

## VLDLR gene

very low density lipoprotein receptor

### Normal Function

The *VLDLR* gene provides instructions for making a protein called a very low density lipoprotein (VLDL) receptor. This protein is active in many different organs and tissues, including the heart, muscles used for movement (skeletal muscles), fatty (adipose) tissue, and the kidneys. The VLDL receptor appears to play a particularly important role in the developing brain.

The VLDL receptor works together with a protein called reelin. Reelin fits into the VLDL receptor like a key in a lock, which triggers a series of chemical reactions within the cell. During early brain development, the reelin signaling pathway helps to guide the movement of immature nerve cells (neuroblasts) to their appropriate locations in the brain.

### Health Conditions Related to Genetic Changes

#### VLDLR-associated cerebellar hypoplasia

At least six mutations in the *VLDLR* gene have been found to cause *VLDLR*-associated cerebellar hypoplasia. These mutations prevent cells from producing any functional VLDL receptor protein. Without this protein, neuroblasts cannot reach the parts of the brain where they are needed. These problems with brain development predominantly affect the cerebellum, which is the part of the brain that coordinates movement. People with *VLDLR*-associated cerebellar hypoplasia have an unusually small and underdeveloped cerebellum, which leads to problems with balance and coordination (ataxia) and impaired speech. Other regions of the brain are also affected, resulting in intellectual disability and the other major features of this condition.

### Other Names for This Gene

- CARMQ1
- CHRMQ1
- FLJ35024
- VLDL receptor
- VLDLR\_HUMAN

- VLDLRCH

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of VLDLR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7436\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7436[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28VLDLR%5BTIAB%5D%29+OR+%28very+low+density+lipoprotein+receptor%5BTIAB%5D%29+OR+%28VLDL+receptor%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%29>)

### Catalog of Genes and Diseases from OMIM

- VERY LOW DENSITY LIPOPROTEIN RECEPTOR; VLDLR (<https://omim.org/entry/192977>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7436>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=VLDLR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=VLDLR[gene]))

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## Genomic Location

The *VLDLR* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

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