CONDITION

Batten disease

Learn how Batten disease affects the nervous system, including its symptoms, inheritance, diagnosis, treatment, complications, and ongoing care.

WHAT IT IS
Rare inherited lysosomal storage disorder

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Seizures, progressive vision loss, loss of learned skills, walking difficulties

WHAT CAUSES IT
Inherited genetic variants

HOW IT IS DIAGNOSED
Genetic testing, sometimes lysosomal enzyme testing

MAIN TREATMENT
Supportive care and enzyme replacement therapy

CAN IT BE CURED?
No, most forms have no cure

IS IT AN EMERGENCY?
Sometimes, seizure lasting 5 minutes or longer

LONG-TERM OUTLOOK
Progressive decline at type-dependent rates

Overview

Batten disease is a group of rare inherited disorders that damage the brain, eyes, and other parts of the nervous system over time. Doctors also call these conditions neuronal ceroid lipofuscinoses , or NCLs .

NCLs are lysosomal storage disorders. Lysosomes are structures inside cells that break down and recycle certain materials. In Batten disease, a genetic change interferes with this process. Material builds up inside cells, and nerve cells gradually stop working normally.

There are several types of Batten disease. They are usually named for the affected gene, such as CLN1, CLN2, or CLN3 disease. The types differ in their usual age of onset, early symptoms, and rate of progression. Although many forms begin during childhood, some first appear in adolescence or adulthood.

The term "Batten disease" once referred mainly to juvenile CLN3 disease. It is now commonly used for the larger group of NCL disorders.

Batten disease matters because it is progressive, meaning symptoms tend to increase over time. Early genetic and enzyme testing can identify the specific type, help families understand what to expect, and determine whether a disease-specific treatment or clinical trial may be available. Earlier diagnosis also allows supportive therapies to begin before problems become severe.

Symptoms

Symptoms vary by NCL type and from person to person. Even relatives with the same genetic change may have different experiences.

Early childhood forms may begin with delayed development or a loss of skills a child had already learned. Later childhood forms may first cause vision changes, learning difficulties, or seizures. Adult-onset disease may begin with movement problems, changes in thinking, or seizures, and vision may be less affected in some adult forms.

Possible symptoms include:

Seizures can take different forms. Some cause shaking and loss of awareness. Others may look like brief staring, sudden muscle jerks, stiffening, or unexplained falls.

Vision loss can be an early sign, especially in CLN3 disease. A child may have trouble seeing in dim light, reading, recognizing objects, or moving through unfamiliar spaces. An eye examination may show damage to the retina, the light-sensitive tissue at the back of the eye.

Symptoms generally progress, but the speed and pattern depend strongly on the NCL type. A specialist familiar with the specific genetic diagnosis can provide more useful information than the general label "Batten disease" alone.

- Seizures
- Progressive loss of vision
- Increasing difficulty with walking, balance, or coordination
- Muscle stiffness, muscle spasms, or involuntary movements
- Declining school performance or difficulty learning new information
- Loss of speech or other communication skills
- Changes in attention, mood, sleep, or behavior

- Difficulty chewing or swallowing
- Loss of bladder or bowel control
- Increasing need for help with daily activities

Causes and risk factors

Batten disease is caused by a disease-related change, or variant, in an NCL-associated gene. These genes provide instructions for proteins involved in cell recycling, movement of materials within cells, or other functions important to nerve cells.

Most NCL types follow an autosomal recessive inheritance pattern. This means a person usually has a disease-causing variant in both copies of the same gene. Each parent commonly carries one altered copy but does not have symptoms.

When two people carry disease-causing variants in the same recessive NCL gene, each pregnancy has its own chance of resulting in an affected child, an unaffected carrier, or a child who inherited neither variant. A genetic counselor can explain the exact probabilities for a particular family.

Some adult-onset forms have different inheritance patterns. For example, CLN4 disease is usually autosomal dominant, meaning one altered gene copy can cause the condition.

The main risk factor is having a disease-causing genetic variant. A family history may raise suspicion, but Batten disease can occur without a known family history. This may happen because carriers usually have no symptoms, the family is small, or the condition was previously misidentified.

Batten disease is not caused by parenting, diet, infection, or something a person did during pregnancy.

Diagnosis

Diagnosis often begins when a child or adult develops an unusual combination of seizures, vision loss, developmental regression, or movement problems. Because these symptoms can have many causes, evaluation usually involves several tests.

Genetic testing is central to diagnosis. A blood or saliva sample can be checked for variants in multiple NCL genes. Finding disease-causing variants can confirm the NCL type and help guide treatment, family testing, and counseling.

For suspected CLN1 or CLN2 disease, blood-based tests can measure the activity of specific

lysosomal enzymes. Low activity of palmitoyl-protein thioesterase 1, or PPT1, supports CLN1 disease. Low activity of tripeptidyl peptidase 1, or TPP1, supports CLN2 disease. Genetic testing is generally used to confirm the result.

Other parts of the evaluation may include:

A skin or other tissue biopsy was used more often in the past to look for characteristic storage material under a microscope. It is now less commonly needed when genetic and enzyme testing provide a clear diagnosis.

When a familial variant is known, genetic counseling can address carrier testing and reproductive options. These may include testing during pregnancy or testing embryos created through in vitro fertilization. These decisions are personal and should be discussed without pressure.

- A neurological examination of movement, reflexes, coordination, and development
- An eye examination, sometimes with retinal imaging or electrical testing of retinal function
- An electroencephalogram, or EEG, to measure the brain's electrical activity
- Magnetic resonance imaging, or MRI, to look for changes in the brain
- Developmental, cognitive, speech, and movement assessments
- Swallowing and nutrition evaluations when eating becomes difficult

Treatment and management

There is not yet a cure for most forms of Batten disease. Care focuses on slowing disease where a specific therapy exists, controlling symptoms, maintaining function, and supporting quality of life.

Cerliponase alfa , sold as Brineura, is an enzyme replacement therapy for CLN2 disease caused by TPP1 deficiency. In the United States, it is approved to slow the loss of walking ability in symptomatic children aged 3 years and older with late-infantile CLN2 disease.

Cerliponase alfa replaces the missing TPP1 enzyme. It is delivered into fluid-filled spaces in the brain through a surgically implanted reservoir and catheter. It may slow decline but does not restore skills that have already been lost or treat other NCL types. Treatment requires regular specialist visits and monitoring for device problems, infection, and allergic reactions.

Supportive treatment is tailored to current symptoms and may involve:

If swallowing is no longer safe or a person cannot take in enough nutrition, the care team may

discuss tube feeding. This decision considers safety, comfort, nutritional needs, and the family's goals.

Palliative care can be added at any stage. It is not limited to end-of-life care. It can help manage pain and other symptoms, coordinate services, support decision-making, and address emotional or spiritual concerns.

Clinical trials are studying gene therapies and other approaches for several NCL types. Eligibility depends on the specific gene, age, symptoms, and study requirements. Participation is voluntary, and possible benefits and burdens should be reviewed carefully with the research team.

- Anti-seizure treatment and an individualized seizure action plan
- Physical therapy to support mobility, positioning, and joint movement
- Occupational therapy for daily activities and adaptive equipment
- Speech therapy and communication devices
- Swallowing assessment and nutrition support
- Medicines or therapy for muscle stiffness, abnormal movements, sleep, mood, or discomfort
- Low-vision services and orientation or mobility training
- Educational accommodations and individualized school support
- Respiratory care and help clearing secretions when needed

Complications

As Batten disease progresses, loss of mobility can lead to falls, joint stiffness, contractures, pressure injuries, and reduced bone strength. Contractures occur when muscles or tendons shorten and limit movement.

Swallowing problems can cause dehydration, poor nutrition, or aspiration. Aspiration means food, liquid, or saliva enters the airway. It can lead to lung infection.

Seizures may become more frequent or difficult to control. Communication loss can also make pain, infection, constipation, or emotional distress harder to recognize.

Other possible complications include sleep disruption, breathing problems, dental difficulties, loss of independence, and increasing need for full-time care. The condition can also affect the emotional, social, and financial well-being of the entire family.

Planning ahead for equipment, home accessibility, school services, respite care, and future

medical decisions can reduce avoidable stress.

Follow up

Regular follow-up is usually coordinated by a neurologist or metabolic specialist. Depending on the person's needs, the team may include genetics specialists, eye doctors, rehabilitation therapists, dietitians, speech-language pathologists, pulmonologists, palliative care clinicians, educators, and social workers.

Routine visits may review seizures, vision, movement, communication, swallowing, nutrition, sleep, breathing, comfort, and caregiver needs. Medication effects and equipment fit should also be reassessed. People receiving cerliponase alfa need additional monitoring of the implanted delivery system and treatment-related reactions.

Contact the care team promptly for a meaningful change in seizure pattern, repeated choking, reduced drinking, new breathing symptoms, unexplained decline, or signs of pain. Fever, scalp redness, swelling, drainage, or fluid leakage near an implanted reservoir requires urgent medical advice.

Seek emergency help for:

During a convulsive seizure, protect the person from nearby hazards, place them on their side when possible, and time the seizure. Do not restrain them or put anything in their mouth.

Families may benefit from genetic counseling after diagnosis and again when considering future pregnancies or testing adult relatives. Caregivers should also ask about mental health support, respite services, and patient organizations familiar with NCL disorders.

- A seizure lasting 5 minutes or longer, unless the person's emergency plan gives different instructions
- Repeated seizures without recovery between them
- Severe trouble breathing, blue or gray lips, or choking that does not clear
- Unresponsiveness, a serious fall, or a significant head injury
- A sudden severe reaction during or after an infusion

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