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Cherubism

National Cancer Institute

Source

National Cancer Institute. *Cherubism*. NCI Thesaurus. Code C84630.

A rare autosomal dominant inherited disorder usually caused by mutations in the SH3BP2 gene. It is characterized by a prominent lower part of the face due to bilateral replacement of the mandibular or maxillary bones by lesions. The lesions contain osteoclast-like cells admixed with spindle-shaped mononuclear stromal cells. With time, the lesions become more fibrotic and less osteoclast-rich.