

Familial Paroxysmal Kinesigenic Dyskinesia

Subjects: Genetics & Heredity

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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.

Keywords: genetic conditions

1. Introduction

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.

2. 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.

3. 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.

3.1. The Gene Associated with Familial Paroxysmal Kinesigenic Dyskinesia

- PRRT2
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4. Inheritance

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.

5. Other Names for This Condition

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

References

1. Callenbach PM, van den Boogerd EH, de Coo RF, ten Houten R, Oosterwijk JC, Hageman G, Frants RR, Brouwer OF, van den Maagdenberg AM. Refinement of the chromosome 16 locus for benign familial infantile convulsions. *Clin Genet*. 2005 Jun;67(6):517-25.
2. Chen WJ, Lin Y, Xiong ZQ, Wei W, Ni W, Tan GH, Guo SL, He J, Chen YF, Zhang QJ, Li HF, Lin Y, Murong SX, Xu J, Wang N, Wu ZY. Exome sequencing identifies truncating mutations in PRRT2 that cause paroxysmal kinesigenic dyskinesia. *Nat Genet*. 2011 Nov 20;43(12):1252-5. doi: 10.1038/ng.1008.
3. Kato N, Sadamatsu M, Kikuchi T, Niikawa N, Fukuyama Y. Paroxysmal kinesigenic choreoathetosis: from first discovery in 1892 to genetic linkage with benign familial infantile convulsions. *Epilepsy Res*. 2006 Aug;70 Suppl 1:S174-84.
4. Kikuchi T, Nomura M, Tomita H, Harada N, Kanai K, Konishi T, Yasuda A, Matsuura M, Kato N, Yoshiura KI, Niikawa N. Paroxysmal kinesigenic choreoathetosis (PKC): confirmation of linkage to 16p11-q21, but unsuccessful detection of mutations among 157 genes at the PKC-critical region in seven PKC families. *J Hum Genet*. 2007;52(4):334-341. doi: 10.1007/s10038-007-0116-7.
5. Mehta SH, Morgan JC, Sethi KD. Paroxysmal dyskinesias. *Curr Treat Options Neurol*. 2009 May;11(3):170-8.
6. Rochette J, Roll P, Szepietowski P. Genetics of infantile seizures with paroxysmal dyskinesia: the infantile convulsions and choreoathetosis (ICCA) and ICCA-related syndromes. *J Med Genet*. 2008 Dec;45(12):773-9. doi:10.1136/jmg.2008.059519. Review.
7. Silveira-Moriyama L, Gardiner AR, Meyer E, King MD, Smith M, Rakshi K, Parker A, Mallick AA, Brown R, Vassallo G, Jardine PE, Guerreiro MM, Lees AJ, Houlden H, Kurian MA. Clinical features of childhood-onset paroxysmal kinesigenic dyskinesia with PRRT2 gene mutations. *Dev Med Child Neurol*. 2013 Apr;55(4):327-34. doi:10.1111/dmcn.12056.
8. van Rootselaar AF, Schade van Westrum S, Velis DN, Tijssen MA. The paroxysmal dyskinesias. *Pract Neurol*. 2009 Apr;9(2):102-9. doi: 10.1136/jnnp.2009.172213. Review.

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