## 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