



## CLN3 gene

CLN3, battenin

### Normal Function

The *CLN3* gene provides instructions for making a protein called battenin. This protein is primarily located in the membranes of lysosomes and endosomes, which are compartments within the cell that digest and recycle materials. The protein can also be found in the membrane that surrounds the cell and in the membrane of the Golgi apparatus, which is a cell structure that modifies newly produced enzymes and other proteins. The function of battenin in these cellular structures is unclear.

Studies have associated battenin with many cellular processes, including recycling of worn-out cell parts and unneeded proteins (autophagy), maintenance of the relative acidity (pH) of lysosomes, the movement of molecules from the cell surface into the cell (endocytosis), transportation (trafficking) of proteins to where they are needed in the cell, self-destruction of cells (apoptosis), cell growth and division (proliferation), and maintenance of the body's water balance (osmoregulation). It is uncertain whether any of these varied functions is the primary role of battenin, or if they represent downstream effects.

### Health Conditions Related to Genetic Changes

#### CLN3 disease

More than 65 mutations in the *CLN3* gene have been found to cause CLN3 disease. CLN3 disease is an inherited disorder that primarily affects the nervous system. Children with this condition develop worsening vision impairment, intellectual disability, movement problems, speech difficulties, and seizures.

One *CLN3* gene mutation, found in more than 90 percent of cases, deletes about 1,000 DNA building blocks (base pairs) in the gene. This mutation, which is usually called the 1 kilobase (kb) deletion, removes a piece of the *CLN3* gene and leads to the production of an abnormally short protein that is probably broken down quickly. As a result, there is a severe reduction in the amount of functional battenin in cells. Other mutations also reduce the amount or impair the function of battenin. It is not known how the loss of this protein causes the signs and symptoms of CLN3 disease.

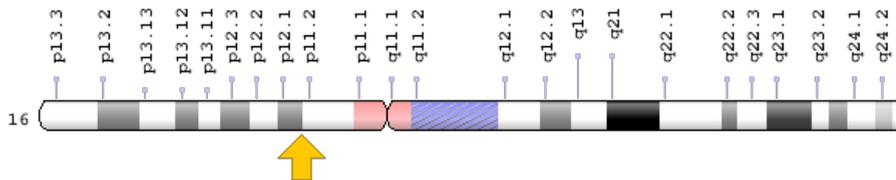
CLN3 disease is characterized by the accumulation of proteins and other substances in lysosomes. These accumulations occur in cells throughout the body; however, nerve cells seem to be particularly vulnerable to their effects. The accumulations can cause cell damage leading to cell death. The progressive death of nerve cells in the brain and other tissues leads to the neurological signs and symptoms of CLN3 disease. Additionally, it is thought that cardiac cell damage and death due

to lysosomal accumulations contribute to the heart problems in people with CLN3 disease. However, it is unclear how mutations in the *CLN3* gene are involved in the buildup of substances in lysosomes.

### Chromosomal Location

Cytogenetic Location: 16p12.1, which is the short (p) arm of chromosome 16 at position 12.1

Molecular Location: base pairs 28,466,653 to 28,492,302 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- BATTENIN
- BTN1
- BTS
- ceroid-lipofuscinosis, neuronal 3
- CLN3\_HUMAN
- JNCL
- MGC102840

### Additional Information & Resources

#### Educational Resources

- Beyond Batten Disease Foundation: CLN3 gene  
<https://beyondbatten.org/research/cln3-gene/>
- Molecular Biology of the Cell (fourth edition, 2002): Lysosomes Are the Principal Sites of Intracellular Digestion  
<https://www.ncbi.nlm.nih.gov/books/NBK26844/#A2365>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CLN3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- CLN3 GENE  
<http://omim.org/entry/607042>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_CLN3.html](http://atlasgeneticsoncology.org/Genes/GC_CLN3.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=CLN3%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:2074](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:2074)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:1201>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1201>
- UniProt  
<https://www.uniprot.org/uniprot/Q13286>
- University College London: CLN3 Gene Mutation Database  
<https://www.ucl.ac.uk/ncl-disease/ncl-resource-gateway-batten-disease/mutation-and-patient-database/mutation-and-patient-datasheets-2>

### **Sources for This Summary**

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