

# Padaczka uogólniona - dyskineza napadowa

## Kod Orpha: 79137 Kod OMIM: 609446

### 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

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#### [\\*Źródło](#)

orphanet

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.