

CHM gene

CHM Rab escort protein

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.

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

Tests Listed in the Genetic Testing Registry

- Tests of CHM ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1121\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1121[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29%29>)

Catalog of Genes and Diseases from OMIM

- CHM RAB ESCORT PROTEIN; CHM (<https://omim.org/entry/300390>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1121>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CHM\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CHM[gene]))

References

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Genomic Location

The *CHM* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

Last updated July 1, 2013