Isolated hyperCKemia

Description

Isolated hyperCKemia is a condition characterized by elevated levels of an enzyme called creatine kinase in the blood. In affected individuals, levels of this enzyme are typically 3 to 10 times higher than normal. While elevated creatine kinase often accompanies various muscle diseases, individuals with isolated hyperCKemia have no muscle weakness or other symptoms. Some people with this condition have abnormalities of muscle cells that can be seen with a microscope, such as unusual variability in the size of muscle fibers, but these changes do not affect the function of the muscle.

Frequency

The prevalence of isolated hyperCKemia is unknown. Because the condition has no symptoms, it is likely that some cases never come to medical attention.

Causes

Isolated hyperCKemia is one of a group of conditions called caveolinopathies, which are 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). Creatine kinase is released when muscle cells are broken down. Although no muscle weakness occurs in isolated hyperCKemia, destruction of some muscle cells may lead to the elevated blood levels of creatine kinase that characterize this condition.

In addition to isolated hyperCKemia, *CAV3* gene mutations can cause other caveolinopathies including CAV3-related distal myopathy, limb-girdle muscular dystrophy, rippling muscle disease, 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.

Learn more about the gene associated with Isolated hyperCKemia

- CAV3

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 isolated hyperCKemia or another caveolinopathy. Rare cases result from new mutations in the gene and occur in people with no history of caveolinopathies in their family.

Other Names for This Condition

- Elevated serum CPK
- Elevated serum creatine phosphokinase
- H-CK
- Idiopathic hyperCKemia
- Idiopathic persistent elevation of serum creatine kinase

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Caveolinopathy (https://www.ncbi.nlm.nih.gov/gtr/conditions/C5679790/)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- CREATINE PHOSPHOKINASE, ELEVATED SERUM (https://omim.org/entry/123320)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28isolated+hyperckemia%5

BTIAB%5D%29+OR+%28elevated+serum+cpk%5BTIAB%5D%29+OR+%28idiopathic+hyperckemia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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