

## 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