Achondroplasia

National Cancer Institute

Source

National Cancer Institute. Achondroplasia. NCI Thesaurus. Code C34345.

An autosomal dominant disorder caused by mutation(s) in the FGFR3 gene, encoding fibroblast growth factor receptor 3. The condition is characterized by inappropriate cartilage growth plate differentiation and deficient endochondral growth, manifest clinically with severe rhizomelic short stature, short limbs, characteristic facies with frontal bossing and midface hypoplasia.