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.

Causes

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 Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Progressive myoclonus epilepsy with ataxia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2676254/

Research Studies from ClinicalTrials.gov

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

Other Diagnosis and Management Resources

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

Additional Information & Resources

Health Information from 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

- MalaCards: prickle1-related progressive myoclonus epilepsy with ataxia
  https://www.malacards.org/card/prickle1_r
  elated_progressive_myoclonus_epilepsy_with_ataxia
- Merck Manual Home Edition for Patients and Caregivers: Myoclonus
  https://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/
  movement-disorders/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/

Clinical Information from GeneReviews

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

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%2
  9+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

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

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