Opis choroby *

Definicja

A form of acromelic dysplasia characterized by the distinctive radiological sign of angel-shaped middle phalanges, a typical metacarpophalangeal pattern profile (mainly affecting first metacarpals and middle phalanges of second, third and fifth digits, which all appear short), epiphyseal changes in the hips and, in some, abnormal dentition and delayed bone age.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
ASPED
ASPED

Kod ORPHA
63442

Kod OMIM
105835

Kod ICD10
Q78.8

Kod ICD11
LD24.8Y

---

*Źródło

orphanet