Opis choroby *

Definicja

A subtype of Metachromatic leukodystrophy characterized by progressive psychomotor regression with an insidious onset after the age of 16 years, most often beginning with intellectual and behavioral changes, such as memory deficits or emotional instability. The clinical picture is dominated by gradual cognitive, later also motor, decline, taking a protracted course with periods of waxing and waning. Decerebration and death occur within decades after disease onset.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Arylsulfatase A deficiency, adult form
MLD, postać dorosła
Niedobór arylsulfatazy A, postać dorosła
MLD, adult form

Kod ORPHA
309271

Kod OMIM
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Kod ICD10
E75.2

Kod ICD11
5C56.02

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*Źródło

orphanet