## Open Peer Review on Qeios

## Rhizomelic dysplasia, Patterson-Lowry type

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Rhizomelic</u> <u>dysplasia, Patterson-Lowry type</u>. ORPHA:2831* 

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.