Potassium-aggravated myotonia

Potassium-aggravated myotonia is a disorder that affects muscles used for movement (skeletal muscles). Beginning in childhood or adolescence, people with this condition experience bouts of sustained muscle tensing (myotonia) that prevent muscles from relaxing normally. Myotonia causes muscle stiffness that worsens after exercise and may be aggravated by eating potassium-rich foods such as bananas and potatoes. Stiffness occurs in skeletal muscles throughout the body. Potassium-aggravated myotonia ranges in severity from mild episodes of muscle stiffness to severe, disabling disease with frequent attacks. Unlike some other forms of myotonia, potassium-aggravated myotonia is not associated with episodes of muscle weakness.

Frequency

This condition appears to be rare; it has been reported in only a few individuals and families worldwide.

Causes

Mutations in the \( \text{SCN4A} \) gene cause potassium-aggravated myotonia. The \( \text{SCN4A} \) gene provides instructions for making a protein that is critical for the normal function of skeletal muscle cells. 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 positively charged atoms (ions), including sodium, into skeletal muscle cells. The SCN4A protein forms channels that control the flow of sodium ions into these cells.

Mutations in the \( \text{SCN4A} \) gene alter the usual structure and function of sodium channels. The altered channels cannot properly regulate ion flow, increasing the movement of sodium ions into skeletal muscle cells. The influx of extra sodium ions triggers prolonged muscle contractions, which are the hallmark of myotonia.

Inheritance Pattern

Potassium-aggravated myotonia 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 some cases, an affected person inherits a mutation in the \( \text{SCN4A} \) gene from one affected parent. Other cases result from new mutations in the gene. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- PAM
- sodium channel myotonia
Diagnosis & Management

Genetic Testing Information

• What is genetic testing? 
  https://primer/testings/genetictesting

• Genetic Testing Registry: Potassium aggravated myotonia

Research Studies from ClinicalTrials.gov

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22potassium-aggravated+myotonia%22+OR+%22Myotonic+Disorders%22+OR+%22Myotonia+Fluctuans%22

Additional Information & Resources

Health Information from MedlinePlus

• Drugs and Supplements: Acetazolamide
  https://medlineplus.gov/druginfo/meds/a682756.html

• Health Topic: Muscle Disorders
  https://medlineplus.gov/muscledisorders.html

Genetic and Rare Diseases Information Center

• Potassium aggravated myotonia
  https://rarediseases.info.nih.gov/diseases/4459/potassium-aggravated-myotonia

Additional NIH Resources

• National Institute of Neurological Disorders and Stroke
  https://www.ninds.nih.gov/Disorders/All-Disorders/Myotonia-Information-Page

Educational Resources

• MalaCards: myotonia, potassium-aggravated
  https://www.malacards.org/card/myotonia_potassium_aggravated

• Orphanet: Potassium-aggravated myotonia
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=612

Patient Support and Advocacy Resources

• Muscular Dystrophy Association
  https://www.mda.org/

• Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/muscular.html
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28potassium-aggravated +myotonia%5BTIAB%5D%29+OR+%28myotonia+fluctuans%5BTIAB%5D%29 +OR+%28myotonia+permanens%5BTIAB%5D%29+OR+%28%28acetazolamide%5BTIAB%5D%29+AND+%28myotonia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- MYOTONIA, POTASSIUM-AGGRAVATED
  http://omim.org/entry/608390

Medical Genetics Database from MedGen

- Potassium aggravated myotonia

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16832098

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8308722
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1143902/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9771789
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2170305/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8058156

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16193245

Reprinted from Genetics Home Reference:

Reviewed: April 2007
Published: October 29, 2019