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

Synonimy

Grupa fenomenów

Open spina bifida Spina bifida aperta

Kod ORPHA

Kod OMIM

Kod ICD10

268369

Q05.7

Kod ICD11

LA02.1

*Źródło

orphanet