



CAV1 gene

caveolin 1

Normal Function

The *CAV1* gene provides instructions for making a protein called caveolin-1. This protein appears to have diverse functions in cells and tissues throughout the body.

Caveolin-1 is the major component of caveolae, which are small pouches in the membrane that surrounds cells. Caveolae have multiple functions, some of which are not well understood. They are known to be involved in the transport of molecules from the cell membrane to the interior of the cell (endocytosis), processing of molecules on their way into the cell, maintaining the cell structure, and regulating chemical signaling pathways. Studies suggest that caveolae are particularly numerous in adipocytes, which are cells that store fats for energy. Adipocytes make up most of the body's fatty (adipose) tissue. In these cells, caveolae appear to be essential for the normal transport, processing, and storage of fats.

Caveolin-1 is also found in many other parts of cells, where it regulates various chemical signaling pathways. Through these pathways, caveolin-1 is involved in regulating cell growth and division (proliferation), the process by which cells mature to perform specific functions (differentiation), cell survival and the self-destruction of cells (apoptosis), and cell movement. The functions of caveolin-1 likely differ depending on the type of cell and the part of the cell where the protein is found.

Health Conditions Related to Genetic Changes

Congenital generalized lipodystrophy

At least one mutation in the *CAV1* gene has been found to cause congenital generalized lipodystrophy (also called Berardinelli-Seip congenital lipodystrophy) type 3. This rare condition is characterized by an almost total absence of adipose tissue and a very muscular appearance. A shortage of adipose tissue leads to multiple health problems, including high levels of fats called triglycerides circulating in the bloodstream (hypertriglyceridemia) and diabetes mellitus. Additional features of congenital generalized lipodystrophy type 3 include poor growth and short stature.

The identified mutation replaces a single protein building block (amino acid) with a premature stop signal at position 38 of the caveolin-1 protein (written as Glu38Ter or E38X.) This mutation occurs in both copies of the *CAV1* gene in each cell, and it prevents cells from producing any functional caveolin-1. It is unclear how a lack of this protein leads to the particular features of congenital generalized lipodystrophy type 3. However, the absence of caveolin-1 likely disrupts the normal development and function of adipocytes, which would prevent fats from being stored normally in

adipose tissue. A lack of body fat underlies many of the signs and symptoms of this condition.

Pulmonary arterial hypertension

Other disorders

In addition to congenital generalized lipodystrophy (described above), mutations in the *CAV1* gene have been found to cause several other forms of lipodystrophy, which all involve a loss of adipose tissue. At least two mutations have been identified in people with atypical partial lipodystrophy, a condition characterized by the loss of fat under the skin of the face and upper body. Affected individuals also have hypertriglyceridemia and clouding of the lens of the eyes starting at birth (congenital cataracts). The *CAV1* gene mutations that cause this condition are present in one copy of the gene in each cell. They reduce the amount of caveolin-1 that is produced within cells.

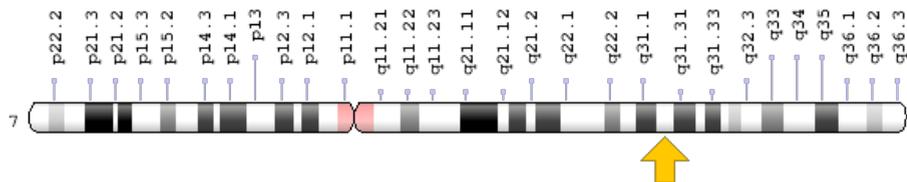
Two other mutations in the *CAV1* gene have been found to cause neonatal-onset generalized lipodystrophy syndrome. The signs and symptoms of this condition include an overall loss of body fat except in the buttocks and on the palms of the hands and soles of the feet. Additional features include thin skin with visible blood vessels; a large, triangular head; and slow weight gain in childhood. The *CAV1* gene mutations that cause this condition also occur in one copy of the gene in each cell and reduce the amount of caveolin-1 produced in cells.

Because *CAV1* gene mutations can cause several different forms of lipodystrophy, it is clear that it plays an essential role in the development of adipose tissue. However, researchers are still working to determine how a shortage of caveolin-1 results in a loss of body fat and the associated features of these conditions.

Chromosomal Location

Cytogenetic Location: 7q31.2, which is the long (q) arm of chromosome 7 at position 31.2

Molecular Location: base pairs 116,525,009 to 116,561,185 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BSCL3
- CAV
- caveolin-1 isoform alpha
- caveolin-1 isoform beta
- caveolin 1, caveolae protein, 22kDa
- cell growth-inhibiting protein 32
- CGL3
- LCCNS
- MSTP085
- PPH3
- VIP21

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Transport into the Cell from the Plasma Membrane: Endocytosis
<https://www.ncbi.nlm.nih.gov/books/NBK26870/>

Clinical Information from GeneReviews

- Heritable Pulmonary Arterial Hypertension
<https://www.ncbi.nlm.nih.gov/books/NBK1485>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CAV1%5BTI%5D%29+OR+%28caveolin+1%5BTI%5D%29+OR+%28caveolin-1%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- CAVEOLIN 1
<http://omim.org/entry/601047>
- LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 7
<http://omim.org/entry/606721>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/CAV1ID932ch7q31.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CAV1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:1527
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:857>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/857>
- UniProt
<https://www.uniprot.org/uniprot/Q03135>

Sources for This Summary

- Boscher C, Nabi IR. Caveolin-1: role in cell signaling. *Adv Exp Med Biol.* 2012;729:29-50. doi: 10.1007/978-1-4614-1222-9_3. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22411312>
- OMIM: CAVEOLIN 1
<http://omim.org/entry/601047>
- Cao H, Alston L, Ruschman J, Hegele RA. Heterozygous CAV1 frameshift mutations (MIM 601047) in patients with atypical partial lipodystrophy and hypertriglyceridemia. *Lipids Health Dis.* 2008 Jan 31;7:3. doi: 10.1186/1476-511X-7-3.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18237401>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2276215/>
- Garg A, Kircher M, Del Campo M, Amato RS, Agarwal AK; University of Washington Center for Mendelian Genomics. Whole exome sequencing identifies de novo heterozygous CAV1 mutations associated with a novel neonatal onset lipodystrophy syndrome. *Am J Med Genet A.* 2015 Aug; 167A(8):1796-806. doi: 10.1002/ajmg.a.37115. Epub 2015 Apr 21.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25898808>
- Kim CA, Delépine M, Boutet E, El Mourabit H, Le Lay S, Meier M, Nemani M, Bridel E, Leite CC, Bertola DR, Semple RK, O'Rahilly S, Dugail I, Capeau J, Lathrop M, Magré J. Association of a homozygous nonsense caveolin-1 mutation with Berardinelli-Seip congenital lipodystrophy. *J Clin Endocrinol Metab.* 2008 Apr;93(4):1129-34. doi: 10.1210/jc.2007-1328. Epub 2008 Jan 22.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18211975>
- Quest AF, Lobos-González L, Nuñez S, Sanhuesa C, Fernández JG, Aguirre A, Rodríguez D, Leyton L, Torres V. The caveolin-1 connection to cell death and survival. *Curr Mol Med.* 2013 Feb; 13(2):266-81. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23228128>

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/CAV1>

Reviewed: January 2016
Published: November 12, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services