Opis choroby *

Definicja

Oculomaxillofacial dysostosis is a rare, genetic bone developmental disorder characterized by short stature, orbital region and ocular abnormalities (e.g. asymmetric orbits, anophthalmia, down-slanted and S-shaped palpebral fissures, sparse eyebrows/eyelashes, abnormal eyelids, ectropion, symblepharon, corneal leukoma), abnormal nose (e.g. broad and abnormally modeled nasal root, bridge and tip, lateral deviation), malar hypoplasia, cleft lip/palate, and oblique facial clefts. Intellectual disability, microcephaly, micrognathia and limb anomalies (e.g. hemimelia, abnormal scapular girdle, brachydactyly, syndactyly, broad halluces) have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Richieri-Costa-Gorlin syndrome
Zespół Richieri, Costa i Gorlina

Kod ORPHA
1794

Kod OMIM
-

Kod ICD10
Q75.1

Kod ICD11
LD25.3

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*Źródło

orphanet