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

MedlinePlus Genetics provides information about 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.

Other Names for This Gene

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

- PPH3
- VIP21

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CAV1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=857[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; CAV1 (https://omim.org/entry/601047)
- LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 7; FPLD7 (https://omim.org/entry/606721)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/857)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CAV1[gene])

References

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Genomic Location

The *CAV1* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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