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