Rodzinna encefalopatia z ciałami wtrętowymi z neuroserpiny
Kod Orpha: 85110 Kod OMIM: 604218

Opis choroby *

Definicja
A rare serpinopathy characterized by progressive myoclonus epilepsy and/or pre-senile dementia with prominent frontal-lobe features and relative sparing of recall memory. In addition, other neurological manifestations like cerebellar symptoms and pyramidal signs may be present. Age of onset is variable, the disease having been reported in children as well as elderly patients. Neuropathological examination reveals the typical neuronal inclusions of mutated neuroserpin (Collins bodies).

Dane

Klasyfikacja
Choroba

Synonimy
FENIB
FENIB

Kod ORPHA
85110

Kod OMIM
604218

Kod ICD10
G31.8

Kod ICD11
8A61.41

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.