Multiple epiphyseal dysplasia, Lowry type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Multiple epiphyseal dysplasia, Lowry type. ORPHA:166016

Multiple epiphyseal dysplasia, Lowry type is a rare primary bone dysplasia characterized by small, flat epiphyses (esp. the capital femoral epiphyses), rhizomelic shortening of limbs, cleft of secondary palate, micrognathia, mild joint contractures and facial dysmorphism (incl. mildly upward-slanting palpebral fissures, hypertelorism, broad nasal tip). Additionally reported features include scoliosis, genu valgum, mild pectus excavatum, platyspondyly, dislocated radial heads, brachydactyly, hypoplastic fibulae and talipes equinovarus.