

## Opis choroby \*

### Definicja

A rare, genetic, neuronal ceroid lipofuscinosis disorder characterized by infantile- to early childhood-onset of progressive myoclonic seizures (occasionally accompanied by generalized tonic-clonic seizures) and severe, progressive neurological regression, leading to psychomotor and cognitive decline, cerebellar ataxia, dementia and, frequently, early death. Vision loss may be associated. EEG typically reveals epileptiform activity with predominance in the posterior region and photosensitivity.

### Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	CLN14 disease EPM3 PME typu 3 Postępująca padaczka miokloniczna z powodu niedoboru KCTD7 EPM3 PME type 3 Progressive myoclonic epilepsy due to KCTD7 deficiency Progressive myoclonus epilepsy type 3

Kod ORPHA	Kod OMIM	Kod ICD10
263516	611726	G40.3

Kod ICD11
8A61.41

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### \*Źródło

orphanet