

Opis choroby *

Definicja

A neurodegenerative disease belonging to the inherited cerebellar ataxias mainly characterized by progressive spino-cerebellar ataxia, loss of proprioception, areflexia, and is associated with a marked deficiency in vitamin E.

Dane

Klasyfikacja

Choroba

Synonimy

AVED

Ataksja podobna do ataksji Friedricha

Ataksja z izolowanym niedoborem witaminy E

AVED

Izolowany Niedobór witaminy E

Rodzinny izolowany Niedobór witaminy E

Ataxia with isolated vitamin E deficiency

Familial isolated vitamin E deficiency

Friedreich-like ataxia

Isolated vitamin E deficiency

Kod ORPHA

96

Kod OMIM

277460

Kod ICD10

G11.1

Kod ICD11

8A03.10

*Źródło

orphanet