ISSN: 2581-5989
PubMed - National Library of Medicine - ID: 101738774

International Journal of Dental Science and Innovative Research (IJDSIR)

IJDSIR : Dental Publication Service

Available Online at: www.ijdsir.com

Volume – 6, Issue – 4, August - 2023, Page No. : 70 - 74

Nonfamilial cherubism in an 8-year-old child: A case report

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Citation of this Article: Dr. Inam Uddin, Dr. Abira Chattopadhyay, Dr. Monalisa Das, Dr. Aritra Chatterjee, Dr. Shreya Ganguly, "Nonfamilial cherubism in an 8-year-old child: A case report", IJDSIR- August - 2023, Volume – 6, Issue - 4, P. No. 70 – 74.

Type of Publication: Case Report

Conflicts of Interest: Nil

Abstract

Cherubism is a rare disease affecting the maxillomandibular complex and should be considered in the differential diagnoses of bone diseases affecting the jaws. The molecular pathogenesis of cherubism involves a mutation in the gene encoding SH3 binding protein (SH3BP2) and suspected degradation of the Msx-1 gene. Dental alterations associated with cherubism include early exfoliation of deciduous teeth, impaction and/or displacement of teeth which radiographically seem to float in radiolucent areas, conferring the so-called "floating tooth appearance''. An 8 year-old female child reported to the Department of Oral surgery with a chief complaint of painless, progressive and bilateral enlargement of upper and lower jaws. Extraoral examination revealed bilateral enlargement of the mandible as well as maxilla. Absence of any abnormal pigmentation on the face was noted. On palpation there was absence of any tenderness and consistency of the swelling was hard. On intraoral evaluation few deciduous teeth were seen with others missing. Few permanent teeth were also seen. Cone beam computed

tomography revealed extensive involvement of mandible as well as maxilla. Osteolytic changes were evident radiographically with bilateral involvement of mandible and maxilla. Intraoral biopsy was done and the histopathological report revealed presence of multiple multinucleated giant cells within a background of multiple proliferative mononuclear cells. Few areas of perivascular eosinophilic cuffing in the stroma were also noted.

Keywords: Cherubism, Giant cell lesions, Floating tooth.

Introduction

Cherubism (OMIM#118400) is a rare fibro-osseous disease of childhood affecting the mandible and maxilla. It affects bilaterally leading to facial, dental and ocular abnormalities. The term Cherubism was invented in 1933 by Jones, who found a similarity between these patients and the cherubs which were depicted in the renaissance paintings.[1] The disease has other synonyms that include familial multilocular cystic disease of the jaw; familial fibrous dysplasia; familial multiple giant cell lesions of the jaws.[1,2]

The molecular pathogenesis of cherubism involves a mutation in the gene encoding SH3 binding protein (SH3BP2) and suspected degradation of the Msx-1 gene which is involved in the regulation of mesenchymal interaction during craniofacial morphogenesis. It is believed that the clinical features of cherubism are due to the genetic mutations or incomplete penetrance. [3,4]

Cherubism is found in children in the age group of 2 to 7 years with the exacerbation of symptoms in the first 2 years after diagnosis and of stabilization or regression after puberty. Gender predilection is present with males being more affected than females in a ratio of 2:1. Clinically it manifests as bilateral enlargement of the mandible and/or maxilla, causing a rounded face and swollen cheeks accompanied by upward-looking eyes. In this disorder serum calcium and phosphorus concentrations and TSH, T4 and T3 hormone levels are usually within normal limits but alkaline phosphatase levels might be elevated.[5,6]

Radiographically cherubism can be classified into 3 grades, grade I, bilateral involvement of ascending ramus of mandible; grade II, bilateral involvement of ascending ramus of mandible and maxillary tuberosity; grade III, extensive involvement of mandible and maxilla involving the coronoid and condylar processes of mandible.

Dental alterations associated with cherubism include early exfoliation of deciduous teeth, impaction and/or displacement of teeth which radiographically seem to float in radiolucent areas, conferring the so-called “floating tooth appearance’’. In addition, ectopic tooth eruptions, agenesis of permanent teeth occur in cherubism. Agenesis mainly affects second and third molars due to involution of their germs and root resorption of existing teeth. [5,6]

Case report

An 8-year-old female child reported to the Department of Oral surgery with a chief complaint of painless, progressive and bilateral enlargement of upper and lower jaws. The patient was apparently well until the age of 6 years. The history revealed that the patient had been born as a full-term caesarean delivery and showed no abnormalities until about the age of 6 when she noticed gradual painless bilateral enlargement of lower face which gradually involved the upper face as well. The enlargement continued to progress gradually over the subsequent years. Family history did not reveal any close family members suffering from such a disease. No abnormality was found on clinical examination of the chest, abdomen, cardiovascular and central nervous

system. There was absence of any abnormal pigmentation anywhere else in the body.

Extraoral examination revealed bilateral enlargement of the mandible as well as maxilla. Absence of any abnormal pigmentation on the face was noted. On palpation there was absence of any tenderness and consistency of the swelling was hard. There was no ophthalmic abnormality (Figure-1).

On intraoral evaluation few deciduous teeth were seen with others missing. Few permanent teeth were also seen (Figure-2). Cone beam computed tomography revealed extensive involvement of mandible as well as maxilla. Osteolytic changes were evident radiographically with bilateral involvement of mandible and maxilla which extended upto the infraorbital rim bilaterally (Figure 3-5).

Laboratory investigations showed a hemoglobin level of 11.8 gm/dl (normal 13 to 18 gm/dl), hematocrit value of 38.4% (normal, 40 to 52%), each being slightly low, and an elevated alkaline phosphatase value of 290 IU/L (normal - 85 to 270 IU). Parathyroid hormone level and other lab investigations were within normal limits. The lesion was diagnosed as grade III cherubism.

Intraoral biopsy was done and the histopathological report revealed presence of multiple multinucleated giant cells within a background of multiple proliferative mononuclear cells. Few areas of perivascular eosinophilic cuffing in the stroma were also noted.

A policy of wait and watch was taken for this particular patient and patient was kept on follow up. Definitive Prosthetic rehabilitation was deferred until the completion of the growth for that particular patient.

Discussion

Cherubism is a rare disease affecting the maxillomandibular complex and should be considered in the differential diagnoses of bone diseases affecting the jaws. According to World Health Organization (WHO) cherubism is a nonneoplastic bone disease affecting the jaws. The most accepted theory regarding the pathogenesis of cherubism is its association with an autosomal dominant gene. The SH3BP2 gene which plays an important role in the regulation of osteoblastic and osteoclastic activity during physiological tooth eruption undergoes point mutation in cherubism.[5-8]

The molecular model which has been proposed for the etiopathogenesis of cherubism is based upon the interaction between an altered parathyroid hormone related protein (PTHrP) receptor with the Hox gene Msx-1. Thus SH3BP2 - dependent signal transduction pathways interfere with the jaw morphogenesis leading to cherubism. As a result there is dysregulation of mesenchymal bone formation leading to development of giant cell granulomas.[7,8]

The radiographic techniques used for the diagnosis of suspected cases of cherubism include posteroanterior radiograph of face, orthopantomogram. Computed tomography is also a useful adjunct. Magnetic resonance imaging is also useful to study the expansion of soft tissue, in particular in the aggressive forms. On bone scintigrams, low radioactivity (cold areas) can be sometimes observed which is characteristic of cherubism.[8]

The specific histological finding of cherubism is the presence of eosinophilic perivascular cuffing which, however, is not always present and histology is of limited diagnostic significance. Histopathologically cherubism has similarity with central giant cell lesions as well as with lesions associated with hyperparathyroidism. Histopathologically multinucleated giant cells are found scattered throughout collagenous and vascularized fibrous connective tissue containing collagen fibers. This connective tissue is

richly vascularised and mainly contain giant cells, with the presence of hemosiderin deposits.[8] Southgate et al. found that multinucleated cells in cherubic lesions had phenotypic characteristics of osteoclasts, resorbing bone in vitro.[9] In addition to histopathology, Cytogenetic and molecular studies have being used for diagnosis purpose, such as fluorescence in situ hybridization and quantitative analysis of Msx-1 expression in different tissues.[9,10]

Once the diagnosis is established, therapeutic management should be evaluated. Treatment options include waiting for stabilization and spontaneous remission of the disease. Tooth extraction can be done in areas showing fibrous alterations, cosmetic osteoplasty also can be done of the affected jaws after regression of disease activity. In case of functional impairment, curettage of the lesions and treatment with calcitonin can be done.[5] Curettage has been suggested to be as a good approach since this intervention stimulates bone replacement. The policy of waiting for disease regression, followed by the evaluation of physiological bone remodelling, is the most recommended. Radiation therapy is not considered as a treatment modality for cherubism because of the potential risk of osteoradionecrosis or even malignant transformation resulting in osteosarcoma.[5] Although cherubism was described more than 70 years ago, the scarcity in literature has provided little insight on effective therapies. Prosthodontic rehabilitation can be considered for these patients to restore function and improve the quality of life.

Conclusion

The prognosis for cherubism is good. It does not progress after puberty and as the patient reaches adulthood, the jawbone reaches a more normal configuration. Surgery is not the commonest treatment modality however in certain cases biopsy and surgical intervention can be considered. Prosthodontic rehabilitation is required for restoration of function.

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biochemistry of cherubism. J Clin Pathol. 1998;51(11):831-837.

Legend Figures

Figure 1: Extraoral photograph of the patient

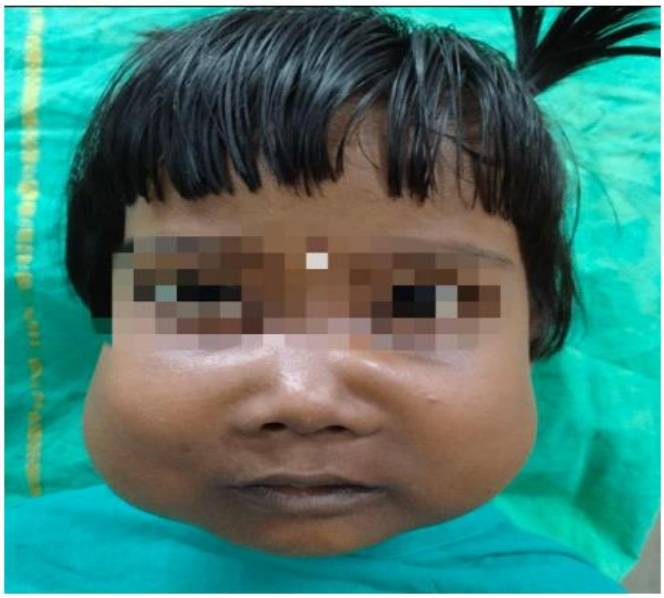

Figure 2: Intraoral photograph showing expansion of the cortex

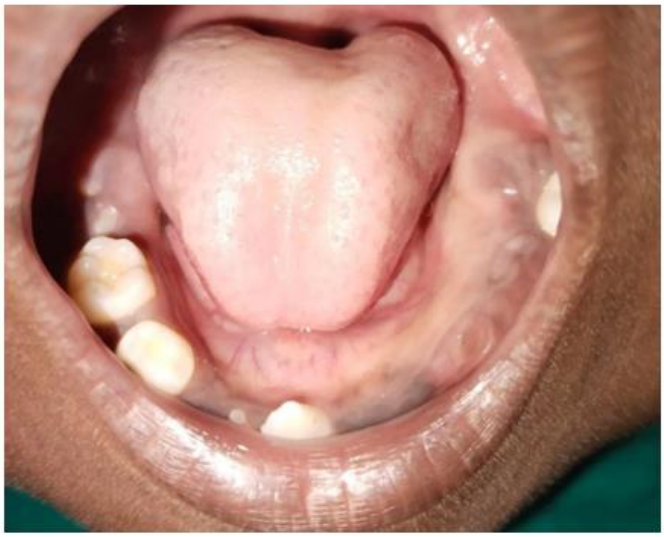

Figure 3: Cone beam computed tomography image of the lesion (Frontal view)

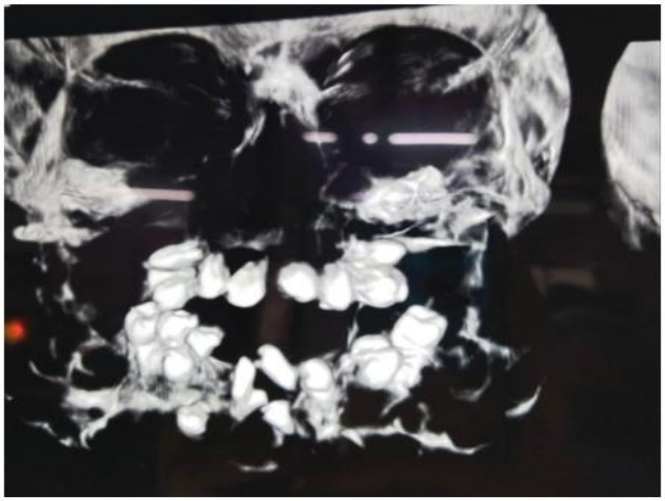

Figure 4: Cone beam computed tomography image of the lesion (Lateral view)

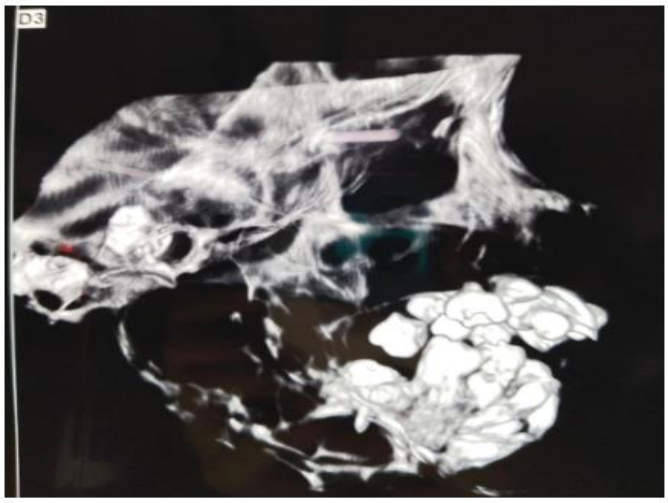

Figure 5: Orthopantomographic image of the lesion