Opis choroby *

Definicja

A rare genetic neurological disorder characterized by progressive spastic paraparesis and delayed gross motor development with an onset in infancy or early childhood. Patients also show variable degrees of intellectual disability, speech delay, and dysarthria. Other reported features include microcephaly, seizures, bifid uvula with or without cleft palate, and ocular anomalies. Brain imaging shows white matter abnormalities in the periventricular and other regions.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive complex SPG due to Kennedy pathway dysfunction
Autosomal recessive spastic paraplegia type 81
Autosomal recessive complex SPG due to Kennedy pathway dysfunction
Autosomal recessive spastic paraplegia type 81

Kod ORPHA
506353

Kod OMIM
618768

Kod ICD10
G11.4

Kod ICD11
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*Źródło

orphanet