

## Opis choroby \*

### Definicja

A rare sporadic human prion disease characterized by rapidly progressive cognitive impairment in combination with variable neurologic signs and symptoms including myoclonus, visual or cerebellar problems, pyramidal or extrapyramidal features, or akinetic mutism. Brain imaging may show high signal intensity in caudate, putamen, and/or cortical regions, and a typical EEG pattern consisting of generalized periodic sharp wave complexes is observed in many cases. The disease is invariably fatal within less than two years. Neuropathologic examination reveals deposition of abnormal prion protein in brain tissue, as well as spongiform change and massive neuronal loss and gliosis.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Sporadic CJD

#### Kod ORPHA

204

#### Kod OMIM

123400

#### Kod ICD10

A81.0

#### Kod ICD11

8E00

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#### \*Źródło

orphanet