

### **Introduction**

“Phillip” is a 17 year old young man with Batten Disease who is blind, has a seizure disorder and is developmentally delayed.

He reads Braille, and uses a white cane and sighted guide for mobility, but shuffles his feet and walks with a crouched gait. As he has gotten older, his hip and knee extension has become more limited and more support has been needed from his sighted guide as his balance has decreased.

He has some tightness in his facial muscles with decreased articulation and intelligibility but this varies. While he occasionally bites his tongue when eating, feeding is being monitored and is otherwise okay at this time.

He needs manual cues to locate the chair and feel the back of the chair when moving from standing to sitting. We have tried a rolling walker with a seat, but he loses the orientation he normally gets from his cane. We have tried floor reaction braces for the crouching to see if we can reduce fatigue. A lightweight manual wheelchair is being requested for community mobility.

### **What is Batten Disease?**

Batten disease is a rare (1 in 20,000 children in the US) and fatal, inherited disorder of the nervous system that usually begins between 5 and 8 years of age. It is the most common form of a group of disorders known as NCLs (Neuronal Ceroid Lipofuscinoses) where there is an excessive 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 a special microscope). The lipopigments build up in cells of the brain and the eye as well as in skin, muscle, and many other tissues, causing the death of nerves cells in the brain, eye and spinal cord.

### **How is a Diagnosis Made?**

Tests for Batten disease include blood or urine tests, skin or tissue sampling, an electroencephalogram (EEG), electrical studies of the eyes, and brain scans.

### **What Medical Concerns Commonly Occur with this Condition?**

The most common medical concerns include vision loss (usually one of the earliest symptoms) and seizures.

### **What Behavioral /Developmental Concerns Occur with this Condition?**

Children with this condition have many of the following conditions:

Mood and personality changes

Clumsiness/ Balance problems

Speech changes

Progressive loss of motor function

Worsening mental impairment

## **What is the Life Expectancy for Children with Batten Disease?**

Batten Disease is often fatal by the late teens or twenties.

## **What Treatments are Available for Children with this Condition?**

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. Current experimental trials using the medicine Cystagon® are being performed at the National Institutes of Health (see below).

While there are no current treatments that can halt or reverse the symptoms of Batten disease, many of its associated conditions can be treated. Seizures can sometimes be reduced or controlled with anticonvulsant drugs and physical therapy, occupational therapy, and speech therapy may help patients retain functioning as long as possible.

## **Where Can I Get More Information?**

Batten Disease Support and Research Association (BDSRA)

<http://www.bdsra.org/>

BDSRA Southeast Chapter (NC, SC, VA) — (919)774-1933

Batten Disease Family Association

<http://www.bdfauk.freemove.co.uk/>

Batten Disease Information from the National Institute of Neurological Disorders and Stroke

<http://www.ninds.nih.gov/disorders/batten/batten.htm>

Batten Disease Diagnostic and Clinical Research Center

<http://dbb.urmc.rochester.edu/labs/pearce/bddcrc/index.htm>

**Current Drug Trial - Cystagon to Treat Infantile Neuronal Ceroid Lipofuscinosis**

<http://clinicaltrials.gov/ct/show/NCT00028262;jsessionid=978150E12B005B6D70F1AA77C89A37B8?order=1>



Produced for WATCH (Wake Area Telehealth Collaborative Helping Children with Special Needs)