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