

FAMILY SUPPORT
ADVOCACY
RESEARCH
SCIENCE
AMBIANCE
BUSINESS
COMMUNITY
GIVING
HOPE
SUPPORT
PARTNERSHIPS
EDUCATION

A YEAR WITH



2014
ANNUAL REPORT

BDSRA...the year in numbers:

BATTEN DISEASE IS
1 of 7,000
RARE DISEASES

1 in 10 Americans have a rare disease — 30 million at any one time.

50% OF THESE DISEASES AFFECT children under



the age of 5

It is estimated that only 5% of rare diseases have an FDA-approved treatment

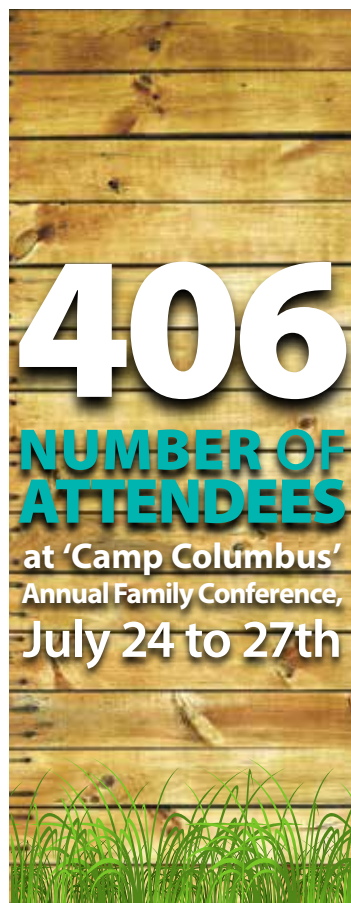


3 BDSRA FAMILIES

relocated to Columbus, Ohio, the **only US site** for an enzyme replacement trial sponsored by BioMarin



US, Canada, Spain, Portugal, Hungary, Ireland, UK, France, Germany, India, Yemen, Iran, Peru, Malaysia, Turkey, South Africa, Serbia, New Zealand, Argentina, Australia, Brazil, Poland, Israel, Saudi Arabia, Belgium and Taiwan.



RESEARCH CONFERENCES SUPPORTED by BDSRA so that 100s of researchers were able to share findings and work together on TREATMENTS



93 FAMILIES answered the BDSRA FAMILY NEEDS ASSESSMENT in partnership with University of MARYLAND SCHOOL OF SOCIAL WORK

\$346K in BDSRA merit review process approved by BDSRA board in conjunction with Hope FOR **8** 4 Bridget, BDSRA-Australia, Batten Disease PROJECTS Family Association (UK), and Noah's Hope

 **708** sharing information on BDSRA'S PRIVATE FACEBOOK PAGE FOR PARENTS AND CAREGIVERS

Research in Review

Research Trials

In 2014, the BDSRA reached out to dozens of researchers to invite Letters of Intent (LOI) or short proposals to preview work they would like to accomplish in the following year with a maximum of \$60,000. Once these LOIs were narrowed to requests for full proposals, nearly 30 researchers from universities, children's hospitals, and industry stepped in to provide peer reviews on a volunteer basis.

Noah's Hope, Drew's Hope, Hope 4 Bridget, BDSRA-Australia and the Batten Disease Family Association in the United Kingdom have partnered with BDSRA to co-fund these important projects.

- **Meaningful Endpoints for Phase III Clinical Trials in Juvenile Batten Disease (CLN3).** Principal Investigator: Heather Adams, Ph.D., University of Rochester. **\$45,000: BDSRA**
- **INCL Gene Therapy Using AAV9 Vectors (CLN1).** Principal Investigator: Steven Gray, Ph.D., University of North Carolina, Chapel Hill. **\$50,000: BDSRA**
- **Antisense Oligonucleotides for the Treatment of Juvenile Neuronal Ceroid Lipofuscinosis (CLN3).** Principal Investigator: Michelle Hastings, Ph.D., Rosalind Franklin School of Medicine and Science. **\$50,000: BDSRA**
- **Stop Codon Read-through and Non-sense Suppression for the Treatment of Infantile and Late-Infantile Neuronal Ceroid Lipofuscinosis (CLN2).** Principal Investigator: Michelle Hastings, Ph.D., Rosalind Franklin School of Medicine and Science. **\$25,000: Noah's Hope, Hope 4 Bridget**
- **Astrocytic Thrombospondin-1 in Juvenile Neuronal Ceroid Lipofuscinosis: Impact on Synaptic Dysfunction (CLN3).** Principal Investigator: Tammy Kielian, Ph.D., University of Nebraska. **\$50,000: Drew's Hope, BDSRA**
- **Crossing the Blood Brain Barrier: Enzyme Replacement Therapy for LINCL (CLN2).** Principal Investigator: Peter Lobel, Ph.D. Rutgers, The State University of New Jersey. **\$50,000: Noah's Hope, Hope-4Bridget, Drew's Hope**

- **NCL Mutation and Patient Database.**

Principal Investigator: Sara Mole, Ph.D., University College London, United Kingdom. **\$15,000: BDSRA and BDFA-UK**

- **Viral Mediated Gene Therapy in Ovine Batten Disease (CLN5 and CLN6).**

Principal Investigator: David Palmer, Ph.D., Lincoln University, Christchurch, New Zealand. **\$50,000: BDSRA – Australia**



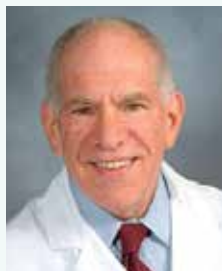
Family Involvement is key to research progress for Batten disease

The involvement of families of affected children and adults in fundamental research related to Batten disease has been crucial to moving discoveries in the lab to translational work at the bedside. Underlying all of this work is the families — their belief that something can be better, perhaps not for their own children, but for children and adults with Batten in the future. Those families who choose to donate blood samples, skin samples, and who courageously donate brains for research purposes have made possible the vast array of research assets available for many Batten researchers.



According to Ryan Geraets, an MD, PhD student at Sanford School of Medicine in Sioux Falls, South Dakota. "Scientists that are granted the opportunity to utilize these samples

are extremely grateful for the participation of patients and their families given the challenges they must endure to be in this research," he said.



Ronald Crystal, MD, professor and chair of the Department of Genetic Medicine of the Weill Medical College of Cornell University, views this as an import-

ant partnership. "Research in the area of developing therapeutics for rare diseases is a partnership in many ways between families of subjects that have these diseases and the scientists that work in that area."



Katherine Sims, MD, Director of the Massachusetts General Hospital NCL Disorders Clinic, says that "in order to better understand the Batten disease disorders, it is critical to have

the best characterization of the clinical features and issues that these patients evidence at the start of and during the course of their disease." Dr. Sims has worked to establish and maintain a biorepository to assist with making these discoveries.



BDSRA Welcomes Families to Columbus for Only United States Trial Site for CLN2 Late Infantile Study

Nationwide Children's Hospital is the US site for BioMarin Pharmaceuticals' clinical trial of enzyme replacement for children with CLN2 Batten disease. With BDSRA's help, three wonderful families from across the country have relocated to Columbus, Ohio, to participate in the study for 2 years. Children receive treatments every two weeks under the supervision of BDSRA's Center of Excellence leader, Dr. Emily de Los Reyes and her colleague, Dr. Lenora Lehwald. Other sites include London, Rome and Hamburg.



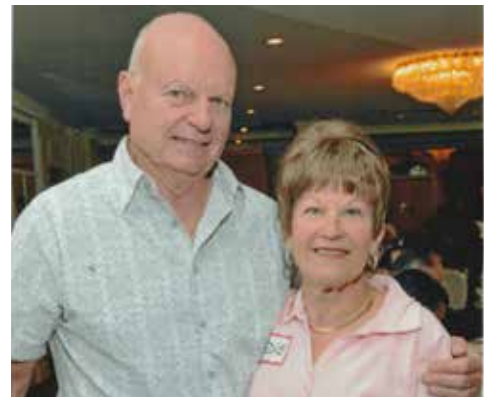
BDSRA Family Support & Education



Top left: Mabel Larson with her grandpa, Dan Moore.

Middle: Ramee Larson with Mabel's siblings, Braden and Nora Larson.

Top Right: Gene and Edie Dockter. Bottom right: Edie and children Ken, Tommy and Laurie, who all had JNCL.



BDSRA Conference

The 2014 BDSRA annual Family Conference, "Camp Columbus," had 406 attendees – a mix of family members, loved ones with Batten disease, siblings, extended family members, scientists, researchers, health care providers and BDSRA staff and volunteers. Here's what one mother had to say:

"I'm so grateful to have had this opportunity. I'm so thankful I got to share it with my dad. We talked through many things and I think we both walked away with a lot of peace." – Ramee Larson

Grief Support

For our Batten families, grief is not time-limited. BDSRA is here to provide support as long as families need it. Bereaved families can connect in various ways: face-to-face at the BDSRA conference "Life Goes On" outings, online in the BDSRA Closed Group for Parents and

Caregivers and by phone and email with BDSRA staff.

"BDSRA was there for me when my Christopher was first diagnosed. They have provided steady support by making connections to exchange equipment and information. BDSRA has not only provided me with sound advice through the grief process, but they continue to help make me feel connected to the organization many years after suffering a loss." – Peggy DiCapua

School Accommodations

Many parents are unfamiliar with how best to inform the team of professionals in the school setting about Batten disease and how it impacts their child or teen. BDSRA staff work with families throughout the school year, providing helpful guidance and informational materials about special school accommodations for the students.

BDSRA Web Site Includes New Resources for Families

2014 was a busy year for BDSRA! As an international non-profit organization that serves patients and families in many parts of the world, we have focused on developing more listings of community resources on a variety of topics to help families. For examples of available resources, visit <http://bdsra.org/patient-and-family-support/>. In addition, visitors can download past issues of the "Illuminator" our quarterly newsletter and other information about our work.

"Since 1987, the BDSRA has helped us with their support and love. Lance Johnston, Executive Director at the time was available whenever we had questions about Batten disease and told us what research doctors were available to us. We were also put in contact with other families, who know what you are going through and also to know you're not alone." – Edie and Gene Docktor

Partnerships in Awareness

BDSRA Represents!

Because Batten research is a global enterprise, BDSRA staff and board members attend events throughout the year to serve on task forces, educate clinicians and lead discussions. This is where we were in 2014:

- Rare Disease Day Events, Washington, DC
- BioMarin Rare Disease Day Employee Exchange, San Rafael, CA
- National Institutes of Health, Rare Disease Clinical Research Network (RDCRN) Council of Patient Advocacy Groups, Bethesda, MD.
- US Food and Drug Administration Conference on Inborn Errors of Metabolism, Silver Spring, MD.
- Global Genes Rare Disease Summit, Huntington Beach, California
- NCL Congress 2014, Cordoba Argentina
- Child Neurological Society Meeting, Columbus, Ohio
- European Working group on JNCL Education and Quality of Life, Oslo, Norway
- NCL-Stiftung Juvenile Batten Research Conference, Hamburg, Germany

Makeup Geek funds Research

Without a shadow of a doubt, Makeup Geek was one of BDSRA's largest supporters in 2014, sending \$40,000 in proceeds of Caitlin Rose eye shadow and a company-wide leadership seminar during the Annual Fund drive. Makeup Geek's president, Marlena Stell, said she has been 'like family' of the Allios of Vacaville, California, whose daughter, Caitlin, lost her battle with Juvenile Batten disease in 2012. To learn more about Makeup Geek and to order your own Caitlin Rose shadow, go to <http://bit.ly/1FGUetU>.



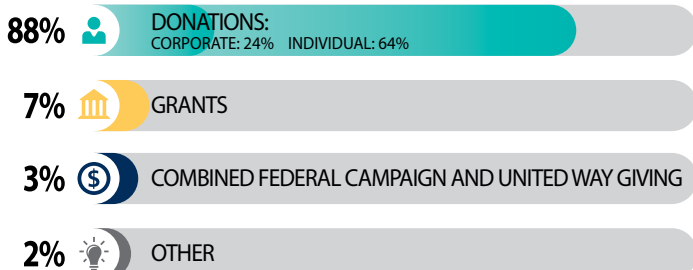
Metro New York Chapter

Garage sales, Bowl-a-Thons and more have helped the BDSRA Metro New York Chapter fund \$250,000 in research via the BDSRA research merit review process.

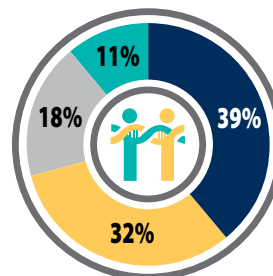


Financials

Revenues



Research Awards and Support Services



SUPPORT PROGRAMS – 39%
 RESEARCH – 32%
 ADMINISTRATION – 18%
 FUNDRAISING – 11%

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