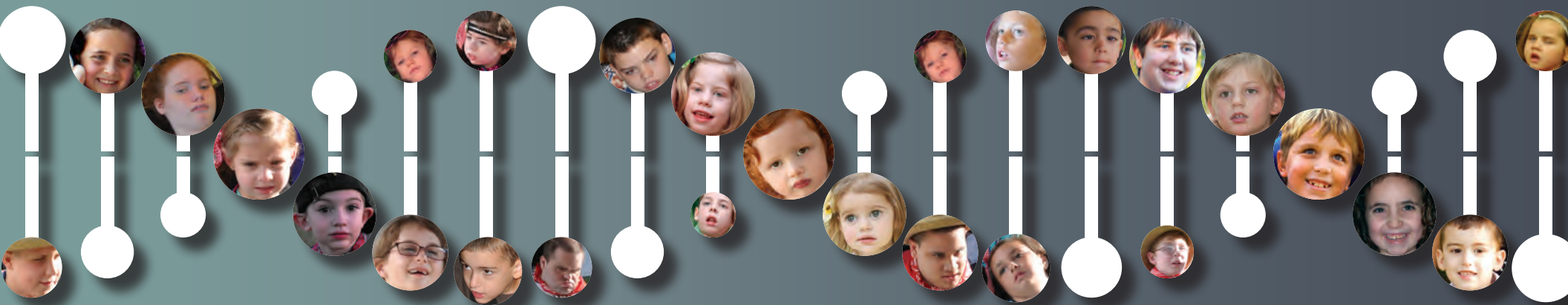


# Connecting Help, Hope and Need

2013 BDSRA ANNUAL REPORT



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## Batten Disease Support & Research Association

BDSRA is dedicated to funding research for treatments and cures, providing family support services, advancing education, raising awareness, and advocating for legislative action. Founded in 1987, by parents seeking to build a network for those diagnosed with Batten disease, BDSRA is now the largest support and research organization dedicated to Batten disease in North America. BDSRA believes that to effectively unravel the mysteries of Batten disease, the worlds of medical science, research, and families must work together toward a common goal: discover treatments and cures while assuring a better quality of life for those living with the disease.



# Letter from the Director

Dear Members and Friends,

This past year has been a time of growth, partnership, and opportunity.

We are excited to present this annual report to highlight the successes we see every day and to thank those who make it possible to serve and carry out our mission. BDSRA is a vital organization that brings families, industry, researchers and health professionals together to unravel the mysteries of Batten disease. Together we are a united force. Whether it be attending a dynamic family conference, a medical meeting, or community gathering, each of us brings a unique talent for sharing the stories of compassion, a passion for knowledge, and hope for discoveries that will ultimately end the tremendous human costs of Batten disease.

The collective vision of the BDSRA board, the membership, and our supporters is to continue the comprehensive programs and projects to discover treatments and cures, while assisting families who face each day with courage, hope, and strength. We are deeply grateful to those who commit their time and financial support to these efforts. The generosity of donors makes it possible to do the work that is so essential in raising awareness and providing information and support.

Highlighted here are stories of our dedicated partners, our latest scientific research awards, and the significant ways we help families. Thank you for all you do to make a difference in the lives of those coping with Batten disease.

**Margie Frazier**  
Executive Director





# BDSRA Boards and Staff

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*1<sup>st</sup> Vice President*

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*Senior Director of Advancement and Outreach*

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*Family Support Officer*

Chris Leonard, PhD  
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*Administrative Manager*

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Katherine Sims, MD  
Massachusetts General Hospital

David Sleat, PhD  
Center for Advanced Biomedical Research



Above: BDSRA Board Members: (front row, left to right) Barbara Wuebbels, Kim Zellmer, Mike Collins, Margie Frazier; (second row) Tony Ferrandino, Rob Geer, Chris Lowden; (back row) David Pearce, Chris Hawkins, Tracy VanHoutan. Not pictured, Craig Benson and Joel Karg.



Left: BDSRA staff: (front row) Becky Hetteberg, Tracy Kirby; Margie Frazier, Julie Conry. Not pictured, Chris Leonard.

# About Batten Disease

**A**s a rare, fatal, inherited disorder of the nervous system, Batten disease has no known treatment or cure. Batten disease is the most common, inherited neurodegenerative disorder found in children, and is also the most common form of a group of disorders called neuronal ceroid lipofuscinoses (NCLs). As a lysosomal storage disorder caused by genetic mutations, Batten disease primarily affects the nervous system. Batten disease occurs in approximately 2-4 births per 100,000 in the United States. There are multiple forms of the disease, which is most commonly diagnosed between infancy and school age. The three most common forms of Batten disease are infantile NCL, late infantile NCL, and juvenile NCL. An adult form of the disease is known as Kuf's. All forms of Batten disease result in a shortened life span, and those diagnosed have extensive needs for comprehensive medical care.

Those affected by the disease suffer progressive neurological impairment, seizures, blindness, and loss of motor skills and the ability to walk, talk, and communicate. The arc of the disease, from diagnosis to symptom management, can vary tremendously for each patient. The diagnostic process to identify Batten disease can often take years, as the disease is frequently misidentified in the early stages.

## Batten Disease Centers of Excellence

BDSRA has partnerships with major medical centers designated as Centers of Excellence for Batten Disease. Physicians, researchers, and healthcare teams are at the forefront in Batten disease management and resources.

- Nationwide Children's Hospital – Columbus, Ohio
- Massachusetts General Hospital – Boston, Massachusetts
- University of Rochester Medical Center – Rochester, New York
- Oregon Health and Science University-Doernbecher Children's Hospital – Portland, Oregon
- Hospital De Ninos De La Provincia De Cordoba – Argentina-Cemeco
- Texas Children's Hospital – Houston, Texas
- Institute for Mother and Child Healthcare of Serbia, Belgrade





# Programs, Services, Support and Outreach

Since the founding of BDSRA in 1987 by parents of children with Batten disease, providing programs, services and support to families has been a central focus of the mission of the organization. **Currently, BDSRA is the only family support organization in North America, the United States and Canada dedicated to serving newly diagnosed patients, families coping with care, and those who have lost a loved one to Batten disease.** With sister organizations in New Zealand, Australia, Spain, South America, and South Africa, BDSRA extends its support model to the global community for families and patients. With active BDSRA Chapters in states across the U.S. and in many nations, families in their local and regional communities connect, assist, gather and support each other. They continue to be significant partners in raising awareness, raising funds, and advancing the mission of the organization.

## Top 5 Most Requested Resources from BDSRA

- 1 Social support/Connecting to other Batten families**
- 2 Carrier testing**
- 3 Special education concerns and accommodations**
- 4 Disease specific information for newly diagnosed patients and families**
- 5 Respite care and equipment needs**

**Communicating with families** is a vital link that BDSRA provides in a world that increasingly depends on technology and web based and virtual systems for connections and dialogue. Through a closed facebook page with nearly 650 members, BDSRA facilitates a forum for parents, caregivers, and family members to discuss, share, and listen to one another about the daily challenges of Batten disease. This virtual community operates 24-hours a day to break the isolation experienced by Batten families and offer a lifeline to those seeking support and understanding.

Through specialized services such as educational consulting, carrier testing, one-on-one referral and guidance to newly diagnosed families, networking with community resources for respite care and equipment needs, and social and bereavement support, BDSRA strives every day to connect parents and caregivers to the information they need.

**The Annual Family Conference** welcomed 389 people to Nashville in July, 2013, for networking, research updates, and programming for siblings, grandparents, parents, and caregivers. This highly valued experience brings families together for emotional support, learning, sharing stories, and gaining information about research, medical care, and scientific discovery. Empowering families to make decisions and choices as they cope with Batten disease remains a primary goal for the organization and its advocates.

The BDSRA website [www.bdsra.org](http://www.bdsra.org), open facebook page, facebook.com/bdsra, and email update system enables the organization to provide timely and meaningful information across a range of topics for the Batten community. A quarterly newsletter, the Illuminator, delivered electronically and posted on the website, also provides numerous articles and updates about research, events, governance, advocacy, and programs.

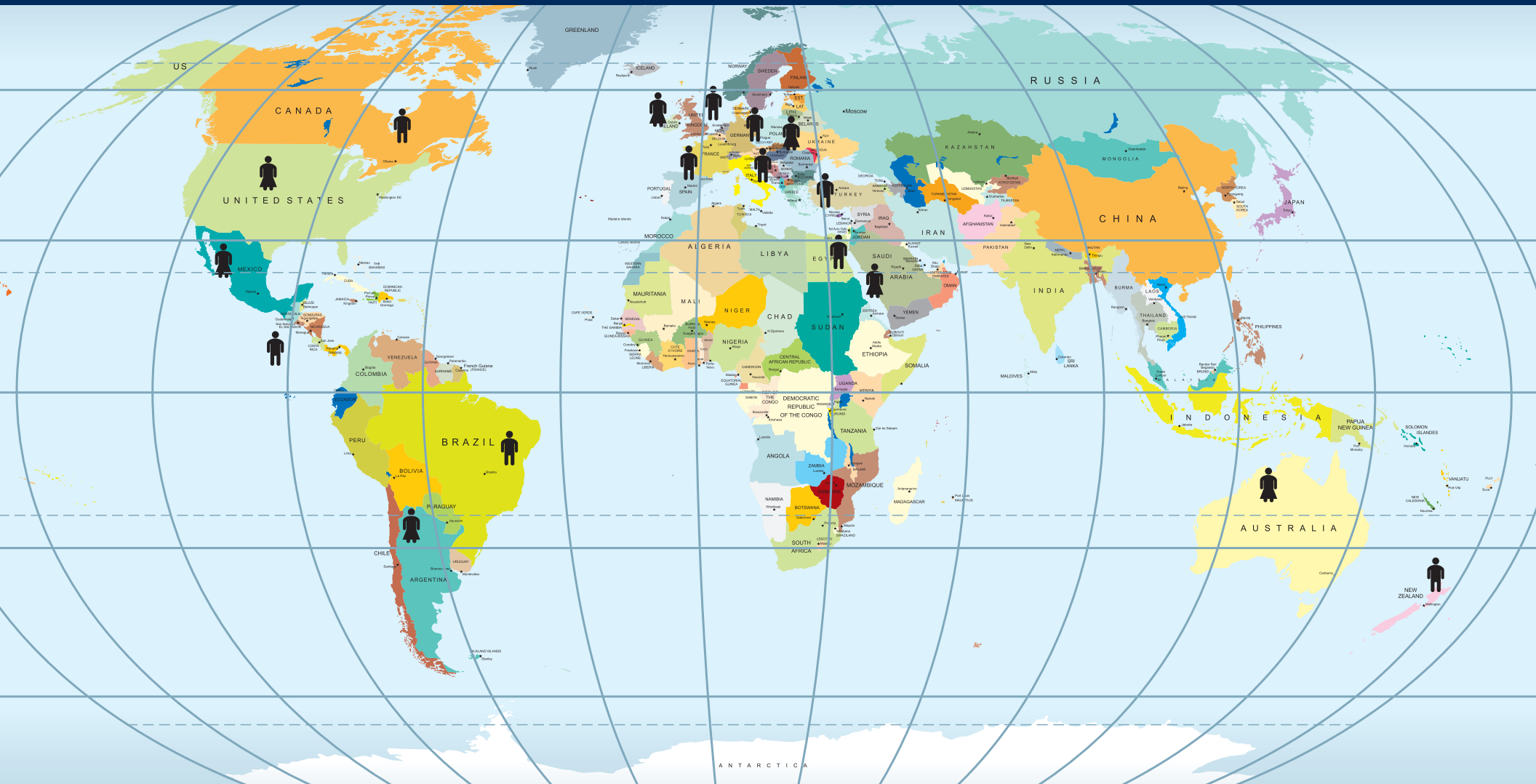
Specialized publications, brochures, information flyers, and fact sheets are available from the BDSRA national office addressing medical and educational topics and general information about Batten disease.



FORGET ME NOT

**Each day, we remember our community of families who miss their loved ones lost to Batten disease.** In 2013, there were 55 children known to BDSRA who passed away. We are deepened, but not diminished. We remember with sadness and hope, smiles and courage. BDSRA services and support continue to be part of each family's life as well after their loved one has passed. Through calls and help with community grief service referrals, we make known the need for families to remain whole and supported in their loss.





**BDSRA is a global organization, serving patients with Batten disease and their families throughout the world. In 2013, we touched lives in countries including the United States, Canada, Costa Rica, United Kingdom, Brazil, France, Ireland, Serbia, New Zealand, Argentina, Poland, Germany, Mexico, Oman, Australia, Saudi Arabia and Israel.**

# Funding Our Mission

**B**DSRA funding for services, research grants, and outreach is accomplished primarily through charitable donations. Families coping with Batten disease inspire volunteers across the U.S. and North America to dedicate hundreds of hours to support the cause of BDSRA, through events, through fundraising activities, and awareness building in their home communities. Whether it's a 5K, golf tournament, garage sale, fun run, or lemonade stand, our supporters are tireless in their efforts, and generous with their time to bring diverse groups of people together in the fundraising enterprise. We are grateful for the ongoing support, time, and efforts that volunteers devote throughout the year to raising crucial funds for research and services. At the end of each day, this generosity enables BDSRA to help families and empower families to help each other.

## Giving Highlights

**ANNUAL FUND** – The BDSRA Annual Fund is organized to generate special gifts to make services and programs available to those affected by Batten disease, and to fund research initiatives in the areas of highest priority. These gifts ensure a solid financial base for BDSRA to carry out the mission of the organization and serve the growing number of families linked by a Batten disease diagnosis. Annual fund gifts are also a meaningful way to honor or remember loved ones affected by Batten disease and those who care for them.



**WORKPLACE GIVING** – Throughout many cities and states in the U.S., generous donors contribute to BDSRA through workplace giving programs such as the CFC and United Way. In 2012 and 2013, more than \$18,825 was designated to Batten disease by individuals in these programs. Many donors also double their donations through the matching gift programs their companies provide for employees.



**RESEARCH FUNDING** – BDSRA manages a diverse pool of restricted funding to support research into the diagnosis, cause, and potential treatments for Batten disease. Every year these funds are distributed to researchers and scientists through a rigorous peer-review process that focuses on scientific merit and translational potential. Individual donations are leveraged through a vital set of collaborative partnerships with family foundations and international Batten groups to

advance the best scientific projects and studies. In 2012 and 2013, approximately \$1.3 million was allocated for research initiatives to scientists across the globe engaged in Batten disease research.



**BATTEN AWARENESS WEEKEND** – The first weekend in June every year is designated as Batten Awareness Weekend. Families and their advocates and supporters bring the message of hope and the awareness of the disease to thousands of people and inspire charitable gifts to BDSRA. Through social media campaigns, home town fundraisers, school and church events, and awareness projects, volunteers explain their connection to Batten disease and the importance of raising funds for support and research.

**BEQUEST AND ESTATE GIFTS** – Designating BDSRA in an estate plan or will is a simple way to create a sustaining legacy to support Batten disease services and research into the future and honor a special individual who has inspired others in facing the disease. Legacy funds can be named in honor or memory of a loved one. A named fund can also be dedicated and restricted for special purposes, such as research or programs, that the donor has a special interest in aligned within the mission of BDSRA.



**BDSRA CHAPTER AND COMMUNITY FUNDRAISERS AND EVENTS** – On many weekends during the year in the U.S. and in many countries, energetic and passionate volunteers gather to devote time and talents to the cause of raising funds for Batten disease research and services. Special events and community based fundraisers organized through BDSRA Chapters, neighborhoods,

families, and friends of those with Batten disease generate crucial revenue and resources. In 2013, BDSRA Chapters in the U.S. and Canada raised and donated \$120,117 for Batten disease. With this support, partnership, and charitable base, BDSRA is able to continue its work to accomplish important goals in research, advocacy, and service. The dedication of so many donors and volunteers makes it possible to keep searching for new scientific breakthroughs as families hold on to hope.



Sophie and her piggy bank championed the 2013 Annual Fund.





**CORPORATE AND FOUNDATION GRANTS** – Special projects and focused programs are made possible for BDSRA by the support of special grants from foundations and corporations. In 2013, BDSRA was awarded a Genzyme Patient Advocacy Leadership Award (PAL) to create multicultural practice guidelines for Batten disease. The Genzyme global grant program supports outreach programs by nonprofits that work on behalf of patients living with lysosomal storage disorders. With the guidelines project, BDSRA hopes to improve the diagnostic efficiency and disease management for Batten patients by producing practice guidelines for medical professionals around the globe. The Wawa Corporate Charities program of Pennsylvania also supported this project.

**MEMORIAL AND HONORARY GIFTS** – Designating special gifts to BDSRA to honor or memorialize a loved one affected by Batten disease has a long tradition within the organization. In 2013, 67 percent of all charitable dollars donated to BDSRA were in memory or honor of a special person. We strive every day to bring the inspiration, courage, and hope of so many to the work that is done on behalf of families and children. BDSRA is committed to this important link between past and future, so that all who have faced Batten disease are remembered and their stories shared.





# Batten Disease Advocacy and Awareness

For the last two years, BDSRA staff, board members, and parent advocates have traveled to Washington, D.C. to meet with legislators and appear before committees to push for accelerated drug discovery, increased services for those with rare disease, and improved FDA processes and other initiatives important to our families. Along with the Rare Disease Legislative Advocates, we join with other patient advocacy groups to speak for those who cannot.



BDSRA board, staff, and volunteers have also advocated across state legislatures during Rare Disease Day and Batten Awareness Weekend to raise awareness and increase research related funding for NCL studies. Parents of children with Batten disease have testified to committees in numerous state legislatures on a range of issues, including access to medications, accessibility, and support for services to those caring for patients with life limiting illness.

In 2013, Batten disease advocates appeared on program panels of the National Institutes of Health and National Institute of Medicine to discuss medical and ethical ramifications of gene therapy in rare disease research. In February 2014, our Batten team from BDSRA, Taylor's Tale and Noah's Hope visited over 40 Congressional offices on Rare Disease Day to talk about the care needs of Batten children and push for drug discovery.



BDSRA staff welcomed Noah Coughlan to Columbus, Ohio, during his second coast to coast run in October, 2013. Left to right: Julie Conry, Tracy Kirby, Lance Johnston, Noah Coughlan, Margie Frazier, and Becky Hetteberg gather during a reception in Ohio for families to greet Noah when he passed through the Buckeye state. In a span of 108 days, Noah Coughlan covered 3,100 miles. He departed from Half Moon Bay, California, in July and arrived in Boston, Massachusetts in November in a cross country journey to raise awareness for Batten disease.





# BDSRA – Connecting Help, Hope and Need

Trina and Bob Rodgers of Boise, Idaho followed a typical path for many couples after meeting and getting married 14 years ago. Both born and raised in the Boise Valley, their focus on family and service kept them busy caring for daughters Paige, 11, and Hannah, 9. Their hours were filled with activities that surrounded the two sisters and their involved and happy school days. That changed abruptly in 2011, when the family received the devastating news that Hannah had Juvenile Batten disease.

In the three years since Hannah's diagnosis, the Rodgers' have faced enormous challenges in seeking services and providing the best environment they can create for their children. At several turns they sought out the help of BDSRA to assist with school issues, adjustments, and medical needs. In their mind, it takes a village to care for a child with Batten disease, and BDSRA is a big part of that village.

"Some of my biggest challenges being a Batten mom is juggling it all," Trina says. "Trying to keep everyone satisfied and content, while raising a typical child to be a happy, successful adult, while caring for another child who is losing everything."

"Bob's biggest challenge is having more patience than he ever thought he could muster. Paige's biggest challenge having a sibling with Batten disease has to be that she has had to give up being 'just a sister' and become part of the collective family unit caring for a sick child," Trina notes. "Hannah's biggest challenge is feeling like no one likes her and being blind."

Last year the Rodgers' asked BDSRA for help to work with their school and the teachers who were struggling to meet Hannah's needs in the classroom. With no other Batten children in their community, the staff had few resources to turn to. "They had never seen anything like Batten before, and they would try techniques they use for kids with autism, or Down syndrome, none of which worked," Trina recalls.

Working with BDSRA, an educational specialist with Batten disease expertise met with the Boise teachers and staff to provide training and techniques for the classroom. Small changes soon made a huge difference for her daughter. "She hated school, felt she was dumb, less than the other children. But since they have been taught how to care for her and make her feel special and creative, school is a much happier place for her now," Trina says.

"BDSRA has helped us in so many ways, helping us connect to other families going through the same things we are," she adds. "Knowing we are not the only ones dealing with the side effects of the disease has given me great strength."

For her family, these links to others working through similar problems has transformed their daily outlook. "Besides bringing a specialist here to educate the school staff, I am most grateful to BDSRA for helping us connect with fellow Batten parents," Trina says. "No matter how many diseases someone is familiar with, they just can't understand the daily struggles our family goes through like a fellow Batten parent does," she notes. "We are a community coming together for our children and the children yet to be diagnosed."



The Rodgers family: Trina, Bob, and daughters Paige and Hannah

*"Besides bringing a specialist here to educate the school staff, I am most grateful to BDSRA for helping us connect with fellow Batten parents. No matter how many diseases someone is familiar with, they just can't understand the daily struggles our family goes through like a fellow Batten parent does..."*

# Hope 4 Bridget Family Foundation Sees the Power of Partnership

*"We knew we needed to make this our mission, to raise funds to support the researchers investigating Batten disease.*

*We also knew we could not do this alone, so we began networking and conversing and working with BDSRA..."*



The Kennicott family: Sara, Dave, son Harrison and daughter Bridget

Bridget Kennicott was born on November 11, 2004, to proud parents Sara and Dave and big brother Harrison. The Carpentersville, Illinois family felt complete, with their energetic little daughter soon known as "Miss Busy B." She barreled her way into her toddler years, until the family met their first medical puzzle in 2008. With no warning, the Kennicotts faced the frightening experience of carrying their daughter to the emergency room with a seizure. It would be 16 months of testing, consultations, medications, and questions before a diagnosis in April of 2009 could be definitive that Bridget had late infantile Batten disease (LINCL).



According to Sara, "there is no preparation for devastating news like this. We knew that our little girl struggled with health and development, this, however, was far beyond our imagination."

"Our journey with Bridget is to take one day at a time," says Sara. "We realize our efforts to help find a treatment may be too late for Bridget, but we forge ahead to fight for other children like Bridget."

Through their shock, grief, disbelief and pain, the Kennicotts were determined to seek answers and find ways to bolster Batten disease research, which they discovered was underfunded and limited by being categorized as a rare disease. Shortly after Bridget's diagnosis, they created the Hope 4 Bridget Foundation to raise money for research and education in efforts to find a cure for late infantile Batten disease.

"We knew we needed to make this our mission, to raise funds to support the researchers investigating Batten disease," Sara notes. "We also knew we could not do this alone, so we began networking and conversing and working with BDSRA," she says. "Through these relationships we began to learn about the scientists and laboratories that needed funds to further their research that could lead to a treatment for LINCL. Working alongside BDSRA kept us up to date in regard to what research was out there."

As a member of the collaborative of family foundations that join with BDSRA in funding grant recipients through the RFP process, Hope 4 Bridget has provided crucial funding for a range of research in LINCL. Drug discovery, identifying biomarkers, enzyme replacement therapies and developing translational projects for Batten disease have all been supported by foundation grants totaling nearly \$200,000 in the last five years. It is a partnership that has generated tangible advances toward treatment, and it has sustained their hope that someday Batten disease will have a library of treatments available that their daughter does not have.

"In this partnership, we can accomplish so much more when we work toward the common good. We find this comforting – knowing that it is a collective group – there is power in numbers and a common goal," Sara says. "We are not alone out here fighting Batten disease. We have support, love, friendship, and the most relevant resource for finding projects to fund through BDSRA."

"It is a blessing that we can work together in order to find a treatment and ultimately a cure so that no more children will have to suffer through this horrible disease," she adds. "We will always have hope for a cure. We will always have hope for Bridget."





## Searching for Treatments and a Cure

**W**ith the introduction of BDSRA's current Scientific Merit Review Process in 2012, we integrated an assessment system for grant proposals based on scientific merit that reflects the highest standards associated with the U.S. National Institutes of Health, the European Research Council, and industry-standard drug discovery processes.

Since Batten disease research has historically been severely underfunded through public channels, private funding for innovative and translational research has greatly advanced through the active partnerships of BDSRA with private donors, family foundations, and European Batten organizations. Due to the rare nature of Batten disease, funding models are continually challenging. Progress is being made to generate more scientific collaborations and funding for promising researchers to leverage BDSRA grant monies and obtain funding from the NIH and other agencies.

The first research grant from BDSRA was awarded in 1992, and since then more than \$6.5 million has been allocated to scientists and research laboratories across the globe. Funding of basic discovery to identify drug targets, translate those discoveries into treatment, and identify biomarkers capable of measuring the efficacy of potential drugs to treat Batten disease has been a major focus. To turn cellular problems into medical solutions, BDSRA is funding investigations into direct treatments such as enzyme replacement therapy, gene therapy, and drug discovery.

To prepare and advance clinical trials, a major priority for BDSRA in the past two years has been funding a project to expand a Batten patient registry. Currently in use in Germany, Finland, Italy, India, and the U.K., the registry, through BDSRA support, will be expanded to include the United States, Brazil, Argentina, Turkey, France, Norway, and Denmark.

Why is a registry important to Batten disease research? Because the disease is so rare, no single country or organization can marshal sufficient numbers of patients to conduct generalizable clinical and translational research. The geographic isolation of patients has been a major challenge in recruiting patients into clinical trials. Identifying patients and gathering genetic test results and clinical status parameters for the many forms of Batten disease is time intensive and expensive. The DEM-CHILD registry project, led by Angela Schultz, MD, of the Children's Hospital and University Medical Center Hamburg-Eppendorf, with BDSRA funding support, aims to collect the world's largest, clinically and genetically best characterized set of NCL patients.

Scientists and pharmaceutical companies are more likely to conduct research on a given rare disease if they find a patient registry in place. Registries also assist in:

- Understanding the disease
- Collecting natural history data on patients to track symptom progression and type
- Recruiting patients for clinical trials
- Tracking genotype and phenotype associations
- Safety monitoring
- Identifying novel biomarkers and modifiers
- Developing novel therapies for NCLs

## RESEARCH GRANTS AWARDED FOR 2013

BDSRA, with the collaboration of our international and family foundation partners (BDSRA - Australia, Batten Disease Family Association of the United Kingdom, Noah's Hope, (Chicago) and Hope 4 Bridget, (Chicago), awarded \$408,000 in research grants for 2013 that spanned the United States, United Kingdom, Australia, and Germany.

Proposals were funded that focused on CLN1, CLN2, CLN3, CLN5, CLN6, CLN7, and the registry and chart review project that will include data on CLN8, CLN10, and CLN14.

### 2013 FUNDED INVESTIGATORS



**David Palmer, PhD**

Lincoln University, New Zealand

Targets: CLN5, CLN6

Project: "Viral mediated gene therapy in ovine Batten disease"

This study is a gene therapy trial of CLN5 and CLN6 in the affected sheep flocks that reside in New Zealand. This pre-clinical work is directly translational and essential to determining if gene therapy approaches will work in these CLN forms. In addition, as a membrane bound protein, CLN6 may also provide some insight into the potential of gene therapy approaches to CLN3.



**Stephan Storch, PhD**

University Medical Center Hamburg-Eppendorf, Germany

Targets: CLN7

Project: "Phenotypic analysis of a mouse model for CLN7 disease"

The CLN7/MFSD8 gene encodes an integral lysosomal membrane protein, which belongs to the Major Facilitator Superfamily (MFS) of transporters. The function and the substrates of CLN7 are, as yet, unknown. The aim of the project is to characterize pathological changes in the brain and in the kidney in a mouse model for CLN7 disease.



**Erika Augustine, MD**

University of Rochester

Target: CLN3

Project: Rochester CellCept Trial

Third year of funding from BDSRA for a JNCL clinical trial to learn if Mycophenolate (CellCept) is safe and well tolerated in children with Juvenile Neuronal Ceroid Lipofuscinosis.



**Angela Schulz, MD**

University Medical Center Hamburg-Eppendorf, Germany

Target: All forms of NCL

Project: "Retrospective chart analyses to develop a tool for the evaluation of experimental therapies in NCL"

This Batten disease patient registry project seeks to perform large-scale retrospective chart analysis in all forms of NCL. It will use existing clinical scoring systems developed for use in retrospective analyses of disease progression, improve and adapt existing scales

to score follow-up examinations of different kinds, and develop new scales to evaluate the effectiveness of various palliative care measures.

There is a wealth of clinical data on NCL patients globally that needs to be collated and organized into a central database to provide baseline data for future therapy development. This data is critical to involvement from pharmaceutical companies in clinical trials for treatments and drug delivery systems.



**Jonathan Cooper, PhD**

Institute of Psychiatry, King's College London, UK

Target: CLN1, CLN2

Project: "Human cell based models of INCL and LINCL"

This project will develop human cell-based models for NCL to aid investigations to better understand the cellular and molecular basis of these diseases and to generate a platform that can subsequently be developed for drug discovery. This project also extends work on a human neural stem cell (huNSC) models for the juvenile form of NCL (JNCL), for infantile (INCL) and late infantile (LINCL) forms.



**Jeffery Gerst, PhD**

Weizmann Institute of Science, Israel

Target: CLN3

Project: "Development of a cell-based assay for identifying down-stream effectors of CLN3 and use in drug screening"

This funding continues a second year from BDSRA for fellowship funding.

The graduate student in the Gerst lab will be leveraging observations made about the yeast BTN1 gene (the yeast version of CLN3) in mammalian cells to look at the downstream impact of CLN3 mutation on similar functions. The goal of the project is to discover biomarkers that can be used in the screening for small molecules that ameliorate deficiencies in CLN3 function. This will allow investigators to obtain pharmacological leads that may prove effective in the treatment of Batten disease.



## RESEARCH GRANTS AWARDED IN 2012

### David Sleat, PhD

University of Medicine and Dentistry of New Jersey  
Target: CLN1, CLN 2, CLN3, and Adult Batten disease  
Project: "Lysosomal biomarkers in Neuronal Ceroid Lipofuscinosis"

### Sandra Hofmann, MD, PhD

University of Texas Southwestern Medical Center  
Target: CLN1  
Project: "Enzyme replacement therapy for PPT1-related NCL"

### Jeffrey Gerst, PhD

Weizmann Institute of Science, Israel  
Target: CLN3  
Project: "Development of a cell-based assay for identifying down-stream effectors of CLN3 and use in drug screening"  
Funded for 2012 and 2013

### David Palmer, PhD

Lincoln University, New Zealand  
Target: CLN3  
Project: "Molecular dissection of neuro-inflammation in ovine Batten disease"

### Sreeganga Chandra, PhD

Yale University  
Target: Adult Batten disease  
Project: "Molecular basis of DNAJC5-mediated neuronal ceroid lipofuscinosis"

### Mark Schultz, BS

University of Iowa  
Target: CLN3  
Project: "CLN3 regulation of Cdc42 and drug screening for JNCL"

### Jonathan Cooper, PhD

King's College London, UK  
Target: CLN3  
Project: "Cell based systems for drug discovery in JNCL"

### Colleen Stein, PhD

University of Iowa  
Target: CLN3  
Project: "Does restoration of CLN3 to endothelial cells alleviate neurological symptoms in JNCL mice?"

### Angela Schulz, MD

University Medical Center Hamburg-Eppendorf, Germany  
Target: CLN1, CLN2, CLN3, and Adult Batten disease  
Project: "The DEM-Child database network – an online NCL patient registry for clinicians, researchers, and parents"

### BDSRA Co-funders for the 2012 grants were:

- Batten Disease Family Association - UK
- BDSRA - Australia
- Noah's Hope
- Beyond Batten Disease Foundation
- NCL - Stiftung
- Bee for Batten Ireland
- BioMarin
- Hope 4 Bridget

## RESEARCH STUDIES AND CLINICAL TRIALS FOR BATTEN DISEASE

Research studies and clinical trials led by the Department of Genetic Medicine at Weill Cornell Medical College in New York City, by the University of Rochester Batten Center, and by pharmaceutical company BioMarin continue to investigate Batten disease at sites in the United States and Europe.

■ **BioMarin** launched a Phase I/2 clinical trial in September 2013 to determine the safety and efficacy of rhTPP 1 (BMN 190), an enzyme replacement therapy for the treatment of children with late infantile CLN2 disease. BMN 190 is a recombinant human TPP 1 enzyme in development by BioMarin as an enzyme replacement therapy for CLN2 patients. Patient inclusion criteria include diagnosis of CLN2 disease, and mild to moderate disease documented by two domain scores of 3-6 on motor/gait and language domains of the Hamburg scale with a score of at least one point in these two domains, and seizures are stable. Patients who have received stem cells, gene therapy, or enzyme replacement therapy are not eligible for the study. Patients enrolled in the study follow a 12 month protocol of treatment.



The primary study objectives are to evaluate the safety and tolerability of BMN 190 administered to patients with CLN2 and to evaluate effectiveness using a CLN2-specific rating scale score in comparison with natural history data after 12 months of treatment. The study sites are currently in Germany and the United Kingdom. Updates and study information are available at [www.clinicaltrials.gov](http://www.clinicaltrials.gov).

### ■ The Department of Genetic Medicine at Weill Cornell Medical College in New York City

is conducting a gene therapy study for the CLN2 gene. Patients with the diagnosis of late infantile neuronal ceroid lipofuscinosis who score between a 6 and 10 on the LINCL scale in the early stages of the Batten disease are eligible for the study. The purpose of the study is to determine whether gene transfer surgery, in which an experimental drug called AAVrh.10CUhCLN2, is administered to the brain, can be achieved safely and whether the procedure will slow down or halt the progression of the disease. The study site is located in New York City, with study information available at [dem2026@med.cornell.edu](mailto:dem2026@med.cornell.edu).



Weill Cornell Medical College

### ■ The University of Rochester Medical Center

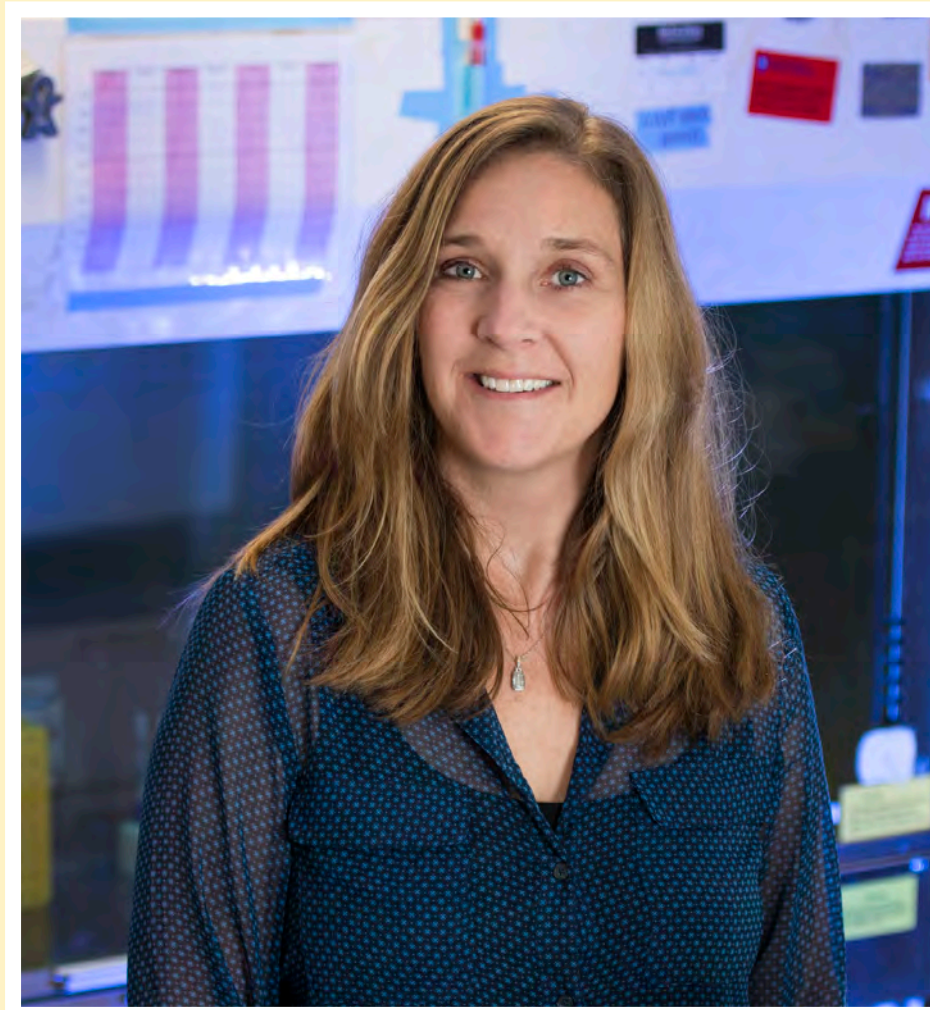
continues a JNCL clinical trial to learn if mycophenolate (CellCept) is safe and well tolerated in children with juvenile neuronal ceroid lipofuscinosis. The JUMP study focuses on evaluating CellCept and its effects on the symptoms of JNCL. These symptoms include unusual movements, seizures, problems with learning or behavior, or difficulty performing daily tasks. To be eligible for the study, which lasts 22 weeks, patients must be diagnosed with JNCL, must be able to walk 10 feet unassisted, be able to swallow liquid medication, have a local doctor who is willing to conduct the study, and be able to complete four study visits to the University of Rochester Medical Center. Study information is available by emailing [batten@urmc.rochester.edu](mailto:batten@urmc.rochester.edu).



# Early Research Funding from BDSRA Drives Innovation for Batten Disease

## SUSAN COTMAN

As a native Ohioan, with both her bachelor's and doctorate degrees from Ohio State University, Susan Cotman had never heard of Batten disease until she moved from Columbus to Boston thirteen years ago. Now an Assistant Professor of Neurology at Harvard Medical School and Assistant in Neuroscience at the Massachusetts General Hospital, Cotman spent nearly a decade at OSU working on projects related to brain development. It was a chance meeting at a conference in 2000 with researcher Marcy



Susan Cotman, PhD

MacDonald that would shape the direction of Cotman's scientific work in fundamental ways toward Batten disease and translational NCL research.

"I was looking for a post-doctoral lab, and met Dr. MacDonald, of Massachusetts General Hospital, who was working on Huntington's disease, but also had an NCL project started in her lab," Cotman recalls. She left Columbus for Boston to pursue her postdoctoral training and the NCL project, which created the foundation for her NCL research discoveries over the last dozen years.

"It was in Dr. MacDonald's lab where I created the first genetically accurate mouse and neuronal culture models of NCL," Cotman said. These are now in use across more than ten research labs worldwide for basic lysosomal biology, lysosomal disease, and neurodegenerative disease studies, she adds.

In 2001, Cotman applied for and received fellowship research funding from BDSRA. It was critical support at a time in her research development that Cotman credits with enabling her projects to grow and expand. She now leads an NCL research laboratory within the Center for Human Genetic Research at Massachusetts General Hospital. The primary research focus of the lab is on expanding the genetic model systems of NCL through mouse and neuronal cells lines and NCL patient-derived induced pluripotent stem (iPS) cells to investigate the damage of disease mutations and the NCL protein functions.

According to Cotman, this work has led to collaboration with research groups around the world to use the NCL models and other tools developed in her lab for both basic and drug development research in NCL and related disorders. The lab is also working with local and world leaders in cutting-edge genetics research to identify novel genetic causes of NCL. This, she adds, is leading to new insights into the overlap between NCL and other genetic diseases, including Parkinson's disease, frontotemporal lobar degeneration, and mitochondrial disease. From 2009 to 2012, BDSRA grants also provided additional funding for the NCL iPS cell studies.

"The role of foundations in helping advance science is critical, and I am proof of that," Cotman notes. "Without the support of BDSRA, the NCL research would not have happened, it was critical for me." For a young investigator, moving into a new disease area can be very overwhelming, she adds. "When you are welcomed into it the way I was welcomed by BDSRA and the Batten community it is invaluable and a huge motivation to choose that path to study," she says. "It solidifies the choice to enter this community as an area of focus for the scientist."

Outside the lab, her role in NCL research includes serving on the BDSRA Scientific Advisory Board and the Board of Scientific Counselors for the NIH National Institute of Arthritis and Musculoskeletal and Skin Diseases. With Dr. Katherine Sims she is also co-director of the Massachusetts General Hospital-Center for Human Genetic Research Joint Program in the NCL Disorders. For Cotman, it is an exciting time in science to be pursuing CLN3 and JNCL studies, and she sees the increasing collaboration between labs as a significant step in moving research forward faster. "The network has really



# Labs

expanded among the science community in NCL,” she notes, “and we want the pace to move forward faster.” The clinical trials emerging for Batten disease are also a great benefit to the basic science labs, she adds. “We see the natural history studies and the benchmarks for success from the clinical trials and this absolutely helps.”

Currently, her funded research projects include studying the function of the CLN3 protein and pathway involving the lysosomes and other compartments within the cell. “The exciting advance is that the iPS cells we have made for CLN3 are allowing us to look through the earliest stages of the disease in patient cells, brain cells and eye cells,” she explains. “We can study the progression of events and design different drugs to throw on the cells.” Cotman received an R01 grant in 2011 through the NIH for \$1 million to investigate the role of CLN3 (the protein encoded by the gene mutated in juvenile NCL) in the endocytic pathway.

## **TAMMY KIELIAN**

For Tammy Kielian, PhD., professor of pathology and microbiology at the University of Nebraska Medical Center, BDSRA funding for research she pursued in 2012 investigating the link between microglia, reactive oxygen species and inflammasome activation in JNCL, has led to a two-year, \$500,000 National Institutes of Health grant for pre-clinical testing of therapeutic compounds for juvenile Batten disease.

Kielian, an accomplished researcher whose lab studies immune responses in the brain, shifted course to pursue Batten disease projects in 2011 when her niece was diagnosed with juvenile Batten disease. “Up until two-and-a-half years ago, I had never heard of Batten disease,” she said. In 2012, she contacted both David Pearce, PhD, at Sanford Children’s Health Research Center in Sioux Falls, South Dakota, and BDSRA, to propose a collaboration. Her research expertise was refocusing on finding answers to the disease that had struck so close to home.

Through access to mouse models with mutated CLN3 genes from Pearce’s lab and project funding from BDSRA, Kielian turned her research passion toward JNCL. She found that the two types of brain cells she studies, the microglia and the astrocyte, activate early in the CLN3 mice. This early activation may contribute to eventual neuron death, according to Kielian.

Through the NIH grant, her lab is now testing two classes of drugs that have an anti-inflammatory activity, which may delay the destructive path of JNCL. The study is utilizing mouse models with Batten disease, to test if their outcomes are improved following treatment. The goal is to prolong the survival of the neurons, and, ultimately, slow the disease itself. Kielian, who has received Nebraska’s Distinguished Scientist Award and an Emerging Inventor Award, spent a decade focused on brain research, never expecting the news that changed her family forever. Her realization that her research expertise might be applicable to juvenile Batten disease has generated an innovative path of discovery that continues to expand through the network of BDSRA scientists and funders.

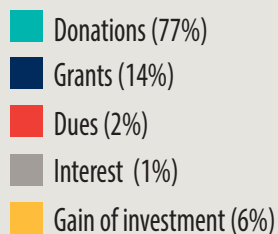
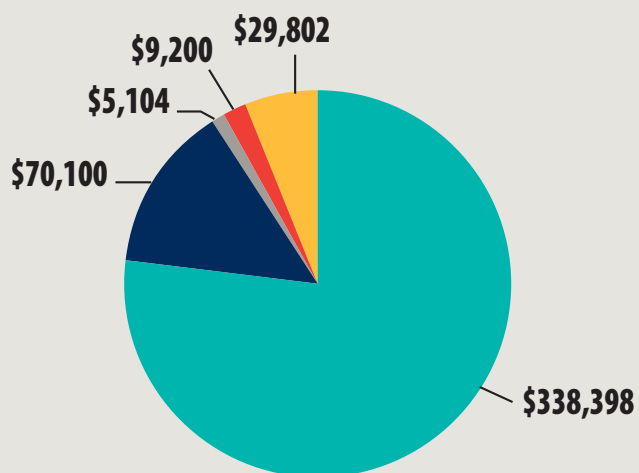


Tammy Kielian, PhD

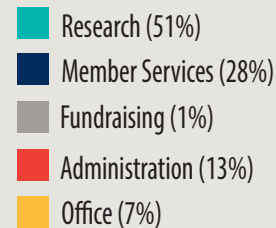
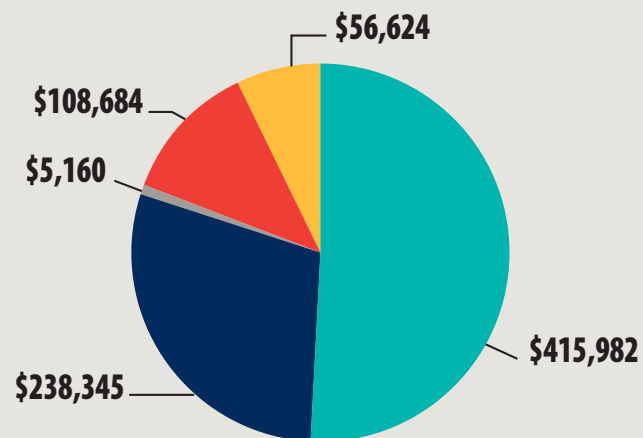
# Financials

The accompanying financial information for 2013 has been summarized from the financial statements of the Batten Disease Support and Research Association. The audited financial statements are available for review at the BDSRA office at 1175 Dublin Road, Columbus, Ohio.

## REVENUES



## RESEARCH GRANTS and SERVICES EXPENSES





# With Gratitude

**BDSRA is grateful for the continued charitable gifts made by many generous donors to the organization to support its mission. Between January 1 and December 31, 2013, our donors made gifts for research, programs, family services and awareness. The Batten Disease Support and Research Association wishes to express its deepest appreciation to those who keep Batten disease in the forefront of their philanthropy. The important work of serving families and searching for treatments and cures would not be possible without this generosity.**

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## Partners and Volunteers

The vital volunteer work and efforts of so many across the U.S. and Canada to raise funds and awareness through events is a significant partnership for BDSRA. We are thankful for all those who host garage sales, lemonade stands, 5k's, walks, auctions, golf tournaments and so many other events throughout the year to contribute to research and services. Our special thanks to the organizers and supporters of these 2013 events for Batten disease.

A Cure for Nick Golf Outing

Asher Bash

Bash 4 Batten's

Batten Blue Print

Battin' for Batten

Battling Batten Disease 5K

Beating Batten 5K Run and Walk

Benefit for Kate

Boston's Battle

Bringing Sammie Smiles

The Calerra Elks Lodge 2703 and  
Becky Lucas

Davies Garage Sale

Golf to Cure Batten Disease

Greater Atlanta Girls' Choir Concert

Haylee Joyce Battin' for Batten Disease  
Softball Tournament

The Lugine Hein Family

Hoops for Hope

Kick Batten's for Carl

Laps for Life Fun Run and Walk

Loopy for a Cause

Mackenzie's Hope Zumbathon

Maugansville Ruritan Club

The Medley Family

The Melissa Froio Foundation

Mike's Beer Bucket Golf Outing

Miles for Michael

No Show Golf Outing

No Show Super Bowl Party

Our Boys 5K/10K

Red Land High School Alumni Association

Run For Broxton

Run the Creek 5K Race and Walk

Running Towards a Cure 5K for Kate

The Surrey Family 6TH Annual Yard Sale

Trottin' for Taylor



# Couple's Fund in Memory of Son Continues to Help Others

Marty and Vicki Lumm might not have traveled the same road as other parents whose children have been diagnosed with serious illness.

But they have common ground.

They understand the helplessness that a mother and father feel as disease wreaks havoc on small bodies. They know, firsthand, that rare disorders don't just strike people you read about in a magazine. They strike your loved ones.

They all share the dream that one day a cure will be found for fatal diseases that rob infants and children of the opportunity to live full lives – play baseball, go to college, get married and have a family of their own.

And, often, they turn to each other for support.

When the Lumms' son Kevin was diagnosed with Batten disease at the age of 10, his illness and treatments presented the family with many emotional and financial problems, his mother said.

A degenerative neurological disorder that typically strikes children, Batten causes seizures, progressive loss of sight and motor skills and developmental regression.

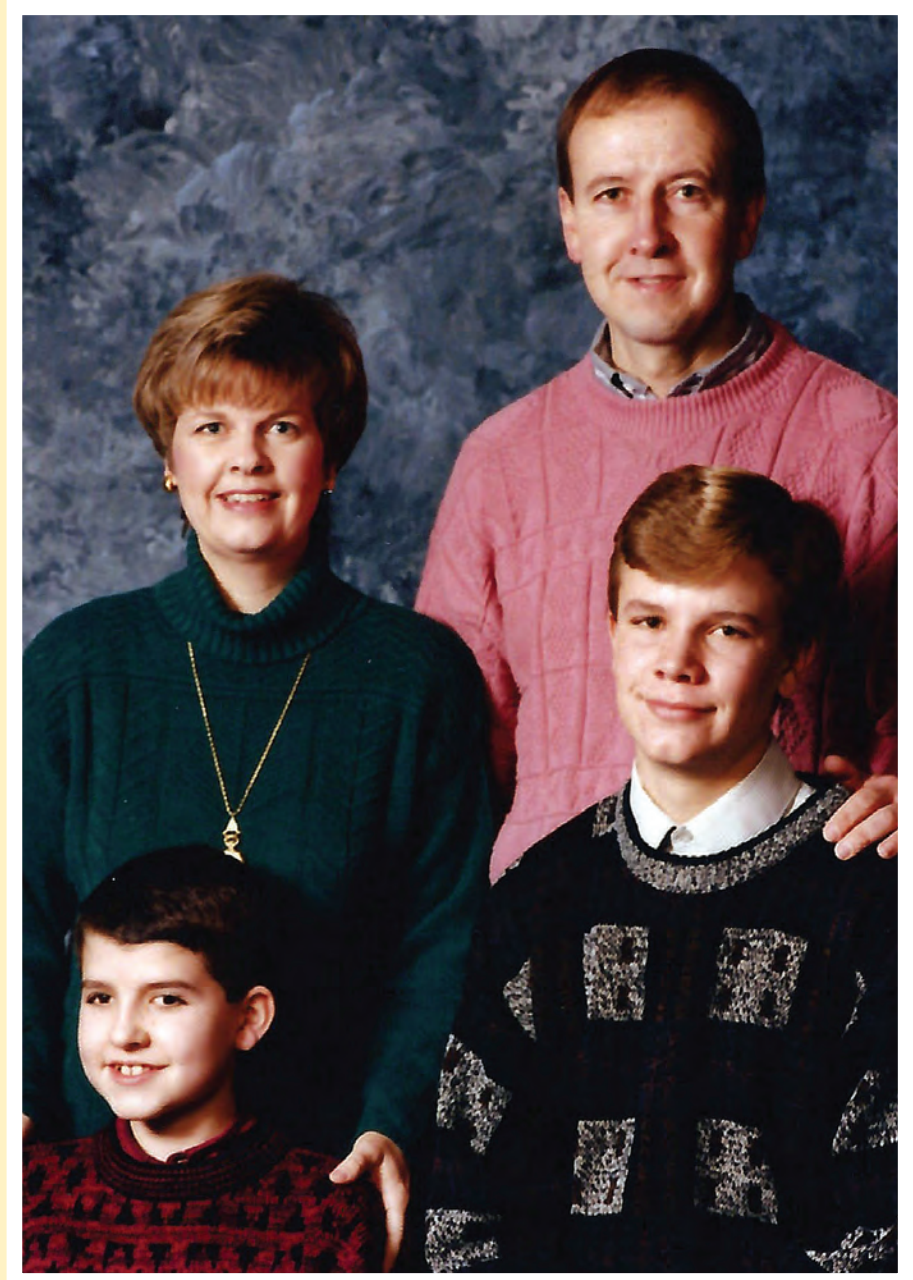
The most devastating fact: there is no cure.

During the years their son lived with the rare disease, the Lumms said they were helped by family and friends, their church, and the Maugansville Ruritan Club. Because of their gratitude for the support they received, following Kevin's death in 2003, they Hagerstown, Maryland couple began looking for ways to honor their son's memory.

"Several members of the Maugansville Ruritan Club suggested starting a memorial fund to assist children with special needs and the Batten Disease Support and Research Association (BDSRA), said Marty Lumm.

That was when the Kevin Lumm Memorial Fund, or Kevin's Fund, officially was established.

In June, 2004, a golf tournament was held as a means of raising money for Kevin's fund, Marty Lumm noted. Since then, it has become an annual event. Throughout the years, Kevin's Fund has raised almost \$119,000 and distributed about \$90,000. Vicki Lumm said that proceeds earned from the golf tournament go to BDSRA to support research and treatments for the disease. A portion also is dedicated locally to assist families of children with special medical problems.



The Lumm family: Vicki and Martin and sons Kevin and Brian



## The fund's namesake

Kevin was born in Hagerstown on June 17, 1983, but was not diagnosed with Batten disease until he was 10 years old, his mother said.

"However, he began having night vision problems at 4 ½ years of age," she said. "Vision doctors originally diagnosed him as having retinitis pigmentosa, but within two years he was almost completely blind. Learning difficulties and seizures began around the age of 10; and after a visit to the Maryland School for the Blind, plus trips to a Johns Hopkins neurologist where a tissue sample was taken, he was diagnosed with neuronal ceroid lipofuscinosis (NCL), commonly called Batten disease."

"Each child born to us had a one in four chance of inheriting the disease," she said. "Kevin's older brother, Brian, is unaffected."

"He learned to use a cane and Braille typewriter at school. At age 17 he had a spinal fusion surgery at Johns Hopkins to correct a 90-degree scoliosis curve. The hospital stay lasted almost three months due to complicating respiratory problems. He received a stomach feeding tube while at Hopkins – as many Batten children end up getting – and stopped eating solid food. "

Kevin passed away at home at the age of 20.

"We were determined to care for him at home and believe he felt safe and comfortable there," Vicki said.

The Lumms' recalled Kevin's love of collecting McDonald's Happy Meal toys, which he would identify by feeling, and said he loved having books read to him. He also participated in the Challenger Baseball and Bowling Leagues for special children.

"His dog Arrow, a toy poodle, kept him company," Vicki said, "and he was very fortunate to have a neighbor a few years younger who played with him. The children at school, the Challenger Leagues, and church were all very kind to him."

The Lumms said plans are underway for this year's golf tournament, which continues to raise money for Kevin's Fund.

Story written by Marie Gilbert, The Herald-Mail  
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