

Leukoencefalopatia- dystonia - neuropatia ruchowa

Kod Orpha: 163684 Kod OMIM: 613724

Opis choroby *

Definicja

Leukoencephalopathy-dystonia-motor neuropathy syndrome is a peroxisomal neurodegenerative disorder characterized by spasmodic torticollis, dystonic head tremor, intention tremor, nystagmus, hyposmia, and hypergonadotrophic hypogonadism with azoospermia. Slight cerebellar signs (left-sided intention tremor, balance and gait impairment) are also noted. Magnetic resonance imaging (MRI) shows bilateral hyperintense signals in the thalamus, butterfly-like lesions in the pons, and lesions in the occipital region, whereas nerve conduction studies of the lower extremities shows a predominantly motor and slight sensory neuropathy.

Dane

Klasyfikacja

Choroba

Kod ORPHA
163684

Kod OMIM
613724

Kod ICD10
E75.2

Kod ICD11
-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl