

Opis choroby *

Definicja

A metabolic disease characterized by anosmia, cataract, early-onset retinitis pigmentosa and possible neurological manifestations, including peripheral neuropathy and cerebellar ataxia. Other features can be deafness, ichthyosis, skeletal abnormalities, and cardiac arrhythmia. It is characterized biochemically by accumulation of phytanic acid in plasma and tissues.

Dane

Klasyfikacja

Choroba

Synonimy

Adult Refsum disease

Dziedziczna neuropatia ruchowa i czuciowa typu 4

Heredopathia atactica polyneuritiformis

HMSN 4

Niedobór oksydazy kwasu fitanowego

Classic Refsum disease

HMSN 4

HMSN IV

Hereditary motor and sensory neuropathy type 4

Hereditary motor and sensory neuropathy type IV

Heredopathia atactica polyneuritiformis

Phytanic-CoA hydroxylase deficiency

Kod ORPHA

773

Kod OMIM

614879

Kod ICD10

G60.1

Kod ICD11

5C57.1

*Źródło

orphanet