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Craniotubular Hyperostosis

National Cancer Institute

Source

National Cancer Institute. *Craniotubular Hyperostosis*. NCI Thesaurus. Code C131430.

A group of sclerosing bone dysplasias characterized by pronounced sclerosis of the cranial vault and the long bones, resulting in increased cortical bone density. Most cases are caused by mutation(s) in the SOST or LRP5 genes, encoding sclerostin and low-density lipoprotein receptor-related protein 5, respectively. Other genes have been implicated, and some cases have been described, but do not yet have identified genetic mutations.