Opis choroby *

Definicja

A rare inherited human prion disease characterized by adult onset of progressive disturbance and loss of circadian rhythms, dysautonomia with increased sympathetic activity, and cognitive impairment with fluctuating vigilance, impaired long-term memory, disorientation, and oneiric states. Motor disturbances include myoclonus, cerebellar ataxia, and pyramidal signs. The disease rapidly leads to a somnolent or comatose state and is typically fatal after 9 or 30 months on average (bimodal course). Neuropathologic examination shows marked neuronal loss and gliosis predominantly in thalamic nuclei and inferior olives, while deposition of abnormal prion protein may be relatively sparse.

Dane

Klasyfikacja
Choroba

Kod ORPHA
466

Kod OMIM
600072

Kod ICD10
A81.8

Kod ICD11
8E02.2

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*Źródło

orphanet