Opis choroby *

Definicja

A rare, genetic leukodystrophy characterized by developmental delay, increased muscle tone leading later to spasticity, mild ataxia, nystagmus, dysarthria, intentional tremor, and mild intellectual disability. Brain imaging reveals supratentorial and infratentorial hypomyelination.

Dane

Klasyfikacja
Choroba

Kod ORPHA
438114

Kod OMIM
616140

Kod ICD10
E75.2

Kod ICD11
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*Źródło

orphanet