Autosomal dominant spastic paraplegia type 6

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant spastic paraplegia type 6. ORPHA:100988

Autosomal dominant spastic paraplegia type 6 (SPG6) is a form of hereditary spastic paraplegia which usually presents in late adolescence or early adulthood as a pure phenotype of lower limb spasticity with hyperreflexia and extensor plantar responses, as well as mild bladder disturbances and pes cavus. Rarely, it can present as a complex phenotype with additional manifestations including epilepsy, variable peripheral neuropathy and/or memory impairment.