Spondyloepimetaphyseal dysplasia, Shohat type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spondyloepimetaphyseal dysplasia, Shohat type. ORPHA:93352

Spondyloepimetaphyseal dysplasia congenita, Shohat type is characterized by severely disproportionate short stature, short limbs, small chest, short neck, thin lips, severe lumbar lordosis, marked genu varum, joint laxity, distended abdomen, mild hepatomegaly and splenomegaly.