CAV3-Related Distal Myopathy

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CAV3-related distal myopathy is one form of distal myopathy, a group of disorders characterized by weakness and loss of function affecting the muscles farthest from the center of the body (distal muscles), such as those of the hands and feet.

genetic conditions

1. Introduction

People with *CAV3*-related distal myopathy experience wasting (atrophy) and weakness of the small muscles in the hands and feet that generally become noticeable in adulthood. A bump or other sudden impact on the muscles, especially those in the forearms, may cause them to exhibit repetitive tensing (percussion-induced rapid contraction). The rapid contractions can continue for up to 30 seconds and may be painful. Overgrowth (hypertrophy) of the calf muscles can also occur in *CAV3*-related distal myopathy. The muscles closer to the center of the body (proximal muscles) such as the thighs and upper arms are normal in this condition.

2. Frequency

The prevalence of *CAV3*-related distal myopathy is unknown. Only a few affected individuals have been described in the medical literature.

3. Causes

CAV3-related distal myopathy is part of a group of conditions called caveolinopathies, which are muscle disorders caused by mutations in the *CAV3* gene. The *CAV3* gene provides instructions for making a protein called caveolin-3, which is found in the membrane surrounding muscle cells. This protein is the main component of caveolae, which are small pouches in the muscle cell membrane. Within the caveolae, the caveolin-3 protein acts as a scaffold to organize other molecules that are important for cell signaling and maintenance of the cell structure.

CAV3 gene mutations result in a shortage of caveolin-3 protein in the muscle cell membrane and a reduction in the number of caveolae. Researchers suggest that a shortage of caveolae impairs the structural integrity of muscle cells, interferes with cell signaling, and causes the self-destruction of cells (apoptosis). The resulting degeneration of muscle tissue leads to the signs and symptoms of *CAV3*-related distal myopathy.

In addition to *CAV3*-related distal myopathy, *CAV3* gene mutations can cause other caveolinopathies including limb-girdle muscular dystrophy, rippling muscle disease, isolated hyperCKemia, and a heart disorder called hypertrophic cardiomyopathy. Several *CAV3* gene mutations have been found to cause different caveolinopathies in different individuals. It is unclear why a single *CAV3* gene mutation may cause different patterns of signs and symptoms, even within the same family.

3.1. The Gene Associated with CAV3-Related Distal Myopathy

- CAV3

4. Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with *CAV3*-related distal myopathy or another caveolinopathy. Rare cases result from new mutations in the gene and occur in people with no history of caveolinopathies in their family.

5. Other Names for This Condition

- distal myopathy, Tateyama type
- MPDT

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