

Batten Disease

Background

Batten disease is the common name for a group of rare, inherited neurodegenerative disorders, also known as neuronal ceroid lipofuscinoses (NCLs). Genetic mutation disrupts the cells' ability to dispose of cellular waste. The buildup of waste within cells leads to the death of neurons (brain cells) and progressive nervous system decline. There are 14 recognized forms of Batten disease, each linked to a specific gene mutation, that differ by severity, average age of onset, and rate of disease progression.

Batten disease is characterized by progressive loss of vision, seizures, loss of cognitive and motor functions, and altered behavior and emotions. Children with Batten disease will usually grow and meet early developmental milestones before they stop progressing and lose acquired skills. Developmental regression can affect posture, walking, writing, dressing, bathing, and feeding. Due to such overlapping symptoms, Batten disease can be misdiagnosed as autism, epilepsy, and other developmental disorders.

Common clinical hallmarks of Batten disease include:

- Progressive visual impairment leading to blindness
- Subtle to more pronounced personality and behavioral changes
- Seizures
- Echolalia (repetitive speech)
- Loss of speech
- Slowed movement
- Loss of coordination and balance
- Spasms, tremors, jerky movements
- Sleep disturbances
- Cognitive decline, dementia

There is no cure for Batten disease. Early diagnosis and treatment aim to slow disease progression and improve quality of life. Treatment includes symptom management, targeted therapies, and supportive care. Given the progressive and debilitating nature of Batten disease, respect for student and family autonomy and dignity are essential. Mental health and family counseling are essential following the diagnosis.



Top Takeaways for School

Batten disease is the most common neurodegenerative disorder in childhood. Staff education is key to understanding the progressive nature of the disease and managing expectations.

The progressive degeneration of physical abilities can affect mobility, positioning, access, and participation. Activities should be balanced to build on the student's strengths while supporting their gross and fine motor abilities.

Changes in vision can quickly progress. Educational interventions may be needed to support learning and participation for visual impairment.

Progressive vision loss and communication decline often lead children to rely more heavily on their sense of hearing for learning, navigation, and comfort. Consider sensory input throughout the school day.

Recommendations for nutrition and mealtime support may be necessary to reduce the child's risk of aspiration when eating or drinking.

Side effects of anti-seizure medication can cause fatigue, inattention, and restlessness that may further affect school performance.

Allow the student to determine their level of participation. Focus on what the student finds enjoyable.

Families may be navigating overwhelming medical information and emotions. Build trust through clear, compassionate communication. Provide regular updates about their child.

Considerations for the Individualized Healthcare Plan (IHP)

- Nursing diagnoses: Risk for injury, impaired physical mobility, disturbed sensory perception, impaired verbal communication, impaired swallowing, cognitive impairment
- Student-specific seizure triggers and prevention tips
- Nutrition interventions and equipment (consider brand/size of feeding tube, tube replacement, water flushes, fluid intake goal and supplements); note school district policy on tube replacement and consider keeping backup feeding tube kit at school if applicable
- Use of specialized equipment, adaptive equipment and orthotics
- Activity, positioning, transferring (consider precautions and/or restrictions)
- Skin check, pressure relief techniques
- Consider emergency action plans (EAPs) and emergency evacuation plans (EEPs) related to special health care needs, including staff education/training

Discussion Starters for the Educational Team

1. Would the student benefit from evaluations or assessments in any of the following areas: physical therapy, occupational therapy, speech and language therapy, assistive technology, adapted physical education, functional behavior, psychology, hearing and vision?
2. Would the student benefit from additional academic support and/or modified education (e.g., copies of notes, extra time, reduced workload, simplified instructions, alternative formats for presentation of material, 504/IEP)?
3. Does the student need additional adult support to access the academic curriculum in the least restrictive environment?
4. Is the physical school environment safely accessible for the student's mobility needs (e.g., entry and exit, ramps, location of classes, access to elevator, doorways)?
5. Does the classroom environment support the student's needs and/or equipment (e.g., desk/seating options, maneuverability space, electrical outlets, flash pass for bathroom or nurse)?
6. Does the student require activity precautions to prevent injury?
7. Will staff receive education/training to implement the student-specific emergency plan?

Resources

Kennedy Krieger Institute: Batten Disease Clinic
kennedykrieger.org/patient-care/centers-and-programs/batten-disease-clinic

Batten Disease Support, Research, and Advocacy Foundation (BDSRA)
bdsrafoundation.org/

Beyond Batten Disease Foundation
beyondbatten.org/



For more information, please scan the QR code or visit: KennedyKrieger.org/SHNIC

