



ELOVL4 gene

ELOVL fatty acid elongase 4

Normal Function

The *ELOVL4* gene provides instructions for making a protein that is found primarily in the retina, the specialized light-sensitive tissue that lines the back of the eye. Within the retina, the ELOVL4 protein is produced in specialized light receptor cells (photoreceptors). The ELOVL4 protein is also found in the brain and skin, but less is known about its activity (expression) in these structures.

Inside photoreceptor cells, this protein is located in a cell structure called the endoplasmic reticulum that is involved in protein production, processing, and transport. The ELOVL4 protein plays a role in making a group of fats called very long-chain fatty acids. The protein helps add carbon molecules to long-chain fatty acids, making them very long-chain fatty acids. The function of the very long-chain fatty acids produced by the ELOVL4 protein is unknown.

Health Conditions Related to Genetic Changes

Stargardt macular degeneration

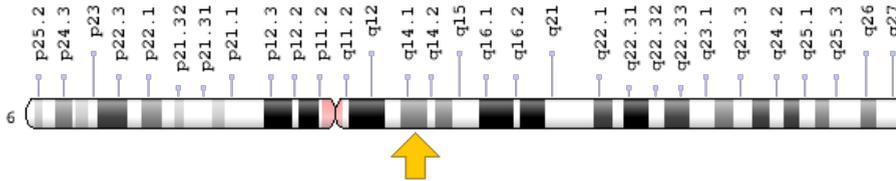
At least three mutations in the *ELOVL4* gene have been found to cause Stargardt macular degeneration. These mutations create a premature stop signal in the instructions used to make the ELOVL4 protein. As a result, the protein cannot be retained in the endoplasmic reticulum of photoreceptor cells. Instead, the ELOVL4 protein forms clumps (aggregates). These aggregates cannot make very long-chain fatty acids and may interfere with cell functions, ultimately leading to cell death. The loss of photoreceptor cells causes progressive vision loss in people with Stargardt macular degeneration. Mutations in the *ELOVL4* gene are a rare cause of this condition.

Age-related macular degeneration

Chromosomal Location

Cytogenetic Location: 6q14.1, which is the long (q) arm of chromosome 6 at position 14.1

Molecular Location: base pairs 79,914,814 to 79,947,553 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- elongation of very long chain fatty acids (FEN1/Elo2, SUR4/Elo3, yeast)-like 4
- elongation of very long chain fatty acids protein 4
- ELOV4_HUMAN

Additional Information & Resources

Educational Resources

- Webvision: The Organization of the Retina and Visual System (2007): Photoreceptors
<https://www.ncbi.nlm.nih.gov/books/NBK11522/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ELOVL4%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+2520+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- ELONGATION OF VERY LONG CHAIN FATTY ACIDS-LIKE 4
<http://omim.org/entry/605512>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ELOVL4%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:14415
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6785>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6785>
- UniProt
<https://www.uniprot.org/uniprot/Q9GZR5>

Sources for This Summary

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