

## 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

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### \*Źródło

orphanet