Opis choroby *

Definicja

A rare, genetic, primary bone dysplasia syndrome characterized by bilateral, painless swelling of the face extending from the mandible to the inferior orbital margins (cherubism), epilepsy, gingival fibromatosis (possibly obscuring teeth), and intellectual disability. Other associated variable features include hypertrichosis, stunted growth, juvenile rheumatoid arthritis, and development of ocular abnormalities (e.g. pigmentary retinopathy, optic disc pallor, Axenfeld anomaly). Radiological images typically show bilateral multifocal radiolucency involving the body, angle and ramus of the mandible and coronoid process.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cherubism-gingival fibromatosis-intellectual disability syndrome
Cherubizm - zwłóknienie dziąseł - niepełnosprawność intelektualna

Kod ORPHA
3019

Kod OMIM
266270

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet