Autosomal recessive spastic paraplegia type 26

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spastic paraplegia type 26. ORPHA:101006

Autosomal recessive spastic paraplegia type 26 (SPG26) is a rare, complex type of hereditary spastic paraplegia characterized by the onset in childhood/adolescence (ages 2-19) of progressive spastic paraplegia associated mainly with mild to moderate cognitive impairment and developmental delay, cerebellar ataxia, dysarthria, and peripheral neuropathy. Less commonly reported manifestations include skeletal abnormalities (i.e. pes cavus, scoliosis), dyskinesia, dystonia, cataracts, cerebellar signs (i.e. saccadic dysfunction, nystagmus, dysmetria), bladder disturbances, and behavioral problems. SPG26 is caused by mutations in the B4GALNT1 gene (12q13.3), encoding Beta-1, 4 N-acetylgalactosaminyltransferase 1.