

Opis choroby *

Definicja

A rare genetic bone development disorder characterized by parietal foramina in association with hypoplasia of the clavicles (short abnormal clavicles with tapering lateral ends, with or without loss of the acromion). Additional features may include mild craniofacial dysmorphism (macrocephaly, broad forehead and frontal bossing). No dental abnormalities were reported.

Dane

Klasyfikacja

Zespół wad wrodzonych
Parietal foramina with cleidocranial dysplasia
Otwór ciemieniowy z dyzostozą obojczykowo-
czaszkową

Synonimy

Kod ORPHA

251290

Kod OMIM

168550

Kod ICD10

Q74.0

Kod ICD11

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*Źródło

orphanet