Metaphyseal Chondrodysplasia, Jansen Type

National Cancer Institute

Source

National Cancer Institute. Metaphyseal Chondrodysplasia, Jansen Type. NCI Thesaurus. Code C131868.

A form of metaphyseal chondrodysplasia caused by mutation(s) in the PTH1R gene, encoding parathyroid hormone/parathyroid hormone-related peptide receptor. This condition is characterized by severe short stature, short bowed limbs, clinodactyly, prominent upper face, and a small mandible. Hypercalcemia and hypophosphatemia due to PTH resistance can appear later in childhood.