Opis choroby *

Definicja

A rare genetic neurological disorder characterized by the association of hypomyelinating leukodystrophy with spondylometaphyseal dysplasia. Patients present in infancy with absent or delayed ability to walk independently, slowly progressive motor deterioration, spasticity, ataxia, proximal weakness, and joint contractures. Additional manifestations include mild cognitive impairment, short stature, scoliosis, enlarged and deformed joints, dysarthria, nystagmus, visual defects, and mildly dysmorphic features, among others. Mode of inheritance is X-linked recessive.

Dane

Klasyfikacja
Choroba

Synonimy
H-SMD
Hypomyelination-spondyloepimetaphyseal dysplasia syndrome
Leukoencephalopathy-SEMD syndrome
Leukoencephalopathy-metaphyseal chondrodysplasia syndrome
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Hypomyelination-spondyloepimetaphyseal dysplasia syndrome
Leukoencephalopathy-SEMD syndrome
Leukoencephalopathy-metaphyseal chondrodysplasia syndrome

Kod ORPHA
83629

Kod OMIM
300232

Kod ICD10
G37.8

Kod ICD11
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*Źródło

orphanet