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

Synonimy

Podtyp kliniczny

Arylsulfatase A deficiency, adult form

MLD, postać dorosła

Niedobór arylsulfatazy A, postać dorosła

MLD, adult form

Kod ORPHA

Kod OMIM

Kod ICD10

309271

E75.2

Kod ICD11 5C56.02

*Źródło

orphanet