Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
Lison syndrome
Parapareza spastyczna - bielactwo nabyte - przedwczesne siwienie - charakterystyczna twarz
SPG23
Zespół Lisona
SPG23
Spastic paraparesis-vitiligo-premature graying-characteristic facies syndrome

Kod ORPHA
101003

Kod OMIM
270750

Kod ICD10
G11.4

Kod ICD11
8B44.01

---

*Źródło

orphanet