Opis choroby *

Definicja

A rare, severe, genetic, neurometabolic disease characterized by infantile-onset of progressive neurodevelopmental regression, optic atrophy with nystagmus and diffuse white matter disease. Affected individuals usually have central hypotonia that progresses to limb spasticity and hyperreflexia, eventually resulting in a vegetative state. Recurrent chest infections are frequently associated and seizures (usually generalized tonic-clonic) may occasionally be observed. Brain magnetic resonance imaging shows diffuse bilateral symmetric abnormalities in the cerebral periventricular white matter, with variable lesions in other areas but sparing the basal ganglia.

Dane

Klasyfikacja
Choroba

Synonimy
MMDS4
MMDS4

Kod ORPHA
457406

Kod OMIM
616370

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet