Batten Disease
(Neuronal Ceroid Lipofuscinosis)

Batten disease is a fatal, inherited disorder of the nervous system. It begins in childhood.

SYMPTOMS

In some cases, the early signs are subtle. They may take the form of:

- Personality and behavior changes.
- Slow learning.
- Clumsiness.
- Stumbling.

Problems (*symptoms*) of Batten disease are linked to a buildup of substances called lipopigments in the body's tissues. Lipopigments are made up of fats and proteins. The lipopigment that is built up in Batten disease is called ceroid lipofusin. This is similar to the aging pigment in the body. Vision loss is often an early sign. So Batten disease may be first suspected during an eye exam. Often, an eye specialist or other physician may refer the child to a neurologist.

DIAGNOSIS

Diagnostic tests for this disease include:

- Blood or urine tests.
- Skin or tissue sampling.
- Brain scans.
- An EEG (*electroencephalogram*).
- Electrical studies of the eyes.

TREATMENT

As yet, no specific treatment is known that can halt or reverse the symptoms of this disease. But convulsions (*seizures*) can sometimes be reduced or controlled with anticonvulsant drugs. Other medical problems can be treated appropriately as they arise. Physical therapy and occupational therapy may help patients retain functioning as long as possible.

- Over time, affected children suffer:
  - Mental impairment.
  - Worsening seizures.
  - Progressive loss of sight and movement (*motor*) skills.
- Eventually, children with Batten disease become:
  - Blind.
  - Bedridden.
  - Demented.
- Batten disease is often fatal by the late teens or twenties.

RESEARCH IS BEING DONE

The biochemical defects that underlie several NCLs (Neuronal Ceroid Lipofuscinosis) have recently been discovered. An enzyme called “palmitoyl-protein thioesterase” has been shown to be not active enough in the infantile form of Batten disease. This condition is now referred to as CLN1. In the late infantile form (CLN2), a deficiency of an acid protease has been found as the cause of this condition. The acid protease is an enzyme that hydrolyzes proteins. A mutated gene has been identified in juvenile Batten disease (CLN3). But the protein for which this gene codes has not been identified. Also, research scientists are working with NCL animal models to improve understanding and treatment of these disorders. One research team, for example, is testing the usefulness of transplanting bone marrow in a sheep model. Other investigators are working to develop mouse models. Mouse models will make it easier for scientists to study the genetics of these diseases.

Research is continually being done to learn more about this problem.

Information provided by The National Institute of Neurological Disorders and Stroke.

Document Released: 09/09/2003 Document Re-Released: 03/16/2010
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