Hypokalemic periodic paralysis

Description

Hypokalemic periodic paralysis is a condition that causes episodes of extreme muscle weakness typically beginning in childhood or adolescence. Most often, these episodes involve a temporary inability to move muscles in the arms and legs. Attacks cause severe weakness or paralysis that usually lasts from hours to days. Some people may have episodes almost every day, while others experience them weekly, monthly, or only rarely. Attacks can occur without warning or can be triggered by factors such as rest after exercise, a viral illness, or certain medications. Often, a large, carbohydrate-rich meal or vigorous exercise in the evening can trigger an attack upon waking the following morning. Although affected individuals usually regain their muscle strength between attacks, some develop persistent muscle weakness later in life.

People with hypokalemic periodic paralysis typically have reduced levels of potassium in their blood (hypokalemia) during episodes of muscle weakness. Researchers are investigating how low potassium levels may be related to the muscle abnormalities in this condition.

Frequency

Although its exact prevalence is unknown, hypokalemic periodic paralysis is estimated to affect 1 in 100,000 people. Men tend to experience symptoms of this condition more often than women.

Causes

Mutations in the *CACNA1S* or *SCN4A* gene can cause hypokalemic periodic paralysis. These genes provide instructions for making proteins that play essential roles in muscles used for movement (skeletal muscles). For the body to move normally, skeletal muscles must tense (contract) and relax in a coordinated way. Muscle contractions are triggered by the flow of certain positively charged atoms (ions) into muscle cells. The CACNA1S and SCN4A proteins form channels that control the flow of these ions. The channel formed by the CACNA1S protein transports calcium ions into cells, while the channel formed by the SCN4A protein transports sodium ions.

Mutations in the *CACNA1S* or *SCN4A* gene alter the usual structure and function of calcium or sodium channels. The altered channels are "leaky," allowing ions to flow

slowly but continually into muscle cells, which reduces the ability of skeletal muscles to contract. Because muscle contraction is needed for movement, a disruption in normal ion transport leads to episodes of severe muscle weakness or paralysis.

A small percentage of people with the characteristic features of hypokalemic periodic paralysis do not have identified mutations in the *CACNA1S* or *SCN4A* gene. In these cases, the cause of the condition is unknown.

Learn more about the genes associated with Hypokalemic periodic paralysis

- CACNA1S
- SCN4A

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.

Other Names for This Condition

- Familial hypokalemic periodic paralysis
- HOKPP
- HypoKPP
- HypoPP
- Primary hypokalemic periodic paralysis
- Westphall disease

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Hypokalemic periodic paralysis, type 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3714580/)
- Genetic Testing Registry: Hypokalemic periodic paralysis (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0238358/)
- Genetic Testing Registry: Hypokalemic periodic paralysis, type 2 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750061/)

Genetic and Rare Diseases Information Center

- Hypokalemic periodic paralysis (https://rarediseases.info.nih.gov/diseases/6729/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Hypokalemic periodic paralysis%22)

Catalog of Genes and Diseases from OMIM

- HYPOKALEMIC PERIODIC PARALYSIS, TYPE 1; HOKPP1 (https://omim.org/entry/170400)
- HYPOKALEMIC PERIODIC PARALYSIS, TYPE 2; HOKPP2 (https://omim.org/entry/613345)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Hypokalemic+Periodic+Paralysis%5BMAJR%5D%29+AND+%28hypokalemic+periodic+paralysis%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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