

## 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