

## 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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#### Kod ORPHA

83629

#### Kod OMIM

300232

#### Kod ICD10

G37.8

#### Kod ICD11

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