Autosomal recessive spastic paraplegia type 64

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spastic paraplegia type 64. ORPHA:401810

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.