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

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*Źródło

orphanet