Autosomal recessive spastic paraplegia type 44

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spastic paraplegia type 44. ORPHA:320401

Autosomal recessive spastic paraplegia type 44 (SPG44) is a very rare, complex form of hereditary spastic paraplegia characterized by a late-onset, slowly progressive spastic paraplegia associated with mild ataxia and dysarthria, upper extremity involvement (i.e. loss of finger dexterity, dysmetria), and mild cognitive impairment, without the presence of nystagmus. A hypomyelinating leukodystrophy and thin corpus callosum is observed in all cases and psychomotor development is normal or near normal. SPG44 is caused by mutations in the GJC2 gene (1q41-q42) encoding the gap junction gamma-2 protein.