

## 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 arylosulfatazy 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