

## Opis choroby \*

### Definicja

A rare, genetic, neurological disorder characterized by childhood to adolescent-onset of action myoclonus, generalized tonic-clonic seizures, and slowly progressive, moderate to severe cognitive impairment that may lead to dementia. EEG reveals progressive slowing of background activity and epileptic abnormalities and brain MRI shows cerebellar and brainstem atrophy.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

EPM8

EPM8

PME typu 8

Postępująca padaczka miokloniczna z powodu niedoboru CERS1

PME type 8

Progressive myoclonic epilepsy due to CERS1 deficiency

Progressive myoclonus epilepsy type 8

#### Kod ORPHA

424027

#### Kod OMIM

616230

#### Kod ICD10

G40.3

#### Kod ICD11

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#### \*Źródło

orphanet