Opis choroby *

Definicja

A rare, lethal, congenital, chondrodysplasia disorder characterized by dumbbell-shaped long bones with markedly shortened diaphyses and metaphyseal irregularities associated with a 'Swiss cheese' appearance of the cartilage matrix, as well as distinctive changes in the growth plate and resting cartilage, resulting in death in the neonatal period. There have been no further descriptions in the literature since 1983.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2347

Kod OMIM
245190

Kod ICD10
Q77.8

Kod ICD11
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*Źródło

orphanet