

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, dysmetria, 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
Gerstmann i Strausslera

Kod ORPHA

356

Kod OMIM

137440

Kod ICD10

A81.8

Kod ICD11

8E02.1

*Źródło

orphanet