Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 54 (SPG54) is a rare, complex form of hereditary spastic paraplegia characterized by the onset in early childhood of progressive spastic paraplegia associated with cerebellar signs, short stature, delayed psychomotor development, intellectual disability and, less commonly, foot contractures, dysarthria, dysphagia, strabismus and optic hypoplasia. SPG54 is caused by mutations in the DDHD2 gene (8p11.23) encoding phospholipase DDHD2.

Dane

Klasyfikacja
Choroba

Synonimy
SPG54
SPG54

Kod ORPHA
320380

Kod OMIM
615033

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet