Blomstrand Type Chondrodysplasia

National Cancer Institute

Source

National Cancer Institute. Blomstrand Type Chondrodysplasia. NCI Thesaurus. Code C131420.

An autosomal recessive lethal condition caused by inactivating mutation(s) in the PTH1R gene, encoding parathyroid hormone/parathyroid hormone-related peptide receptor. This condition is characterized by short limbs, polyhydramnios, hydrops fetalis, facial anomalies, increased bone density, and advanced skeletal maturation.