

Opis choroby *

Definicja

Focal epilepsy-intellectual disability-cerebro-cerebellar malformation is a rare, genetic neurological disorder characterized by early infantile-onset of seizures, borderline to moderate intellectual disability, cerebellar features including dysarthria and ataxia and cerebellar atrophy and cortical thickening observed on MRI imaging. Seizures are typically focal (with prominent eye blinking, facial and limb jerking), precipitated by fever and often commence with an oral sensory aura (anesthetized tongue sensation). When not properly controlled by anti-epileptic medication, weekly frequency and persistence into adult life is observed.

Dane

Klasyfikacja

Choroba

Synonimy

Focal epilepsy-intellectual disability-dysarthria-ataxia syndrome
Padaczka ogniskowa - niepełnosprawność intelektualna - dyzartria - ataksja

Kod ORPHA

352587

Kod OMIM

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Kod ICD10

Q04.8

Kod ICD11

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*Źródło

orphanet