

Opis choroby *

Definicja

A rare neural tube closure defect characterized by a skin defect with exposed neural tissue in the area of the spinal column, with or without a protruding sac at the location of the defect. Signs and symptoms are variable depending on the content (only meninges or also spinal cord tissue), location, and severity of the lesion, but may include motor, sensory, and/or sphincter dysfunction, hydrocephalus, and/or skeletal anomalies (e. g. scoliosis, hemivertebrae), among others.

Dane

Klasyfikacja

Grupa fenomenów

Synonimy

Open spina bifida

Spina bifida aperta

Kod ORPHA

268369

Kod OMIM

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Kod ICD10

Q05.7

Kod ICD11

LA02.1

*Źródło

orphanet