

## LDLR gene

low density lipoprotein receptor

### Normal Function

The *LDLR* gene provides instructions for making a protein called the low-density lipoprotein receptor. This receptor binds to particles called low-density lipoproteins (LDLs), which are the primary carriers of cholesterol in the blood. Cholesterol is a waxy, fat-like substance that is produced in the body and obtained from foods that come from animals.

Low-density lipoprotein receptors sit on the outer surface of many types of cells, where they pick up LDLs circulating in the bloodstream and transport them into the cell. Once inside the cell, the LDL is broken down to release cholesterol. The cholesterol is then used by the cell, stored, or removed from the body. After low-density lipoprotein receptors drop off their cargo, they are recycled back to the cell surface to pick up more LDLs.

Low-density lipoprotein receptors play a critical role in regulating the amount of cholesterol in the blood. They are particularly abundant in the liver, which is the organ responsible for removing most excess cholesterol from the body. The number of low-density lipoprotein receptors on the surface of liver cells determines how quickly cholesterol is removed from the bloodstream.

### Health Conditions Related to Genetic Changes

#### Familial hypercholesterolemia

Mutations in the *LDLR* gene cause a form of high cholesterol called familial hypercholesterolemia. More than 2,000 mutations have been identified in this gene. Some of these genetic changes reduce the number of low-density lipoprotein receptors produced within cells. Other mutations disrupt the receptor's ability to remove LDLs from the blood. As a result, people with mutations in the *LDLR* gene have very high blood cholesterol levels. As the excess cholesterol circulates through the bloodstream, it is deposited abnormally in tissues such as the skin, tendons, and arteries that supply blood to the heart (coronary arteries). A buildup of cholesterol in the walls of coronary arteries greatly increases a person's risk of having a heart attack.

Most people with familial hypercholesterolemia inherit one altered copy of the *LDLR*

gene from an affected parent and one normal copy of the gene from the other parent. These cases are associated with an increased risk of early heart disease, typically beginning in a person's forties or fifties. Rarely, a person with familial hypercholesterolemia is born with two mutated copies of the *LDLR* gene. This situation occurs when the person has two affected parents, each of whom passes on one altered copy of the gene. The presence of two *LDLR* gene mutations results in a more severe form of hypercholesterolemia that usually appears in childhood.

### Other Names for This Gene

- FHC
- LDL receptor
- LDLCQ2
- LDLR\_HUMAN
- Low density lipoprotein (LDL) receptor
- low density lipoprotein receptor (familial hypercholesterolemia)

### Additional Information & Resources

#### Tests Listed in the Genetic Testing Registry

- Tests of LDLR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3949\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3949[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28LDLR%5BTI%5D%29+OR+%28low+density+lipoprotein+receptor%5BTI%5D%29+OR+%28LDL+receptor%5BTI%5D%29%29+NOT+%28low+density+lipoprotein+receptor+related%5BTIAB%5D%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D%29>)

#### Catalog of Genes and Diseases from OMIM

- LOW DENSITY LIPOPROTEIN RECEPTOR; LDLR (<https://omim.org/entry/606945>)

#### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3949>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=LDLR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=LDLR[gene]))

## References

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## Genomic Location

The *LDLR* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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