

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

*Źródło

orphanet