Opis choroby *

Definicja

A rare primary bone dysplasia syndrome characterized by growth retardation with proportionate short stature, cortical thickening and medullary stenosis of the long bones, delayed anterior fontanelle closure, hypocalcemia due to congenital hypoparathyroidism and facial dysmorphism, including prominent forehead, microphthalmia, and micrognathia. Additional manifestations include ocular and dental anomalies (e.g. corneal opacity, hyperopia, optic atrophy, tortuous retinal vessels, dental caries, enamel defects) and, occasionally, hypoplastic nails and neonatal liver disease. Inheritance may be autosomal dominant or autosomal recessive, with more severe growth retardation, small hands and feet, intellectual disability, microcephaly and recurrent bacterial infections being observed in the latter.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Kenny syndrome
Zespół Kenny'ego

Kod ORPHA
2333

Kod OMIM
127000

Kod ICD10
Q87.1

Kod ICD11
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*Źródło

orphanet