Opis choroby *

Definicja

A rare genetic neurological disorder characterized by multiple lateral meningoceles, distinctive facial dysmorphism (including hypertelorism, downslanting palpebral fissures, posteriorly rotated ears, micrognathia, and high, narrow palate, among others), and skeletal abnormalities (e. g. vertebral anomalies, wormian bones, short stature, and scoliosis). Multiple additional features may present, such as conductive hearing impairment, hypotonia, and connective tissue and urogenital abnormalities. Cognition is usually normal.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Lehman syndrome
Zespół Lehmana

Kod ORPHA
2789

Kod OMIM
130720

Kod ICD10
Q87.5

Kod ICD11
LA07.Y

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*Źródło

orphanet