

## Opis choroby \*

### Definicja

Autosomal recessive spastic paraplegia type 20 (SPG20) is a type of complex hereditary spastic paraplegia characterized by an onset in infancy of progressive spastic paraparesis associated with distal amyotrophy, psuedobulbar palsy, motor and cognitive delays, mild cerebellar signs (dysarthria, dysdiadochokinesia, mild intention tremor), short stature and subtle skeletal abnormalities (pes cavus, mild talipes equinovarus, kyphoscoliosis). SPG20 is due to mutations in the *SPG20* gene (13q13.1), which encodes the protein spartin.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Childhood-onset spastic paraparesis-distal muscle wasting syndrome

Paraplegia spastyczna o początku w wieku dziecięcym - zanik mięśni dystalnych

SPG20

Zespół Troyera

SPG20

Troyer syndrome

#### Kod ORPHA

101000

#### Kod OMIM

275900

#### Kod ICD10

G11.4

#### Kod ICD11

8B44.01

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#### \*Źródło

orphanet