CASE REPORT

Cone beam computed tomography features of cherubism: A case report
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Abstract
Cherubism is an uncommon benign hereditary disease of autosomal dominant inheritance, affecting the maxilla and mandible. This condition presents with bilateral enlargement of jaws, where bone is replaced by fibrovascular tissue. This condition is usually painless and presents in children during 2–5 years of age. The clinician should be aware of clinical and radiographic features of this condition for a better differential diagnosis, as it mimics the other giant cell lesions radiologically. The current case report elucidates cherubism in a 15-year-old male patient, presented with bilateral mandibular enlargement, highlighting the cone beam computed tomography features.

Key words: Cherubism, cone beam computed tomography, diagnosis

Introduction
Jones in 1933, first described cherubism which is characterized by a marked fullness of the cheeks and occasional turning of eyes in an upward direction giving the child an angelic appearance.[1] World Health Organization has classified, cherubism under a group of nonneoplastic bony lesions which affects maxilla and mandible.[2] According to Anderson and McCleden cherubism displays a autosomal dominant inheritance pattern with 100% penetrance in males and up to 70% penetrance in females.[3] Although sporadic cases with no familial history have been described.[4] Mangion and Tiziani et al. described that mutation of the gene located on chromosome 4p16.3 is responsible for cherubism, also a point mutation in SH3BP2 gene was detected.[5]

Characteristic features of cherubism are painless swelling of the jaw bones with proportioned involvement of the maxilla and mandible. The lesion start regressing after puberty and display spontaneous involution in adulthood.[6] Radiographically, it presents as multiple radiolucencies with a ground-glass pattern in the jaws, resembling other giant cell lesions. As there is spontaneous involution of the lesion, active treatment of cherubism is usually not required, albeit, limited surgery can be performed for improvement in esthetics or function.[6]

Case Report
A 15-year-old male child, reported to the outpatient department with the chief complaint of painless swelling of the lower jaw since 10 years. The child was normal at birth; the swelling started around 5 years of age, and gradually increased in size. There was no significant past medical history.

On examination bilateral swellings were noted over the mandibular body and ramus region, giving a chubby facial appearance, overlying skin was normal in color and texture [Figure 1]. Palpation revealed firm nontender swellings without any raise in local temperature. Bilateral nontender enlargement of submandibular lymph nodes was noted.

Intraorally there was crowding seen in maxillary arch, and missing 43, 37, 38, 47, and 48 were noted, the intraoral soft tissues appeared normal.

Orthopantomogram (OPG) revealed developed 11, 12, 13, 14, 15, 16, 17, 21, 22, 23, 24, 25, 26, 27 and developing 18 and 28 in maxillary arch. Developed 31, 32, 33, 34, 35, 36, 37, 41, 42, 43, 44, 45, 46, 47 and missing 38 and 48 were noted in mandibular arch. Impaction was noted with 28, 37, 43, and 47. There was a multilocular radiolucency noted in mandible extending from canine to sigmoid notch bilaterally. There was thinning of lower border of mandible noted in region involved by radiolucency [Figure 2].

Cone beam computed tomography (CBCT) findings
CBCT showed a radiolucent multilocular expansile lesion involving the mandible, extending from parasympyseal region to sigmoid notch bilaterally sparing the condyles. The periphery of lesion appeared to be irregular and the internal of lesion showed radiopaque septas in between. Expansion and thinning of buccal and lingual cortical plates were noted, expansion was more prominent on lingual aspect. Perforation...
of buccal and lingual cortices was also noted at some regions. Root resorption is noted with 36 and 46. Impaction is noted with 37, 43, and 47.

There is another radiolucent lesion noted in left maxilla, posteriorly, extending from 26 to the posterior wall of maxillary sinus. Mild expansion of buccal and palatal cortex is noted in this region. There is displacement of floor of maxillary sinus in upward direction with partial opacification of maxillary sinus [Figures 3-5]. Root resorption is noted with 26 and 27, impaction is noted with 28.

The lesion on maxillary left side was not apparent on two-dimensional (2D) imaging OPG, which became clearly evident on three-dimensional (3D) imaging CBCT.

Discussion
Cherubism is an uncommon fibro-osseous disease of childhood characterized by degradation of bone which is replaced by fibrous tissue, and it results in prominence of the cheeks. The disease has also been referred as, bilateral giant cell tumor and familial or hereditary fibrous dysplasia.[7]

The pathogenesis of this disease is controversial till date, its relationship with trauma, infection, and hemorrhage has never been confirmed.[8] Cherubism has been associated with some syndromes, which includes Multiple giant cell lesion syndrome, Neurofibromatosis Type 1, and Ramon syndrome.[9]

Children affected with cherubism, are normal at birth, its manifestations start during first 5 years, progresses slowly, and stops around 12–15 years of age. At puberty, the bony lesions of cherubism starts retrogressing spontaneously.[7] Jaw lesions

Figure 1: Extraorally bilateral swelling noted over mandibular body and ramus

Figure 2: Orthopantamogram showing bilateral multilocular lesion involving the body and ramus of the maxilla

Figure 3: Axial section of cone beam computed tomography showing bilateral multilocular swelling, causing expansion and perforation of buccal and lingual cortices

Figure 4: Sagittal section of cone beam computed tomography showing lesions of maxilla and mandible with resorption of teeth on left side
of cherubism are generally painless and symmetrical and do not involve other parts of the skeleton.\(^6\) Maxillary involvement is less common as compared to mandible and is characterized by opacification of the inferior part of the maxillary sinus, with anterior displacement of dental follicles. Although cherubism is a self-limiting entity which abates with age, it occasionally can cause severe orbital abnormalities.\(^10\)

Our patient showed displacement of teeth, impacted teeth and root resorption, which are classic radiographic findings of cherubism.\(^6\)

A grading system was proposed by Ramon and Engelberg for cherubism, which is as follows:\(^7\)

- Grade 1: Involvement of ramus of mandible bilaterally.
- Grade 2: Grade 1 along with diffuse mandibular involvement and bilateral involvement of maxillary tuberosities.
- Grade 3: Massive involvement of the maxilla and mandible sparing the condyles.
- Grade 4: Grade 3 and in addition involvement of orbital floor resulting in orbital compression.

In the current case, the grade of cherubism was in between 1 and 2 as there was diffuse involvement of mandible sparing the condyles and involvement of posterior maxilla on the left side.

According to Marck and Kudryk conventional radiography can only provide a limited detail as it is 2D. On the other hand, 3D imaging like CBCT, can provide an exact extent of the lesions, which otherwise would not be apparent due to anatomic complexity of the jaws and superimposition in 2D images. Furthermore, the 3D formatting feature helps in the diagnosis [Figure 6].\(^{11}\)

For our patient the OPG was taken as conventional radiograph, and CBCT was taken as advanced imaging technique. Expansion, thinning, and perforation of cortical plates were noted in images bilaterally. The conventional and advanced CBCT radiographic findings of the current case were consistent with the reports in literature.\(^6,12\)

As cherubism is a self-limiting condition, treatment is indicated when function and esthetics are impaired. According to some authors calcitonin therapy may limit the disease and preclude surgery.\(^{11}\) Cherubism generally have a good prognosis as the condition starts regressing at puberty, with resolution of bony lesions resulting in a normal jaw configuration.\(^4\)

Familial gigantiform cementoma, Brown tumor of hyperparathyroidism, and Fibrous dysplasia shows similar features radiographically, thus are included in differential diagnosis of cherubism.

**Conclusion**

Cherubism is an entity which presents with an appearance of a cherub like face, awareness regarding this disease entity is important and it must be considered in the differential diagnosis of children and young adults reporting with bilateral mandibular swelling.

Radiographic evaluation is must as clinical presentation, and distribution of the lesions may define the diagnosis. CBCT plays a very important role in lesion assessment as it provides detailed information regarding the involvement of the jaw bones, with a precise spectrum of the lesion. Once diagnosis is validated, frequent follow-ups should be scheduled for the patient and surgery, if required should be postponed till puberty so there will be less chances of recurrence with a better prognosis.

Oral and maxillofacial radiologist plays an important role in diagnosis of this disease, so a radiologist must be thorough with the knowledge of the clinical and radiographic changes observed in patients with cherubism.

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**Figure 5:** Coronal section of cone beam computed tomography showing lesions in mandible bilaterally and maxilla on left side.

**Figure 6:** Three dimensional reconstruction.
References


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