Familial paroxysmal nonkinesigenic dyskinesia

Description

Familial paroxysmal nonkinesigenic dyskinesia is a disorder of the nervous system that causes episodes of involuntary movement. Paroxysmal indicates that the abnormal movements come and go over time. Nonkinesigenic means that episodes are not triggered by sudden movement. Dyskinesia broadly refers to involuntary movement of the body.

People with familial paroxysmal nonkinesigenic dyskinesia experience episodes of abnormal movement that are brought on by alcohol, caffeine, stress, fatigue, menses, or excitement or develop without a known cause. Episodes are not induced by exercise or sudden movement and do not occur during sleep. An episode is characterized by irregular, jerking or shaking movements that range from mild to severe. In this disorder, the dyskinesia can include slow, prolonged contraction of muscles (dystonia); small, fast, "dance-like" motions (chorea); writhing movements of the limbs (athetosis); and, rarely, flailing movements of the limbs (ballismus). The dyskinesia also affects muscles in the torso and face. The type of abnormal movement varies among affected individuals, even among affected members of the same family. Individuals with familial paroxysmal nonkinesigenic dyskinesia do not lose consciousness during an episode. Most people do not experience any neurological symptoms between episodes.

Individuals with familial paroxysmal nonkinesigenic dyskinesia usually begin to show signs and symptoms of the disorder during childhood or their early teens. Episodes typically last 1 to 4 hours, and the frequency of episodes ranges from several per day to one per year. In some affected individuals, episodes occur less often with age.

Frequency

Familial paroxysmal nonkinesigenic dyskinesia is a very rare disorder. Its prevalence is estimated to be 1 in 5 million people.

Causes

Mutations in the *PNKD* gene can cause familial paroxysmal nonkinesigenic dyskinesia. The function of the protein produced from the *PNKD* gene is unknown, although it is thought to play an important role in normal brain function. The PNKD protein may help control the release of chemicals in the brain called neurotransmitters, which allow nerve

cells (neurons) to communicate with each other.

The PNKD protein is similar to a protein that helps break down a chemical called methylglyoxal. Methylglyoxal is found in alcoholic beverages, coffee, tea, and cola. Research has demonstrated that this chemical has a toxic effect on neurons. It remains unclear if the *PNKD* gene is related to the breakdown of methylglyoxal or another substance in the body. How mutations in the *PNKD* gene lead to the signs and symptoms of familial paroxysmal nonkinesigenic dyskinesia is also unknown.

In some families with familial paroxysmal nonkinesigenic dyskinesia, the condition is not caused by a mutation in the *PNKD* gene. Researchers suspect that mutations in one or more other genes that have not been identified can cause the condition.

Learn more about the gene associated with Familial paroxysmal nonkinesigenic dyskinesia

- PNKD

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is typically sufficient to cause the disorder. In all reported cases caused by *PNKD* gene mutations, an affected person has inherited the mutation from one parent. A small number of people with the altered gene have not developed signs and symptoms of the condition, a situation known as reduced penetrance.

Other Names for This Condition

- Familial paroxysmal choreoathetosis
- Mount-Reback syndrome
- Nonkinesigenic choreoathetosis
- Paroxysmal dystonic choreoathetosis
- Paroxysmal nonkinesigenic dyskinesia
- PDC
- PNKD

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Paroxysmal nonkinesigenic dyskinesia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1869117/)
- Genetic Testing Registry: Paroxysmal nonkinesigenic dyskinesia 2 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1970149/)

Genetic and Rare Diseases Information Center

- Paroxysmal non-kinesigenic dyskinesia (https://rarediseases.info.nih.gov/diseases/8722/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Familial paroxysmal nonkinesigenic dyskinesia%22)

Catalog of Genes and Diseases from OMIM

- PAROXYSMAL NONKINESIGENIC DYSKINESIA 1; PNKD1 (https://omim.org/entry/118800)
- PAROXYSMAL NONKINESIGENIC DYSKINESIA 2; PNKD2 (https://omim.org/entry/611147)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28familial+paroxysmal+nonkinesigenic+dyskinesia%5BTIAB%5D%29+OR+%28pnkd%5BTIAB%5D%29+OR+%28paroxysmal+nonkinesigenic+dyskinesia%5BTIAB%5D%29+OR+%28paroxysmal+dystonic+choreoathetosis%5BTIAB%5D%29+OR+%28familial+paroxysmal+choreoathetosis%5BTIAB%5D%29+OR+%28mount-reback+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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