Opis choroby *

Definicja

A complex hereditary spastic paraplegia characterized by progressive lower limbs weakness and spasticity, upper limbs weakness, dysarthria, hypomimia, sphincter disturbances, peripheral neuropathy, learning difficulties, cognitive impairment and dementia. Magnetic resonance imaging shows thin corpus callosum, cerebral atrophy, and periventricular white matter changes.

Dane

Klasyfikacja
Choroba

Synonimy
Nakamura-Osame syndrome
Paraplegia spastyczna - niepełnosprawność intelektualna - cienkie ciało modzelowate
SPG11
Zespół Nakamura i Osame
SPG11
Spastic paraplegia-intellectual disability-thin corpus callosum syndrome

Kod ORPHA
2822

Kod OMIM
604360

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet