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 mięsni 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