Pycnodysostosis

National Cancer Institute

Source

National Cancer Institute. Pycnodysostosis. NCI Thesaurus. Code C131187.

An autosomal recessive disorder caused by loss-of-function mutation(s) in the CTSK gene, encoding cathepsin K, an enzyme involved in bone resorption by osteoclasts. This condition is characterized by some or all of the following: osteosclerosis, short stature, pituitary hypoplasia with growth hormone deficiency, and cerebral demyelination.