

Opis choroby *

Definicja

A rare genetic neurological disorder characterized by sensorineural hearing loss, sensory neuropathy, behavioral abnormalities, and dementia. Occurrence of seizures has also been reported. Age of onset is between adolescence and adulthood. The disease is progressive, with fatal outcome typically in the fifth to sixth decade.

Dane

Klasyfikacja

Choroba

Synonimy

HSAN1E

HSAN1E

HSN1E

Zespół dziedzicznej czuciowej neuropatii,
czuciowo-nerwowej utraty słuchu i demencji

HSN1E

Hereditary sensory neuropathy-sensorineural
hearing loss-dementia syndrome

Kod ORPHA

456318

Kod OMIM

614116

Kod ICD10

G60.8

Kod ICD11

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*Źródło

orphanet