

# Cherubism: A rare case report

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## Abstract

Cherubism is a rare congenital disease resulting in malformation of the jaw. It occurs before the age of 5 years and regress spontaneously after puberty. It can result into enlargement of the jaw bone, tooth displacement, facial disfigurement and psychological trauma to patient. Hence, the understanding about the condition, its progression and management is necessary.

**Key words:** Cherubism, childhood disease, giant cell lesion, jaw growth, malocclusion

## INTRODUCTION

Cherubism (MIM ID 118400) is a rare childhood condition presents before the age of 5 years and regress spontaneously after puberty.<sup>[1,2]</sup> Until now, around 300 cases of Cherubism have been reported.<sup>[1]</sup> This inherited autosomal dominant disorder affecting jaw characterized by replacement of normal bone by proliferation of fibrovascular tissue containing multinucleated giant cells. It was first described by Willium Jones in 1933.<sup>[2]</sup> Perivascular fibrosis leading to mesenchymal disorder and decreased oxygenation is widely accepted theory for its pathogenesis.<sup>[3]</sup> This condition is also known as familial fibrous dysplasia of jaws, familial multilocular disease of the jaws, multilocular cystic disease of the jaw.<sup>[1,4,5]</sup> Most of the cases showed familial history, but some cases presented without familial histories of disorder.<sup>[2]</sup>

## CASE REPORT

An 8-year-old girl patient presented with slowly growing painless, bilaterally symmetrical swelling of the jaw [Figure 1]. Enlargement involved whole jaw. Enlarged jaw was hard to palpate. Family history was not significant. Mouth opening was adequate with “V” shaped palate. Submandibular lymph nodes were bilaterally palpable. Skin over the swelling was normal, intact and freely movable. Teeth were displaced resulting in to malocclusion and anterior cross bite. There was deep carious lesions present with primary mandibular second molars and right primary first molar showed internal resorption,

her oral hygiene was poor [Figures 2 and 3] There was an expansion of both buccal and lingual cortical plates, with normal overlying mucosa in mandible but maxilla was unaffected [Figures 2 and 4]. Orthopantomograph and anteroposterior view of skull radiograph revealed bilateral, multilocular, radiolucent lesion with thinning of cortical plates and displacement of unerupted teeth in mandible [Figures 5 and 6]. Hematological investigations were within the normal limits. On the basis of clinical and radiographic examination, the diagnosis of Cherubism was made. Restoration of carious teeth and professional oral prophylaxis was done. Since the disorder is spontaneously regress after puberty hence no surgical intervention was undertaken and follow-up evaluation was advised with oral hygiene instructions.

## DISCUSSION

Cherubism is a childhood disorder affecting male predominantly. Here, we have reported a case of Cherubism in a girl, which is uncommon. According to World Health Organization classification, Cherubism belongs to the non-neoplastic bone lesions group that involves the mandible.<sup>[3]</sup> The prevalence in male is 100% when compared with female 50-70% i.e., 2:1 ratio.<sup>[2,4]</sup> It has been identified that Chromosome 4p16.3, between D4S127 and 4p-telomere and named SH3BP2 (SH3-binding protein) is responsible for Cherubism.<sup>[6,7]</sup> This disorder characteristically presents as gradually enlarging painless bilateral swelling of mandible and sometimes affecting maxilla, it causes rounding of the face and swollen cheeks accompanied by upward-looking



**Figure 1:** Photograph of patient showing frontal view of face with symmetrical bilateral mandibular swelling



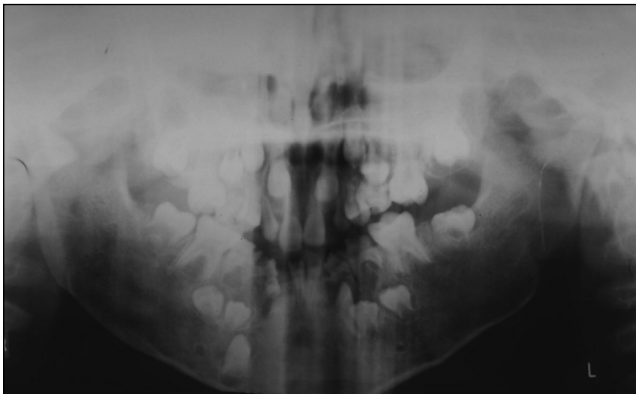
**Figure 2:** Bilateral mandibular swelling with normal intact mucosal, teeth malposition, carious mandibular primary second molars



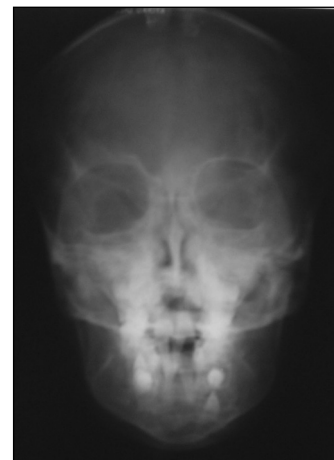
**Figure 3:** Photograph showing anterior cross bite and labially placed canines



**Figure 4:** Photograph showing normal maxilla



**Figure 5:** Orthopantomagraph showing bilateral multilocular lesions



**Figure 6:** Anteroposterior view of radiograph showing bilateral multilocular lesions in mandible

of eyes. This condition gives the patient the appearance of “cherubs” depicted in baroque artwork, hence the name Cherubism.<sup>[2,4]</sup> Even though the condition is bilateral, unilateral involvement has been documented.<sup>[5]</sup> The first radiographic sign of Cherubism are commonly found in the region of the mandibular angle.<sup>[1]</sup> Mandible is primarily affected than maxilla (60%). It has been observed that

children will be normal at birth, but at around 14 months to 5 years of age, a symmetric enlargement of the jaw begins to until puberty and then regress spontaneously.<sup>[7]</sup> Silva *et al.* reported an unusual extreme case of Cherubism, where it showed aberrant symmetrical orofacial swelling, eyes being pushed upward and appreciable weight loss in 9

months of observation.<sup>[8]</sup> Perforation of the cortex noted in some cases.<sup>[9]</sup>

Dental abnormalities associated with this disorder can be tooth malposition, root resorption, abnormal shaped teeth and impacted teeth, loss of teeth, ectopic eruption and agenesis of permanent teeth mainly second and third molars due to involution of their germs. These abnormalities result in malocclusion, problems of phonation and swallowing. These abnormalities are due to substitution of bone into a fibrous tissue, resulting in oclusopathologies.<sup>[1,2,4,7]</sup> Constant finding in these cases is enlargement of submandibular and cervical lymph nodes.<sup>[4]</sup>

In general, Cherubism is limited to craniofacial region; however, there is reported case of extra cranial involvement affecting the ribs.<sup>[1]</sup> Respiratory problems are usually absent in these patients, but occasionally manifest as upper airway obstruction caused by backward displacement of tongue or obliteration of nasal airway.<sup>[1]</sup>

Radiographically, it is characterized by expansive radiolucent, multilocular lesions clearly delineated by cortical bone and distributed bilaterally in the posterior quadrants of the mandible and or maxilla. Bone alteration generally starts in the region of angle and ascending ramus of the mandible, involvement of condyle is rare, teeth shows floating tooth appearance. In maxilla, it starts in maxillary tuberosity region. In more severe case, infiltration of the orbital cavities may cause exophthalmia and limited eye movements. Facial sinus generally appears obliterated.<sup>[4]</sup>

Based on area of involvement Ramon and Engelberg have proposed four gradings:

- Grade I : Bilateral involvement of the ascending ramus of mandible.<sup>[3]</sup>
- Grade II : Bilateral involvement of the ascending ramus of mandible and maxillary tubercity
- Grade III: Massive involvement of the maxilla and mandibles except for the condylar process,
- Grade IV: Grade III plus involvement of the floor of the orbits, causing orbital compression.

Cherubism grading system according to Motamedi (1998) and Raposo-Amaral (2007).<sup>[1]</sup>

- Grade I: Lesions of the mandible without signs of root resorption
  - Class 1 solitary lesion of the mandibular body
  - Class 2 multiple lesions of the mandibular body
  - Class 3 solitary lesion of the ramus
  - Class 4 multiple lesions of the rami
  - Class 5 lesions involving the mandibular body and rami.

- Grade II: Lesions involving the mandible and maxilla without signs of root resorption
  - Class 1 lesions involving the mandible and maxillary tuberosities
  - Class 2 lesions Involving the mandible and anterior maxilla
  - Class 3 lesions involving the mandible and entire maxilla.
- Grade III: Aggressive lesions of the mandible with signs of root resorption
  - Class 1 solitary lesion of the mandibular body
  - Class 2 multiple lesions of the mandibular body
  - Class 3 solitary lesion of the ramus
  - Class 4 multiple lesions of the mandibular rami
  - Class 5 lesions involving the mandibular body and rami.
- Grade IV: Lesions involving the mandible and maxilla and showing signs of root resorption
  - Class 1 lesions involving the mandible and maxillary tuberosity
  - Class 2 Lesions involving the mandible and anterior maxilla
  - Class 3 lesions involving the mandible and entire maxilla
- 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, massively growing, aggressive and extensively deforming

Juvenile lesions involving the maxilla, mandible and orbits.

Syndromes associated with Cherubism are: neurofibromatosis type-1, Noonan-like/multiple giant cell lesion syndrome, Ramon syndrome and Jaffe-Campanacci syndrome.<sup>[3]</sup>

Differential diagnosis includes, craniofacial variety of fibrous dysplasia (McCune — Albright syndrome), central giant cell reparative granuloma, brown tumors, true giant cell tumor, infantile hyperostosis, familial gigantism cementoma, ameloblastoma, myxoma,<sup>[3,5]</sup> lesions containing giant cells such as hyperparathyroidism and osteomalasia.<sup>[4]</sup>

Biochemical investigations can show normal limits of serum calcium and phosphorus concentrations, thyroid stimulating hormone, follicle stimulating hormone, luteinizing hormone, T4 and T3 levels but elevated levels of alkaline phoshatase.<sup>[4]</sup> Histologically, the lesion presents with numerous multinucleated giant cells scattered throughout a fibrous connective tissue.<sup>[2]</sup>

Diagnosis of the condition can be based on history, clinical examination, radiographic (orthopantomographs, posteroanterior radiographs, telerradiograph), computed tomography (helps to check disease progression, provides a realistic picture), histological and biochemical investigations.<sup>[4,9]</sup> Prenatal identification of Cherubism can be done by deoxyribonucleic acid analysis from extract of fetal cells obtained by amniocentesis, which can be performed at about 15-18 weeks of gestation or chorionic villus sampling at about 10-12 weeks of gestation.<sup>[10]</sup>

### Management

Since, this condition is regress spontaneously after puberty, wait and observe policy can be applied. After regression of disease, tooth extraction in areas showing fibrous alterations (floating tooth), cosmetic osteoplasty of the affected jaws or physiological bone remodeling can be done. In case of functional impairment, curettage of the lesion and treatment with calcitonin can be advised. Dental prosthesis may be needed to establish proper chewing. Orthodontic treatment is appropriate after growth is complete and after the regression of condition.<sup>[11]</sup> Psychological counseling for facial disfigurement can be advised. In extreme cases with functional impairment surgical intervention should be performed at the earliest. Radiotherapy has been suggested because of the potential risk of osteoradionecrosis or malignant transformation.<sup>[4]</sup>

### CONCLUSION

Cherubism is a rare autosomal dominant disorder resulting in enlargement of jaws. It occurs before age of 5 years and regress spontaneously after puberty. It can presents with several dental abnormalities. Hence, it is necessary for dentist to know about the condition.

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