

ADVANCE FOR NURSES

Battling Batten Disease

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By Maureen Gavin, BSN, RN-BC, CDDN, Milen Velinov, MD, PhD & Edie Dockter

Edie Dockter's story: My husband, Gene, and I are the parents of three children with Batten disease. Our children were born in 1963, 1965 and 1969. Our oldest child, Laurie, showed symptoms for Batten disease in 1969, when she was 6 years old. She started having problems with coordination, speech, and cognitive skills. After seeing many doctors and undergoing numerous medical tests, she was finally diagnosed at age 10. At that time, we were told our daughter was one of the few children with a diagnosis of neuronal ceroid lipofuscinoses (NCL).

In 1973, the same year Laurie was diagnosed, our older son, Tommy, started showing signs of the disease. He was 8 years old at that time. Laurie died in 1987 at age 24, and Tommy died in 1994 at age 29. Our youngest child, Ken, started showing signs of Batten disease at the age of 12, in 1981.

Our three children were dying of a rare disease we knew nothing about, and we could not find any information about Batten disease in the early '80s. But in 1987, we attended the first Conference on Batten Disease and met Krystyna Wisniewski, PhD, MD. She invited me and Kathy Potterfield, whose son also had Batten disease, to help establish the Batten Disease Registry. As parents of children with Batten disease, we could help other families by sharing our stories, listening to their concerns and answering the many questions parents have.

Since 1987, the Batten Disease Registry has added more than 1,200 patients. The registry works closely with its parent organization, the Batten Disease Support and Research Association (BDSRA) to reach out to families with new diagnoses. The registry identifies families with Batten disease and provides a computerized central data bank of clinical information for clinicians and researchers.

As for my youngest child Ken, his regression was very slow. As he witnessed his siblings deteriorating, he would ask if this was going to happen to him. Over time, his coordination and speech deteriorated, he lost his sight at age 22, and he eventually required the use of a wheelchair. He is now 41 years old, lives in a nursing home and attends a United Cerebral Palsy program. Ken is one of the oldest surviving Batten disease patients.



Laurie, Tommy and Ken Dockter, siblings who were each diagnosed with Batten disease, in the early 1970s.

What is Batten Disease?

Batten disease is the common name for a group of diseases called the neuronal ceroid lipofuscinoses (NCL). It is an inherited fatal neurodegenerative disease, caused by an abnormal buildup of ceroid lipopigments in the tissues of the brain, eyes, skin and muscles. A gene mutation causes a deficiency of enzymes, which triggers lysosomal dysfunction, resulting in an accumulation of lipid substances collectively known as ceroid lipofuscin. Batten disease was named after the British physician, Fredrick Batten, who first described it in 1903.

Signs & Symptoms

Batten disease begins and progresses slowly after a period of good health. The clinical course includes progressive loss of vision, a marked decline in cognitive function, progressive loss of communication skills, personality and behavior changes, progressive loss of motor skills, decrease in muscle mass, loss of ability to ambulate, hallucinations and eventually death.

One of the first signs of Batten disease is often progressive vision loss. Batten disease may be suspected when retinitis pigmentosa is seen during an ophthalmological exam. The symptoms progress over several years, with an increase in seizures, cognitive and mental impairment, and progressive loss of sight and motor skills. As the disease progresses, the affected child's

abilities regress; the child can no longer perform tasks he or she could previously do.

Due to the unusual presentation of symptoms and the complicated diagnostic process, Batten disease is rarely diagnosed immediately after onset of symptoms. It often takes many years from the onset of the first symptoms to correctly diagnose the disease.

Diagnostic Work-Up

The process of diagnosing and then classifying the type of Batten disease (NCL) is an involved process. The initial diagnostic work-up consists of a complete clinical evaluation and detailed medical history, a blood test, and/or a skin biopsy for ultrastructural electromicroscopy (EM). In Batten disease, the lipopigments form deposits with unique shapes (granular osmophilic deposits -- or grods -- curvilinear bodies, or fingerprint inclusion bodies) that can be viewed under an electron microscope. The symptoms; age of onset of vision changes, seizures and cognitive delays; and the type of inclusion bodies (seen in EM) will help determine and prioritize the type of biochemical enzyme testing and/or DNA mutation screening performed.

Additional tests include an EEG (electroencephalogram) to rule out seizures, an ERG (electroretinogram) and a VER (visual evoked response test) to check for vision changes, and MRI or CT scan.

Genetics

Although Batten disease is rare, it may affect more than one person in a family. Batten disease is a recessive inherited disease; both parents must carry the gene mutation and pass it on to their child. A child must inherit one copy of the gene mutation from each parent to be affected.

If a child inherits only one gene mutation, he will be a carrier and can, but not necessarily will, pass the disease on to his or her children. If both parents carry one gene mutation for Batten disease, each of their children faces a one in four chance of being affected, and a one in two chance of being a carrier. A carrier can have an affected child only if his/her partner is also a carrier. Carrier and prenatal testing is available only if the specific gene mutation is known.

The current genetic classification includes approximately eight genes that have identified mutations associated with Batten disease. The symptoms of the different types of Batten disease start at different ages and progress at different rates. In the rare adult form of NCL, Kuf's disease, the ophthalmological studies are normal.

Treatment

Presently, there is no known cure or treatment for Batten disease. The goal of care is to maintain the child's independence and to provide a positive stimulating educational environment. Physical and occupational therapy can help maintain an optimal level of functioning for as long as possible.

As the disease progresses and vision acuity decreases, use contrasting colors and large fonts, low-vision aids, talking books and clocks, large-print books, voice recognition software and computer programs. For safety, adequate lighting and night lights should be used. Simplify routines, and use adaptive equipment as needed.

If muscle spasms or pain develop, use medication and/or physical therapy. Seizures may be controlled or reduced with anticonvulsants. If swallowing becomes difficult, assistance may be needed with feeding. The family may have to make a decision regarding the use of a feeding tube.

Older children may become frustrated and depressed as they become aware of their loss of abilities. Support groups can assist children and families cope with this devastating illness. Some families have established websites for support and to promote public awareness of Batten disease, e.g., www.taylorstale.com.

Research

Research is ongoing to improve diagnostic tools, provide early prenatal diagnosis, develop therapeutic strategies, identify new forms of NCL, and maintain registries for diagnostic research purposes and statistical analysis. The National Batten Disease Registry, at the New York State Institute for Basic Research in Developmental Disabilities, identifies families with Batten disease and provides a computerized central data bank of clinical information for clinicians and researchers.



Maureen Gavin, BSN, RN-BC, CDDN; Milen Velinov, MD, head of the Comprehensive Genetic Disease Program at Richmond County at the Institute for Basic Research in Developmental Disabilities; and Edie Dockter.

There are several ongoing areas of research. Currently, research trials are being conducted of Cystagon (cysteamine), to treat the infantile form of Batten disease, and of enzyme replacement therapy. An NIH-funded gene therapy trial is being conducted, and Phase 1 clinical trials involving stem cell transplant therapy (www.stemcellsinc.com/clinicaltrials) are in progress.

Resources & Support

Batten Disease Support and Research Association (BDSRA) is an international support and research networking organization. Its motto is "a light in a world of darkness." Contact information: www.bdrsa.org (click here for a list of local chapters) or or 800-448-4570. The 2010 BDSRA Conference will be held July 29-Aug. 1, 2010 in Oakbrook, IL. The Batten Disease International Awareness Days are June 5-6, 2010.

The National Batten Disease Registry, at the New York State Institute for Basic Research in Developmental Disabilities, was founded in 1987 by Krystyna Wisniewski, PhD, MD, with the assistance of Kathy Potterfield and Edie Dockter. The registry identifies families with Batten disease and provides a computerized central data bank of clinical information for clinicians and researchers. Contact information: 800-952-9628 or edith.dockter@omr.state.ny.us.

Authors' note: This article is dedicated to the memory of Krystyna Wisniewski, PhD, MD, whose research significantly advanced our knowledge of Batten disease.

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