

Izolowany brak rdzenia kręgowego

Kod Orpha: 268868 Kod OMIM:

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.