ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 02 | February - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

FAMILIAL FIBRO OSSEOUS LESIONS OF THE JAWS

Dental Science

K. Saraswathi Gopal Head of the department, Department of Oral Medicine and Radiology, Faculty of Dentistry, Meenakshi Academy of higher education and research, Chennai.

V. S. Madhu Preetha* Post graduate, Department of Oral Medicine and Radiology, Faculty of Dentistry, Meenakshi Academy of higher education and research, Chennai. *Corresponding Author

M. Ragu Ganesh Associate Professor, Department of Oral Medicine and Radiology, Faculty of Dentistry, Meenakshi Academy of higher education and research, Chennai.

ABSTRACT

Cherubism is a rare, self-limiting, autosomal dominant inheritance, first described by Jones in 1933. It is benign symmetrical fibro-osseous disease limited to maxilla and mandible caused by mutation of SH3BP2 gene on chromosome 4p16.3. Aggressive form of the disease is usually seen in young children, non-aggressive form is seen in teenagers and quiescent form is usually seen in older patients. Regression occurs during puberty when the disease stabilizes after the growth period leaving some facial deformity and malocclusion. Here we present a case of 15year-old female patient who presented with bilateral bony enlargement of jaws and she was diagnosed based on clinical and radiographical findings and treated accordingly.

KEYWORDS

Benign, genetic, self-limiting, fibro osseous, jaws.

INTRODUCTION

Benign fibro osseous lesions are diverse collection of diseases which majorily includes fibrous dysplasia, ossifying fibroma and osseous dysplasia[1]. A practical and direct approach based on patient's history, intra oral presentations, clinical presentation, radiological and histological findings will help in accurate diagnosis of the lesion. Since most of the fibro osseous lesions have similar histological findings, it is very important to rely on clinical and radiological findings to provide an accurate and definitive diagnosis, which will ensure proper treatment and good prognosis[2].

CASE REPORT

A 15year old female patient reported with chief complaint of swelling and pain in left middle third of the face for past 2 months. Patient gives history of swelling in the left middle half of the face which gradually increased in size to attain the present size. Patient gives h/o mild to moderate intermittent type of pain in the left side. Patient gives h/o frequent obstruction of left nose and h/o COLD which subsided under medication. Patient also gives h/o fullness of middle and lower third of the face (round and chubby face) since childhood. Patient's family history revealed, her father has similar facial features of fullness of middle and lower third half of the face. On local examination, hypertropia of eyes present, post nasal drip present, lifting of left ala of nose and left nasal obstruction present.

On extra oral examination, presence of diffuse enlargement of right and left middle and lower third of the face involving the malar region, mandibular body, ramus, angle region causing fullness of the face. Obliteration of the left nasolabial fold evident and asymmetrical lifting of left ala of nose. On palpation, the swellings are bony hard in consistency, tender on palpation on left maxillary sinus region with no secondary changes.

On intra oral examination, presence of diffuse swelling in the left side of the hard palate extending from palatal rugae to posterior part of the hard palate, the mucosa over the swelling appears pale pink in color in accordance with the adjacent mucosa with evidence of single well defined papule present palatal to 26 measuring approximately 3*3mm in its greatest dimension, the mucosa surrounding the papule appears greyish with no secondary changes. Evidence of buccal and lingual cortical plate expansion of right mandibular alveolus in relation to 46,47 region causing obliteration of right lower buccal vestibule, with no tenderness and no secondary changes. On palpation of the swelling of the hard palate on left side, it is hard in consistency, non-tender with no secondary changes. Evidence of diffuse swelling in the lower left buccal vestibule in relation to 33,74 region measuring 2*2cm in which is hard in consistency, non-tender with no secondary changes.

Hard tissue examination revealed, total number of teeth present are 25, clinically missing 17,27,34,35,37,47, palatally placed 14, rotated 13, buccally placed 25, retained deciduous 74,75,85, lingually placed 46 and evidence of diffuse chalky /frosty white flecks on the labial and buccal surface of all teeth with yellowish brown stains/bands evident in the middle and incisal third of labial surface of upper and lower anterior teeth- giving mottled appearance.

Patient was subjected to blood investigations, which revealed parathyroid hormone level 37.21 pg/ml and alkaline phosphatase level 723 IU/L, which are in normal range. Based on the patient's history, family history, general and clinical examination of the craniofacial region, a provisional diagnosis of benign fibro osseous lesions of maxilla and mandible was given with differential diagnosis of cherubism, fibrous dysplasia (cranio facial type-polystotic form) and giant cell lesions (Central giant cell granuloma) was given. Other diagnosis include malocclusion, partially edentulous 17,27,37,47, retained deciduous teeth 74,75,85, generalized enamel hypoplasia (Dental fluorosis) and internal derangement of TMJ- Anterior disc displacement with reduction of left TMJ seen.

INVESTIGATIONS:

RADIOGRAPHICAL INVESTIGATIONS:

MAXILLA OCCLUSAL -CROSS SECTIONAL RADIOGRAPH revealed.,

Rotated-13, palatally erupted 14, resorption of apical third of the roots in relation to 21,22, buccally erupted 25 and altered bony trabeculae pattern with multiloculated radiolucent areas surrounded by sclerotic borders in the palate.

MANDIBULAR OCCLUSAL- CROSS SECTIONAL RADIOGRAPH revealed.,

Lingually placed -34,46, buccally erupted 35 rotated 44, buccal and lingual cortical plate expansion in relation 45,46 region and alteration in bony trabeculae pattern with areas of multiple loculated radiolucent areas.

OPG

Zone-1 reveal-

No of teeth-27, rotated-13, superimposition of 13 and 14, resorption of apical third of root in relation to 21,22, open apex- 25, impacted -34,35, retained deciduous-74,75, superimposition of retained deciduous 85 and 46.

Zone-2,3,4,5,6 reveal-

Multilocular radiolucencies in right and left maxillary sinus noted. Alteration in bony trabeculae pattern in maxilla, mandible and TMJ with areas of multilocular expansile radiolucencies surrounded by sclerotic borders of bone in the zygoma, maxilla, mandibular body, angle and ramus, coronoid process noted. Hyoid appears normal.

LATERAL CEPHALOGRAM
Alteration in the bony trabeculae pattern noted in the maxilla and mandible with multilocular radiolucencies surrounded by sclerotic borders evident.

CT WITH CONTRAST -HEAD AND NECK
Predominantly expansile multiloculated radiolucent lesions with thin sclerotic scalloped cortical margins seen involving bilateral ramus and posterior body of the mandible. Similar lesions with central ground glass attenuation seen involving postero-lateral walls, anterior walls of bilateral maxillary sinuses causing mild obliteration of maxillary antrum (LT>RT). The lesion in the left maxillary sinus is seen to extend into left nasal cavity and superior alveolar arch causing left nasal cavity obstruction. Left maxillary mild sinusitis and deviated nasal septum to right noted. So an impression of benign fibro osseous lesions/fibrous dysplasia / odontogenic keratocysts was given.

Patient was further subjected to incisional biopsy and final diagnosis of cherubism was given based on paitent's family history, general and oral examination, radiographical and serological findings, a final diagnosis of Cherubism was given. Further, bilateral maxillary contouring and enucleation was done under GA.

DISCUSSION
Cherubism has historically been considered a variant of fibrous dysplasia, but in reality is likely a distinct entity. Familial fibrous dysplasia of jaws, disseminated juvenile fibrous dysplasia, Familial multilocular cystic disease of jaws are the other names of cherubism[3]. Cherubism is usually described by the appearance of multilocular, symmetrical expansile lesions of the mandible and the maxilla that typically appear at the age of 2 to 7 years, continue through puberty and may or may not continue to progress with age. Cherubism is a rare disorder and the precise incidence is unknown[4]. It is inherited in an autosomal dominant pattern and has variable penetrance, with onset in early childhood. Approximately 200 cases have been reported by medical journals with the majority being males, but there are many cases where females are also involved[5].

Grading systems for cherubism have been suggested to describe location and severity of lesions.

- **Grade I:** Fibro-osseous bilateral and symmetrical expansions in the rami of the mandible without root resorption
- **Grade II**: More severe involvement of the ramus and body of the mandible and the tuberosity region of the maxillae
- **Grade III**: Involvement of maxilla and mandible in their entirety with considerable facial deformity
- **Grade IV:** Lesions involving the mandible and maxilla and showing signs of root resorption
- **Grade V**: The rare, massively growing, aggressive, and extensively deforming juvenile cases involving the maxilla and mandible, and may include the coronoid and condyles
- **Grade VI**: The rare, aggressive and deforming juvenile lesions which involve the maxilla, mandible and the orbits.

Clinical Presentation
It is bilateral, usually symmetric, with a slight upward turning of eyes noted. Displacement of the globe and retraction of the eyelids can result in exposure of a rim of the sclera. The disease also invade the retrobulbar spaces and cause displacement of the optic nerves and result in proptosis. The orbital effects of this fibro osseous lesion are due to this displacement and not to direct invasion of the globe and surrounding extraocular muscles. In addition, submandibular lymph node enlargement may be present and it is the only fibro osseous lesion where lymph node enlargement will be seen[6]. The teeth in the affected regions may be loose, and delayed tooth eruption. Normal bone remodeling activity may resume after puberty. The fibrous cystic lesions will be found in the trabecula of the Coronoid process, the ramus of mandible, the body of mandible and the maxilla region. Radiographically- TEETH HANGING IN AIR appearance is noted.

PATHOGENESIS
Cherubism has also been found combined with other genetic disorders including Noonan syndrome, Ramon syndrome, and fragile X syndrome. The mutation of the SH3BP2 gene,will result in increased production of over active proteins from this gene. The SH3BP2 gene is found on the smaller arm of chromosome 4 at position 16.3. The SH3BP2 protein is involved with chemical signaling to immune system cells known as macrophages and B cells. The effects of SH3BP2 mutations are still under study, but researchers believe that the abnormal protein disrupts critical signaling pathways in cells associated with the maintenance of bone tissue and in some immune system cells. The overactive protein likely causes inflammation in the jaw bones and triggers the production of osteoclasts, which are cells that break down bone tissue during bone remodeling. Osteoclasts also sense the increased inflammation of the mandible and maxilla and are further activated to break down bone structures. Bone loss and inflammation lead to increased fibrous tissue and cyst growth. An excess of these bone-eating cells contributes to the destruction of bone in the upper and lower jaws. A combination of bone loss and inflammation likely underlies the cyst-like growths characteristic of cherubism[7].

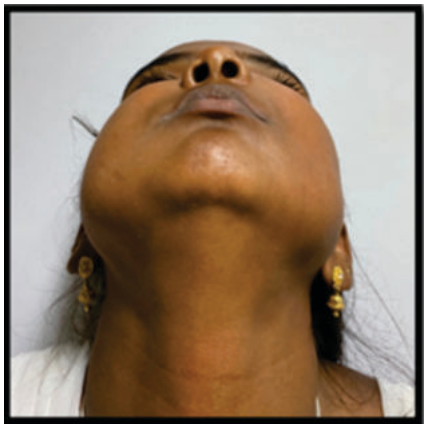
INVESTIGATIONS
Serum levels of calcium, parathyroid hormone (PTH), parathyroid hormone related peptide (PTHrP), calcitonin and alkaline phosphatase (ALP) are typically within normal range. Serum levels for alkaline phosphate may be increased during the active stages of cherubism[8].

HISTOLOGY
Cherubism lesions resemble giant cell tumors because they contain many giant-cells and mononuclear or stromal cells. The fibrotic lesions are non-neoplastic. Cherubism cannot be diagnosed by histology alone because they are not distinguishable from other giant cell lesions of bone[9].

TREATMENT
Cherubism changes and improves over time. Generally, moderate cases are watched until they subside or progress into the more severe range. Severe cases may require surgery to eliminate bulk cysts and fibrous growth of the maxilla and mandible. Partial resection, contour resection, curettage or a combination of these. Orthodontic treatment may be used to erupt permanent teeth that have been unable to descend due to lesions and cysts being in their path of eruption. Patients with orbital issues of diplopia, eye proptosis, and visual loss will require ophthalmologic treatment. Radiation therapy, interferon therapy and calcitonin are the other treatment options[10].

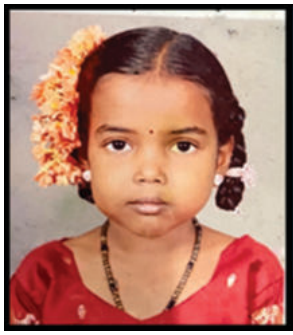
PROGNOSIS
In general, cherubism does not have a poor prognosis. It has been noted that the condition does not progress beyond puberty. As the patient grows to adulthood, the jawbone lesions tend to resolve, and a progressively more normal jaw configuration is noted[11].

PROFILE (FIGURE-1)



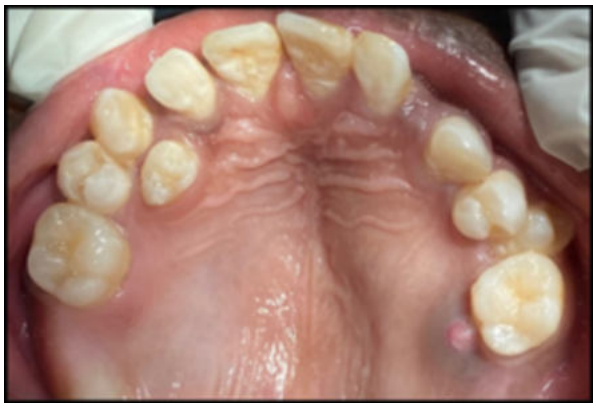
WORM'S EYE VIEW (FIGURE-2)

PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr

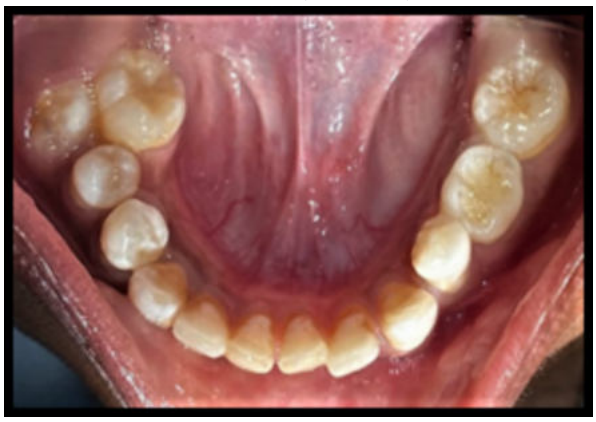


Patient At 3yrs Of Age (Figure-3)

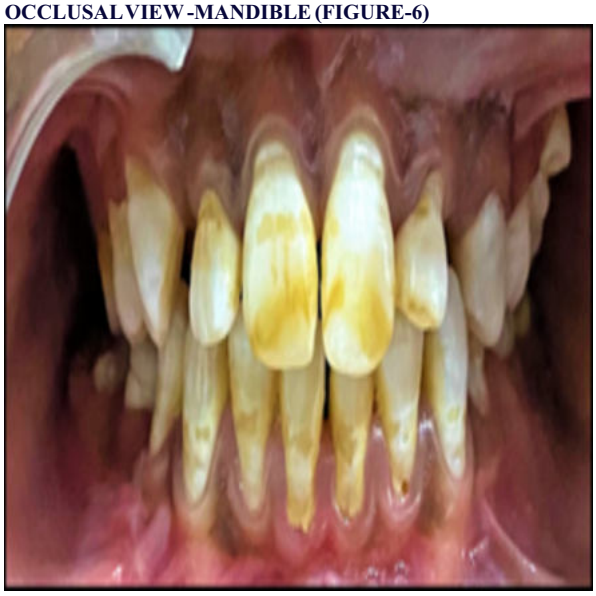
Patient's Father (Figure-4)



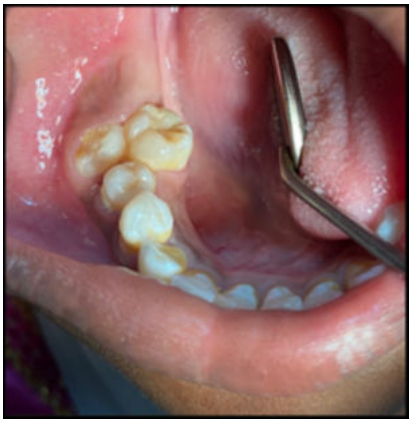
OCCLUSAL VIEW -MAXILLA (FIGURE-5)



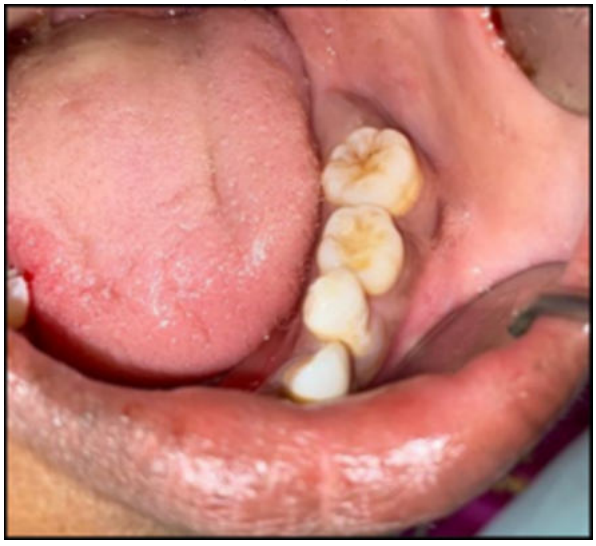
OCCLUSAL VIEW -MANDIBLE (FIGURE-6)



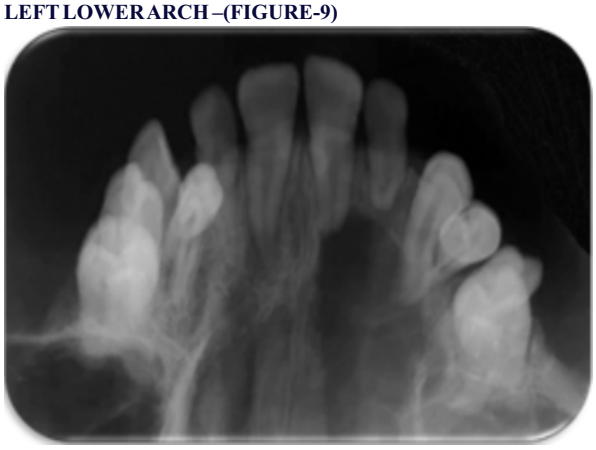
INTRA ORAL (FIGURE-7)



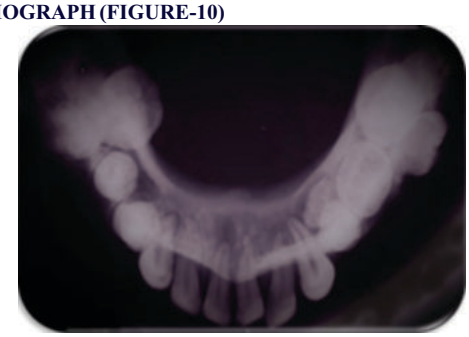
RIGHT LOWER ARCH –(FIGURE-8)



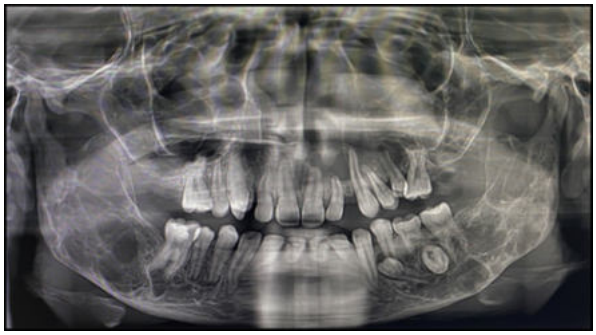
LEFT LOWER ARCH –(FIGURE-9)



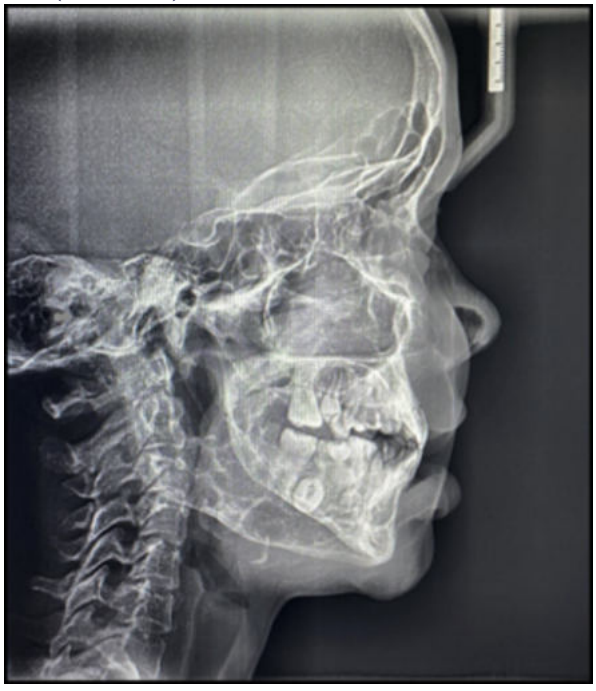
MAXILLARY OCCLUSAL -CROSS SECTIONAL RADIOGRAPH (FIGURE-10)



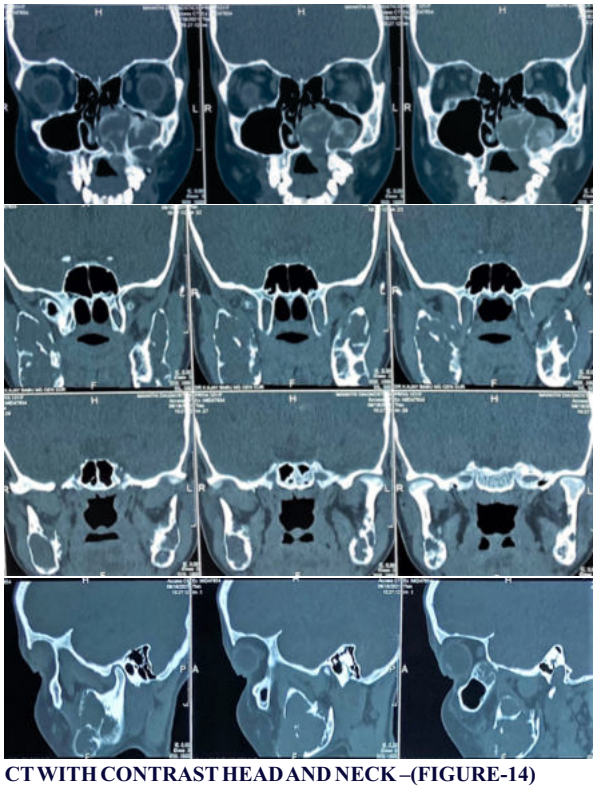
MANDIBULAR OCCLUSAL -CROSS SECTIONAL RADIOGRAPH (FIGURE-11)



OPG (FIGURE-12)



LATERAL CEPHALOGRAM (FIGURE-13)



CT WITH CONTRAST HEAD AND NECK –(FIGURE-14)

REFERENCES

1. Eversole R, Su L, ElMofty S. Benign fibro-osseous lesions of the craniofacial complex a review. Head and neck pathology. 2008 Sep 1;2(3):177-202.
2. Nelson BL, Phillips BJ. Benign fibro-osseous lesions of the head and neck. Head and neck pathology. 2019 Sep;13(3):466-75.
3. Crusoé-Rebello I, Torres MG, Burgos V, Oliveira C, Santos JD, Azevedo RA, Campos PS. Hybrid lesion: central giant cell granuloma and benign fibro-osseous lesion. Dentomaxillofacial Radiology. 2009 Sep;38(6):421-5.
4. Abramovitch K, Rice DD. Benign fibro-osseous lesions of the jaws. Dental Clinics. 2016 Jan 1;60(1):167-93.
5. Mainville GN, Turgeon DP, Kauzman A. Diagnosis and management of benign fibro-osseous lesions of the jaws: a current review for the dental clinician. Oral diseases. 2017 May;23(4):440-50.
6. McCarthy EF. Fibro-osseous lesions of the maxillofacial bones. Head and neck pathology. 2013 Mar 1;7(1):5-10.
7. Argyris PP, Gopalakrishnan R, Hu Y, Reichenberger EJ, Koutlas IG. Clinicopathologic and molecular characteristics of familial cherubism with associated odontogenic tumorous proliferations. Head and neck pathology. 2018 Mar;12(1):136-44.
8. Holley TJ, Giannini PJ, Narayana N, Desa VP. Early detection of cherubism with eventual bilateral progression: a literature review and case report. Oral surgery, oral medicine, oral pathology and oral radiology. 2019 Mar 1;127(3):e77-83.
9. Beaman FD, Bancroft LW, Peterson JJ, Kransdorf MJ, Murphey MD, Menke DM. Imaging characteristics of cherubism. American Journal of Roentgenology. 2004 Apr;182(4):1051-4.
10. Singh A, Sharma A, Chandra N. Non-familial cherubism. Contemporary clinical dentistry. 2011 Apr;2(2):131.
11. Niranjan B, Shashikiran ND, Singla S, Kasetty S. Non-hereditary cherubism. Journal of oral and maxillofacial pathology: JOMFP. 2014 Jan;18(1):84.