

Non-familial cherubism

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Abstract

Cherubism is a rare, self-limiting, non-neoplastic fibro-osseous disorder of the jaws, usually seen in pediatric population. It is characterized by painless bilateral swelling of the jaws that gives the patient a typical cherubic appearance. Here, we describe the clinical, radiographic, histologic and computed tomographic features of cherubism in a 6-year-old boy.

Keywords: Cherubism, computed tomography, fibro-osseous disorder, giant cell lesions

Introduction

Cherubism is a rare, hereditary, autosomal dominant, benign, fibro-osseous bone disease of childhood that causes a progressive but generally self-limiting, diffuse, bilateral and multicentric expansion of the mandible and/or maxilla and has a characteristic radiographic and histopathologic appearance.^[1] The condition is usually observed in patients aged 2–5 years. The disease progresses or becomes increasingly evident at the time of puberty and then shows gradual regression thereafter. The signs and symptoms of disease depend on the severity of the condition, and range from clinically and radiologically undetectable features to severe deformity of the jaws, upright palate, respiratory distress, impairment of vision and hearing.^[1,2]

Case Report

A 6-year-old male child of non-consanguineous parents presented with a history of painless progressive swelling of bilateral cheeks of 2 years duration. The swelling was small initially, which gradually increased to its present size. There was no history of similar disease in any of the siblings or in the parents of the affected child. On clinical examination, the child had mild swelling of bilateral cheeks, more prominent on the right side, which was seen to involve angle of the

mandible. There was no local tenderness and mouth opening was adequate [Figure 1].

Intraorally expansion was seen on right angle region of mandible involving buccal cortex. There was no evidence of any decayed or missing teeth.

Panoramic radiograph revealed variably expansile, multiloculated osteolytic lesions involving angle and bilateral rami of the mandible with sparing of condyles [Figure 2].

Computed tomography (CT) with thin axial and coronal section was performed and multiplanar images were reconstructed and three-dimensional images were obtained. CT confirmed the presence of multiloculated cystic lesions affecting the angle and rami of mandible. The lesions caused marked expansion of the bones, with a multifocal cortical breakthrough [Figures 3 and 4].

Due to lack of family history of similar disease, an incisional biopsy was performed intraorally under general anesthesia and the specimen was obtained from the right side of the mandible and sent for histopathologic examination.

Histopathologic examination

Gross examination: Multiple thick fragments of grayish brown

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Figure 1: Patient's profile

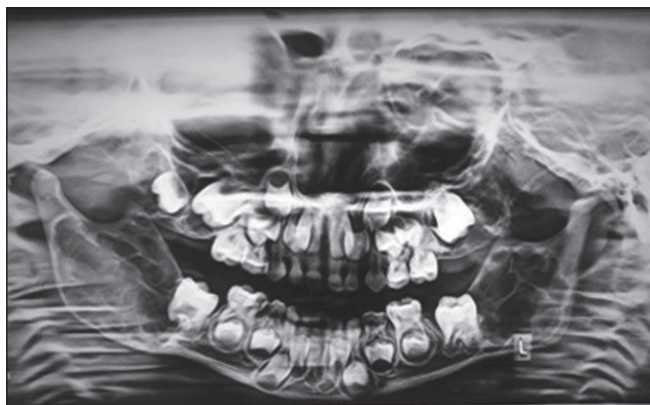


Figure 2: Panoramic radiograph showing multiloculated osteolytic lesions involving the jaw.

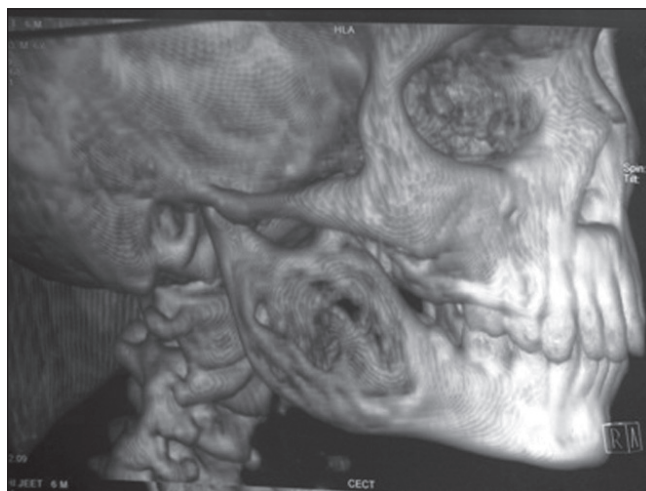


Figure 3: Computed tomographic image of right side of the jaw.

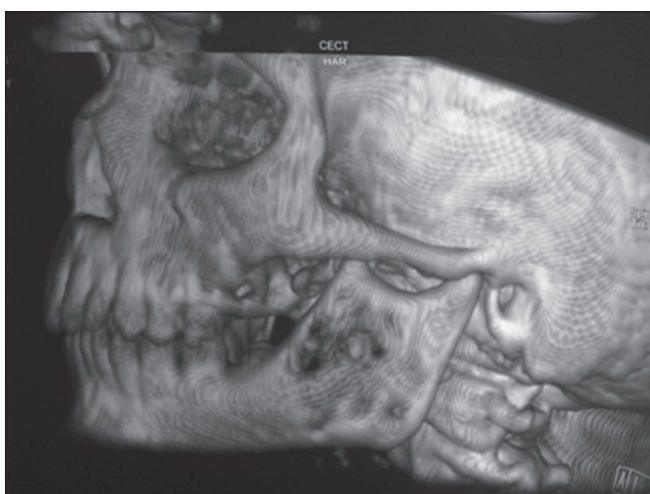


Figure 4: Computed tomographic image of left side of the jaw.

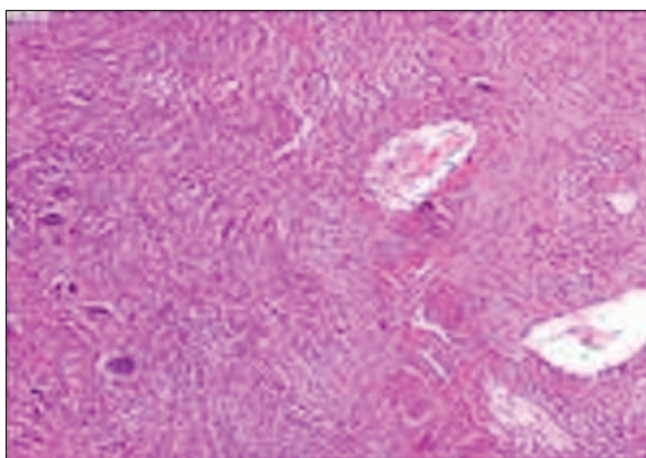


Figure 5: Low-power photomicrograph showing highly vascular fibrous stroma at places showing loose bundles. Many multinucleate giant cells are scattered on the stroma. Vascular spaces are thin walled and dilated (HandE, $\times 40$).

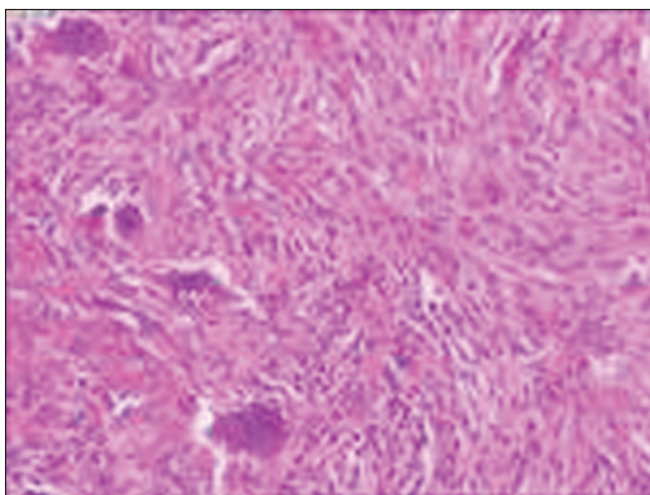


Figure 6: Higher magnification showing spindly fibrous cells. Multinucleate giant cells contain variable number of nuclei and are sparse as compared to giant cell granuloma or giant cell tumor. Osteoid formation is not present. Few lymphocytes are present in the background (H and E, $\times 100$).

firm tissue, together measuring $2 \times 2 \times 1.5$ cm, were received. Whole tissue was processed.

Microscopic examination: Multiple sections stained with hematoxylin and eosin (HandE) were examined. Lesion was made up of highly vascular fibrous stroma containing spindle fibroblasts at places arranged compactly. There were scattered multinucleate giant cells possessing 3–20 nuclei. No osteoid tissue formation was seen. Vascular spaces were dilated, but thick-walled blood vessels surrounded by collagenous tissue were not identified. Diagnosis of giant cell lesion compatible with cherubism was considered [Figures 5 and 6].

Therefore, a diagnosis of cherubism was made based on the characteristic radiological findings, along with compatible histopathologic features.

Due to expected tendency of these lesions to regress spontaneously, no surgical intervention was undertaken and the patient was kept on follow-up.

Discussion

Cherubism is a fibrous osseous condition characterized by firm, painless swelling of the jaws that are said to give the patient a “cherubic appearance”. This disorder was first described in 1933 by Jones as a “familial multiocular cystic disease involving mandible and maxilla”, but the term “cherubism” was later coined to describe the classical characteristics of full round cheeks, which resembled those of the cherubs immortalized by Renaissance art.^[1,3]

It is inherited as an autosomal dominant trait which appears to have 100% penetrance in males and 50–70% penetrance in females.^[4] There was no history of a similar disease in any of the family members of our patient, thus adding to the list of non-familial cases that have been reported in literature.^[1,5] The disease progresses until puberty and shows involution in adulthood. The affected child is normal at birth and develops the disease in the second or third year of life. Clinically, cherubism is characterized by progressive swelling of the face, with marked increase in the fullness of cheeks and jaws, resulting in a round face, sometimes retrusion of the lower eyelid that produces a visible sclera beneath the iris causing “eye raised at heaven” look.^[6,7] Usually, the mandible is affected more than the maxilla.

Abnormalities in the configuration of the dental arch or dental eruption, premature exfoliation of deciduous or permanent teeth, resorption of roots, and malformation are frequently observed in cases of cherubism.^[8]

The radiographic appearance of cherubism is unique because of its diffuse, bilateral, multiocular nature which begins at the angle of the mandible and extends into the ramus and body.^[1] These multiocular areas of diminished densities are later replaced by irregular patchy sclerosis with progressive calcifications. The presence of numerous unerupted teeth and the destruction of the alveolar bone may displace the teeth, producing an appearance referred to as “floating tooth syndrome”.^[2] There is a classical but not specific “ground

glass appearance” as a result of tightly compressed trabecular pattern.^[1] In our case, this was evident on right side of the mandible.

CT scan provides a clear delineation of the extent of the disease which is difficult on radiographs due to an overlapping of the facial bones. CT investigation is the best method for demonstrating expansile lesions and soft tissue components in these lesions.

Radiographs and CT scan are sufficient for the diagnosis of cherubism. Histologically, the lesion contains numerous multinucleated giant cells scattered throughout a fibrous connective tissue.^[1]

Since the lesion undergoes spontaneous regression, the surgical intervention is usually delayed until after puberty. However, in patients with functional or cosmetic problems or emotional disturbances, surgical intervention can be considered.

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