Opis choroby *

Definicja

A rare primary bone dysplasia characterized by severe intrauterine and postnatal growth retardation and short stature in association with craniofacial dysmorphism (such as large forehead, triangular face, low-set ears, and micro-retrognathism) and osteochondrodysplastic lesions. Radiographic findings include epiphyseal maturation delay, abnormal metaphyses, a narrow thorax, small pelvis, and short and broad metacarpal bones and phalanges. There have been no further descriptions in the literature since 1996.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mievis-Verellen-Dumoulin syndrome
Zespół Mievisa, Verellena i Dumoulina

Kod ORPHA
2867

Kod OMIM
601350

Kod ICD10
Q87.1

Kod ICD11
LD24.2Y

---

*Źródło

orphanet