

## Welcome Our Newest Team Members

We are excited to announce that **UT Southwestern Medical Center** has joined as the final enrolling BDCRC site. We'd like to extend a warm welcome to the principal investigator, Dr. Saima Kayani, and her team!

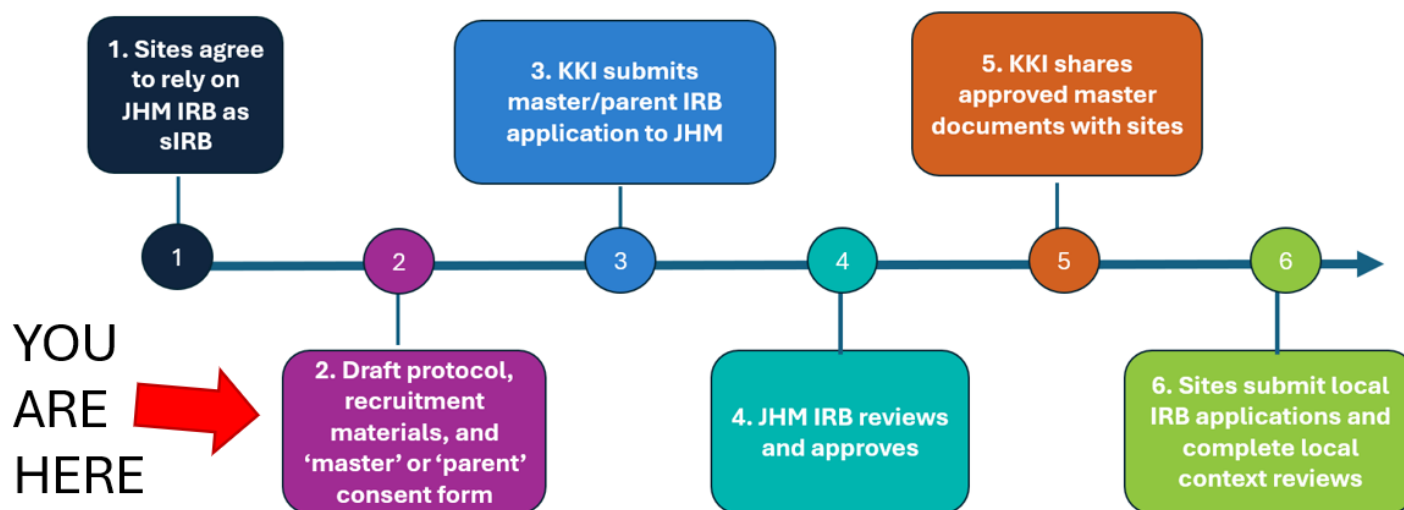
---

## Study Start-Up: Where Are We Now?

The first step in getting our projects off the ground is finalizing the protocol and project materials and submitting a parent application to the **single (central) IRB** for review (Johns Hopkins Medicine IRB). Once approved, participating sites can move forward with their local context review with their IRB and complete single IRB reliance processes, as applicable.

**Curious about the workflow?** Check out the graphic below for the process and timeline!

### Single IRB (sIRB) Application Process



#### Progress on Step 2

- **Protocol**
  - Feedback has been collected and revisions are underway. After internal revisions are complete, the protocol will move through review steps with **RDCRN DMCC, NIH Project Scientists, and JHM IRB.**
- **Recruitment Group**
  - The recruitment workgroup has drafted recruitment materials for the initial IRB submission, which are currently being reviewed internally. BDCRC members are invited to comment directly in the materials in [Box](#) by **January 9<sup>th</sup>**. Use your federated login or RDCRN credentials to login to Box.

#### Want to learn more about the sIRB process?

Participating sites can also learn more about the sIRB process by visiting the JHM [Participating Sites Responsibilities & FAQ](#).

## Help Shape Our Shared Vision for the BDCRC

As we continue building a strong and collaborative BDCRC community, we want to capture what matters most to each of you - take a few moments to share your goals in the [Shared Vision Form](#).



## Send Us Your Suggestions!

Have an idea to improve the consortium, our newsletter, or upcoming initiatives? We want to hear from you!

Share your thoughts, big or small, using the [Suggestion Box](#).



# Inside the Consortium

*Your look at the people and projects driving this consortium*



## Monthly Feature: Meet the BDCRC Pilot Core

### Who We Are

The Pilot Study Core is a key part of the Batten Disease Clinical Research Consortium (BDCRC). We fund innovative pilot studies that tackle top research priorities and advance early diagnosis, management, and treatment.



Dr. Liu Lin Thio  
*Washington University in St. Louis*



Dr. Ineka Whiteman  
*Batten Disease Support, Research, and Advocacy (BDSRA) Foundation*

## What We're Working On

The Pilot Study Core solicits, rigorously evaluates, and funds high-quality translational and clinical pilot studies. Each year, we select at least one project aligned with the top *Research Priorities for the Neuronal Ceroid Lipofuscinoses*, identified by Batten disease stakeholders (*Lancet Neurology*). Our process is systematic and NIH-based, ensuring that funded studies are rigorous, feasible, impactful, and innovative. We provide end-to-end support—from project launch through completion and beyond—helping investigators succeed at every stage.

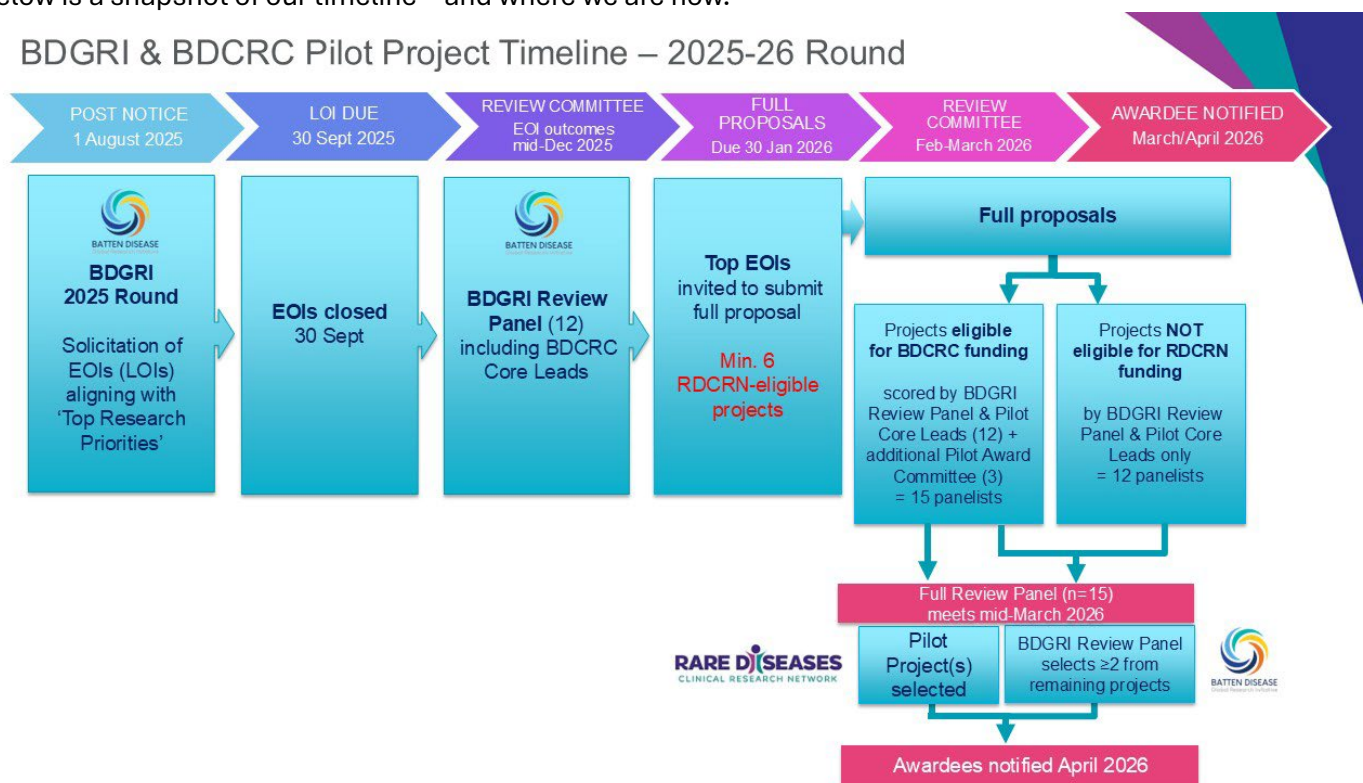
We partner with the Batten Disease Global Research Initiative (BDGRI) to solicit and review applications. Together, the BDGRI Review Panel and the BDCRC Pilot Review Panel review applications and award one or more recipients (up to \$50,000 per funding round).

Here's how it works:

1. **Initial Proposal Submission** – Applicants submit preliminary proposals as Expressions of Interest (EOIs).
2. **Shortlisting** – A review panel of 12 members review EOIs. A selected group of applicants is invited to advance.
3. **Full Proposal Submission** – Shortlisted applicants submit detailed proposals.
4. **Final Review and Selection** – Review panels review full proposals and select Pilot Award recipient(s). Remaining proposals considered for BDGRI funding.

Below is a snapshot of our timeline—and where we are now.

### BDGRI & BDCRC Pilot Project Timeline – 2025-26 Round



## Why It Matters

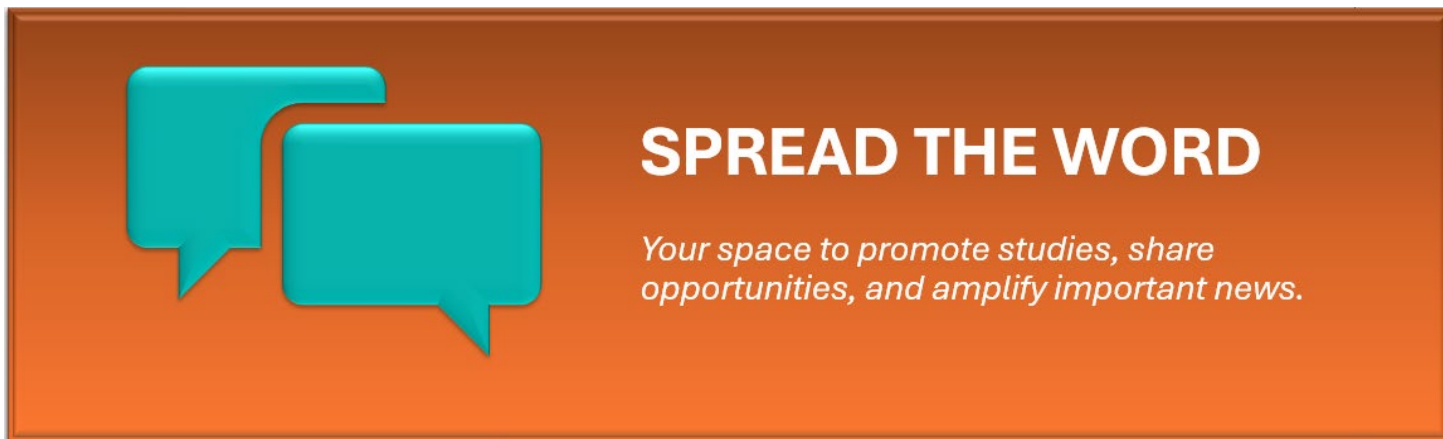
These proof-of-concept studies pave the way to new lines of research, growth in knowledge about the NCLs, and better outcomes for families affected by Batten disease.

## How You Can Connect

Have a bold idea? We welcome proposals from early-career and experienced investigators—even those new to Batten research. Contact [Dr. Liu Lin Thio](#) or [Dr. Ineka Whiteman](#) to learn more.

## Fun Fact or Quote

“Our favorite part of this work? Seeing innovative ideas take shape and knowing they could genuinely change the future for families affected by Batten disease.”



## Help Us Recruit the Next Generation of NCL Clinician-Scientists!

You can help spread the word! Consider any of the following:

1. **Update your email signature, see template below**

1.  Applications are open for the Batten Disease Clinical Research Consortium [Early Career](#)

**Investigator Grant!** 

2. **Post or re-post on social media**

- a. [X \(Twitter\)](#)
- b. [LinkedIn](#)
- c. [Facebook](#)

d. Or create your own post using this template:

*New funding opportunity for clinically oriented fellows and early career faculty!*

#BattenDisease Clinical Research Consortium (BDCRC) Early Career Investigator Grants

Supports one year of innovative research to advance care for individuals living with

[#NeuronalCeroidLipofuscinoses](#) (NCLs)

Learn more and [apply](#)

3. **Share within your networks**

Please share widely! If you plan to post to any listservs, kindly notify [Dr. Nick Abreu](#) to avoid duplicate efforts.

## Have a study to highlight or materials to share?

Send us a brief summary and any IRB-approved recruitment flyers or links you'd like featured in the next issue. Email your materials to [BDCRC@kennedykrieger.org](mailto:BDCRC@kennedykrieger.org).

# Extending Our Reach

*Showcasing the work of BDCRC members beyond the consortium—through conferences, collaborations, and community impact.*

## **Bespoke Gene Therapy Offers New Hope for Children with Ultrarare SLC6A1 Disease**

Dr. Emily de los Reyes, *Nationwide Children's Hospital*

Researchers at Nationwide Children's Hospital have delivered a first-in-human, bespoke gene therapy for 8-year-old Maxwell, who has an ultrarare SLC6A1-related neurodevelopmental disorder — marking a major step forward for precision medicine in rare disease. By using an AAV9 vector to deliver a healthy copy of the SLC6A1 gene directly to the central nervous system, the team aims to treat the underlying cause of the disorder rather than just managing symptoms. This work is made possible through collaboration among scientists, clinicians, families and rare disease foundations.

Learn more [here](#).



Image provided courtesy of Nationwide Children's Hospital. ©NationwideChildren'sHospital/Stephanie Costilo

## Chicago doctor dedicated to changing lives of kids with rare neurological diseases

Dr. Elizabeth Berry-Kravis, *Rush University Medical Center*

Dr. Berry-Kravis has been featured by local news for her pioneering work in research and experimental treatments for conditions like Fragile X Syndrome and Krabbe disease, offering immense hope and real futures for families facing devastating diagnoses. Read more [here](#).



## Hot off the Press

*Explore the newest research shaping the future of Batten disease studies.*

### Recent Publications with BDCRC Member Contributors

**Longitudinal Exploration of Auditory Sensory-Perceptual Processing in CLN3 Disease (Juvenile Neuronal Ceroid Lipofuscinosis (Batten disease)): A High-Density Auditory Evoked Potential (AEP) Study.**

EK Bojanek, ER Lang, HR Adams, J Vermilion, EF Augustine, T Brima, S Nasimjonova, EG Freedman, JJ Foxe. *bioRxiv* (2025) [<https://doi.org/10.1101/2025.11.19.689311>]

## Mark Your Calendar

### Upcoming events

#### BDCRC & Partner Events

- **WORLD Symposium** — February 2-6, 2026, San Diego, CA
- **RDCRN Semi-Annual Meeting** — February 25-26, 2026, Washington, DC
- **Rare Disease Day at NIH** — February 27, 2026, Bethesda, MD
- **Stengel 200 CLN3 Conference** — August 23–27, 2026, Røros, Norway
- **BDSRA Foundation Annual Family Conference** — July 10–12, 2026, Chicago, IL

- **10th Annual NCL Translational Research Conference— November 4–6, 2026, Boston, MA**

### **Other Key Conferences**

- **Child Neurology Society (CNS) Annual Meeting — Oct 14–17, 2026**
- **American Society of Human Genetics (ASHG) Annual Meeting — Oct 20–24, 2026**
- **European Society of Gene & Cell Therapy (ESGCT) Annual Congress — Oct 27–30, 2026**
- **National Society of Genetic Counselors (NSGC) Annual Conference — Nov 10–14, 2026**
- **Society for Neuroscience (SfN) — Neuroscience 2026 — Nov 14–18, 2026**

## Share Your News

Have exciting news? Share your achievements so we can spotlight your success.



**Your accomplishments deserve recognition—let’s celebrate together!**

Have a new grant, award, promotion, or publication? Share your individual or team achievements with us. Email your news to [BDCRC@kennedykrieger.org](mailto:BDCRC@kennedykrieger.org) for inclusion in the next newsletter.

*Happy Holidays*

Thank you for your hard work and dedication within the  
Batten Disease Clinical Research Consortium.

We wish you a joyful holiday season and look forward to a  
successful and impactful 2026 together.