

[Open Peer Review on Qeios](#)

Sclerosteosis

National Cancer Institute

Source

National Cancer Institute. *Sclerosteosis*. NCI Thesaurus. Code C131133.

An autosomal recessive form of craniotubular hyperostosis due to loss-of-function mutation(s) in the *SOST* gene, encoding sclerostin. Clinical features include tall stature, enlarged jaw and facial bones, and cranial nerve compression leading to hearing loss and facial palsy. About two-thirds of patients have syndactyly and/or nail malformations. Increased intracranial pressure due to the thickened calvaria and skull base can occur.