Cherubism with idiopathic gingival enlargement: A rare case report

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Abstract:
Cherubism is a congenital childhood disease of autosomal dominant inheritance. It is a benign, familial giant cell lesion characterized by gradually progressive painless swelling of the jaws. Idiopathic gingival enlargement is a rare condition and may be associated with some uncommon syndromes. This case report describes an 11-year-old patient with unusual clinical form of gingival enlargement, cherubic facial appearance. Clinical examination revealed the presence of the hyperplastic gingiva, which completely covered all teeth. The bilateral swelling of mandible and the appearance of the sclera beneath the iris suggested cherubism. The diagnosis was confirmed by histopathological examination, which revealed multinucleated giant cells. Computed tomography scan showed multiple osteolytic zones in the mandible. A full mouth gingivectomy was performed in four stages. Lesion healed successfully, and no recurrence observed after 1-year follow-up. There was a marked improvement in esthetics and function through the surgical excision of the overgrowth.

Key words: Cherubism, idiopathic gingival enlargement, gingivectomy

INTRODUCTION
Cherubism is an extremely rare disorder, which usually involves maxilla and mandible, and it results in unilateral/bilateral swelling. It is characterized by a marked fullness of the cheeks and jaws, with an eye to heaven appearance. The appearance was similar to reminiscent angelic cherubs. It is a congenital childhood disease of autosomal dominant inheritance and is also designated as familial multinodular cystic disease of jaws since multinucleated giant cells are detected histopathologically. Genetic analysis revealed cherubism is because of the mutation in the gene encoding SH3-binding protein (SH3BP4) and degradation of MSV1 gene during craniofacial morphogenesis. The different clinical manifestations are due to the incomplete penetrance or changes secondary to a mutation.

Hereditary gingival enlargement is a rare disease with unknown etiology, and usually appears in early childhood. Though the etiology is still unknown, an inherited genetic predisposition remains the major factor. The idiopathic gingival enlargement also appears to be a part of several syndromes.

In this case report, we present a patient who has a rare combination of gingival enlargement and cherubism with her clinical, radiological and histopathological findings.

CASE REPORT
A female child aged 11 years reported to the Department of Periodontia, of JKKN Dental College with the chief complaint of overgrown gums in both upper and lower jaw. The patient history reveals that her complaints started at 3 years of age and enlargement increased progressively covering the crowns of teeth [Figure 1]. Clinical examination revealed the presence of gingival enlargement and facial dysmorphism, with no relevant medical and familial history. There was no eye complaint, but there was upturning of eyes. On examination, psychomotor skills were normal. Computed tomography (CT) brain scan shows absence of neurological involvement. No skin pigmentation or other pathology was evident. Serum alkaline phosphatase level was normal.

Intra orally, gingival enlargement without interdental papilla was evident. Gingival enlargement completely covers the teeth and...
obscures the lip closure [Figure 2]. The enlargement was non-erythematous and on palpation it was firm and leathery in consistency.

Panaromic radiograph showed a bilateral multilocular radiolucency involves the body and ramus of the mandible, with sparing of condyle and thinning of the inferior border of the mandible was also evident. It also revealed the progressing osteolytic lesion pushing the permanent molar tooth bud to the inferior border of the mandible and also showed missing of lower right permanent canine tooth bud and upper left premolar tooth bud [Figure 3].

Gingival biopsy reveals multinucleated giant cells scattered throughout the connective tissue [Figure 4].

Bone biopsy was taken in the right retromolar region of the mandible under local anesthesia. The section shows vascular fibrous stroma with scattered multinucleated giant cells. The

Figure 1: The patient with bilateral swelling of cheeks and upward gaze of eyes

Figure 2: Gingival enlargement covers the teeth and prevents lip closure

Figure 3: OPG showing bilateral multilocular radiolucency

Figure 4: Gingival biopsy showing multinucleated giant cells

Figure 5: Bone biopsy showing vascular fibrous stroma with scattered multinucleated giant cells

Figure 6: Three-dimensional frontal view
fibrous stroma is arranged in whirled pattern, which is abetting on the bony trabeculae. The giant cells are showing prominent nucleus and nucleolus. Few areas of the blood vessels show the collagen cuffing, but it is not prominent [Figure 5]. No malignant changes were seen.

Three dimensional frontal view of mandible and Axial view of the CT of mandible reveals bilateral expansile multilocular osteolytic zones, extending from the body of the ramus to the parasymphysis on both the sides, with thinning of the inferior border of the mandible. The hypodense areas were more prominently seen on the right side of the mandible [Figures 6 and 7].

**DISCUSSION**

Cherubism was first reported by Jones[1] in 1993 as it is a disease of early childhood and consists of a painless mandibular enlargement with or without the maxillary involvement. The cause of cherubism is usually unknown but can be due to the developmental disturbance of the forming mesenchyme. According to the Novack and Faccio,[6] cherubism is caused by the enhanced tumor necrosis factor-α production by myeloid cells due to an activating mutation of SH3BP2.[2]

Gingival enlargement may be due to a variety of causes such as inflammation, leukemic infiltration, and drugs.[7] Another form of gingival fibromatosis may be occur by itself or as a part of a syndrome and appears in early childhood and is known as idiopathic or hereditary gingival fibromatosis.[8]

The present case was characterized by gingival enlargement and cherubism. With regard to cherubism, the present case shows the clinical characteristics of cherubism including bilateral mandibular enlargement, high arched palate, and teeth malposition’s with the upward gaze or eyes to heaven with diffuse enlargement of lower half of the face. According to Meng et al. 2005,[9] cherubism is usually diagnosed in 6–10 years of age and the present case was diagnosed during the first decade of life. Peñarrocha et al.[10] in 2006 reported that radiographic lesions of cherubism are multiple, well defined multilocular radiolucency’s and may contain teeth that float in these areas. The panoramic radiograph revealed the presence of multilocular radiolucency and the axial view of CT scan of the mandible showed multiple osteolytic areas, which confirmed the diagnosis as cherubism. Gingival biopsy showed a highly vascular fibrous stroma with unevenly distributed multinucleated giant cells. This was in accordance with Lannon et al. in 2001.[11]

Thus, the present case was diagnosed as cherubism depending on the clinical presentation, physical examination, radiographic and histological variables.

Ramon[12] in 1967 described two cases with cherubism, gingival enlargement, mental deficiency and hypertrichosis and suggested that both hereditary gingival enlargement and cherubism were different expression of the same disease. According to Ramon[12] et al. gene dosage effect or alteration in multiple genes may be responsible for syndromic form of gingival fibromatosis. Later Pina-Neto et al.[13] in 1986 reported cherubism, gingival fibromatosis, epilepsy, mental deficiency along with juvenile rheumatoid arthritis. In the present case, hypertrichosis, epilepsy, rheumatoid arthritis was not reported. Our case showed similar characteristic as Ramon’s case including cherubism, gingival fibromatosis, but mental retardation was absent.
The patient was explained about the procedure and informed consent was obtained. In the present case, gingivectomy was performed in sextants under local anesthesia. Oral hygiene instructions were given, and maintenance recalls were established. The wound healing is uneventful. At 1-year follow-up period teeth eruption was noted in the upper and lower anterior and also marked esthetic improvement was observed [Figures 8 and 9].

Thus, the knowledge of the clinical and radiographic alteration observed in patients with cherubism and its relation to gingival fibromatosis is important as the dentist might be the first professional sought for the diagnosis of the diseases.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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