Spastic paraplegia-neuropathy-poikiloderma syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spastic paraplegia-neuropathy-poikiloderma syndrome. ORPHA:2821

Spastic paraplegia-neuropathy-poikiloderma syndrome is a complex form of hereditary spastic paraplegia characterized by spastic paraplegia, demyelinating peripheral sensorimotor neuropathy, poikiloderma (manifesting with loss of eyebrows and eyelashes in childhood in addition to delicate, smooth, and wasted skin) and distal amyotrophy (presenting after puberty). There have been no further descriptions in the literature since 1992.