Opis choroby *

Definicja

A rare, genetic, human prion disease characterized by adult-onset neurodegenerative manifestations associated with a movement disorder and psychiatric/behavioral disturbances. Patients typically present personality changes, aggressiveness, manias, anxiety and/or depression in conjunction with rapidly progressive cognitive decline (presenting with dysarthria, apraxia, aphasia, and eventually leading to dementia) as well as ataxia (manifesting with gait disturbances, unsteadiness, coordination problems), Parkinsonism, myoclonus, and/or chorea. Additional features may include generalized spasticity, seizures, urine incontinence and pyramidal abnormalities.

Dane

Klasyfikacja
Choroba

Synonimy
Early-onset prion disease with prominent psychiatric features
Choroba prionowa o wczesnym początku z wyraźnymi objawami psychicznymi
HDL1
HDL1

Kod ORPHA
157941

Kod OMIM
603218

Kod ICD10
G10

Kod ICD11
8A01.11

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*Źródło

orphanet