BDSRA Board Approves Grant Funding for 2013 Research Cycle

BDSRA, with the collaboration of our international and family foundation partners (BDSRA-Australia, BDFA of the United Kingdom, Noah’s Hope (Chicago) and Hope4Bridget (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. Two studies were second year-funding for projects that began in 2012. The 2013 RFP process reviewed 27 proposals totaling $2.2 million in requests spanning projects in the basic biology of Batten disease to drug discovery. All proposals are peer reviewed by NCL experts, incorporating evaluation systems used by the pharmaceutical industry.

Significance of the project to advancing NCL research, strategy to accomplish research objectives, expected outcomes, and potential for translation are all evaluated within the peer review system now used by BDSRA.

To identify drug targets, funded grants were approved for research that investigates the molecular basis of Batten disease (Dr. Palmer and Dr. Storch) and investigations into drug discovery (Dr. Cooper, Dr. Gerst). Dr. Erika Augustine has received continued funding for the CellCept trial for juvenile Batten disease at the University of Rochester. To continue the expansion and effectiveness of a Batten disease patient registry, the DEM-Child database network project was also funded for a second year (Schulz).

The 2014 Request for Proposals (RFP) will be open and published September 19, 2014. The announcement will be published on the BDSRA website and by email to the Batten disease research community. BDSRA will accept submitted grant requests from researchers until December 12, 2014. Peer review will follow the December deadline.

(continued on page 2)
**Grant Funding**  
(continued from the front cover)

BDSRA continues to be committed to advancing basic and translational research to find treatments and cures through the RFP process and congratulates those who received funding for 2013.

**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.

**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  

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.

**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.
BDSRA Funds Crucial International Project to Expand a Batten Disease Patient Registry

One of the most important steps toward finding effective medical treatments for Batten disease and advancing clinical research is to map out the ways in which the forms of the disease affect patients: the symptoms most commonly seen, how those symptoms progress, trends among and between genetic types, and what complications develop. To achieve this means collecting as much data as possible about all patients throughout the world affected by the disease and assembling it in a single database. By creating a framework to collect, analyze, and distribute this data, called a patient registry, the scientific community can accelerate basic discoveries from bench to bedside.

A patient registry database for Batten disease will describe for the first time the natural history and progression of the disease over a person's lifespan and provide insights on new symptoms that might have been previously overlooked or unidentified. The more patients who have information entered into a registry, the greater the chances are that research will produce results that benefit all affected.

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 Schulz, MD, of Germany’s 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 also 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
- Recruiting patients for clinical trials
- Tracking genotype and phenotype associations
- Safety monitoring
- Identifying novel biomarkers and modifiers
- Developing novel therapies for NCLs

The collection of Batten patient data will also assist in achieving earlier diagnosis and proactive interventions for those affected, increasing quality of life and supportive care.

“The DEM-CHILD NCL Patient Database Consortium is deeply thankful for the financial support of BDSRA for this project,” Dr. Schulz said. “In the consortium, NCL clinicians from 11 countries have agreed to collaborate internationally in collecting and analyzing NCL patient data,” she added.

“This extensive collection and analysis of clinical data of all NCL forms is the first worldwide attempt to do this. It will be of significant importance for the validation of experimental therapies,” she said.

Batten patient families can share information about the existence of the registry project with their medical team to make them aware of the opportunity to participate in the registry.
FAMILY INVOLVEMENT IS KEY TO RESEARCH PROGRESS FOR BATTEN DISEASE IN LABS AND CLINICS

Science leaders value partnerships

The involvement of families of affected children in fundamental research related to Batten disease has been crucial to moving discoveries in the lab to translational work at the bedside. This research issue underscores the work we have funded together and watched grow into important clinical trials. 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.

For the scientists, medical researchers, and others who focus efforts on unlocking the mysteries of Batten disease, the support and involvement of families is fundamental to their work.

“In order to identify and test new therapeutic agents for rare diseases, various research models are needed. These models can range from mice, rats and sheep to monkeys,” according to Ryan Geraets, an MD, PhD student at Sanford School of Medicine in Sioux Falls, South Dakota. “The best models, however, for disease research can often come from the patients themselves in the form of tissue and blood samples. 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.

“In the work that has been accomplished by Susan Cotman, PhD, assistant professor of neurology at the Center for Human Genetic Research at Massachusetts General Hospital, the ability to work with patients and donated samples has been key to major discoveries.

“There is no better approach to studying Batten disease than one that is closely linked to patients and family members. Our research aimed at identifying and better understanding the key steps that lead to Batten disease is driven by the information that we learn from research study participants,” she noted. “For example, DNA, skin, and blood samples provide us with genetic and other biological information, linked to clinical information, to narrow in on factors that we can target for designing potential treatments and help us discover the best ways to measure success. Skin cells, obtained through skin biopsy from participating subjects, are also being ‘reprogrammed’ to become a type of stem cell called an induced pluripotent stem cell, or iPS cell,” she explained. “These iPS cells are key tools we and others are now using for drug screening and biomarker development. We believe that the more we can use these types of tools for early stage drug development for Batten disease, the better we will ultimately be at designing successful treatments. I am thankful for those who make this possible through participation in research projects.”
Ronald Crystal, MD, professor and chair of the Department of Genetic Medicine of the Weill Medical College of Cornell University, views this as an important 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,” he said. “Families help the cause by participating in screening and clinical studies, by being involved in follow-up studies, by spreading the word of work that is being carried out to further the research and by fund raising to support the research,” he noted.

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.”

“This is important both for our understanding of the disorders but also for judging the efficacy of our therapies and management treatments,” she said. “As the bench scientists work on understanding the disease process at the cellular level, having patient cells available for direct study and for assessment of potential therapeutic agents is critical.”

Dr. Sims has worked to establish and maintain a biorepository to assist with making reagents available to the research community. “We collect, store and distribute DNA [extracted from blood and tissues], lymphoblasts [transformed cells made from blood lymphocytes], and plasma and fibroblasts,” she notes, which is essential for many research studies in all forms of Batten disease.

For those interested in becoming involved in Batten disease research, there are several ways to contribute:

- Answering questionnaires from BDSRA and research partners
- Donating to the BDSRA research fund
- Providing blood and skin samples from family members at the annual Family Conference
- Asking about brain donation as part of end-of-life pre-planning. Contact Margie Frazier, mfrazier@bdsra.org or Becky Hetteberg, bhetteberg@bdsra.org to discuss steps required for brain donation. Because brain donation for Batten disease research involves your area funeral director, our research partner in Los Angeles and local hospitals, in a brief window of time at death, pre-planning helps make the process effective and less stressful for families.

Receive the latest updates, learn more about Batten disease and find out how to help at www.bdsra.org.
BioMarin Hosts BDSRA for Rare Disease Day Event

Novato, California based pharmaceutical company BioMarin, which has launched a Phase 1 and 2 clinical trial to determine the safety and efficacy of an enzyme replacement therapy for the treatment of CLN2 disease, hosted BDSRA at company headquarters in March to observe Rare Disease Day.

Batten families have long supported research labs around the U.S. to accelerate the development of treatments like ERT, or Enzyme Replacement Therapy, currently being tested by BioMarin at clinical trial sites in Hamburg, Germany, and London.

“It was thrilling to know that BDSRA’s early support of translational research has brought us this far,” said Margie Frazier, executive director of BDSRA, who met with BioMarin’s team on March 6th. She shared BDSRA’s history and research goals at BioMarin’s annual Rare Disease Week event for employees. After a day-long tour of the Novato research labs with BioMarin scientists, and updates about the progress being made in clinical trials, she met with others in rare disease who have benefitted from BioMarin’s work, including MPS IV or Morquio syndrome.

“The BioMarin Rare Disease event was the greatest,” Frazier said. “So many of the BioMarin team talked with me about their excitement to help Batten families whose children are affected by Late Infantile Batten disease,” Frazier noted. “Many had worked on initial phases of development of the enzyme and shared their pride in being on a project with so much promise of making a difference for children.”
**BDSRA RESEARCH AND ADVOCACY UPDATES**

February was a month of building incredible connections for BDSRA at the Lysosomal Disease Network’s (LDN) 10th Annual WORLD Summit Symposium in California and the RDLA Rare Disease Week in Washington, D.C. Both meetings and events brought major patient advocate groups, researchers, and rare disease leaders together to share advances in science.

Energy and expectations were high at LDN in San Diego February 10-13 as more than 1,100 people assembled to hear presentations, discuss research findings and expand collaborations on solving the scientific challenges of lysosomal storage disorders and diseases, of which Batten disease is a part. BDSRA staff Margie Frazier and Julie Conry participated in meetings and briefings as well as representing the organization as an exhibitor with other patient groups. Some highlights from the week included networking with other rare patient advocacy groups to learn about innovations in research and service for families with the MLD Foundation, Tay-Sachs, Gaucher, and Fabry Disease organizations.

BDSRA was also represented at a program for patient advocates hosted by Genzyme focused on clinical trial development for lysosomal storage disorders, drug discovery, and drug protocols. The take home message was how important the development of patient registries and natural history studies are for pharma to make progress with rare disease work. There is an urgent need for families and patients to provide information through the patient registry projects for their diseases, including Batten. Richard A. Moscicki, M.D., Deputy Center Director of Science Operations at the FDA’s Center for Drug Evaluation and Research, also informed conference attendees about the new focus of the FDA on patient-informed drug development to obtain direct input from those who will use them. More meetings are now set up between FDA reviewers working on drug testing and patients and patient advocate groups. He reported that 36% of all fast-track approved drugs the FDA approved in 2013 targeted orphan diseases.

On Capitol Hill for Rare Disease Day, BDSRA was well represented by Margie Frazier and parent advocates Tracy VanHoutan, Donna Fogle, and Sharon King of Taylor’s Tale and Chris Leonard. Noah Coughlin also traveled from the west coast, this time by plane, to participate in the programs and events around Washington organized through RDLA.

RDLA (Rare Disease Legislative Advocates) and Global Genes and the National Institutes of Health organized four days of panel discussion on drug development and discoveries for rare diseases that culminated in World Rare Disease Day February 28th. On February 26, Batten disease advocates visited more than 40 Congressional offices to talk with senators and legislators about the need for drug discovery and better services for families. For special questions on rare and genetic diseases, contact the Office of Rare Diseases Research and NIH Information Center (GARD) at GARDINFO@NIH.GOV, call toll free to 888-205-2311 or link to rarediseases.info.nih.gov/GARD.
NEWS FROM OUR SUPPORTERS

NEW JERSEY EVENT BOOSTS BATTEN RESEARCH

Gregg and Paula Froio and dozens of volunteers opened the door for the 4th annual Road to a Cure Monte Carlo Night in Berlin, New Jersey on February 22nd. More than 340 people attended the dinner and evening of entertainment, which is the 15th year the Froio family has organized a large scale event through the Melissa Froio Foundation to support Batten disease research honoring their late daughter Melissa. Melissa was diagnosed with late infantile Batten disease in 1999. The Froio's formed the foundation to raise money for Batten disease research. This year the event generated $18,000. The Froio's start organizing the event in October, with most of the raffle gifts donated by family, friends, and supporters. BDSRA representatives Margie Frazier and Julie Conry along with Donna Fogle of Jacksonville, Florida and Kim and Greg Fuller, of New Jersey, also attended.

RUNNERS RALLY IN NORTH CAROLINA

More than 330 runners and walkers participated in the 2014 Run the Creek 5K in Charlotte, North Carolina in March. The Highland Creek Homeowners Association organizes the race and proceeds are donated to BDSRA for Batten disease. Local volunteers and community supporters include Chris Hawkins and Scott Campbell, who have worked to develop local events over the past several years to raise money for Batten disease research and services. Awards are given at the race for first, second, and third place finishers.
INTERNATIONAL PARTNERS SUPPORT BDSRA USA IN RESEARCH PROJECTS

Through collaborations with the Batten Disease Family Association in the United Kingdom and BDSRA-Australia, important leverage to fund research grants is made possible to expand the number of researchers who can be funded each year through the BDSRA RFP process. Volunteer efforts across Australia and New Zealand generated $130,000 in 2013 to support research in various aspects of Batten disease. In the land “down-under” supporters organized garage sales, fun-runs, morning teas, barbeques, and auctions to raise awareness and funds for research. In the UK, collaboration across patient advocacy groups for Batten disease is demonstrated through BDFA’s annual financial support for research projects such as the patient registry and drug target projects in the labs of Dr. Cooper at King’s College and Dr. Gerst at the Weizmann Institute of Science in Israel.

VOLUNTEERS FOR BDSRA HELP WITH TRANSLATION

As an organization with many international and global ties to serve and support families around the world, BDSRA is often contacted by people speaking many languages from many countries. In the last year, assistance was requested from families and medical providers in Costa Rica, Brazil, France, Ireland, Serbia, Canada, the United Kingdom, Argentina, Poland, Germany, Mexico, Saudi Arabia, Australia, Israel, Oman, and New Zealand. Important help is provided to the national office from volunteers who generously donate their time and talents to translate and communicate across borders.

Nancy Peruyero, from Miami, Florida, mom to Nicolas, who has late infantile Batten disease, and Rachel Herrel, a former Spanish teacher in Cincinnati, provide frequent “on-the-spot” translation services for BDSRA when requests arrive from Spanish-speaking families and professionals. Henry Dolin, a sophomore at Ohio State University, often assists with French translation. The help from caring individuals like Nancy, Rachel, and Henry personalize a challenging process, way beyond what any computer software can provide. Merci beaucoup and gracias to all.

Left to right: Nancy Peruyero and Nicolas; Rachel Herrel; Henry Dolin
BDSRA Board Meeting Update

The BDSRA board of directors met January 24, 25, and 26 in Columbus at the national office, the first of two meetings held each year, to reconnect and plan. The second meeting is held each July prior to the annual Family Conference. Highlights from the January board session:

- The board welcomed new members Barbara Wuebbels and Dave Pearce, and returning re-elected members Tracy VanHoutan and Rob Geer.

- The board acknowledged and thanked outgoing board members Chris Daniak and Debbie Ham for their service to the BDSRA organization and Batten community.

- Board members approved the 2014 budget, which includes funding for the annual conference, family support services, web site improvements and continuation of the scientific merit review process, or RFP for research grant allocation.

- The board approved funding for five research projects in the U.S., Germany, UK and Australia.

- The board approved a 2014-2015 timeline for the research RFP release. The RFP call for proposals will be announced in September, 2014, with grant approvals made in the spring of 2015 to better align with the family conference and recruiting for reviewers.

- Board members reviewed the latest information on chapter calls completed in the fall with new guidelines and improved information.

- The board reviewed the service, communication, and development goals for 2014.
Meet Your BDSRA Board Member:
Barbara Wuebbels, RN

Barbara Wuebbels, RN, was recently elected to the BDSRA board in the November elections. She resides in Phoenix, Arizona and is the Vice-President of Patient Advocacy and Medical Affairs for Audentes Therapeutics. She is a founding member of the Wellness Community of Central Arizona, a non-profit that provides free counseling and support to patients and families with cancer.

She serves on the Arizona Biomedical Research Commission, in which she was appointed by the governor to review scientific research requests submitted by researchers at universities and organizations within the state. Her involvement with the Batten community in the U.S. and Europe began through her previous work at BioMarin pharmaceutical company as head of the global patient advocacy program. She has extensive industry experience with Batten disease patients due to her role at BioMarin in setting up the first clinical trial for CLN2 overseas. She has worked in the orphan drug area for 14 years and is very knowledgeable about clinical trials and research.

“I fell in love with the BDSRA community at my first family meeting while I was working at BioMarin and was sad to think I might lose contact with them after I left,” she said. “Being elected to the Board is a high honor and having the opportunity to remain part of the BDSRA community makes me very happy. BDSRA truly exemplifies what it means to be a family with their inclusion of grandparents, healthy sibs, parents who have lost a child, board members, and others.”

“I hope in serving as a board member to be able to help BDSRA through my contacts and experience in industry and in working with other patient advocacy groups.”
Since January 2014, nine graduate students from the University of Maryland-Baltimore School of Social Work have been busy working with BDSRA staff to develop a multi-faceted and comprehensive family needs assessment for Batten disease parents and primary caregivers. This research project consists of detailed interviews with parents, guardians and primary caregivers who have affected children, as well as a broader on-line survey for parents, guardians, and primary caregivers living in the United States and Canada.

The results of the interviews and survey will help shape future BDSRA programming and services, making them more relevant and timely for Batten families. The project has also helped a group of students new to Batten disease and rare disease get a clearer picture of the physical and psychosocial needs of families.

According to Karen Hopkins, PhD, Associate Professor of Social Work at the University of Maryland who is leading the study, this project is a collaboration that benefits both the non-profit being served and the graduate students.

“We take very seriously the need for our students to have real-world research experience by the time they graduate from our program,” Hopkins said. “By partnering with BDSRA, nine of our most promising students have learned about Batten disease, the needs of families and helped managers better understand service delivery to families who rely on them.”
**Children’s Book Selection**

“How Do You Doodle? Drawing My Feelings and Emotions”  
by Elise Gravel

How Do You Doodle? is a drawing book for children to help them get in touch with their emotions and learn how to express them through artistic expression. It can be used with a parent or adult mentor or in a professional setting with a counselor or therapist. This book helps youngsters to work through difficult feelings and develop a better realization and understanding of emotional responses to stressful situations. Recommended for elementary school-age children, ages 6 to 10.

**Adult Book Selection**

“Caring for Children Who Have Severe Neurological Impairment: A Life With Grace”  
by Julie Hauer, M.D.

Global impairment of the central nervous system, whether stable or progressive, is often called severe neurological impairment (SNI). A parent is a child’s best expert and advocate in obtaining the care needed, and many parents become highly skilled in managing their child’s care. This guide provides information to help parents increase their knowledge and improve their caregiving skills.

The author advocates shared decision making between family caregivers and healthcare providers. She details aspects of medical care such as pain, sleep, feeding and respiratory problems that will be particularly useful to parents. Tables and key points summarize discussions for clear, quick reference, while case studies and stories illustrate how different families approach decision making, communication, care plans and informed consent. Parents and other caregivers will find this book to be indispensable — as will bioethicists and clinicians in pediatrics, neurology, physical and rehabilitative medicine, palliative care, and others who care for children with neurological and neuromuscular disorders. Dr. Hauer offers hope and practical coping strategies in equal measure.

If you know of a good book to recommend for other families, please email Becky Hetteberg at bhetteberg@bdsra.org with your suggestions.
Gear up for Batten Awareness Weekend
June 7 & 8

Connection is the theme of Batten Awareness Weekend for 2014 on June 7 & 8.

By starting new connections, expanding existing connections, and building a network of connections, families, friends, and supporters can raise awareness and funds for Batten disease research and services.

Using social media opportunities is a quick and simple way to send the message, inspire a donor, and educate others about the challenges of rare disease. Here are a few ideas:

- Visit www.bdsra.org to download special posters and information for your Batten Awareness Weekend activities.
- Increase visitors to the www.bdsra.org website by adding the website link to your email account signature, which encourages others to see and view the link. Then send an email about Batten Disease Awareness weekend to friends, family, co-workers and neighbors with the web address.
- Update your Facebook status to encourage your FB community to learn about Batten Disease Awareness weekend by visiting www.bdsra.org.
- Add BDSRA information to the small text box underneath your FB profile, and share your story.
- Send a tweet about Batten Disease Awareness Weekend and any fundraisers being held in your community.
- Post photos and stories about the brave families who face Batten disease every day and encourage people to “like” and “share” the posts for supporting Batten Awareness Weekend.

Share your ideas to Connect for Batten Awareness Weekend by emailing jconry@bdsra.org
Conference online registration is open

We are looking forward to seeing everyone in Columbus, Ohio this July.

- Conference dates are Thursday, July 24th to Sunday, July 27th, 2014.

- Conference online registration is open until July 1st, 2014. Go to www.etouches.com/bdsra2014 to register.

- Hotel registration for conference at the Columbus Airport Marriott is open until July 7th, 2014. Register at http://bit.ly/1mL5zAk.

- For questions, contact Tracy Kirby at 800-448-4570 ext. 13 or tkirby@bdsra.org

For families seeking financial support to attend conference, one source for potential support is contacting the local Developmental Disability Council. There are Developmental Disability (DD) Planning Councils that serve each state and territory in the U.S. and under the umbrella of “Family Support Services” financial help may be available.

To see if family support funds might be available in local regions through the DD council, visit: www.nacdd.org/Councils.html

BDSRA IS ALL ABOUT FAMILIES CONNECTING WITH EACH OTHER

Help us update the BDSRA family directory. If you would like to be included, please send your name, address, contact information and affected person’s name and NCL type to tkirby@bdsra.org.

This information is for the directory only. BDSRA does not sell or share the directory with outside companies or organizations.
For Catrina and Jeff Nelson, losing their 6-year-old daughter Charlee on March 15th to late infantile Batten disease has been like a bad dream. In the midst of their grief, the family stood in the private office of Utah Governor Gary Herbert the next day as Herbert signed “Charlee’s Law,” in a quiet ceremony. Also known as HB 105, Charlee’s Law will allow those with intractable epilepsy to first acquire written permission from a neurologist and then apply for a waiver to import cannabis oil. The Utah Legislature formally passed the bill March 13 after Charlee and her parents arrived on the floor of the House and Senate to witness the vote.

According to Catrina and Jeff, Charlee held on until the legislation was passed after many weeks of severely declining health and complications. After battling mononucleosis in October and surgery for a feeding tube, their daughter’s strength slowly diminished. A period in the hospital ended on March 4 when her parents brought Charlee home to hospice care.

She had declined by the time hearings were starting on the bill, but as one of 50 children on a waiting list in Utah to have access to nonintoxicating cannabis oil for seizure control, Charlee became the symbol for compassionate use as parents of children with severe epilepsy conditions lobbied the Utah House and Senate.

“"We were very surprised when the legislators called and offered to name the bill for her," Catrina said. "We did not expect it. What better way to honor her after she’s gone."

“We were contacted right before the vote was scheduled and asked to attend, so we took her to the statehouse,” Catrina recalled. “As sick as she was, she had real energy and was happy when she was on the floor waiting for the vote. There was a standing ovation in the Senate and it was like she knew that was for her. She held on to do this.”

The Nelsons never set out to be active lobbyists for the legislation, but became involved through their association with the local Epilepsy Society. The Nelsons spent years before Charlee’s NCL1 diagnosis dealing with their daughter’s debilitating seizures that started when she was three. “We were desperate to find some solution before we knew what her diagnosis was,” Catrina said. While they know that the law would not have saved their daughter’s life, the potential to extend it with a better quality and less suffering would have been some comfort, they noted.

That she leaves behind a symbolic legacy is a tribute to their daughter’s ability to inspire so many people, Catrina said. The little girl who loved to sing, dance, and sing Twinkle, Twinkle Little Star was able to put a face on what it really meant to need a medical miracle, her mother said, which was her greatest gift for so many others facing severe seizure-inducing conditions.

The Nelson family: Catrina and Jeff Nelson holding Charlee, with Jericho and Cashlee

Charlee Nelson Inspires Utah Legislation Named in Her Honor
in loving memory

EZEKIEL TODD HOLT, son of Andrew and Bree Holt, Toledo, OH
Born: 11-19-09 | Died: 3-23-14
Late Infantile

AUSTIN SAMUEL HEIN, son of Ron and Lugine Hein, Columbia, MO
Born: 10-30-93 | Died: 3-21-14
Juvenile

CHARLEE MARCELLA NELSON, daughter of Jeff and Catrina Nelson, West Jordan, UT
Born: 1-30-08 | Died: 3-15-14
Late Infantile

SKYLER JORDAN ROWE, granddaughter of Terrie Rhoden, Argillite, KY
Born: 4-26-06 | Died: 3-8-14
Late Infantile

NOLAN HENRY JAMES LEIS, son of Chad and Shannon Leis, Bangor, WI
Born: 5-24-03 | Died: 2-25-14
Late Infantile

CHAD ALAN DOVEL, son of Charles and Debbie Dovel, Hamburg, IA
Born: 6-3-89 | Died: 2-9-14 | Juvenile

ANDREW RYAN GRADY, son of Chris and Valerie Grady, Upper St. Clair, PA
Born: 5-18-04 | Died: 1-19-14
Late Infantile

CHRISTOPHER MCDONOUGH, son of Larry and Barb McDonough, Orland Park, IL
Born: 12-7-85 | Died: 1-10-14
Juvenile

EMILY ANNE SMITH, daughter of Hugh and Pamela Smith, Chattanooga, TN
Born: 11-10-87 | Died: 12-28-13
Juvenile

CARSON SHELDON HUELIN, son of Lori-Ann Keeping and Sheldon Huelin, Brantford, Ontario, Canada
Born: 4-29-04 | Died: 12-24-13
Late Infantile

TORRIE NICOLE HENDERSON, daughter of Edward and Ruth Henderson and Carol and Brad Bowker, Granite City, IL
Born: 8-17-81 | Died: 12-05-13
Juvenile

KORRY ELOTA SHARP, daughter of Mark and Georgia Sharp, Vacaville, CA
Born: 6-03-93 | Died: 11-17-13 | CLN5

NOAH CORBIN LACY, son of Scott and Aimee Lacy, South Charleston, WV
Born: 12-21-05 | Died: 11-16-13
Infantile

AIRON HENRY BOLEY, son of Don and Brandy Boley, Davisville, WV
Born: 6-13-02 | Died: 11-03-13
Infantile

CONNOR EVAN ALLBEE, son of Derek and Christy Allbee, Reno, NV
Born: 9-14-94 | Died: 10-20-13
Juvenile

RYAN MAYNARD, son of Joel and Chris Maynard, Colorado Springs, CO
Born: 5-15-85 | Died: 10-12-13
Juvenile

BDSRA takes great care to memorialize those individuals who have passed away from Batten disease. If a person is omitted from this page, it is because BDSRA was not notified of the death, did not have permission to publish, or did not have confirmed information.
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The Batten Disease Support and Research Association has been remembered many times in the past three months by families and friends affected by Batten disease. Thank you for your generous contributions. This support for the vital mission of research and services for families is crucial to finding a cure. We are also grateful to those supporters who have participated in events and volunteered their time to advance our efforts for Batten disease research and family services. We also acknowledge in this donor list all the generous contributions made through the 2013 Annual Fund Campaign.

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