Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 55 (SPG 55) is a rare, complex type of hereditary spastic paraplegia characterized by childhood onset of progressive spastic paraplegia associated with optic atrophy (with reduced visual acuity and central scotoma), ophthalmoplegia, reduced upper-extremity strength and dexterity, muscular atrophy in the lower extremities, and sensorimotor neuropathy. SPG55 is caused by mutations in the C12ORF65 gene (12q24.31) encoding probable peptide chain release factor C12orf65, mitochondrial.

Dane

Klasyfikacja
Choroba

Synonimy
SPG55
SPG55

Kod ORPHA
320375

Kod OMIM
615035

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet