

## Opis choroby \*

### Definicja

Rhizomelic dysplasia, Patterson-Lowry type is a rare primary bone dysplasia characterized by short stature, severe rhizomelic shortening of the upper limbs associated with specific malformations of humeri (including marked widening and flattening of proximal metaphyses, medial flattening of the proximal epiphyses, and lateral bowing with medial cortical thickening of the proximal diaphyses), marked coxa vara with dysplastic femoral heads and brachimetacarpalia.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

2831

#### Kod OMIM

601438

#### Kod ICD10

Q78.8

#### Kod ICD11

LD24.A

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### \*Źródło

orphanet