DISEASE:
Leukoencephalopathy-dystonia-motor neuropathy syndrome

NAME:	Leukoencephalopathy-dystonia-motor neuropathy syndrome
DESCRIPTION:	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.
ORPHACODE:	163684
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SCP2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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