Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 57 (SPG57) is an extremely rare, complex type of hereditary spastic paraplegia, characterized by onset in infancy of pronounced leg spasticity (leading to the inability to walk independently), reduced visual acuity due to optic atrophy, and distal wasting of the hands and feet due to an axonal demyelinating sensorimotor neuropathy. SPG57 is caused by mutations in the TFG gene (3q12.2) encoding protein TFG, which is thought to play a role in ER microtubular architecture and function.

Dane

Klasyfikacja
Choroba

Synonimy
SPG57
Paraplegia spastyczna spowodowana częściowym niedoborem TFG
SPG57
Spastic paraplegia due to partial TFG deficiency

Kod ORPHA
431329

Kod OMIM
615658

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet