

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

*Źródło

orphanet