Opis choroby *

Definicja

A rare neuronal ceroid lipofiscinosis 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