Autosomal recessive spastic paraplegia type 74

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spastic paraplegia type 74. ORPHA:468661

Autosomal recessive spastic paraplegia type 74 is a rare, genetic, spastic paraplegia-optic atrophy-neuropathy-related (SPOAN-like) disorder characterized by childhood onset of mild to moderate spastic paraparesis which manifests with gait impairment that very slowly progresses into late adulthood, hyperactive patellar reflex and bilateral extensor plantar response, in association with optic atrophy and typical symptoms of peripheral neuropathy, including reduced or absent ankle reflexes, lower limb atrophy and distal sensory impairment. Reduced visual acuity and pes cavus are frequently reported.