

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

*Źródło

orphanet