Opis choroby *

Definicja

A rare central nervous system malformation characterized by congenital absence of the spinal cord, usually associated with segmental bony spinal anomalies. Neurologic deficits depend on the affected segments and the functioning of the residual spinal cord. Typically, the spinal cord appears normal above the defect and bulky, thickened, and low-lying caudally. Clinical presentation includes varying degrees of motor weakness (associated with deformities of the lower limbs) and neurogenic bladder dysfunction.

Dane

Klasyfikacja
Wada morfologiczna

Kod ORPHA
268868

Kod OMIM
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Kod ICD10
Q06.0

Kod ICD11
LA07.2

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*Źródło

orphanet