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

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*Źródło

orphanet