Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome. ORPHA:168443

Spondyloepimetaphyseal dysplasia-hypotrichosis syndrome is a rare primary bone dysplasia disorder characterized by congenital hypotrichosis associated with rhizomelic short stature (more pronounced in upper limbs than lower limbs), limited hip abduction and mild genu varum. Flared and irregular metaphyses, delayed and irregular epiphiseal ossification and pear-shaped vertebral bodies are characteristic radiologic findings.