ADCY5-related Dyskinesia

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ADCY5-related dyskinesia is a movement disorder; the term "dyskinesia" refers to abnormal involuntary movements. The abnormal movements that occur in *ADCY5*-related dyskinesia typically appear as sudden (paroxysmal) jerks, twitches, tremors, muscle tensing (dystonia), or writhing (choreiform) movements, and can affect the limbs, neck, and face.

Keywords: genetic conditions

1. Introduction

The abnormal movements associated with *ADCY5*-related dyskinesia usually begin between infancy and late adolescence. They can occur continually during waking hours, and frequently also disturb sleep. The involuntary movements often occur when changing position, such as from sitting to standing, or when deliberately making other movements.

Severely affected infants may experience weak muscle tone (hypotonia) and delay in development of motor skills such as crawling and walking; later, these individuals may have difficulties with activities of daily living and may eventually require a wheelchair. In more mildly affected individuals, the condition has little impact on walking and other motor skills, although the abnormal movements can lead to clumsiness or difficulty with social acceptance in school or other situations.

In some people with *ADCY5*-related dyskinesia, the disorder is generally stable throughout their lifetime. In others, it slowly gets worse (progresses) in both frequency and severity before stabilizing or even improving in middle age. Anxiety, fatigue, and other stress can temporarily increase the severity of the signs and symptoms of *ADCY5*-related dyskinesia, while some affected individuals may experience remission periods of days or weeks without abnormal movements. Life expectancy is not usually affected by *ADCY5*-related dyskinesia, and most people with this condition have normal intelligence.

2. Frequency

At least 400 people have been diagnosed with *ADCY5*-related dyskinesia, but its prevalence is unknown. The disorder is thought to be underdiagnosed because its features can resemble those of other conditions such as cerebral palsy or epilepsy.

3. Causes

As its name suggests, *ADCY5*-related dyskinesia is caused by mutations in the *ADCY5* gene. This gene provides instructions for making an enzyme called adenylate cyclase 5. This enzyme helps convert a molecule called adenosine triphosphate (ATP) to another molecule called cyclic adenosine monophosphate (cAMP). ATP is a molecule that supplies energy for cells' activities, including muscle contraction, and cAMP is involved in signaling for many cellular functions. Some *ADCY5* gene mutations that cause *ADCY5*-related dyskinesia are thought to increase adenylate cyclase 5 enzyme activity and the level of cAMP within cells. Others prevent production of adenylate cyclase 5. It is unclear how either type of mutation leads to the abnormal movements that occur in this disorder.

3.1. The gene associated with ADCY5-related dyskinesia

- ADCY5

4. Inheritance

This condition 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 the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

5. Other Names for This Condition

- familial dyskinesia with facial myokymia
- FDFM

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