

Opis choroby *

Definicja

Generalized epilepsy-paroxysmal dyskinesia syndrome is characterised by the association of paroxysmal dyskinesia and generalised epilepsy (usually absence or generalised tonic-clonic seizures) in the same individual or family. The prevalence is unknown. Analysis in one of the reported families led to the identification of a causative mutation in the *KCNMA1* gene (chromosome 10q22), encoding the alpha subunit of the BK channel. Transmission is autosomal dominant.

Dane

Klasyfikacja

Choroba

Synonimy

GEPD

GEPD

Kod ORPHA

79137

Kod OMIM

609446

Kod ICD10

G40.3

Kod ICD11

8A61.2Y

*Źródło

orphanet