Opis choroby *

Definicja

A rare, neural tube defect characterized by localized longitudinal division of the spinal cord with an interposed osseous, cartilaginous or fibrous septum and double dural sac, typically occurring at the thoracic or lumbar level. Local vertebral segmental defects, syringomyelia, meningocele and intraspinal tumors may be associated. Variable clinical presentation includes pain, scoliosis, asymmetry and weakness of the lower limbs, neurological deficits, sphincter dysfunction, and various cutaneous abnormalities overlying the spine, such as hypertrichosis, dimple, hemangioma, subcutaneous mass or pigmented nevus.

Dane

Klasyfikacja
Wada morfologiczna

Synonimy
SCM type 1
SCM type I
Split cord malformation type 1
Diastematomyelia

Kod ORPHA
1671

Kod OMIM
222500

Kod ICD10
Q06.2

Kod ICD11
LA07.1

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*Źródło

orphanet