FAM126A gene

family with sequence similarity 126 member A

Normal Function

The *FAM126A* gene provides instructions for making a protein called hyccin, which is active (expressed) throughout the nervous system. Researchers believe that hyccin is involved in the formation of myelin, which is the covering that protects nerves and promotes the efficient transmission of nerve impulses. Hyccin is also active in the lens of the eye, the heart, and the kidneys; however, the protein's function in these tissues is unclear.

Health Conditions Related to Genetic Changes

Hypomyelination and congenital cataract

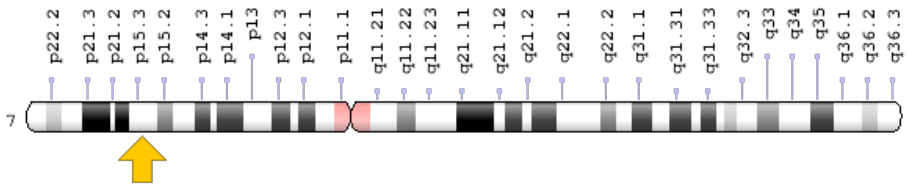
At least four mutations in the *FAM126A* gene have been found to cause hypomyelination and congenital cataract. Most mutations delete a large portion of the gene or create a premature stop signal in the instructions for making hyccin. These mutations prevent the production of any functional protein. One *FAM126A* gene mutation allows some protein to be produced. This mutation replaces the protein building block (amino acid) leucine with the amino acid proline at position 53 in the hyccin protein (written as Leu53Pro or L53P). People with the Leu53Pro mutation tend to have milder symptoms than those with mutations that prevent the production of any protein.

Any disruption in the production of hyccin impairs its role in the formation of myelin, leading to neurological problems such as intellectual disability and walking difficulties. It is unclear how a lack of hyccin causes a clouding of the lens (cataract) in both eyes that is typically present from birth in affected individuals. The neurological problems and cataracts are the characteristic features seen in people with hypomyelination and congenital cataract.

Chromosomal Location

Cytogenetic Location: 7p15.3, which is the short (p) arm of chromosome 7 at position 15.3

Molecular Location: base pairs 22,895,848 to 23,014,133 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DRCTNNB1A
- family with sequence similarity 126, member A
- HCC
- HLD5
- HYCC1
- HYCCI_HUMAN
- hyccin

Additional Information & Resources

Clinical Information from GeneReviews

- Hypomyelination and Congenital Cataract
  https://www.ncbi.nlm.nih.gov/books/NBK2587

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28hyccin%5BTIAB%5D%29+OR+%28DRCTNNB1A%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+720+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- FAMILY WITH SEQUENCE SIMILARITY 126, MEMBER A
  http://omim.org/entry/610531

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FAM126A%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:24587
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:84668
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/84668
- UniProt
  https://www.uniprot.org/uniprot/Q9BYI3

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