Partnering with the Science, Research and Health Communities

Every year, hundreds of children and their families face an uncertain future as they cope with the devastating diagnosis of Batten disease. As a rare, inherited disorder of the nervous system, Batten disease has no known treatment or cure. Those affected with Batten disease suffer progressive neurological impairment, seizures, blindness, loss of motor skills, speech and sadly, life, taken too soon.

Batten disease or Neuronal Ceroid Lipofuscinosis (NCL) is a family of lysosomal storage disorders caused by autosomal recessive genetic mutations resulting in the build-up of fats and proteins, or lipofuscins in the body which cause cell death. There are more than 10 identified forms of the disease, which are most commonly diagnosed between infancy and school age. Kufs, Parry and ANCL disease are known adult forms of Batten. Though recent improvements in genetic testing have made diagnosing Batten disease much quicker and more reliable, families tell of very long diagnostic journeys, often years after the first signs of seizures or loss of sight. In a recent needs assessment completed by BDSRA, more than 30 different diagnoses were reported by families before the final Batten diagnostic determination. Autism, seizure disorder, epilepsy, PDD and others are common early diagnoses. According to the Centers for Disease Control, 2-4 births per 100,000 in the U.S. are affected by Batten disease, though some researchers in the field suggest these numbers are low.

BDSRA: Advancing the Cause

The Batten Disease Support & Research Association (BDSRA) is a nonprofit organization headquartered in Columbus, Ohio, dedicated to funding research for treatments and cures, advancing education, providing family support services, advocating within state and federal governments on behalf of families, and raising awareness of the disease and its impact. Founded in 1987, BDSRA is the largest support and research organization dedicated to Batten disease in North America. BDSRA facilitates support groups, educational initiatives, access to cell and tissue banks, research programs, a family conference, and programming for scientists, researchers and physicians.
Resources for Health Professionals

As part of its mission, BDSRA believes that in order to effectively unravel the mysteries of Batten disease, the worlds of medical science, laboratory research, caregivers, and families must work together toward a common goal: discovering treatments and cures.

Funding A Cure
To date, BDSRA, along with collaborating partners around the world, has funded more than $6 million in research at dozens of institutions. Funded researchers are working to harness cutting-edge technologies, including new developments in gene therapy, enzyme replacement therapy, drug repurposing and others. Foundational work funded by BDSRA has led to clinical trials both in the U.S. and Europe. Research and medical findings are reported each year at the BDSRA annual family conference during poster sessions and presentations. Every two years, BDSRA helps to fund the NCL International Research Congress, falling on even-numbered years.

Merit Review Process
BDSRA facilitates an annual merit review process and invites letters of intent (LOI) across a broad range of Batten research areas of all forms of the disease. Awards are made each year at the family conference in July and typically are funded in the $30,000 to $60,000 range.

Batten Disease Registry
BDSRA is part of a novel global registry linking families, physicians and scientists. Our aim is to help collect the world’s largest clinically and genetically best characterized set of patients with each form of NCL. In addition to the U.S., the registry and natural history database includes information from partners in Germany, United Kingdom, Finland, Italy, India, Brazil, Argentina, Turkey, France, Norway and Denmark.

Research Tools and Collaborations
Significant research tools in the form of small and large animal models, IPS cell lines, and other bio-repository resources have been developed and characterized in the Batten disease area. BDSRA staff is happy to link researchers who are working in emerging research platforms in lysosomal diseases to our research family.

Scientific and Medical Advisory Board
Our scientific and medical advisory board works collaboratively to advise the BDSRA board and staff on issues of importance to the field and to our daily work to improve the lives of families who face an uncertain future.