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
Podtyp kliniczny

Synonimy
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
263516

Kod OMIM
611726

Kod ICD10
G40.3

Kod ICD11
8A61.41

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*Źródło

orphanet