Opis choroby *

Definicja

A rare, genetic, primary bone dysplasia disease characterized by usually moderate, postnatal short stature, progressive genu vara deformity, a waddling gait, and radiological signs of metaphyseal dysplasia (i.e. irregular, sclerotic and widened metaphyses), in the absence of biochemical abnormalities suggestive of rickets disease. Intermittent knee pain, lordosis, and delayed motor development may also occasionally be associated.

Dane

Klasyfikacja
Choroba

Kod ORPHA
2501

Kod OMIM
250400

Kod ICD10
Q78.5

Kod ICD11
LD24.7

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*Źródło

orphanet