

# Chondrodysplazja przynasadowa typu Spahra

## Kod Orpha: 2501 Kod OMIM: 250400

### 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

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.