Opis choroby *

Definicja

A rare inherited human prion disease characterized by adult onset of slowly progressive cerebellar ataxia, with dementia developing relatively late in the disease course (classic ataxic phenotype). Patients may present with gait disturbances and frequent falls, dysarthria, dysphagia, nystagmus, dysmetry, and eventually pancerebellar syndrome, myoclonus, spasticity, severe dementia, and mutism. The disease is invariably fatal after five years on average. Neuropathological hallmark is the presence of numerous multicentric prion protein plaques in the cerebral and cerebellar cortex.

Dane

Klasyfikacja
Choroba

Synonimy
Subacute spongiform encephalopathy, Gerstmann-Straussler type
Podostra gąbczasta forma encefalopatii, typu Gerstmanna i Strausslera

Kod ORPHA
356

Kod OMIM
137440

Kod ICD10
A81.8

Kod ICD11
8E02.1

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*Źródło

orphanet