Craniodiaphyseal Dysplasia

National Cancer Institute

Source

National Cancer Institute. Craniodiaphyseal Dysplasia. NCI Thesaurus. Code C131429.

An autosomal dominant or recessive form of craniotubular hyperostosis due to mutation(s) in the SOST gene, encoding sclerostin. This condition is characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones, which is so severe that the resulting facial distortion is referred to as 'leontiasis ossea'; the bone deposition results in progressive stenosis of craniofacial foramina and can lead to severe neurologic impairment in childhood.