Autosomal recessive spondylometaphyseal dysplasia, Mégarbané type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spondylometaphyseal dysplasia, Mégarbané type. ORPHA:401979

Autosomal recessive spondylometaphyseal dysplasia, Mégarbané type is a rare, primary bone dysplasia characterized by intrauterine growth retardation, pre- and postnatal disproportionate short stature with short, rhizomelic limbs, facial dysmorphism, a short neck and small thorax. Hypotonia, cardiomegaly and global developmetal delay have also been associated. Several radiographic findings have been reported, including ribs with cupped ends, platyspondyly, square iliac bones, horizontal and trident acetabula, hypoplastic ischia, and delayed epiphyseal ossification.