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