

## CASE REPORT

**Cherubism: A Variant Case**Vinayak Kumar Mantu<sup>1</sup>, V.K Prajapati<sup>2</sup>, Ruchi Mitra<sup>3</sup>, Anuj Sharma<sup>4</sup>**ABSTRACT**

**Introduction:** Cherubism, a form of osteolytic genetic disorder presents in childhood and tends to regress spontaneously after puberty. It is characterized by painless expansion of mandible or maxilla or both.

**Case Report:** On radiography, the lesions exhibit bilateral multinuclear radiolucent areas. Histopathology reveals multinucleated giant cells in the background of proliferating fibrous connective tissue. Children are normal at birth and jaw is noticed to be expanding within the first few years of life progressing to further enlargement until adolescence. It is genetically inherited, although non familial cases have also been reported. The present case report describes a variant case of cherubism in 11 year old male child with bilateral facial swelling and protruding mass over his gums.

**Conclusion:** Cherubism is a rare osseous disorder found in children and adolescents. Orbital involvements in cherubism may develop beyond puberty after stabilization or regression of the lesion of the jaws.

**Keywords:** Cherubism, osteolytic, genetic, non familial

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**INTRODUCTION**

Cherubism first described by Jones in 1933, is relat-

ed genetically inherent polyostotic fibrous dysplasia which causes a characteristic facial deformity due to degeneration of maxilla and mandible bony tissue and its replacement with higher amounts of fibrous tissue.<sup>1</sup> It is a characteristic familial disorder that presents as an autosomal dominant trait with 100% and 50-70 % penetrance in males and females respectively.<sup>2</sup> Initially called as 'familial fibrous dysplasia of the jaws', but recent genetic investigations have proved it as to be separate entity at the molecular level. Wait and watch approach has been advocated by many authors. The disease starts early in life manifesting itself fully in the second decade of life and is almost regressed in the third decade. Here we, present a case report on Cherubism in eleven year old boy.

**CASE REPORT**

An eleven (11) year old male child, a resident of Gogunda, Rajasthan reported to the department of Oral and Maxillofacial Pathology with the chief complaint of Bilateral swelling of face and protruding mass over his gums which covered all his teeth leading to difficulty in chewing food and awkward appearance of face. The history of present illness revealed that the bilateral swelling was first noticed by his parents when the patient was about seven years of age. The swelling gradually started increasing in size from the age of seven years and was painless since these years. No significant medical and dental history was seen. None of his siblings or anyone from maternal or paternal side has similar history.

On clinical examination the face had mild degree of bilateral swelling with greater asymmetry on right side. No associated skin changes were seen. The face was looking fuller on right side with loss of nasolabial sulcus bilaterally. No visual impairment was evident. None of the submandibular lymph node were enlarged. Mobility of condyle was in normal range. Swelling was of diffused nature extending anteroposteriorly between posterior border of ramus along the inferior border to midline. Superoinferiorly extended between outer canthus of eye to lower border of mandible. Intraoral examination revealed that on palpation, swelling was bony hard, painless and ill defined. There was

extensive gingival growth covering all over his teeth leading to condition mimicking edentulous arch. The growth was soft to firm in consistency and was painless on palpation. There was ill defined buccal and lingual vestibule V shaped arches with shallow palatal vault. In maxillary arch only incisal edge of 21,15 and coronal  $\frac{1}{4}$  of 26 was found. Bleeding point was seen in relation to 15. In mandibular arch only incisal edges of 36, 31,41,42 44 were visible. Premature loss of primary teeth and impaired development and eruption of permanent teeth was seen. (Figure-1,2)

Bilateral multilocular cystic lesions of the jaws. Displacement of inferior alveolar canal Displaced teeth with "floating tooth syndrome" seen on radiographic examination. Bone shows generalized radiolucencies. Sharply defined round or oval radiolucent areas develop, which may be lobulated. The lamina dura around the teeth is partially lost with ground glass appearance (Figure -3)

All biochemical studies were in normal range.

H&E stained section shows highly cellular and vascular C.T. Many thin walled blood capillaries and presence of perivascular eosinophilic cuffing, few giant cells, extravasated RBCs seen. Bone tissue shows an irregular resorptive edge (Figure-4).

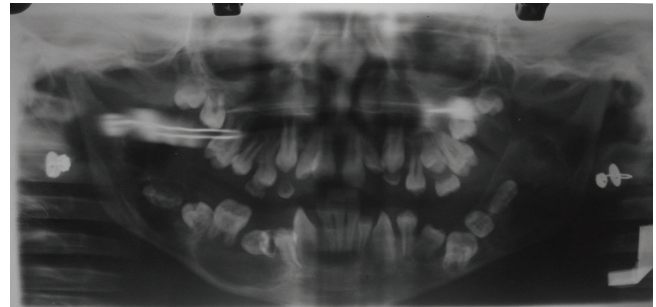
Since the lesion undergoes spontaneous regression, the surgical intervention is usually delayed until puberty. However in patient with functional and cosmetic problems or emotional disturbance some surgical intervention can be considered. But due to its gingival coverage gingivectomy was the line of treatment to reduce the patient's discomfort and recall and revisit every 6 months.

## DISCUSSION

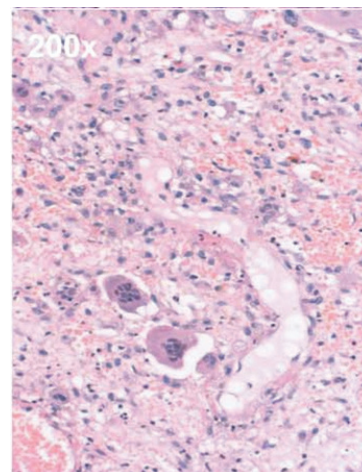
Cherubism is so named because of angel like appearance of patients (chubby and upward look). It was in 1986 when Burkhardt and Berthold examined the tissue taken from a boy affected with cherubism using electron microscope and immunohistology. They found a close relationship between cherubism and giant cell granuloma of the jaws. However extraoral lesions have been reported by some of the authors as osteolytic foci in other bones (neck of femur, ribs), café au lait spots on the skin and associated fibrous dysplasia. The disease may sometimes be associated with cystic foci eg in metacarpal bones, carpal bones, ribs, humerus, pelvis, tibia and femur.<sup>5</sup> In the present case pelvic Xray detected no abnormality similar to another case of a 12 year old male child.<sup>6</sup>



**Figure-1:** Extraoral bilateral facial swelling; **Figure-2:** Intraoral gingival growth extending till the incisal edge



**Figure-3:** Panoramic radiograph



**Figure-4:** Under High power magnification view

McCleden and Anderson reviewed 65 cases in 21 families and reported that cherubism is autosomal dominant inherited disorder.<sup>8</sup> The first signs of disease manifestation are generally observed at about 2 years of age followed by accelerated growth from 8 to 9 yrs and spontaneous regression after puberty.<sup>5</sup> The phenotype ranges from no clinical manifestation to severe maxillary and mandibular overgrowth with respiratory, speech, vision and swallowing problems. Intra-orally it presents as a hard, non-tender swelling palpable in the affected area. Sub-mandibular and upper cervical lymphadenopathy are common. In the present case bilateral facial swelling of face was not so prominent as compared to protruding mass over his gums which covered all his teeth leading to difficulty in chewing food and esthetics. The protruding mass over

the gums making only incisal edges of few teeth visible is a variant case in Cherubism with not much cases reported yet in literature. The pathogenesis of cherubism has been proposed as molecular, with the mutation in the gene encoding SH3BP2 and possible degradation of the Msx-1 gene, which is involved in the regulation of mesenchymal interaction during craniofacial morphogenesis. The most accepted theory related to the pathogenesis of cherubism is its association with an autosomal dominant gene, i.e., family inheritance. However, in our case report no hereditary criteria could be established, for which an autosomal recessive pattern of inheritance could be suggested.<sup>8</sup>

Histologic examination of the lesions usually reveals numerous multinucleated giant cells. These multinucleated cells show strong positivity for monoclonal antibody 23c6 and tartrate-resistant acid phosphatase, which is characteristic of osteoclasts. In the present study the multinucleated giant cells with pre-vascular eosinophilic was seen.<sup>2</sup>

The condition regresses till 30 years of age, when lesion are not detectable frequently. Maxilla is more affected than mandible in this lesion<sup>8</sup> which is similar to the present case.

In general, the treatment of cherubism is to biopsy the lesion, extract any ectopic and impacted teeth and correct it surgically when appropriate. Some surgeons wait and approach to observe the condition because it is assumed that the lesion resolves automatically on its own in the third decade of the life.

The cherubic appearance in a young patient may be highly unpleasant esthetically to ones personality, hence surgical reconstruction and correction of deformity is recommended. Nowadays surgical curettage is not an effective therapy in patients with aggressive signs and symptoms.<sup>9</sup> Liposuction has been proposed to reduce the mass of the lesion in particular cases. Curettage alone or in combination with surgical contouring for cosmetic purposes has been considered the treatment of choice and some authors have reported a massive growth of the lesion after surgery. Some authors point medical therapy in the form of calcitonin as a possibility to curtail the disease and obviate the need for surgery, but only a few references are available in the literature.<sup>10</sup> In the present case gingivectomy to reduce the difficulty in speech and mastication and surgical correction was considered as the treatment plan.

## CONCLUSION

Cherubism is a rare osseous disorder found in children

and adolescents. Orbital involvements in cherubism may develop beyond puberty after stabilization or regression of the lesion of the jaws. Patients with cherubism should be routinely evaluated by an ophthalmologist and follow up is necessary to rule out any other pathology. Nowadays, genetic tests should be used for the final diagnosis of cherubism. In this case, there was no positive inheritance and therefore, it is a non-familial or a sporadic case but, the present case was variant as clinically the facial swelling was not much prominent as compared to the gingival involvement. This finding has not been much discussed in literature. Hence there is need for careful histological, biochemical and radiological diagnosis to establish a final diagnosis.

## REFERENCES

1. Jones WA. Familial multilocular cystic disease of the jaws. *Am J Cancer*. 1933;17:946–950.
2. Pindborg JJ, Kramer IRH. In: WHO histological typing of odontogenic tumors, jaw cysts and allied lesions 1st ed. Geneva: Springer- Verlag 1971:18–9.
3. Davis GB, Sinn DP, Watson SW. Case 43, part II: Cherubism. *J Oral Maxillofac Surg* 1983; 41:119–20.
4. Burkhardt A, Berthold H. Cherubismus-klinische und morphologischebeobachtung. *Dtsch Z Mund Kiefer Gesichtschir*. 1986;10:257–263
5. Carvalho SilvaE, Carvalho Silva GC, VieiraTC. Cherubism: clinicoradiographic features,treatment, and long term follow-up of 8 cases.*J Oral maxillofac surg* 2007;65:517-22.
6. Singh P, Singh A, Raju MS. Cherubism A case report.*Int J Case Reports and Images* 2013; 4:260-65.
7. Anderson DE, McCledon JL. Cherubism-hereditary fibrous dysplasia of the jaws, Part-I. Genetic Considerations. *Oral Surg Oral Med Oral Path* 1962; 15:5–15.
8. Ozkan Y, Varol A, Turker N, Aksakalli N, Basa S. Clinical and radiological evaluation of cherubism: a sporadic case report and review of the literature. *Int J Pediatr Otorhinolaryngol* 2003; 67:1005–12.
9. Penarrocha M, Bonet J, Minguez JM, Bagan JV, Vera F, Minguez I. Cherubism—a clinical, radiographic and histopathologic comparison of 7 cases. *J Oral MaxillofacSurg* 2006;64:924–30.
10. Teixeira VP, Dedivitis RA, Guimarães AV, Benetton AA, França CM. Cherubism—case report and literature review. *Rev De Clín Pesq Odontol* 2004;1:23–5.