Autosomal recessive spastic paraplegia type 23

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spastic paraplegia type 23. ORPHA:101003

Autosomal recessive spastic paraplegia type 23 (SPG23) is a rare, complex type of hereditary spastic paraplegia that presents in childhood with progressive spastic paraplegia, associated with peripheral neuropathy, skin pigment abnormalities (i.e. vitiligo, hyperpigmentation, diffuse lentigines), premature graying of hair, and characteristic facies (i.e. thin with ''sharp'' features). The SPG23 phenotype has been mapped to a locus on chromosome 1q24-q32.