

Unverricht-Lundborg disease

Description

Unverricht-Lundborg disease is a rare inherited form of epilepsy. Affected individuals usually begin showing signs and symptoms of the disorder between the ages of 6 and 15.

Unverricht-Lundborg disease is classified as a type of progressive myoclonus epilepsy. People with this disorder experience episodes of involuntary muscle jerking or twitching (myoclonus) that increase in frequency and severity over time. Episodes of myoclonus may be brought on by physical exertion, stress, light, or other stimuli. Within 5 to 10 years, the myoclonic episodes may become severe enough to interfere with walking and other everyday activities.

Affected individuals also usually have seizures involving loss of consciousness, muscle rigidity, and convulsions (tonic-clonic or grand mal seizures). Like the myoclonic episodes, these may increase in frequency over several years but may be controlled with treatment. After several years of progression, the frequency of seizures may stabilize or decrease.

Eventually people with Unverricht-Lundborg disease may develop problems with balance and coordination (ataxia), involuntary rhythmic shaking called intention tremor because it worsens during movement, difficulty speaking (dysarthria), depression, and a slow, mild decline in intellectual functioning.

People with Unverricht-Lundborg disease typically live into adulthood. Depending on the severity of the condition and a person's response to treatment, life expectancy may be normal.

Frequency

Progressive myoclonus epilepsy is a rare condition. Unverricht-Lundborg disease is believed to be the most common cause of this type of epilepsy, but its worldwide prevalence is unknown. Unverricht-Lundborg disease occurs most frequently in Finland, where approximately 4 in 100,000 people are affected.

Causes

Mutations in the *CSTB* gene cause Unverricht-Lundborg disease. The *CSTB* gene

provides instructions for making a protein called cystatin B. This protein reduces the activity of enzymes called cathepsins. Cathepsins help break down certain proteins in the lysosomes (compartments in the cell that digest and recycle materials). While the specific function of cystatin B is unclear, it may help protect the cells' proteins from cathepsins that leak out of the lysosomes.

In almost all affected individuals, Unverricht-Lundborg disease is caused by an increase in size of the *CSTB* gene. One region of the *CSTB* gene has a particular repeating sequence of 12 DNA building blocks (nucleotides). This sequence is normally repeated two or three times within the gene and is called a dodecamer repeat. Most people with this disorder have more than 30 repeats of the dodecamer sequence in both copies of the *CSTB* gene. A small number of people with Unverricht-Lundborg disease carry other mutations.

The increased number of dodecamer repeats in the *CSTB* gene seems to interfere with the production of the cystatin B protein. Levels of cystatin B in affected individuals are only 5 to 10 percent of normal, and cathepsin levels are significantly increased. These changes are believed to cause the signs and symptoms of Unverricht-Lundborg disease, but it is unclear how a reduction in the amount of cystatin B leads to the features of this disorder.

[Learn more about the gene associated with Unverricht-Lundborg disease](#)

- **CSTB**

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Baltic myoclonic epilepsy
- Baltic myoclonus
- Baltic myoclonus epilepsy
- EPM1
- Lundborg-Unverricht syndrome
- Mediterranean myoclonic epilepsy
- Myoclonic epilepsy of Unverricht and Lundborg
- PME
- Progressive myoclonic epilepsy
- Progressive myoclonus epilepsy 1

- ULD
- Unverricht-Lundborg syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Unverricht-Lundborg syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0751785/>)

Genetic and Rare Diseases Information Center

- Unverricht-Lundborg disease (<https://rarediseases.info.nih.gov/diseases/3876/unverricht-lundborg-disease>)

Patient Support and Advocacy Resources

- Disease InfoSearch (<https://www.diseaseinfosearch.org/>)
- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Unverricht-Lundborg disease%22](https://clinicaltrials.gov/search?cond=%22Unverricht-Lundborg+disease%22))

Catalog of Genes and Diseases from OMIM

- MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG (<https://omim.org/entry/254800>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Unverricht-Lundborg+Syndrome%5BMAJR%5D%29+AND+%28Unverricht-Lundborg+disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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