

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