Autosomal spastic paraplegia type 30

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal spastic paraplegia type 30. ORPHA:101010

Autosomal spastic paraplegia type 30 is a form of hereditary spastic paraplegia characterized by either a pure spastic paraplegia phenotype, usually presenting in the first or second decade of life, with spastic lower extremities, unsteady spastic gait, hyperreflexia and extensor plantar responses, or as a complicated phenotype with the additional manifestations of distal wasting, saccadic ocular movements, mild cerebellar ataxia and mild, distal, axonal neuropathy.