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# X-linked spondyloepimetaphyseal dysplasia

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked spondyloepimetaphyseal dysplasia. ORPHA:93349*

X-linked spondyloepimetaphyseal dysplasia is a rare, genetic primary bone dysplasia disorder characterized by disproportionate short stature with mesomelic short limbs, leg bowing, lumbar lordosis, brachydactyly, joint laxity and a waddling gait. Radiographs show platyspondyly with central protrusion of anterior vertebral bodies, kyphotic angulation and very short long bones with dysplastic epiphyses and flared, irregular, cupped metaphyses.