



## 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/genetic-testing](#)
- Genetic Testing Registry: Episodic kinesigenic dyskinesia 1  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C4552000/>

### 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

- GeneReview: PRRT2-Associated Paroxysmal Movement Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK475803>

## **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](https://www.malacards.org/card/episodic_kinesigenic_dyskinesia_1)
- MalaCards: episodic kinesigenic dyskinesia 2  
[https://www.malacards.org/card/episodic\\_kinesigenic\\_dyskinesia\\_2](https://www.malacards.org/card/episodic_kinesigenic_dyskinesia_2)
- Merck Manual Consumer Version: Movement Disorders  
<https://www.merckmanuals.com/home/brain,-spinal-cord,-and-nerve-disorders/movement-disorders/overview-of-movement-disorders>
- Orphanet: ICCA syndrome  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=31709](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](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>

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