

Opis choroby *

Definicja

A rare primary bone dysplasia characterized, radiologically, by short, stubby long bones, severely angulated femurs and lesser bowing of other long bones (mild, moderate or no bowing), short and wide iliac wings with horizontal acetabular roofs, platyspondyly and a narrow thorax, clinically manifesting with severe, disproportionate short stature. Regression of femora angulation is observed with advancing age.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

1801

Kod OMIM

211350

Kod ICD10

Q79.8

Kod ICD11

LD24.C

*Źródło

orphanet