

## 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