Opis choroby *

Definicja

A rare, neural tube closure defect characterized by partial lacking of bone fusion, resulting in sac-like protrusions of the brain and the membranes that cover it through the openings in the skull. Protruding tissue may be located on any part of the head, but most often affects the occipital area. Depending in the size nad location, encephalocele are often associated with neurological problems including intellectual disability, seizures, vision impairment, ataxia, and hydrocephalus.

Dane

Klasyfikacja
Wada morfologiczna

Kod ORPHA
199647

Kod OMIM
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Kod ICD10
Q01.9

Kod ICD11
LA01

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*Źródło

orphanet