Autosomal recessive distal osteolysis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive distal osteolysis syndrome. ORPHA:2776

Autosomal recessive distal osteolysis syndrome is an early-onset distal osteolysis characterised by severe resorption of the hands and feet and absence of the distal and middle phalanges. It has been described in a son and daughter born to consanguineous parents. Other manifestations include distal muscular hypertrophy, flexion contractures, short stature, mild intellectual deficit and characteristic facies (maxillary hypoplasia, exophthalmos, and a broad nasal tip). It is transmitted as an autosomal recessive trait.