PRICKLE1-related progressive myoclonus epilepsy with ataxia

PRICKLE1-related progressive myoclonus epilepsy with ataxia is a rare inherited condition characterized by recurrent seizures (epilepsy) and problems with movement. The signs and symptoms of this disorder usually begin between the ages of 5 and 10.

Problems with balance and coordination (ataxia) are usually the first symptoms of PRICKLE1-related progressive myoclonus epilepsy with ataxia. Affected children often have trouble walking. Their gait is unbalanced and wide-based, and they may fall frequently. Later, children with this condition develop episodes of involuntary muscle jerking or twitching (myoclonus), which cause additional problems with movement. Myoclonus can also affect muscles in the face, leading to difficulty swallowing and slurred speech (dysarthria).

Beginning later in childhood, some affected individuals develop tonic-clonic or grand mal seizures. These seizures involve a loss of consciousness, muscle rigidity, and convulsions. They often occur at night (nocturnally) while the person is sleeping.

PRICKLE1-related progressive myoclonus epilepsy with ataxia does not seem to affect intellectual ability. Although a few affected individuals have died in childhood, many have lived into adulthood.

Frequency

The prevalence of PRICKLE1-related progressive myoclonus epilepsy with ataxia is unknown. The condition has been reported in three large families from Jordan and northern Israel and in at least two unrelated individuals.

Genetic Changes

PRICKLE1-related progressive myoclonus epilepsy with ataxia is caused by mutations in the PRICKLE1 gene. This gene provides instructions for making a protein called prickle homolog 1, whose function is unknown. Studies suggest that it interacts with other proteins that are critical for brain development and function.

Mutations in the PRICKLE1 gene alter the structure of prickle homolog 1 and disrupt its ability to interact with other proteins. However, it is unclear how these changes lead to movement problems, seizures, and the other features of PRICKLE1-related progressive myoclonus epilepsy with ataxia.

Inheritance Pattern

Some cases of PRICKLE1-related progressive myoclonus epilepsy with ataxia are inherited in an autosomal recessive pattern, which means both copies of the gene in
each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other cases of *PRICKLE1*-related progressive myoclonus epilepsy with ataxia are considered autosomal dominant because one copy of the altered gene in each cell is sufficient to cause the disorder. These cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

**Other Names for This Condition**
- EPM1B
- PME with ataxia
- *PRICKLE1*-related progressive myoclonic epilepsy with ataxia
- progressive myoclonic epilepsy 1B
- progressive myoclonus epilepsy with ataxia

**Diagnosis & Management**

**Genetic Testing**
- Genetic Testing Registry: Progressive myoclonus epilepsy with ataxia

**Other Diagnosis and Management Resources**
- GeneReview: *PRICKLE1*-Related Progressive Myoclonus Epilepsy with Ataxia
  https://www.ncbi.nlm.nih.gov/books/NBK9674

**General Information from MedlinePlus**
- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
Additional Information & Resources

**MedlinePlus**
- Health Topic: Epilepsy  
  https://medlineplus.gov/epilepsy.html
- Health Topic: Movement Disorders  
  https://medlineplus.gov/movementdisorders.html

**Additional NIH Resources**
- National Institute of Neurological Disorders and Stroke: Epilepsy Information Page  
  https://www.ninds.nih.gov/Disorders/All-Disorders/Epilepsy-Information-Page
- National Institute of Neurological Disorders and Stroke: Myoclonus Fact Sheet  
  https://www.ninds.nih.gov/Disorders/All-Disorders/Myoclonus-Information-Page

**Educational Resources**
- Disease InfoSearch: Progressive myoclonus epilepsy with ataxia  
  http://www.diseaseinfosearch.org/Progressive+myoclonus+epilepsy+with+ataxia/9167
- MalaCards: prickle1-related progressive myoclonus epilepsy with ataxia  
  http://www.malacards.org/card/prickle1_rrelated_progressive_myoclonus_epilepsy_with_ataxia
- Merck Manual Home Edition for Patients and Caregivers: Myoclonus  

**Patient Support and Advocacy Resources**
- American Epilepsy Society  
  https://www.aesnet.org/
- Citizens United for Research in Epilepsy (CURE)  
  https://www.cureepilepsy.org/
- National Organization for Rare Disorders (NORD): Progressive Myoclonus Epilepsy  
  https://rarediseases.org/rare-diseases/progressive-myoclonus-epilepsy/

**GeneReviews**
- PRICKLE1-Related Progressive Myoclonus Epilepsy with Ataxia  
  https://www.ncbi.nlm.nih.gov/books/NBK9674

**ClinicalTrials.gov**
- ClinicalTrials.gov  
  https://clinicaltrials.gov/ct2/results?cond=%22progressive+myoclonus+epilepsy+with+ataxia%22+OR+%22Myoclonic+Epilepsies%2C+Progressive%22
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28progressive+myoclonus+epilepsy+%5Bti%5D+AND+ataxia+%5Bti%5D%29+OR+%28epm1b%5BTIAB%5D%29+OR+%28PRICKLE1+%5Btiab%5D+AND+epilepsy+%5Btiab%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- EPILEPSY, PROGRESSIVE MYOCLONIC, 1B
  http://omim.org/entry/612437

Sources for This Summary

  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2668041/


Reprinted from Genetics Home Reference:

Reviewed: December 2011
Published: June 12, 2018