Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 53 (SPG53) is a very rare, complex type of hereditary spastic paraplegia characterized by early-onset spastic paraplegia (with spasticity in the lower extremities that progresses to the upper extremities) associated with developmental and motor delay, mild to moderate cognitive and speech delay, skeletal dysmorphism (e.g. kyphosis and pectus), hypertrichosis and mildly impaired vibration sense. SPG53 is due to mutations in the VPS37A gene (8p22) encoding vacuolar protein sorting-associated protein 37A.

Dane

Klasyfikacja
Choroba

Synonimy
SPG53
SPG53

Kod ORPHA
319199

Kod OMIM
614898

Kod ICD10
G11.4

Kod ICD11
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*Źródło

orphanet