## Opis choroby *

Definicja

A rare leukodystrophy characterized by infantile onset of lower limb spasticity and severe developmental delay associated with delayed myelination and periventricular white matter abnormalities. Other reported signs and symptoms include microcephaly, optic atrophy, nystagmus, ataxia, or seizures.

Dane

**Klasyfikacja**
Choroba

Synonimy
C11ORF73-related autosomal recessive hypomyelinating leukoencephalopathy
Leukodystrofia hipomielinizująca z powodu niedoboru hikeshi
Hypomyelinating leukodystrophy due to hikeshi deficiency

**Kod ORPHA**
495844

**Kod OMIM**
616881

**Kod ICD10**
G93.8

**Kod ICD11**
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*Źródło

orphanet