Metaphyseal anadysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Metaphyseal anadysplasia. ORPHA:1040

Metaphyseal anadysplasia is a very rare form of metaphyseal dysplasia characterized by short stature, rhizomelic micromelia and a mild varus deformity of the legs evident from the first months of life, that is associated with radiological features of severe metaphyseal changes (irregularities, widening and marginal blurring) in long bones, most prominent in proximal femurs, and generalized osteopenia, and that usually spontaneously resolves by the age of three years. Severe autosomal dominant and milder recessive variants have been observed.