Opis choroby *

Definicja

A rare genetic progressive myoclonic epilepsy characterized by childhood onset of progressive dysarthria, myoclonus, ataxia, seizures, and cognitive decline. The disease takes a protracted course with patients surviving into adulthood, developing signs and symptoms like psychosis with outbursts of prolonged agitation and screaming, spasticity and hyperreflexia, confusion, mutism, and incontinence. There are no visual disturbances. Muscle biopsy shows numerous periodic acid-Schiff-positive inclusions, so-called Lafora bodies.

Dane

Klasyfikacja
Choroba

Kod ORPHA
324290

Kod OMIM
616640

Kod ICD10
G40.3

Kod ICD11
8A61.41

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*Źródło

orphanet