



CHM gene

CHM, Rab escort protein 1

Normal Function

The *CHM* gene provides instructions for producing the Rab escort protein-1 (REP-1), which is active (expressed) throughout the body. As an escort protein, REP-1 attaches (binds) to one of a number of Rab proteins. Following a chemical modification, REP-1 then directs the Rab protein to the membrane of one of the cell's compartments (organelles). While attached to the membrane, the Rab protein plays a role in directing the movement of proteins and organelles within cells (intracellular trafficking). After the Rab protein has reached its destination, it is released by REP-1 which then attaches to another Rab protein to begin the process again.

Health Conditions Related to Genetic Changes

Choroideremia

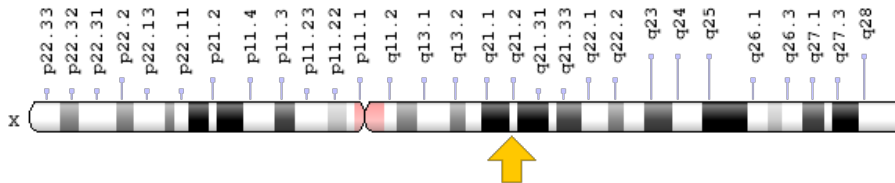
More than 140 mutations in the *CHM* gene have been found to cause choroideremia. Nearly all of these mutations lead to the production of an abnormally small, nonfunctional REP-1 protein. Other gene mutations result in a decrease in the protein's function or delete part or all of the gene and abolish REP-1 protein production. A lack of normal REP-1 disrupts the ability of Rab proteins to aid in intracellular trafficking. The immobility of proteins and organelles within the cell cause the cell to die prematurely.

The REP-1 protein is active (expressed) throughout the body, as is a similar protein, REP-2. Research suggests that when REP-1 is absent or not functioning properly, REP-2 can perform the protein escort duties of REP-1 in many of the body's tissues. Very little REP-2 protein is present in the light sensitive-tissue at the back of the eye (the retina), however, so it cannot compensate for the loss of REP-1 in this tissue. Loss of REP-1 function and subsequent misplacement of Rab proteins within the cells of the retina causes the progressive vision loss characteristic of choroideremia.

Chromosomal Location

Cytogenetic Location: Xq21.2, which is the long (q) arm of the X chromosome at position 21.2

Molecular Location: base pairs 85,861,180 to 86,047,558 on the X chromosome (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- choroideremia
- choroideremia (Rab escort protein 1)
- DXS540
- FLJ38564
- GGTA
- HSD-32
- MGC102710
- RAE1_HUMAN
- REP-1
- REP-1, Rab escort protein 1
- TCD

Additional Information & Resources

Educational Resources

- Eureka Bioscience Collection: Rab GTPases: Key Regulators of Membrane Trafficking
<https://www.ncbi.nlm.nih.gov/books/NBK6035/>
- Molecular Biology of the Cell (fourth edition, 2002): Membrane protein attachment by a fatty acid chain or a prenyl group
<https://www.ncbi.nlm.nih.gov/books/NBK26878/figure/A1893/>
- Molecular Cell Biology (fourth edition, 2000): Covalently Attached Hydrocarbon Chains Anchor Some Proteins to the Membrane
<https://www.ncbi.nlm.nih.gov/books/NBK21570/#A617>

Clinical Information from GeneReviews

- Choroideremia
<https://www.ncbi.nlm.nih.gov/books/NBK1337>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CHM%5BTIAB%5D%29+OR+%28REP-1%5BTIAB%5D%29%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

- CHM GENE
<http://omim.org/entry/300390>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CHM.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CHM%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:1940
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:1121>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1121>
- UniProt
<https://www.uniprot.org/uniprot/P24386>

Sources for This Summary

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

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