

Opis choroby *

Definicja

Fiskerstrand type peripheral neuropathy is a slowly-progressive Refsum-like disorder associating signs of peripheral neuropathy with late-onset hearing loss, cataract and pigmentary retinopathy that become evident during the third decade of life.

Dane

Klasyfikacja

Choroba

Synonimy

PHARC syndrome

Neuropatia obwodowa, typ Fiskerstranda

Zespół PHARC

Peripheral neuropathy, Fiskerstrand type

Polyneuropathy-deafness-ataxia-retinitis

pigmentosa-cataract syndrome

Kod ORPHA

171848

Kod OMIM

612674

Kod ICD10

G60.1

Kod ICD11

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*Źródło

orphanet