CAVIN1 Gene

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caveolae associated protein 1

genes

1. Normal Function

The *CAVIN1* gene provides instructions for making a protein called cavin-1. This protein is found in cells and tissues throughout the body. It is most abundant in several types of cells: osteoblasts, which are cells that build bones; muscle cells; and adipocytes, which are cells that store fats for energy. Adipocytes make up most of the body's fatty (adipose) tissue.

Studies suggest that cavin-1 plays an essential role in forming and stabilizing 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. Caveolae are particularly numerous in adipocytes, where they appear to be essential for the normal transport, processing, and storage of fats.

Within cells, cavin-1 is also found in the nucleus and in the fluid that surrounds the nucleus (the cytoplasm). In addition to its role in caveolae, studies suggest that this protein is involved in repairing damage to the outer cell membrane, cell growth and division (proliferation), cell movement, stopping cell division in older cells (senescence), and regulating various chemical signaling pathways. The functions of cavin-1 likely differ depending on the type of cell and the part of the cell where the protein is found.

2. Health Conditions Related to Genetic Changes

2.1. Congenital Generalized Lipodystrophy

More than 10 mutations in the *CAVIN1* gene have been found to cause congenital generalized lipodystrophy (also called Berardinelli-Seip congenital lipodystrophy) type 4. 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 4 include muscle weakness,

delayed development, joint abnormalities, a narrowing of the lower part of the stomach (pyloric stenosis), and severe abnormalities of the heart rhythm (arrhythmias) that can lead to sudden death.

All of the identified *CAVIN1* gene mutations prevent cells from producing any functional cavin-1. A lack of this protein probably impairs the formation of caveolae. Researchers suspect that a shortage of these important structures on the cell membrane disrupts many cell functions. However, it is unknown specifically how the absence of cavin-1 leads to a loss of body fat and the other health problems associated with congenital generalized lipodystrophy type 4.

3. Other Names for This Gene

- CAVIN
- cavin-1
- CGL4
- FKSG13
- polymerase I and transcript release factor
- PTRF
- RNA polymerase I and transcript release factor
- TTF-I interacting peptide 12

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