Eiken Type Chondrodysplasia

National Cancer Institute

Source

National Cancer Institute. Eiken Type Chondrodysplasia. NCI Thesaurus. Code C131811.

An autosomal recessive skeletal dysplasia caused by mutation(s) in the PTH1R gene, encoding parathyroid hormone/parathyroid hormone-related peptide receptor. This condition is characterized by severely delayed skeletal maturation, as well as by abnormal modeling of the bones in the hands and feet, abnormal persistence of cartilage in the pelvis, and mild growth retardation. Calcium and phosphate concentrations are normal.