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 <i>ENTPD1</i> gene (10q24.1), encoding ectonucleoside triphosphate diphosphohydrolase 1.

Dane

Klasyfikacja Choroba Synonimy

SPG64

SPG64

Kod ORPHA

Kod OMIM

Kod ICD10

401810

615683

G11.4

Kod ICD11 8B44.01

*Źródło

orphanet