

Opis choroby *

Definicja

Beemer-Ertbruggen syndrome is a lethal malformation syndrome reported in 2 brothers of first-cousin parents that is characterized by hydrocephalus, cardiac malformation, dense bones, and unusual facies with down-slanting palpebral fissures, bulbous nose, broad nasal bridge, micrognathia and a long upper lip. There have been no further descriptions in the literature since 1984.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Lethal hydrocephalus-cardiac malformation-dense bones syndrome
Śmiertelny zespół wodogłowia, wad serca i gęstych kości

Kod ORPHA

1237

Kod OMIM

209970

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet