Opis choroby *

Definicja

A subtype of Metachromatic leukodystrophy characterized by rapidly progressive psychomotor regression with an onset before 30 months of age after a period of apparently normal development. Manifestations developing during the course of the disease are impaired feeding and swallowing due to pseudobulbar palsies, seizures, painful spasms, muscle weakness, ataxia, paralysis, dementia, and loss of speech, vision, and hearing, quickly resulting in complete loss of motor and cognitive skills, and decerebration. Death occurs within the first decade of life.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Arylsulfatase A deficiency, late infantile form
MLD, postać niemowlęca późna
Niedobór arylsulfatazy A, postać niemowlęca późna
MLD, late infantile form

Kod ORPHA
309256

Kod OMIM
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Kod ICD10
E75.2

Kod ICD11
5C56.02

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*Źródło

orphanet