



## EFHC1 gene

EF-hand domain containing 1

### Normal Function

The *EFHC1* gene provides instructions for making a protein called EF-hand domain containing protein 1 (EFHC1). The EFHC1 protein interacts with another protein that acts as a calcium channel, allowing positively charged calcium atoms (calcium ions) to cross the cell membrane. The movement of these ions is critical for normal signaling between nerve cells (neurons) in the brain and other parts of the nervous system. The role of the EFHC1 protein is not well understood, although it is thought to help regulate the balance of calcium ions inside the cell (calcium homeostasis). Studies also show that the EFHC1 protein may stimulate the self-destruction of cells (apoptosis).

### Health Conditions Related to Genetic Changes

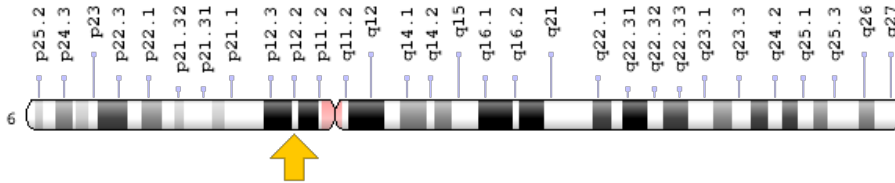
#### Juvenile myoclonic epilepsy

Mutations in the *EFHC1* gene have been identified in a small number of people with juvenile myoclonic epilepsy. This condition typically begins in childhood or adolescence and causes recurrent myoclonic seizures, which are characterized by rapid, uncontrolled muscle jerks. Affected individuals can also have other types of seizures called generalized tonic-clonic seizures (or grand mal seizures) and absence seizures. Most gene mutations associated with juvenile myoclonic epilepsy replace single protein building blocks (amino acids) in the EFHC1 protein. The function of the altered protein is thought to be reduced. Although it is unclear how *EFHC1* gene mutations lead to juvenile myoclonic epilepsy, researchers have suggested that a decrease in EFHC1 protein function reduces apoptosis, leading to more neurons than normal, and disrupts calcium homeostasis. Together, these changes may lead to overstimulation of the neurons, causing seizures characteristic of juvenile myoclonic epilepsy.

## Chromosomal Location

Cytogenetic Location: 6p12.2, which is the short (p) arm of chromosome 6 at position 12.2

Molecular Location: base pairs 52,420,196 to 52,497,198 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- EF-hand domain (C-terminal) containing 1
- EF-hand domain-containing protein 1
- EFHC1\_HUMAN
- myoclonin-1

## Additional Information & Resources

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28EFHC1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- EF-HAND DOMAIN (C-TERMINAL)-CONTAINING PROTEIN 1  
<http://omim.org/entry/608815>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_EFHC1.html](http://atlasgeneticsoncology.org/Genes/GC_EFHC1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=EFHC1%5Bgene%5D>

- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:16406](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:16406)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:114327>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/114327>
- UniProt  
<https://www.uniprot.org/uniprot/Q5JVL4>

### Sources for This Summary

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<http://omim.org/entry/608815>
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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15258581>

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<https://ghr.nlm.nih.gov/gene/EFHC1>

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