Diagnosing Batten disease
Vision loss is an early sign of Juvenile, or CLN3 Batten disease, that may present without any other symptoms. Because the ophthalmologist may be the first physician to see children with vision loss, they can play a vital role in diagnosing the CLN3/Juvenile form of Batten disease — one of the autosomal recessively inherited lysosomal diseases known as neuronal ceroid lipofuscinos (NCL). The funduscopic exam can detect abnormalities within the eye that occur in this form of Batten disease such as:

- Bull’s eye maculopathy
- Optic disc pallor
- Optic atrophy
- Progressive vascular attenuation
- Pigmentary retinopathy

About the Batten Disease Support & Research Association
The Batten Disease Support & Research Association (BDSRA) is a nonprofit organization headquartered in Columbus, Ohio, dedicated to funding research for treatments and cures, advancing education, providing family support services, advocating within systems and promoting awareness of the disease and its impact. Founded in 1987, BDSRA is the largest support and research organization dedicated to all forms of Batten disease in North America. BDSRA facilitates support groups, plans educational initiatives, provides access to cell and tissue banks, funds research studies, hosts an annual national conference for families and coordinates educational programming for professionals.
Signs and Symptoms of CLN3/Juvenile Batten disease

Batten disease is an autosomal recessive, life-limiting disease of the nervous system that presents primarily in childhood. Early symptoms of this disorder typically appear between the ages of 5 to 10 years of age, when parents, caregivers, and teachers begin to notice vision loss, and/or onset of seizures in a previously healthy and normally developing child. Other soft signs can be personality or behavioral changes, learning challenges, clumsiness or falling without cause.

As the disorder progresses, affected children experience severe visual impairment, uncontrolled seizures, loss of gross and fine motor abilities, and cognitive decline, to the point of becoming bedridden, blind, and with full dementia. The life span of a child with CLN3 is usually in the late teens or early twenties.

Batten disease is sometimes difficult to diagnose, and families who have a child with the disease report they have experienced an arduous diagnostic journey, having received many inaccurate diagnoses along the way.

There are 14 identified forms of the disease, which are most commonly diagnosed between infancy and school age. There are also less common adult forms of Batten disease including Kufs, Parry and ANCL disease. According to the U.S. Centers for Disease Control, the incidence of Batten disease is 2-4 cases per 100,000 live births.

Helping the Newly Diagnosed Family with CLN3/Juvenile Batten disease

Because new developments in Batten disease research are common, making sure children with CLN3 get swift and accurate diagnoses is very important. As possible treatments come through the pipeline, children’s ability to participate in trials, receive special support services, engage with other families and be assured of high quality care make all the difference in their quality of life. The ophthalmologist should assure that genetic testing is completed and that families have excellent referrals (often in children’s hospital clinics) for pediatric neurology, genetic counseling, social work services for community supports and OT/PT care as a start. The BDSRA can be very helpful in making these connections with clinicians and welcome your call or email. For detailed information for families, see bdsra.org.

Journal Articles & Sources

- **Surv Ophthalmol.**
- **Juvenile neuronal ceroid lipofuscinosis (JNCL) and the eye.**
  Bozorg S1, Ramirez-Montalegre D, Chung M, Pearce DA
- **The Neuronal Ceroid Lipofuscinoses (Batten Disease), Second Edition.**
  Edited by Sara E. Mole, Ruth E. Williams, and Hans H. Goebel.
- **www.slideshare.net/bdsra**