Autosomal recessive spastic paraplegia type 48

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spastic paraplegia type 48. ORPHA:306511

Autosomal recessive spastic paraplegia type 48 is a form of hereditary spastic paraplegia usually characterized by a pure phenotype of a slowly progressive spastic paraplegia associated with urinary incontinence with an onset in mid- to late-adulthood. A complex phenotype, with the additional findings of cognitive impairment, sensorimotor polyneuropathy, ataxia and parkinsonism, as well as thin corpus callosum and white matter lesions (seen on magnetic resonance imaging), has also been reported.