Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 64 is an extremely rare and complex form of hereditary spastic paraplegia (see this term), reported in only 4 patients from 2 families to date, characterized by spastic paraplegia (presenting between the ages of 1 to 4 years with abnormal gait) associated with microcephaly, amyotrophy, cerebellar signs (e.g. dysarthria) aggressiveness, delayed puberty and mild to moderate intellectual disability. SPG64 is due to mutations in the ENTPD1 gene (10q24.1), encoding ectonucleoside triphosphate diphosphohydrolase 1.

Dane

Klasyfikacja
Choroba

Synonimy
SPG64
SPG64

Kod ORPHA
401810

Kod OMIM
615683

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet