Gemignani syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Gemignani syndrome. ORPHA:2074

Gemignani syndrome is a rare neurodegenerative disease characterized by slowly progressive ataxia, amyotrophy of the hands and distal arms, spastic paraplegia, progressive sensorineural hearing loss, hypogonadism and short stature. Additional features include generalized cerebellar atrophy and peripheral nervous system anomalies. Small cervical spinal cord, intellectual/language disability and localized vitiligo have also been reported. There have been no further descriptions in the literature since 1989.