Opis choroby *

Definicja

A subtype of Metachromatic leukodystrophy characterized by progressive psychomotor regression with an onset between 30 months and 16 years of age, often beginning with behavioral abnormalities or deterioration of school performance. Further manifestations are ataxia, gait disturbances, reduced deep tendon reflexes, spasticity, seizures, paralysis, dementia, and loss of speech, vision, and hearing, eventually resulting in complete loss of motor and cognitive skills, and decerebration. The rate of deterioration is variable with possible survival up to the third decade of life.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Arylsulfatase A deficiency, juvenile form
MLD, postać młodzieńcza
Niedobór arylsulfatazy A, postać młodzieńcza
MLD, juvenile form

Kod ORPHA
309263

Kod OMIM
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Kod ICD10
E75.2

Kod ICD11
5C56.02

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*Źródło

orphanet