

**Batten Disease** is named after the British pediatrician who first described it in 1903. Also known as Spielmeyer-Vogt-Sjogren-Batten Disease, it is the most common form of a group of disorders called **neuronal ceroid lipofuscinoses (or NCLs)**. Although Batten Disease is usually regarded as the juvenile form of NCL, it has now become the term to describe all forms of NCL. The basic cause, progression, and the outcome are the same. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills. Eventually, children with Juvenile Batten Disease become blind, bedridden, and unable to communicate. Juvenile Batten Disease is always fatal by the late teens or twenties. Batten Disease is not contagious or, at this time, preventable.

### **What are the forms of NCL/Batten Disease?**

There are four main types of NCL, including two forms that begin earlier in childhood and a very rare form that strikes adults. The symptoms are similar but they become apparent at different ages and progress at different rates.

**Infantile NCL** (Santavuori-Haltia disease): begins between about 6 months and 2 years of age and progresses rapidly. Affected children fail to thrive and have abnormally small heads (microcephaly). Also typical are short, sharp muscle contractions called myoclonic jerks. Initial signs of this disorder include delayed psychomotor development with progressive deterioration, other motor disorders, or seizures. The infantile form has the most rapid progression and children live into their mid childhood years.

**Late Infantile NCL** (Jansky-Bielschowsky disease) begins between ages 2 and 4. The typical early signs are loss of muscle coordination (ataxia) and seizures along with progressive mental deterioration.. This form progresses rapidly and ends in death between ages 8 and 12.

**Juvenile NCL (Batten Disease)** begins between the ages of 5 and 8 years of age. The typical early signs are progressive vision loss, seizures, ataxia or clumsiness. This form progresses less rapidly and ends in death in the late teens or early 20s, although some may live into their 30s.

**Adult NCL** (Kufs Disease or Parry's Disease) generally begins before the age of 40, causes milder symptoms that progress slowly, and does not cause blindness. Although age of death is variable among affected individuals, this form does shorten life expectancy.

### **How many people have these disorders?**

Batten Disease and other forms of NCL occur in an estimated 2 to 4 of every 100,000 births in the United States. These disorders appear to be more common in Finland, Sweden, other parts of northern Europe; and Newfoundland, Canada. The disease has been identified worldwide. Although NCLs are classified as rare diseases, they often strike more than one person in families that carry the defective gene.

### **How are NCLs inherited?**

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.

### **What causes these diseases?**

Symptoms of Batten Disease and other 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 biochemical defects causing NCLs have not been identified. Some scientists suspect these abnormal deposits result from a shortage of enzymes normally responsible for the breakdown of lipopigments. According to this theory, diseased cells produce inadequate amounts of enzymes or manufacture defective enzymes that function poorly. As a result, the cells cannot process enough of the lipopigments that occur within them, and the lipopigments accumulate. However, scientists have not pinpointed what specific enzymes are at fault or determined how the stored lipopigments damage nerve cells.

Other scientists believe that abnormal lipopigment buildup may result from a glitch in the cell's production or processing. For example, diseased cells could be producing too much of a normally needed lipoprotein.

### **Is there any treatment?**

As yet, no specific treatment is known that can halt or reverse the symptoms of Batten Disease or other NCLs. Scientists pursue medical research that could someday yield an effective treatment.