

Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 61 (SPG61) is a rare, complex form of hereditary spastic paraplegia characterized by an onset in infancy of spastic paraplegia (presenting with the inability to walk unsupported and a scissors gait) associated with a motor and sensory polyneuropathy with loss of terminal digits and acropathy. SPG61 is due to a mutation in the *ARL6IP1* gene (16p12-p11.2) encoding the ADP-ribosylation factor-like protein 6-interacting protein 1.

Dane

Klasyfikacja

Choroba

Synonimy

SPG61

SPG61

Kod ORPHA

401780

Kod OMIM

615685

Kod ICD10

G11.4

Kod ICD11

8B44.01

*Źródło

orphanet