Case Report

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Cherubism- A Case Report with Review of Literature

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ABSTRACT

Background Cherubism is a rare, non neoplastic, self limiting, fibroosseous disease characterized by painless expansion of mandible or maxilla or both, usually found in children between 2-5 years of age giving characteristic cherubic appearance to the patient. Radiographically, the lesions exhibit bilateral multinuclear radiolucent areas. Histopathology reveals multinucleated giant cells in the background of proliferating fibrous connective tissue. The present case report describes cherubism in a 15 yr. old female and briefly reviews literature on this report.

Keywords: Autosomal dominant, cherubism, giant cells, fibrous dysplasia.

INTRODUCTION

Cherubism is a non-neoplastic hereditary bone lesion characterized by a bilateral, painless swelling of the maxilla and mandible resulting in a fullness of the cheeks and retraction of the lower eyelids giving an upward turned appearance of the eyes comparable to a cherub angel [1]. It is an autosomal dominant inheritance presenting an 80% of familial pattern.

Typically the mandible is primarily affected and in 60% of the cases maxilla is also involved [2]. Children are normal at birth. At the time of 14 months to five years of age, a symmetric enlargement of the jaws begins, progressing until puberty [3]. It is genetically inherited [1, 2], although non familial cases have also been reported [3–5] in the literature. The authors present a case of a fifteen year old female with cherubism and compare the findings and clinical approach with current literature.

CASE REPORT

A 15 year old female (Figs. 1, 2) visited the outpatient department of oral and maxillofacial surgery, Rungta college of dental sciences and research Kohka, Bhilai , with complaint of pain and swelling in both sides of her face. She first noticed the swelling 10 years back which gradually increased to the current status. The patient reported facial asymmetry with gross chubby appearance. Both horizontal and vertical dimensions of the face were increased. The skin of the mandibular region and cheek region was stretched and superficial veins were prominent. Bilateral submandibular lymph nodes were enlarged, non tender, about 1.5 cm in size. On ophthalmic examination, orbital floor was raised pushing the eyeballs which seemed to be looking upwards. Lateral canthus of the eyes was inferiorly displaced on both sides giving antimongoloid slant to the face with ectropion of the lower eyelids. Intra-oral examination revealed both maxillary and

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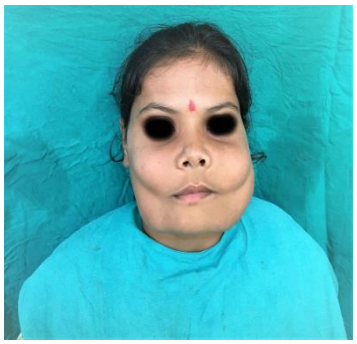

Fig 1: Frontal profile.

Fig 2: Lateral profile.

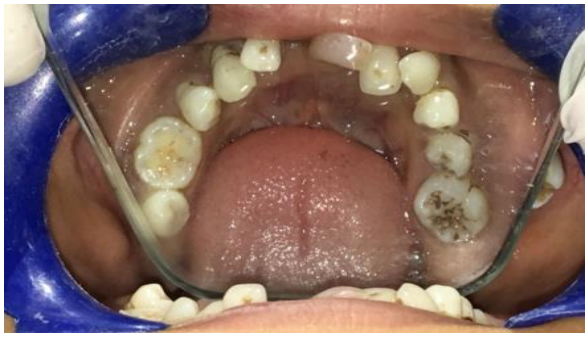

Fig 3: Intra oral view.

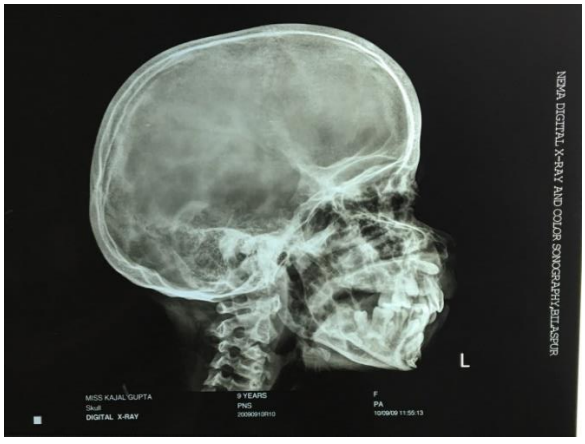

Fig 4: Lateral skull radiograph.

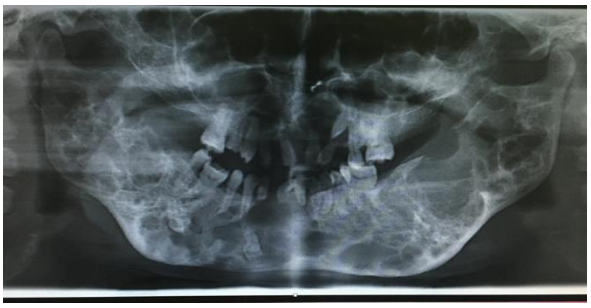

Fig 5: Orthopantomogram.

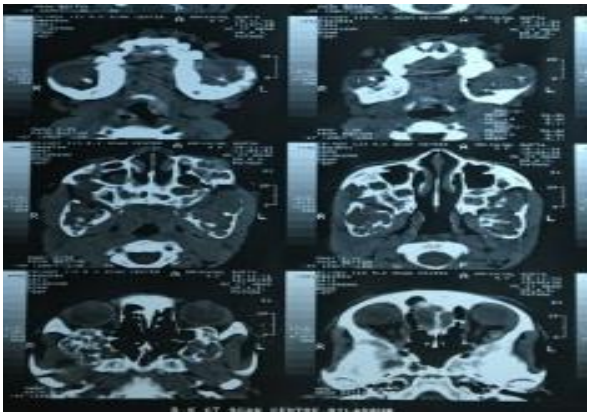

Fig 6: Axial CT Scan slices.

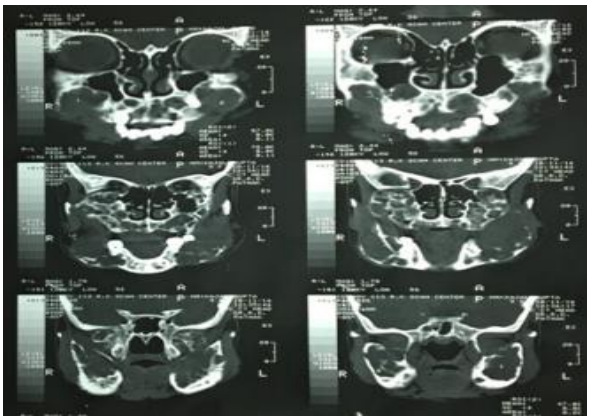

Fig 7: Coronal CT Scan slices.

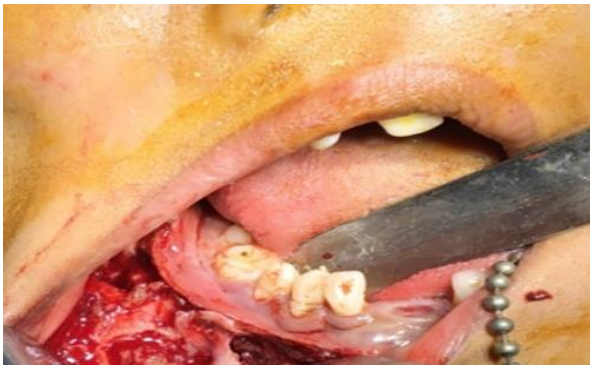

Fig 8: Intraoperative pic.

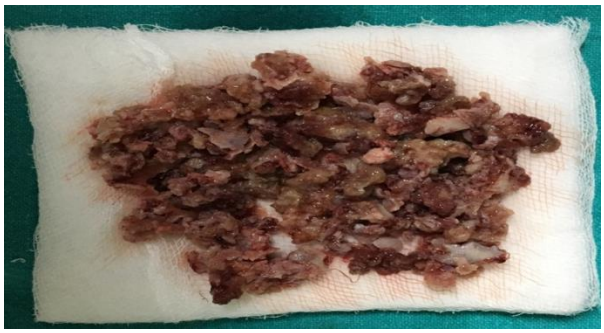

Fig 9: Excised tissue.

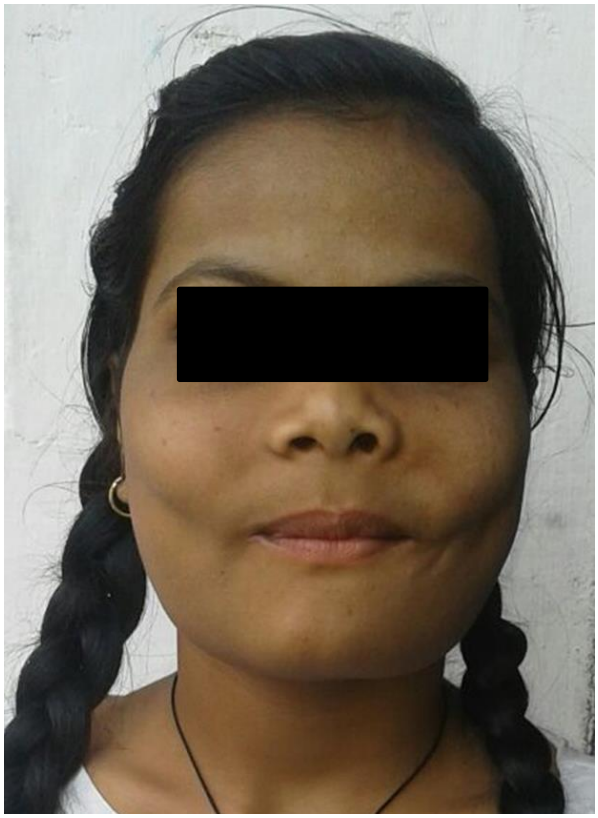

Fig 10:4 Month Postoperative view.

mandibular arch to be maloccluded. Mandibular arch showed two prominent swellings; first being in the incisor area, second larger one in the left canine premolar region (Fig 3). Swelling was reddish, firm and non tender on palpation. There was significant expansion of maxillary and mandibular alveolar ridges both buccally and palatally/lingually. On radiological examination in Lateral Skull Xray and OPG, there appeared bilateral haziness of maxillary sinus, bilateral, diffuse, multilocular radiolucencies in maxilla and mandible, zygomatic and frontal bone (Fig 4,5). CT scan reconfirmed the mixed tumor (Fig 6,7) Histopathological examination showed multiple multinucleated giant cells together with ovoid to spindle shaped cells within a fine fibrillar collagenous stroma, numerous small vessels with large endothelial cells and perivascular capillary cuffing confirming the diagnosis of cherubism. Curretage of both maxillary and mandibular lesions was carried out under General Anaesthesia after a complete work up (Fig. 8, Fig 9). Patient showed remarkable stable improvement in esthetics as visible in the four month post-operative visit. (Fig 10)

DISCUSSION AND LITERATURE REVIEW

Jones [6], was the first to describe cherubism as a familial multilocular cystic disease of the jaws. In the recent literature review, cherubism is associated genetically [1, 2] as an autosomal dominant disorder in which normal bone is replaced by cellular fibrous tissue and immature bone. Among the craniofacial component, mandible is the most severely affected bone which deteriorates the aesthetic balance of the face. Bilateral swelling of the cheeks, mandibular and maxillary enlargement causes orbital manifestation and tendency of eyes looking up to the sky. In a study [1] cherubism has been defined as a genetically determined alteration of tooth germ development which indicates familial occurrence and observed its correlation with a mutation in gene SH3BP2.

Cherubism received its name because of angel like appearance of the patients (Chubby and upward directed look). The clinical presentation of the disease in this patient was similar to the findings in other follow up studies. Burkhardt and Berthold (1986) examined the tissue taken from a boy afflicted with cherubism using light and electron microscopy and immunohistology and found a close relationship of cherubism with giant cell granuloma of the jaws [7].

The histological examination showed fibrohistiocytic tissue with multinuclear histiocytic giant cells thinly disseminated throughout the fibrous tissue. Immunohistochemistry analysis demonstrates tartarate resistant acid phosphatase revealing osteoclastic character of these giant cells.

Although cherubism appears to be inherited as an autosomal dominant trait with 70% in females and

90% in males, other pattern of inheritance and association with syndromes has been reported [8–10]. Around puberty the condition begins to regress until by 30 years of age, when lesions frequently are not detectable. The literature shows that the lesion tends to occur twice as often in the mandible than in the maxilla [11–13]. 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. In a general population, large and aggressive lesions are less common, than suggested by literature. Multiple lesions however, occur more frequently than previously assumed.

Cherubism has been classified according to the severity grades from the Seward and Hankey [14] system:

Grade I: Involvement of bilateral mandibular molar region and ascending rami, mandible body or mentis.
Grade II: Involvement of bilateral maxillary tuberosities as well as the lesion of grade I, diffused whole mandible.
Grade III: Massive involvement of the entire maxilla and mandible except the condyles.
Grade IV: Involvement of both jaws with condyles.

In general, the treatment of cherubism is to biopsy the lesion, extract any ectopic and impacted teeth and correct it surgically when appropriate [1, 3–5, 15]. 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. In patients with aggressive signs and symptoms, surgical curettage is not an effective therapy [16].Mild forms of cherubism without facial dysmorphology, dental and ocular involvement may not require treatment as cherubism is expected to regress spontaneously after puberty. Management in these cases consists of longitudinal observation. During the growth phase of the lesions, annual clinical and radiographic examination with a panoramic or other appropriate radiograph is suggested. Follow-up every 2 to 5 years is advisable after the disease becomes quiescent. Expansile fibrous lesions in severe cases may regress well after adolescence [6, 11]. In other instances actively expanding lesions in suspected cherubism may still be diagnosed in adults [12]. In our patient, a decision to intervene surgically was made as the patient repeatedly approached requesting for the same as she did not have a socially acceptable appearance.

Surgical intervention is indicated when aesthetic or functional concerns arise including nasal obstruction, proptosis or facial deformity. Definitive surgical procedures should be performed after puberty when the lesions are quiescent. Severe aesthetic or functional problems may justify intervention prior to puberty. Options for surgical management include partial resection, contour resection, curettage or a combination of these. Orbital surgery may be required in rare cases when tumor tissue invades the floor of the orbits to a degree where it leads to the displacement of globes or loss of vision is suspected due to optic atrophy [19].

Conservative treatment is appropriate until functional or emotional disturbances demand surgical intervention. Medical therapy in the form of calcitonin is theoretically appropriate but clinical evidence is lacking so as to endorse its application though calcitonin has been shown to cause inhibition of bone resorption in cherubic tissue in vitro. However, curettage in the beginning of resorption phase has been successful with/without bone graft [4, 6].

Development of dentition should always be closely monitored. Roots may resorb or tooth displacement can occur in cases where lytic lesions develop around erupting secondary dentition or teeth may appear missing or to float in the lesion [11]. The problem of early loss of deciduous teeth, absence, delayed development or eruption of the permanent teeth is difficult to treat and no satisfactory solution is available. Space maintainers are used while waiting for the permanent teeth to erupt. Malocclusion is a major concern. Surgical exposure of impacted teeth is sometimes necessary (Figure 5).

CONCLUSION

Cherubism is a rare benign bone disease with autosomal dominant inheritance. The signs and symptoms depend on the severity of condition and range from no clinically or radiographically

detectable features to grotesquely deforming mandible or maxilla with respiratory embarrassment, impaired vision and hearing. Interventional surgery during growth may psychologically support the patient. A routine monitoring of growth and dentition into adulthood combined with appropriate management at each stage is recommended.

CONFLICTS OF INTEREST

The authors declare they have no potential conflict of interests regarding this article.

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