

Opis choroby *

Definicja

Craniometadiaphyseal dysplasia, wormian bone type is an extremely rare craniotubular bone dysplasia syndrome described in fewer than 10 patients to date. Clinical manifestations include macrocephaly, frontal bossing, malar hypoplasia, prominent mandible and dental hypoplasia. Other skeletal anomalies include abnormal bone modeling in tubular bones, multiple wormian bones and deformities of chest, pelvis and elbows. An increased risk of fractures is noted.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

85184

Kod OMIM

269300

Kod ICD10

Q78.8

Kod ICD11

LD24.1Y

*Źródło

orphanet