

“The support I get from BDSRA has allowed me to survive the initial diagnosis, maintain hope for the future of our Batten affected kids, and feel I am understood in this journey toward the unknown.”

- Marlo Urbina, mother of Sofia

Pictured: Sofia Urbina (CLN3, right) and her BFF (left).



# THE BATTEN DISEASE SUPPORT AND RESEARCH ASSOCIATION



# FROM OUR BOARD PRESIDENT

Dear Friends,

Looking back on the year of contributions, dedication and incredible progress in the future of Batten disease, it's clear that one of my greatest sources of pride is being part of this incredible community. In spite of the challenges we face as parents of children with Batten disease and as advocates of this mission, we are fortunate to have each other and the power of many is apparent when you look at what we have accomplished together.

Many of you know my son, Jacob, who is one of the loves of my life and has a unique, uncategorized form of Batten disease. He is the reason why I turned to BDSRA during the most difficult time in my life and he is the motivation behind my commitment to creating a brighter future for all children with Batten. My family's personal experience with the services and refuge that BDSRA offered is a gift that is hard to measure. And it is the reason why I have chosen to serve this organization.

I have a close-up view of the progress and impact BDSRA is creating for families and I am continually amazed by what we can do when we pool our experience, ideas and resources. My gratitude for the staff, donors and network of supporters is abundant. I see families reaching out to each other in the closed Facebook group, which has continued to grow as a haven and resource of shared experiences; people working hard to create momentum with the scientific community and industry partnerships; camaraderie among advocates for the needs and legacy of our children with Batten disease.

To see how BDSRA has been developing as a leader in our global ecosystem is inspiring and hopeful. I appreciate all that you do for our shared mission. This accelerated track would not have been possible without you. Let's keep building. The upcoming coming year is filled with exciting opportunities to improve lives through education, support, research and advocacy services. Five ways you can support BDSRA right now are in the pages to follow. I invite you to be part of the mission we are living out.

Together our power is magnified.

Sincerely,


**Rob Geer**, Board President, BDSRA

“Together  
our  
power is  
magnified.”



Pictured: Jacob Geer (left, uncategorized form),  
Rob Geer (right).





“There is such a struggle to meet these kids’ needs.”

## SCIENTIFIC RESEARCH AND EDUCATION

### Partnerships Moving Education Forward

Education and schooling can be a challenging road for a Batten parent. It can also be a source of socialization, fun and connection for Batten children, especially those with CLN3 (JNCL). We want to do everything possible to further expert knowledge in this area and share it with parents and educators to help make their child’s experience an effective one. Having more resources created using real-life data is something the entire Batten community is craving. We’ve heard it from BDSRA’s previous needs assessments and in conversations with partner organizations. Those conversations led to the opportunity to participate in a project, with 20 organizations across 7 countries, to create the Juvenile Neuronal Ceroid Lipofuscinosis (JNCL) and Education textbook. This

type of global collaboration is hopeful. This is what we’ve all been working for. We can see missions aligning and people uniting for the purpose of a better educational quality of life for JNCL kids and families.

Many BDSRA parents and family members participated directly in the making of this book by taking an extensive survey about their personal experiences. One of the mothers who contributed to the project is Marjorie Newell of Maple Valley, Washington. Both of Marjorie’s children had CLN3, Michelle and Sean. Michelle was a braille reader, who loved to listen and be with the group. Sean took an interest in the solar system, science and cooking. In school, Marjorie found herself advocating for her child and using all of the resources available to her.

“There is such a struggle to meet these kids’ needs,” said Marjorie. “Most aids have never dealt with this in their career and may never see it again. So you do, as a parent, have to advocate for what your child needs and partner with authoritative voices like your doctor and BDSRA to help you validate what you are telling the school. The support side is so important.”

#### **There are commonalities in what many parents face when working with their school system:**

- Staff who often want to be helpful, but are afraid of Batten disease and don’t know what to do
- Doing nothing because they are unskilled
- Blaming kids for behaviors they can’t manage and see it as willful rather than a part of the disease

The JNCL book looks at all those factors and provides tools and techniques for teachers and aides to help them help our kids, and it gives parents ideas about possibilities in the home and at school for maximizing quality of life. Trends seen in our shared experiences have fueled the content that you’ll get to read when the book is published in 2018. Stay tuned for the launch date and other details by subscribing to BDSRA’s newsletter, the *Illuminator*: <http://bdsra.org/about-bdsra/illuminator-newsletter/>

A full list of international partners and publisher, Erasmus+ can be found on the book outline available on BDSRA’s website: <http://bdsra.org/patient-and-family-support/educational-resources/>

# NEURONAL CEROID LIPOFUSCINOSIS (NCL) RESEARCH CONFERENCE

The 15th Annual International NCL Conference was held in Boston, MA from October 5-9, 2016. Every two years, more than 200 researchers, industry representatives, patient advocacy group leaders and parents attend this event featuring sessions from basic research in cell biology to the latest in clinical trials. BDSRA greatly values this international collaboration opportunity and was a primary sponsor, and hands-on organizer helping to coordinate the event logistics on-site, collateral materials and presentations. Susan L. Cotman, Ph.D., from the Center for Genomic Medicine at Massachusetts General Hospital, was the conference chair and worked on the conference content, approving abstracts and choosing speakers.

With any rare disease, there are only so many researchers and funds to go around. Which makes having conferences like this important to ensure that the researchers are all on the same page with new advancements. It creates a place to collaborate and lead the way toward faster treatments and cures. Those who attended, echoed the sentiment that it was a much needed and valuable conference.

There were six sessions devoted to quality of life, the burden of disease and educational needs. It is important that these real-life needs are communicated with researchers in order for them to develop treatments meaningful to parents. To do so, BDSRA shared survey results collected from plenary sessions at the annual family conference and made a presentation at the event representing the patient voice. Advocating for patients' needs and well-being is a key component to successful research outcomes. A big thank you goes out to the families who have made it possible for BDSRA to be this voice in the scientific community. For nearly 30 years, you have supported this mission financially and with the donation of blood, skin and brain samples.

The energy around the conference was very optimistic with great numbers of industry present, as well as families from Europe and the U.S. The Rare Disease Report was on site covering the event for two days, reporting on Batten research to their audience of more than 100,000, many of whom are clinicians. For interviews, updates and stories from the conference, see the Rare Disease Report updates here <http://bit.ly/2e8c1xw>.

## Emerging Batten Researchers



"Since graduate school, I have developed a passion for understanding mechanisms in cell biology, like autophagy. As a post-doctoral research fellow, I was inspired to work in Batten disease, especially JNCL, because it was becoming evident that NCL proteins, like CLN3, play a role in autophagy, a mechanism to recycle components of cells that have been damaged and to generate cellular energy. Lack of functional CLN3 protein is known to cause defects in autophagy. With more knowledge of the function of CLN3, I hope to contribute to drug development research so that, one day, there will be a treatment."

Uma Chandrachud, PhD  
Postdoctoral Research Fellow  
Center for Human Genetic Research  
Massachusetts General Hospital



"I began researching Batten disease 5 years ago; I knew nothing of Batten disease or lysosomal storage disorders prior to teaming up with Mark Sands at Washington University in St. Louis. I continue to research Batten disease because I want to contribute to the growing body of knowledge that will help patients, families, and clinicians manage this disease, with the hope that we will find a cure. I want my research to have a direct, positive impact on patients' and caregivers' quality of life, and I hope that my research using cannabidiol to treat CLN1 seizures achieves just that."

Joshua Dearborn, PhD  
Postdoctoral Research Associate  
Department of Internal Medicine  
Washington University, School of Medicine



"I have been studying Batten Disease for three years now and what keeps me going is that there is so much to be discovered about the CLN family of genes and any piece of data I am able to generate will help push the field forward. My current work focuses on two areas, the first is elucidating the basic mechanisms of CLN family of proteins by mapping out their protein-protein interactions, and the second is using automated microscopy techniques and high content screening with patient derived fibroblasts to discover new drugs with potential therapeutic benefit in Batten Disease."

Jacob Cain, Ph.D.  
Weimer Lab  
Pediatric and Rare Disease Group  
Sanford Health



"I feel very fortunate to be able to work in Batten Disease. The support that all these families give and receive is truly inspiring for me. This is a hard fight for all of them; we want to give them some hope to keep fighting. They teach us how to persevere, to have courage and never quit. I am really excited about the potential of gene therapy, and glad to be working to develop this into a treatment approach for CLN1. I hope that one day these families can create many more happy memories together."

Alejandra Rozenberg, DVM  
Post-doctoral Research Associate  
Gene Therapy Center, Gray Lab  
University of North Carolina at Chapel Hill

“It creates a place to collaborate and lead the way towards faster treatments and cures.”



# SUPPORT AND COMMUNITY

While BDSRA's support programs are often provided across some geographic distance throughout the year, one of the most impactful times is when this vast network comes together in person for the BDSRA Family Conference. In 2016 it brought 400+ to St. Louis, Missouri from July 14-17. The St. Louis Post-Dispatch covered the event and Nolan Gahlbeck threw out the first pitch at the River City Rascal baseball game!

The expert scientific presentations, sibling outings, programming and sessions for all family members and facets of the disease are some of the things that have people traveling hundreds or even thousands of miles to be together at this one of a kind event. The family members, siblings, friends, volunteers, researchers and industry partners that made this event a success are a huge source of pride for this community.



Pictured: Chloe Harrison (left), Nathan Hillman (CLN3, right) during pet therapy.

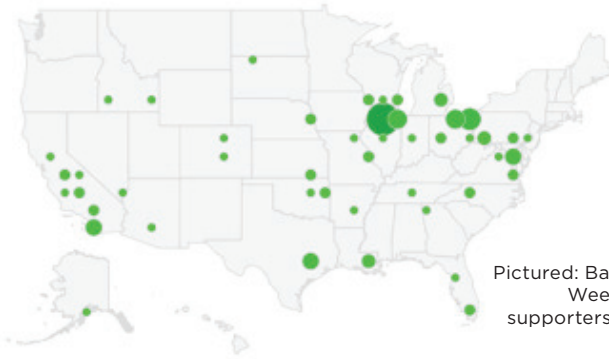
“I am thankful to have a group of caring individuals who can speak from the heart because they have been there.”

- Brian O'Neill (Batten SIB)

# CONNECTED BY A MISSION

2016 marked the first annual Batten Awareness Weekend Virtual 5K fundraiser. During the first weekend of June, we had 253 people across the country running or walking in their hometowns in support of Batten disease. These participants elevated awareness using social media and raised more than \$10,000! No matter where we are geographically, we come together in the spirit of #LoveHopeCure to support our shared mission in memory and in honor affected of loved ones.

## LOVE. HOPE. CURE.



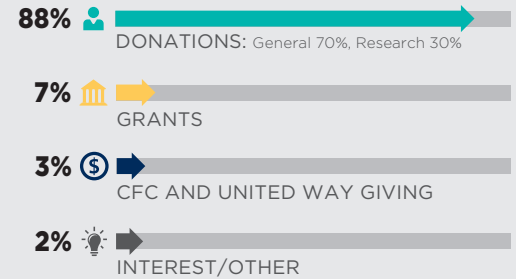
Pictured: Batten Awareness Weekend Virtual 5K supporters across the U.S.



Lita Ciaccio and Friends on Batten Awareness Weekend.

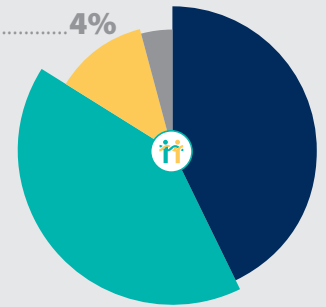
# 2016 FINANCIALS

## Revenues



## Research Awards and Support Services

Research	43%
Support Programs	41%
Administration	12%
Fundraising	4%



## Staff

- Margie Frazier, PhD, LISW-S,** Executive Director, [mfrazier@bdsra.org](mailto:mfrazier@bdsra.org)
- Christina Clark,** Director of Development, [cclark@bdsra.org](mailto:cclark@bdsra.org)
- Tracy Kirby,** Family Liaison and Administrative Lead, [tkirby@bdsra.org](mailto:tkirby@bdsra.org)
- Noreen Murphy, MA,** Patient and Family Education Coordinator, [nmurphy@bdsra.org](mailto:nmurphy@bdsra.org)

## Board of Directors

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# 5 THINGS YOU CAN DO

To Support BDSRA Right Now



**DONATE.** Make it possible to fund our mission, donate today.  
[www.bdsra.org/donate](http://www.bdsra.org/donate)



**ADVOCATE.** Join a core group of supporters and be an advocate.  
[www.bdsra.org/be-a-rare-disease-advocate/](http://www.bdsra.org/be-a-rare-disease-advocate/)



**GIFT.** Make your next birthday a gift to Batten families.  
[www.bdsra.org/birthday](http://www.bdsra.org/birthday)



**EMPLOYER MATCH.** Ask your employer about matching your charitable donation.  
[www.bdsra.org/bdsra-advocacy/funding-our-mission/](http://www.bdsra.org/bdsra-advocacy/funding-our-mission/)



**SHARE.** Share your story on our family profiles page.  
<http://bdsra.org/bdsra-advocacy/share-your-story/>



Pictured: Sam Engman (CLN1)

“We’re thankful to BDSRA for the great support that they have provided, and otherwise making sure that we have the resources that we need to love and take care of our child.”

- Sarah Engman, mother of Sam

# THANK YOU DONORS!



BDSRA extends deep appreciation to our donors that made 2016 possible, it was a year to be proud of. We are humbled by the strength, dedication and what this community is able to create together.

**\$370,000** awarded in research grants in 2016

 **8 families** supported in U.S. clinical trials

Approximately **600 unique posts, 5,000 comments** and **20,000 reactions** in the closed caregiver **Facebook** group.



**16 school districts** supported by BDSRA's **Teach & Be Taught**



BDSRA represented the **patient voice** at more than

**25** meetings and conferences.



**34** countries served

Please subscribe to the BDSRA newsletter, the *Illuminator*, to see all of the donors that support us each month throughout the year. To make a donation, visit our website at [www.bdsra.org/donate](http://www.bdsra.org/donate) or contact Christina Clark at [cclark@bdsra.org](mailto:cclark@bdsra.org) or 614-973-6012. The BDSRA welcomes deferred gifts through your estate plan.

**BATTEN DISEASE**  
Support and Research Association

**800-448-4570 | [www.bdsra.org](http://www.bdsra.org)**