Opis choroby *

Definicja

A rare, non-rhizomelic, chondrodysplasia punctata syndrome characterized, radiologically, by stippled calcifications and disproportionate, short metacarpals and tibiae (with characteristic overshoot of the proximal fibula), clinically manifesting with severe short stature, bilateral shortening of upper and lower limbs, flat midface and nose, in the absence of cataracts and cutaneous anomalies. Neonatal tachypnea, hydrocephalus and mild developmental delay have been seldomly associated. Additional radiologic features include bowed long bones, platyspondyly and/or vertebral clefts.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
79346

Kod OMIM
118651

Kod ICD10
Q77.3

Kod ICD11
LD24.04

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*Źródło

orphanet