

## 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