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

Synonimy
Parietal foramina with cleidocranial dysplasia
Otwór ciemeniowy z dyzostozą obojczykowo-czaszkową

Kod ORPHA
251290

Kod OMIM
168550

Kod ICD10
Q74.0

Kod ICD11
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*Źródło

orphanet