Opis choroby *

Definicja

Primary tethered cord syndrome is a genetic, non-syndromic congenital malformation of the neurenteric canal, spinal cord and column characterized by progressive neurologic deterioration (pain, sensorimotor deficits, abnormal gait, decreased tone or abnormal reflexes), musculoskeletal changes (foot deformities and asymmetry, muscle atrophy, limb weakness and numbness, gait disturbances, scoliosis) and/or genitourinary manifestations (bladder and bowel dysfunction). Midline cutaneous stigmata in the lumbosacral region, such as turfs of hair, skin appendages, dimples, subcutaneous lipomas, skin discoloration or hemangiomas, are frequently associated.

Dane

Klasyfikacja
Wada morfologiczna

Synonimy
Primary tethered spinal cord syndrome

Kod ORPHA
268861

Kod OMIM
-

Kod ICD10
Q06.8

Kod ICD11
LA07.0

---

*Źródło

orphanet