

Opis choroby *

Definicja

A rare neuronal ceroid lipofuscinosis disorder characterized by juvenile-onset of progressive spinocerebellar ataxia, bulbar syndrome (manifesting with dysarthria, dysphagia and dysphonia), pyramidal and extrapyramidal involvement (including myoclonus, amyotrophy, unsteady gait, akinesia, rigidity, dysarthric speech) and intellectual deterioration. Muscle biopsy displays autofluorescent bodies and lipofuscin deposits in brain and, occasionally the retina, upon post mortem.

Dane

Klasyfikacja

Choroba

Synonimy

CLN12 disease

Choroba CLN12

Juvenile parkinsonism-neuronal ceroid lipofuscinosis

Kod ORPHA

314632

Kod OMIM

606693

Kod ICD10

E75.4

Kod ICD11

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*Źródło

orphanet