

Blake Dell'Aringa started his journey in life on January 11, 2002 as a healthy and very active little boy. He was meeting all of his milestones with the exception of a speech delay. We definitely expressed concern for this but were always assured that is was normal for boys to have speech delays. Once Blake turned three years old he qualified to be entered into the school district for this problem and everything was going smoothly. Right before Blake's fourth birthday we noticed a couple balancing issues with Blake. I immediately took him to his pediatrician who then referred us to a neurologist and to also have an MRI done. We hand carried the films from the MRI to our first appointment and without even looking at Blake the neurologist said it doesn't look good. Blake had some atrophy in the cerebellum and it was thought to be degenerative. Our lives were crushed on that day and then it became clear that we had to figure out exactly what we were dealing with. The following week we were admitted into the hospital for a weeks worth of grueling testing. All of the tests were coming back normal and then we had him scheduled to see a pediatric ophthalmologist and that day it was determined that Blake had NCL disease. Next we had to find out which type of the disease Blake had. Later that week an enzyme test confirmed that Blake had Late Infantile NCL disease. All of this happened in June of 2006.

We have some hope as this type of the disease has the most promise for hopefully finding some kind of treatment, which is what keeps us going everyday. Blake has lost his vision and is now unable to walk. He has seizures, which are controlled with medication. We are keeping him stimulated constantly and will continue doing everything that we can to fight this inevitable monster. We feel it is our responsibility to help educate, increase awareness, and raise money to find better treatments and a cure for Batten Disease.

Blake has touched our lives and hearts in so many ways. He has shown us what it is like to love the world through his eyes and challenged us to fight for his life.

Dawn Jaeger



Presently, there is nothing that we can do to change the devastating outcome of Batten Disease. However, current research gives us hope that someday it will yield an effective cure, which will help these children to live. Because this is such a rare disease, money and advancement is slow. It is imperative that we keep funding this type of research not only to cure Batten Disease, but to also help us advance our medical technologies and find cures for other rare diseases and more common disorders.



Blake's Purpose: Fighting for a Cure
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Blake's Purpose works to help fund research for Batten Disease. We are a non-profit, tax-exempt entity pursuant to Section 501(c)3. Federal Tax ID # 26-0456553. 100% of the funds raised will support Batten Disease Research. Your gift is tax deductible.

BATTEN DISEASE SUPPORT & RESEARCH ASSOCIATION

Our Mission

The Batten Disease Support and Research Association was created to provide information, medical referrals and support, along with furthering the efforts of research to find a viable treatment and eventually eliminate Batten Disease. It is the only family support organization in the United States, Canada, New Zealand, Australia, South Africa and many other nations specific to Batten Disease.

www.bdsra.org

Information in this brochure is intended for general education purposes only, and should not be construed as advising on diagnosis or treatment of this or any other medical condition.

Batten Disease



Batten Disease is a rare and incurable genetic disorder that affects an estimated two to four of every 100,000 children born in the United States.



BATTEN DISEASE

Batten Disease was first described in 1826. It is the common name for a group of diseases known as neuronal ceroid lipofuscinosis, and is one of the more common of the neurodegenerative diseases. At this time there is neither treatment nor cure.

There are 5 major forms of batten Disease.

Infantile: onset at 6 mos-2 yrs. Life expectancy 5-10 yrs old.

“Classic” Late Infantile: onset at 2 -5 yrs. Life expectancy 8 -12 yrs old.

“Variant” Late Infantile: onset at 2-5 yrs old. Life expectancy 8-13 yrs.

Juvenile: onset 5-10 yrs. Life expectancy is teens-early twenties.

Adult: onset before 40 yrs. Life expectancy varies.

All of these forms of NCL are defined by age of onset, pathology and gene identification.

Batten Disease is rarely diagnosed immediately and is often mistaken for epilepsy, mental retardation, retinitis pigmentosa, even schizophrenia in adults. An ophthalmologist can observe pathological changes in the retina. This often provides one of the first diagnostic clues. Onset is characterized by beginning vision loss, seizures, clumsiness, personality and behavior changes. Batten disease causes continuing physical and mental deterioration leading to death.

It is a recessive inherited disease meaning both parents must carry the defective gene. A child must inherit a copy of the bad gene from both parents in order to be affected. A child that inherits a bad copy from just one parent will be a carrier.

CAUSES & SYMPTOMS

Childhood NCLs are autosomal recessive disorders; that is, they occur only when a child inherits two copies of the defective gene, one from each parent. When both parents carry one defective gene, each of their children faces one in four chance of developing NCL. At the same time, each child also faces a one in two chance of inheriting just one copy of the defective gene. Individuals who have only one defective gene are known as carriers, meaning they do not develop the disease, but they can pass the gene on to their own children.

Symptoms of Batten Disease/NCLs are linked to a buildup of substances called lipopigments in the body's tissues. These lipopigments are made up of fats and proteins. Their name comes from the technical word lipo, which is short for “lipid” or fat, and from the term pigment, used because they take on a greenish-yellow color when viewed under an ultraviolet light microscope. The lipopigments build up in cells of the brain and the eye as well as in skin, muscle, and many other tissues. Inside the cells, these pigments form deposits with distinctive shapes that can be seen under an electron microscope. Some look like half-moons (or comas) and are called curvilinear bodies, others look like fingerprints and are called fingerprint inclusion bodies and still others resemble gravel (or sand) and are called granular osmophilic deposits (grods). These deposits are what doctors look for when they examine a skin sample to diagnose Batten Disease.

The diseases cause death of neurons (specific cells found in the brain, retina and central nervous system). The reason for neuron death is still not known.

TREATMENT

As yet, no specific treatment is known that can halt or reverse the symptoms of Batten Disease/NCL. However, seizures can be reduced or controlled with anticonvulsant drugs, and other medical problems can be treated appropriately as they arise. At the same time, physical and occupational therapy may help patients retain function as long as possible.

Some reports have described a slowing of the disease in children with Batten Disease who were treated with vitamins C and E and with diets low in vitamin A. However, these treatments did not prevent the fatal outcome of the disease.

Meanwhile, scientists pursue medical research that will someday yield an effective treatment

CURRENT & FUTURE RESEARCH

Identification of the specific genes for Infantile, Late Infantile, Variant Late Infantile and Juvenile Batten Disease/NCL has led to the development of DNA diagnostics, carrier and prenatal tests.

Scientists have discovered that the Infantile and Late Infantile diseases are missing key lysosomal enzymes, i.e. Palmitoyl Protein Thioesterase 1 (PPT1) for Infantile and Tripeptidyl Peptidase 1 (TPP1) for Late Infantile. Knowing that these enzymes are missing is now leading to the development of gene replacement and stem cell transplantation therapies.

Recent studies have shown a link between the Juvenile form and the body's autoimmune system. Although this link is not yet fully understood, it may eventually lead to a treatment.

Currently there are two research trials being conducted in the United States and more research is being done overseas. Stem Cells Inc. is conducting a trial which takes cells derived from the human brain and is injecting them into Battens patients with the hopes that they will survive and migrate throughout the brain producing the missing enzyme (PPT1 & TPPI) which Batten patients are lacking. The other study is being conducted out of Weill Medical College at Cornell University. This experiment is using a method called gene transfer, where the doctors use the delivery of a gene to dispense an experimental drug (AAV2CUhCLN2) into the brain.

Although researchers are making progress, there is still lot of work to be done. Finding a cure for Batten Disease does not only promise a future to patients dealing with NCL, it also is relevant in helping researchers find advancements in other rare genetic, metabolic and degenerative disorders.

