

## DHCR7 gene

7-dehydrocholesterol reductase

### Normal Function

The *DHCR7* gene provides instructions for making an enzyme called 7-dehydrocholesterol reductase. This enzyme is responsible for the final step in cholesterol production in many types of cells. Specifically, 7-dehydrocholesterol reductase converts a molecule called 7-dehydrocholesterol to cholesterol.

Cholesterol is a waxy, fat-like substance that is produced in the body and obtained from foods that come from animals (particularly egg yolks, meat, poultry, fish, and dairy products). It has important functions both before and after birth. Cholesterol plays a critical role in embryonic development by interacting with signaling proteins that control early development of the brain, limbs, genital tract, and other structures. It is also a structural component of cell membranes and myelin, the fatty covering that insulates nerve cells. Additionally, cholesterol is used to make certain hormones and is important for the production of acids used in digestion (bile acids).

### Health Conditions Related to Genetic Changes

#### Smith-Lemli-Opitz syndrome

More than 200 mutations that cause Smith-Lemli-Opitz syndrome have been identified in the *DHCR7* gene. Smith-Lemli-Opitz syndrome is a developmental disorder characterized by distinctive facial features, small head size (microcephaly), intellectual disability or learning problems, and behavioral problems. The most common mutation, which is written as IVS8-1G>C, alters a single DNA building block (nucleotide) in the *DHCR7* gene. This change interferes with the normal processing of 7-dehydrocholesterol reductase, resulting in an abnormally short, nonfunctional enzyme.

Most of the known *DHCR7* mutations change single amino acids in 7-dehydrocholesterol reductase. These mutations reduce the ability of this enzyme to convert 7-dehydrocholesterol to cholesterol. Other mutations insert or delete nucleotides in the *DHCR7* gene or lead to the production of an abnormally short enzyme; these mutations eliminate the activity of the enzyme. Without functional 7-dehydrocholesterol reductase, cells are unable to produce enough cholesterol. In addition, toxic byproducts of cholesterol production (such as 7-dehydrocholesterol) can build up in the blood and other tissues. The combination of low cholesterol levels and an

accumulation of harmful substances likely disrupts the growth and development of many body systems. It is not completely understood, however, how either abnormality leads to the specific features of Smith-Lemli-Opitz syndrome.

## Other Names for This Gene

- 7-DHC reductase
- D7SR
- delta-7-dehydrocholesterol reductase
- DHCR7\_HUMAN
- sterol delta-7-reductase

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of DHCR7 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1717\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1717[geneid]))

## Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DHCR7%5BTIAB%5D%29+OR+%287-dehydrocholesterol+reductase%5BTIAB%5D%29%29+OR+%28%28sterol+delta-7-reductase%5BTIAB%5D%29+OR+%287-DHC+reductase%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

- 7-DEHYDROCHOLESTEROL REDUCTASE; DHCR7 (<https://omim.org/entry/602858>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1717>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DHCR7\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DHCR7[gene]))

## References

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Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25734025>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4343216/>)

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## Genomic Location

The *DHCR7* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

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