Opis choroby *

Definicja

A rare primary bone dysplasia characterized by congenital symmetric or asymmetric shortness and bowing of long bones, resulting in shortness of limbs and limited extension at the knees and elbows. Additional reported features are ''beaten metal'' appearance of the skull, dolichomacrocephaly, ocular hypertelorism, and anterior beaking and bone-within-bone appearance of vertebrae. There have been no further descriptions in the literature since 1993.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Wrodzony zespół wygięcia kości długich, niskiego wzrostu, dolichomakrocefalii i hiperteloryzmu ocznego

Kod ORPHA
2292

Kod OMIM
264050

Kod ICD10
Q68.5

Kod ICD11
LB96.1

---

*Źródło

orphanet