



2015 ANNUAL REPORT

15



Jaylen. Star student. Makes a mean cupcake. Good friend to other BDSRA SIB program kids. Beneath this beautiful exterior lies the deep love and commitment to her brother and sister, Mia and Kaleb, who are part of a clinical trial that shows incredible promise. Because of Jaylen and her family, the work and commitment of BDSRA, industry, and clinicians, there is a brighter future for so many with Batten disease. Inside, we'll share with you how all of these partners work together to make new treatments happen. 2015 was a very good year!

Photo (above): L to R: Mia, Jaylen (middle), and Kaleb

BDSRA and the drug development process



BASIC RESEARCH AND DISCOVERY PHASE

For decades, BDSRA has understood that basic disease-based research has been the foundation, the vast incubator of the treatments and clinical trials we are seeing right now. Understanding mechanisms of cells — the role of inflammation, the pathogenesis of structures and how cells can repair and regenerate, are what fuels innovation and stirs hope for so many around the world.

14 BATTEN DISEASE forms IDENTIFIED

“Although Batten disease is rare, its importance is paramount. By studying the roles of these genes and proteins in this disease it can unravel secrets of the brain, of cell death, and of other diseases.”

– **Rose-Mary Boustany, MD**, Abu Haidar Neuroscience Institute, American University of Beirut Medical Center



HUNDREDS OF
HUMAN BLOOD
AND
TISSUE
SAMPLES
COLLECTED FOR
RESEARCH

PRE-CLINICAL PREPARATION PHASE

As industry and medical researchers prepare to take science from research bench to patient bedside for clinical trials, BDSRA helps with a host of logistical and informational challenges. In 2015, we helped researchers move trials ahead more quickly. With families’ input, we examined meaningful patient outcomes, understandable consenting processes, and more.

Understanding Meaningful Endpoints

“My BDSRA-funded research focuses on meaningful endpoints. That is, if the treatment we are studying in this trial works, how will we know and how will we measure it? My goal is that by talking to parents, coordinating researchers’ input from around the world, and reviewing our natural history data from the last 15 years, we will identify clinical endpoints that are meaningful for trials in juvenile Batten disease.”

– **Heather Adams, Ph.D.**, Neuropsychologist, University of Rochester

BDSRA staff asked if I would lend my voice to a focus group to help BioMarin better understand my experience as Drew’s parent. It’s exciting to see that some of the things our group said are now part of the study.”

– **Katie Ferrandino**, Newtown, PA bereaved Batten mom and Patient Advocate



MAP TO A CURE

"The patient voice is vital in clinical trial planning, from helping us to understand what symptoms are important, to finding the best trial sites, to helping us understand how to support families as they travel from long distances to participate. Their sacrifices and partnership mean so much to progress."

- **Dr. Erika Augustine, MD**, Assistant Professor of Neurology and Pediatrics, University of Rochester Medical Center



THE
**PATIENT
VOICE**
IS
VITAL
IN CLINICAL TRIAL
PLANNING

CLINICAL TRIAL PHASE

As clinical trials get underway, participating families make big sacrifices as they spend long periods of time away from all they know at home. With that in mind, BDSRA advocates for adequate housing and financial support for families, educates trial teams about family needs, and troubleshoots local challenges they have.

"Margie was there at the first phone call and BDSRA is still helping us two years later, navigate the ebbs and flows of being in a clinical trial. It has been so worth it."

- **Michelle O'Leary**, Batten mom

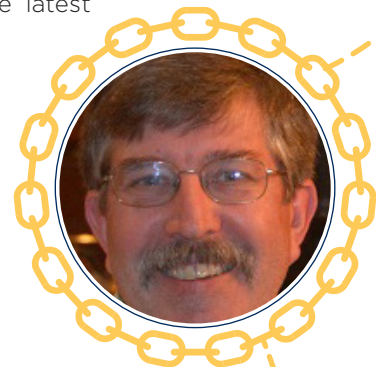


REGULATORY REVIEW PHASE

Potential treatments must undergo the Food and Drug Administration's (FDA) rigorous evaluation process, which scrutinizes everything about them — from the design of clinical trials to the severity of side effects to the conditions under which the drug is manufactured. During this phase, BDSRA helps educate the FDA as well as other governmental agencies on the course and severity of Batten disease, and the needs of families. Patient advocacy groups are often asked to appear before the FDA as part of Advisory Board Meetings and to bring families, care providers and others with them to educate review panels. As the Batten community waits on responses, trial progress, and other treatment developments, communication with families from BDSRA is key.

"We count on BDSRA for news about trials and research. As a Batten parent, as a donor and chapter leader I count on them as my go-to for the latest information."

- **Ron Hein**, bereaved Batten dad and Heart of America Chapter Leader



BECAUSE
OF YOU,
CHILDREN AND FAMILIES
HAVE MORE
HOPE THAN EVER

POST-APPROVAL PROCESS PHASE

After approval by regulating bodies, BDSRA will disseminate information about access to the treatment - where families can go, how they can enroll in treatment regimens and how they work through payment systems. Helping researchers follow patients as they progress with new treatments is vital, so that the natural history of patients through time is captured.

Research and treatments are part of a pipeline infused with support and hope by families.

THE EVOLVING
PARTNERSHIPS
BETWEEN PHARMA
COMPANIES
AND
PATIENT
GROUPS
HAS LED TO
IMPROVED
THERAPIES

“The relationship between pharmaceutical companies and patient advocacy groups is a critical piece in the drug development process. Patient groups provide first-hand knowledge of the disease, desired outcomes from new treatments, and can provide trial information to their members. Companies can assist with educational and financial resources to improve patient education, research updates, and support patients during clinical trials. The evolving partnerships between pharma companies and patient groups has led to improved therapies for patients and shortened drug development timelines.”

- **Barbara Wuebbels, RN, MS**, Member of BDSRA Board of Directors, Vice President of Patient Advocacy, Audentes Therapeutics



The Need for Early Diagnosis and New Born Screening (NBS)

We know that with most diseases, the sooner families know their loved one has a life-limiting illness, the better. As new trials start, as new treatments become available and new methods for symptom management emerge, earlier is better in hopes of keeping Batten disease at bay. We're working with partners in research, advocacy groups and industry to communicate this need and make an earlier difference.

BDSRA staff engaged stakeholders all over the globe in 2015

London, UK Boston, MA Spain Denver, CO
Philadelphia, PA New Zealand Australia Huntington Beach, CA
Portugal Hamburg, Germany St. Louis, MO Indianapolis, IN
Washington, D.C. **Pittsburgh, PA** Las Vegas, NV
Chicago, IL Baltimore, MD

Fundraising and Awareness

BDSRA is grateful for the hard work and time that so many families and volunteers spend organizing fundraisers. Whether it's a 5K, lemonade stand, garage sale, casino night or countless other events, they raise funds for research and support, and bring awareness to their communities.



"This is another way I can work with BDSRA to give back and remember all the kids who have passed away."

- **Laura Hull**, family friend and Columbus Battling Batten Disease 5K Run/Walk co-chair

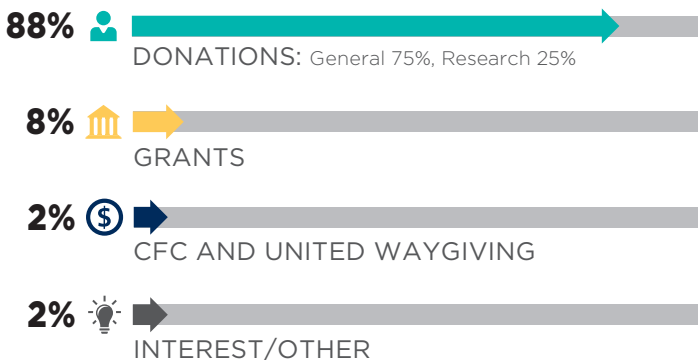
"As a leader in the Batten community, BDSRA is dedicated to driving a mission of hope for improved quality of life, connecting a rare community of families, raising awareness, and funding research for treatments and a cure. This is a vital organization for which we raise funds to honor Ethan's life and memory."

- **Kristine Davies**, bereaved Batten mom and Executive Director, Ethan's Reason

Photo (above): Davies Family: Kristine, Ethan, Maya, Eli, and Mason

Financials

REVENUES



RESEARCH AWARDS AND SUPPORT SERVICES

Support Programs — 55%
 Research — 29%
 Administration — 11%
 Fundraising — 5%



STAFF

Margie Frazier, PhD, LISW-S, Executive Director
mfrazier@bdsra.org

Becky Hetteberg, MA, MSW, LISW-S, Director of Family Support and Education
bhetteberg@bdsra.org

Tracy Kirby, Director of Development
tkirby@bdsra.org

Noreen Murphy, MA, Project Coordinator
nmurphy@bdsra.org

Board of Directors

Rob Geer, President
 Tracy VanHoutan, MBA, Vice President
 Joel Karg, CPA, Treasurer
 Mike Collins, Secretary
 Kristen Drago, RN, BSN, CHPPN

Tony Ferrandino
 Kate Haller, JD
 Donna Fogle
 Bob Jensen
 David Pearce, PhD
 Darlene Royalty
 Barbara Wuebbers RN, MS

Family Support and Advocacy

SUPPORT

Since the founding of BDSRA in 1987, providing programs, services and support to families has been a central focus of the organization. Whether it's carrier testing, educational consulting, guidance to newly diagnosed families, bereavement support, or the annual family conference, we strive to assist families with their needs.

56 NEW FAMILIES SERVED AND WELCOMED TO BDSRA COMMUNITY

"We are here and dedicated to serve families from the point of diagnosis through bereavement."

- **Becky Hetteberg**, BDSRA Director of Family Support and Education



"I am so happy we decided to come to the conference this year. There is so much support from everyone; I wish we had done it sooner."

- **Jill Wellner**, Batten mom



450 CONFERENCE ATTENDEES IN CHICAGO

ADVOCACY

"We must be the change we desire to see. 'Alone, we are small, but together, we stand tall...' This is my quote, my mantra and my passion for advocacy on the local, state and federal levels. It pushes me to continue in the fight when otherwise I may tire. Whenever you or a loved one is affected by a rare disease, the single, most effective thing you can do is advocate. Advocacy is a means by which better services are rendered and necessary changes are made for those who are affected."

- **Marlo Dean**, BDSRA advocate and Batten mom



REMEMBERING can be **HEALING** and **POWERFUL**

6

"ASK AN EXPERT" FACEBOOK CHATS

BATTEN DISEASESM
Support and Research Association

800-448-4570
www.bdsra.org