

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

*Źródło

orphanet