

## Opis choroby \*

### Definicja

A rare lysosomal disease characterized by accumulation of sulfatides in the central and peripheral nervous system due to deficiency of the enzyme arylsulfatase A, leading to demyelination. Three clinical subtypes can be distinguished based on the age of onset: late infantile, juvenile, and adult. Lead symptoms are deterioration in motor or cognitive function or behavioral problems, depending on the subtype, all eventually culminating in a decerebrated state and death after a highly variable disease course and duration. Mode of inheritance is autosomal recessive.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Arylsulfatase A deficiency

MLD

Niedobór arylosulfatazy A

MLD

#### Kod ORPHA

512

#### Kod OMIM

250100

#### Kod ICD10

E75.2

#### Kod ICD11

5C56.02

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#### \*Źródło

orphanet