

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 **Synonimy**

Zespół wad wrodzonych Richieri-Costa-Gorlin syndrome
Zespół Richieri, Costa i Gorlina

Kod ORPHA

1794

Kod OMIM

-

Kod ICD10

Q75.1

Kod ICD11

LD25.3

*[Źródło](#)

orphanet