Case Report

A Case Report of Cherubism

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ABSTRACT

Cherubism is a congenital childhood disease of autosomal dominant inheritance. Here, we present a rare case report of cherubism in a 13 year old male patient came with complaints of painless enlargement of face, puffiness and describing the clinical, radiographic features and confirmed on histopathology.

Keywords: Cherubism, Jaw disease, autosomal dominant inheritance

INTRODUCTION

Cherubism was first described by William Jones in 1933.[1] Cherubism is a rare congenital childhood disease of autosomal dominant inheritance characterized by painless, frequently symmetrical, bilateral enlargement of the jaws, as a result of the replacement of bone with fibrous tissue. [2] This disease also called as "Familial Multiocular cystic disease of jaws". [3]

A molecular pathogenesis of cherubism has been proposed, with the detection of a mutation in the gene encoding SH3 - binding protein 2 (SH3BP2) and possible degradation of the Msx-1 gene which is involved in the regulation of mesenchymal interaction during craniofacial morphogenesis. [4]

Cherubim is usually diagnosed in children aged 2 to 7 years, with the observation of exacerbation of its manifestations within the first 2 years after diagnosis and of stabilization or even regression after puberty. Boys are more affected than girls at the proportion of 2:1. [5]

CASE REPORT

A 13 year old male patient complaint with gradual painless, swollen gums and enlargement of face since 4 years, otherwise he is apparently normal. There was no extra oral secondary skin change. On physical examination, there is no associated pain, temperature, tenderness and hard in consistency. On extra oral examination, diffuse swelling is seen over both side of face. The radiographic findings demonstrated on a

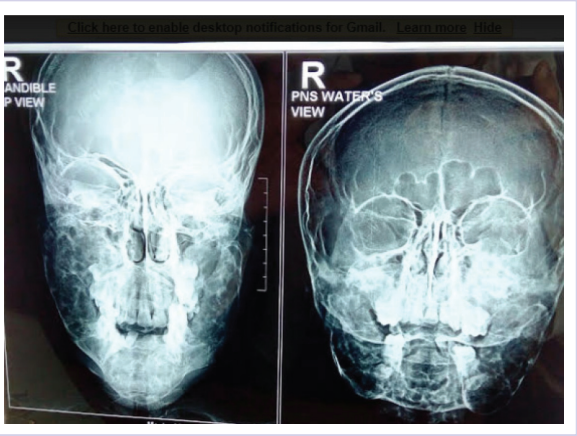

Figure 1: Plain radiography features consist of lucent expanded regions within the maxilla and mandible , with soap bubble appearance, thinning of cortex noted.

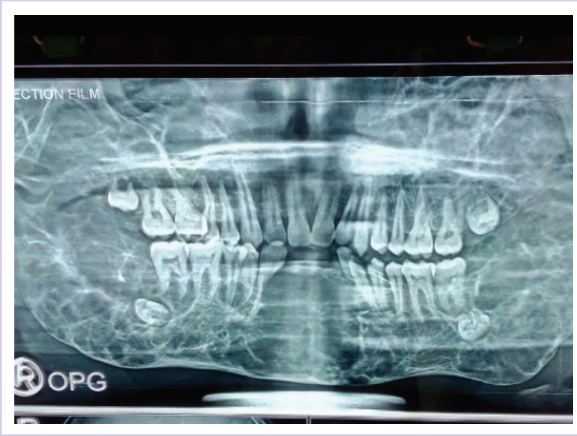

Figure 2: Expansile osteolytic lesions involving mandible on both sides

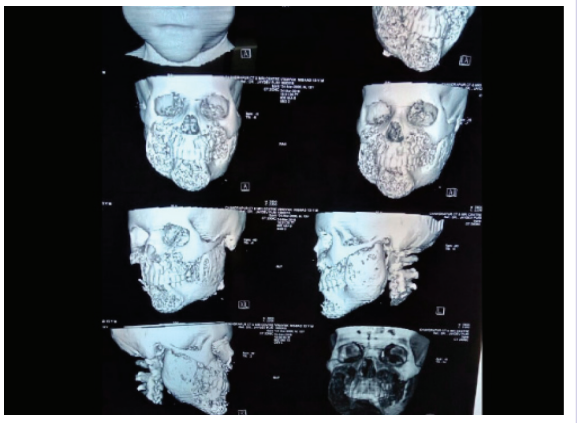

Figure 3: 3D computed tomographic images showing osteolytic lesions in mandible and maxilla

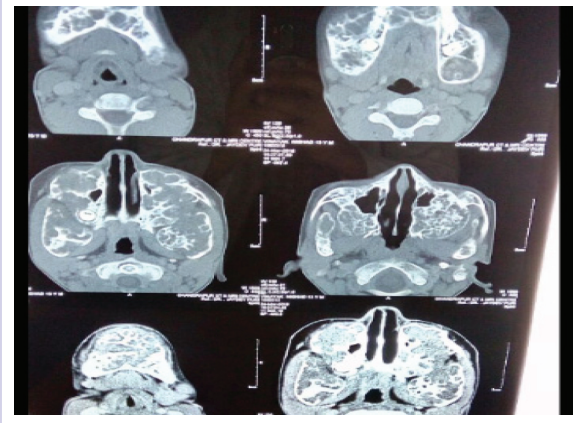

Figure 4: Computed tomographic images showing osteolytic lesions in mandible and maxilla

radiograph (figure 1 and 2), Computed tomography (CT) scan (figure 3&4) of mandible were in keeping with diagnosis of Cherubim. This was confirmed on histology.

DISCUSSION

Cherubism is a rare congenital autosomal dominant disease, characterized by enlarged face due to swelling of the jaws which is bilateral in most cases, bony consistency of the lesion, intact mucosa, dental malocclusion, upward-looking eyes in the case of maxillary involvement, and absence of pain.

In present case, plain radiogaphic finding was showed that lucent expanded regions with in maxilla and mandible, with "Soap-bubble appearance" and thinning of cortex were noted. (Figure 1&2) Paroramic radiographic views are acceptable for the initial diagnosis, but multiplanar and 3 dimensional CT scan are mandatory for optimal visualization of the extent of disease. [6,7]

CONCLUSION

Cherubism is a clinically well- characterized disease which confers to the patient the appearance of a baroque cherub. In cases of a suspicion of cherubism, radiographic examination is essential since the clinical presentation and the location and distribution of the lesions may define the diagnosis. Histopathological examination is complementary.

CONFLICT OF INTEREST :

The authors declared no conflict of interest.

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