Opis choroby *

Definicja

A rare, genetic neurological disorder characterized by early-onset progressive ataxia associated with myoclonic seizures, generalized tonic-clonic seizures (which are often sleep-related), and normal to mild intellectual disability. Dysarthria, upward gaze palsy, sensory neuropathy, developmental delay and autistic disorder have also been associated.

Dane

Klasyfikacja
Choroba

Synonimy
EPM5
EPM5
PME typu 5
PME type 5
Progressive myoclonus epilepsy type 5

Kod ORPHA
402082

Kod OMIM
607459

Kod ICD10
G40.3

Kod ICD11
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*Źródło

orphanet