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

- Eurekah 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+AND+Bdp%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
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1121
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P24386
Sources for This Summary

- OMIM: CHM GENE
  http://omim.org/entry/300390


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