

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	Synonimy
Zespół wad wrodzonych	Lehman syndrome Zespół Lehmana

Kod ORPHA	Kod OMIM	Kod ICD10
2789	130720	Q87.5

Kod ICD11
LA07.Y

*Źródło

orphanet