Opis choroby *

Definicja

A rare leukodystrophy characterized by a spectrum of progressive neurologic manifestations comprising rapidly progressive early-onset nystagmus, spastic tetraplegia, and visual and hearing impairment, resulting in death in early childhood, as well as later onset of slowly progressive complex spastic ataxia with pyramidal and cerebellar symptoms and loss of developmental milestones. Brain imaging shows diffuse hypomyelination of the subcortical and deep white matter, cerebellar atrophy, and diffuse spinal cord volume loss.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive hypomyelinating leukodystrophy-progressive spastic ataxia SPAX8
Autosomalna, recesywna leukodystrofia hipomielinizująca-postępująca ataksja/niezborność spastyczna SPAX8

Kod ORPHA
527497

Kod OMIM
617560

Kod ICD10
E75.2

Kod ICD11
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*Źródło

orphanet