Familial paroxysmal kinesigenic dyskinesia

Familial paroxysmal kinesigenic dyskinesia is a disorder characterized by episodes of abnormal movement that range from mild to severe. In the condition name, the word paroxysmal indicates that the abnormal movements come and go over time, kinesigenic means that episodes are triggered by movement, and dyskinesia refers to involuntary movement of the body.

People with familial paroxysmal kinesigenic dyskinesia experience episodes of irregular jerking or shaking movements that are induced by sudden motion, such as standing up quickly or being startled. An episode may involve slow, prolonged muscle contractions (dystonia); small, fast, "dance-like" motions (chorea); writhing movements of the limbs (athetosis); or, rarely, flailing movements of the limbs (ballismus). Familial paroxysmal kinesigenic dyskinesia may affect one or both sides of the body. The type of abnormal movement varies among affected individuals, even among members of the same family. In many people with familial paroxysmal kinesigenic dyskinesia, a pattern of symptoms called an aura immediately precedes the episode. The aura is often described as a crawling or tingling sensation in the affected body part. Individuals with this condition do not lose consciousness during an episode and do not experience any symptoms between episodes.

Individuals with familial paroxysmal kinesigenic dyskinesia usually begin to show signs and symptoms of the disorder during childhood or adolescence. Episodes typically last less than five minutes, and the frequency of episodes ranges from one per month to 100 per day. In most affected individuals, episodes occur less often with age.

In some people with familial paroxysmal kinesigenic dyskinesia the disorder begins in infancy with recurring seizures called benign infantile convulsions. These seizures usually develop in the first year of life and stop by age 3. When benign infantile convulsions are associated with familial paroxysmal kinesigenic dyskinesia, the condition is known as infantile convulsions and choreoathetosis (ICCA). In families with ICCA, some individuals develop only benign infantile convulsions, some have only familial paroxysmal kinesigenic dyskinesia, and others develop both.

Frequency

Familial paroxysmal kinesigenic dyskinesia is estimated to occur in 1 in 150,000 individuals. For unknown reasons, this condition affects more males than females.

Causes

Familial paroxysmal kinesigenic dyskinesia can be caused by mutations in the PRRT2 gene. The function of the protein produced from this gene is unknown, although it is thought to be involved in the development and function of the brain. Studies suggest
that the PRRT2 protein interacts with a protein that helps control signaling between nerve cells (neurons). It is thought that PRRT2 gene mutations, which reduce the amount of PRRT2 protein, lead to abnormal neuronal signaling. Altered neuronal activity could underlie the movement problems associated with familial paroxysmal kinesigenic dyskinesia.

Not everyone with this condition has a mutation in the PRRT2 gene. When no PRRT2 gene mutations are found, the cause of the condition is unknown.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern. Autosomal dominant inheritance means that one copy of an altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition.

Other Names for This Condition

• dystonia 10
• episodic kinesigenic dyskinesia
• familial paroxysmal dystonia
• paroxysmal kinesigenic choreoathetosis
• paroxysmal kinesigenic dyskinesia

Diagnosis & Management

Genetic Testing Information

• What is genetic testing? /primer/testing/genetictesting

Research Studies from ClinicalTrials.gov

• ClinicalTrials.gov https://clinicaltrials.gov/ct2/results?cond=%22familial+paroxysmal+kinesigenic+dyskinesia%22+OR+%22familial+paroxysmal+dystonia%22+OR+%22paroxysmal+kinesigenic+choreoathetosis%22+OR+%22paroxysmal+kinesigenic+dyskinesia%22

Other Diagnosis and Management Resources

Additional Information & Resources

Health Information from MedlinePlus
• Health Topic: Movement Disorders
  https://medlineplus.gov/movementdisorders.html

Genetic and Rare Diseases Information Center
• Paroxysmal kinesigenic choreoathetosis
  https://rarediseases.info.nih.gov/diseases/8721/paroxysmal-kinesigenic-choreoathetosis

Educational Resources
• MalaCards: episodic kinesigenic dyskinesia 1
  https://www.malacards.org/card/episodic_kinesigenic_dyskinesia_1
• MalaCards: episodic kinesigenic dyskinesia 2
  https://www.malacards.org/card/episodic_kinesigenic_dyskinesia_2
• Merck Manual Consumer Version: Movement Disorders
• Orphanet: Infantile convulsions and choreoathetosis
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=31709
• Orphanet: Paroxysmal kinesigenic dyskinesia
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98809
• University of California, San Francisco Laboratories of Neurogenetics
  http://neugenes.org/test/episodic-disorders/

Patient Support and Advocacy Resources
• Dystonia Medical Research Foundation
  https://dystonia-foundation.org/what-is-dystonia/types-dystonia/paroxysmal/
• The Movement Disorder Society
  https://www.movementdisorders.org/MDS.htm

Clinical Information from GeneReviews
• PRRT2-Associated Paroxysmal Movement Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK475803
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28familial+paroxysmal+kinesigenic-dyskinesia%5BTIAB%5D%29+OR+%28familial+paroxysmal+dystonia%5BTIAB%5D%29+OR+%28paroxysmal+kinesigenic-choreoathetosis%5BTIAB%5D%29+OR+%28paroxysmal+kinesigenic-dyskinesia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- EPISODIC KINESIGENIC DYSKINESIA 1
  http://omim.org/entry/128200

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15857419

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22101681

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16901678

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17387577

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19364451

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19047496
Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23363396

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19289562

Reprinted from Genetics Home Reference:

Reviewed: January 2014
Published: April 15, 2020

Lister Hill National Center for Biomedical Communications
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