## 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

\*Źródło

orphanet