

Non-Familial Cherubism(A Case Report)

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Abstract: Cherubism, a rare non-neoplastic autosomal dominant genetic defect of bone remodelling which predominantly affects the mandible alone or both the mandible and the maxilla, giving a characteristic cherubic appearance to the patient. On radiography, the lesions exhibit bilateral multilocular radiolucent areas. Histopathology reveals multinucleated giant cells in the background of proliferating fibrous connective tissue. Cherubism is a self-limiting condition and usually regresses itself after puberty. The present case report describes cherubism in a 10 year old male child and briefly reviews literature on this report.

Keywords: Cherubism, Mandible, Bilateral, Follow-up.

I. Introduction

Cherubism, a rare autosomal dominant defect affecting mandible or/and maxillary bone remodelling, was first described by William A. Jones in 1933. He described this condition in three Jewish siblings as ‘familial multilocular cystic disease of the jaws. The term ‘cherubism’ is derived from the word ‘cherub’ which means angels with childish full cheeked face often gazing upwards as if ‘eyes to heaven’ as often depicted in Renaissance art. Other synonyms include familial/hereditary fibrous dysplasia, familial multilocular cystic disease of the jaws and familial multiple giant cell lesions of the jaws. To the best of our knowledge only 300 cases from available studies have been reported in the English literature.

Cherubism is defined by the appearance of painless, symmetrical, bilateral jaw swellings that typically first appear at the age of 2 to 7 years. Swelling of submandibular lymph nodes in the early stages contributes to the fullness of the face. As the soft fibrous dysplastic tissue in the lesions expands the protuberant masses can infiltrate the orbital floor and cause the characteristic upward tilting of the eyes, exposing the sclera below the iris. Cherubism lesions are limited to the jaws and in most cases the dysplastic expansile masses begin to regress with the onset of puberty.

We present a case of this rare disease occurring in a 10-year-old child.

II. Case Report

A 10 years old male child presented by his father to our department with the chief complaint of painless bilateral swelling of the cheeks. History revealed that the parents of the child observed the swellings on both the sides of the lower jaw of their child at the age of 5 years that progressively increased to the present size. The child did not have any contributory past medical and dental history. There was no history of such swelling in other siblings or relatives and patient had no other systemic illness. An extraoral clinical examination revealed chubby cheeks and bilateral asymmetry with a solitary swelling present over the left lower-third of face (Fig.1)The swelling was non-tender, bony hard, well-defined & fixed to the underlying structures. A similar kind of swelling was present on the contralateral side with no tenderness and was bony hard in consistency. Submandibular lymph nodes were bilaterally palpable, enlarged and non-tender. Intraorally, a solitary diffuse swelling was seen in the buccal, labial vestibule region in relation to 75. The swelling extends buccally in the mucosa measuring around 3cm in size and extending antero-posteriorly from 73 to 75 region. The color of the mucosa over the swelling was normal with unerupted 75 (Fig. 2a-2b). The swelling was non-tender, bony hard in consistency and well defined. A similar kind of swelling was seen in relation to 85 which was non-tender, and bony hard in consistency. Blood investigations (Complete blood count, serum calcium, serum phosphorus), Thyroid profile, Parathyroid Hormone (PTH) were under normal range. However, serum alkaline phosphatase was slightly elevated. An orthopantomograph (Fig.3) showed bilateral multilocular radiolucency having ‘soap-bubble appearance’ and extending from the molar area and involving the entire ramus of the mandible sparing the condyles along with numerous unerupted and displaced teeth giving ‘Floating tooth’ appearance. The posterior-anterior skull projection revealed multilocularexpansile lesions present bilaterally in the mandible (Fig.4). The mandibular occlusal radiographs revealed expansion of buccal and lingual cortical plates (Fig.5)

The lesion was diagnosed as Familial/Hereditary fibrous dysplasia (Cherubism). According to the grading system proposed by Seward and Hankey in 1957, it was classified as Grade 1. Since it is a self-regressing condition, no treatment was done and the parents of the patient were counseled for the regular follow-up every 6 month to monitor the condition.

III. Discussion

Cherubism or multilocular cystic disease of the jaws was first described as a separate entity in 1933 by William A. Jones in a family with several affected members. He designated the pictorial name “cherubism” because “the full round cheeks and the upward cast of the eyes give the children a peculiarly” cherubic appearance. Because this name so accurately captured the clinical features of the disease, it became the standard nomenclature.² Cherubism, a rare bony dysplasia may be hereditary or sporadic in 60 and 40% of cases respectively.¹ The case reported in this paper may be one of the sporadic types as no previous occurrence was found in the patient’s siblings or relatives. Cherubism is defined by the appearance of symmetrical, multilocular, expansile radiolucent lesions of the mandible and/or the maxilla that typically first appear at the age of 2 to 7 years. The age of onset of swelling in our patient was consistent with those reported in literature. Mengand Yu et al however, outlined 6–10 years as age of onset in a review of 24 cases. Swelling of submandibular lymph nodes in the early stages contributes to the fullness of the face. As the soft fibrous dysplastic tissue in the lesions expands the protuberant masses can infiltrate the orbital floor and cause the characteristic upward tilting of the eyes, exposing the sclera below the iris.² Cherubism lesions are limited to the jaws and in most cases the dysplastic expansile masses begin to regress with the onset of puberty. The familial form of cherubism occurs typically in an autosomal dominant trait with mutations in the SH3- domain binding protein 2 (SH3BP2) on chromosome 4p16.⁴ Cherubism is reported to be associated with Noonan syndrome, Fragile X Syndrome, Gingival Fibromatosis With Psychomotor Retardation, Neurofibromatosis Type 1, Craniosynostosis, Ramon syndrome, Jaffe- Campanacci syndrome, sleep apnea syndrome.^{1,3,5,7} Mineral metabolism has been observed to be normal in patients with cherubism. Serum levels of calcium, phosphorus and alkaline phosphatase are typically within normal range. Haematological and blood chemistry investigations carried out in our patient was consistent with reports in the literature.

Radiographically, cherubism is characterized by bilateral, multilocular, radiolucent lesions of the mandible and/or maxilla. Bone alterations generally start in the angle and ascending ramus of the mandible. Maxillary involvement is less frequent and less extensive. Sparing of mandibular condyles was earlier considered a hallmark of this condition; however condylar involvement has also been reported.⁵ The destruction of the alveolar cavity may displace the teeth, producing a radiographic appearance referred to as ‘floating tooth syndrome’.¹¹ In terms of classification, Seward and Hankey in 1957 categorized cherubism into four grades based on the severity and location of the disease as follows:

- Grade I- Bilateral involvement of mandible body, and ramus sparing the condyles.
- Grade II- Bilateral involvement of maxillary tuberosity in addition to grade 1.
- Grade III- Massive involvement of the entire maxilla and mandible except the condyles.
- Grade IV- Involvement of both jaws plus the condyles.

According to this classical grading system our patient belongs to Grade 1 cherubism in which there is bilateral involvement of mandible body, and ramus sparing the condyles. Histologically, eosinophilic cuffing appears to be specific for cherubism. However, these deposits are not present in many cases and their absence does not exclude the diagnosis of cherubism. Older, resolving lesions of cherubism show an increase in fibrous tissue, a decrease in the number of giant cells and formation of new bone.¹¹

Cherubism, as with other genetic disease is not curable. However, it is a self-limiting condition that regress spontaneously after puberty.⁵ Follow-up every 2 to 5 years is advisable after the disease becomes quiescent. Surgical intervention is indicated when aesthetic or functional concerns arise including nasal obstruction, proptosis or facial deformity. Liposuction has been proposed to reduce the mass of the lesion in particular cases. Curettage alone or in combination with surgical contouring for cosmetic purposes has been considered the treatment of choice.⁷

IV. Conclusion

Cherubism is a rare osseous disorder of children and adolescents. Although the radiologic characteristics are not pathognomic, the diagnosis is strongly suggested by the bilateral, symmetric involvement of mandible and/or maxilla. We have reported cherubism a very rare incurable genetic defect in a 10-year old boy who is still being followed up in our department as treatment is based on natural and clinical course of the disease.

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Figure 1. Characteristic appearance of cherubic patient



Figure 2(a) intraoral examination of teeth2(b) a diffuse swelling is seen in the buccal, labial vestibule region with unerupted 75



Figure 3. Orthopantomogram showing bilateral multilocular radiolucencies in the mandible. The condyle is characteristically spared bilaterally.



Figure 4. The posterior-anterior projection of skull shows multilocular lesions present bilaterally in the mandible along with multiple unerupted teeth.



Figure 5(a) (b). The mandibular occlusal radiographs revealed expansion of the cortical plates: (a) buccal and lingual cortical plate expansion in relation to left side of mandible; (b) buccal cortical plate expansion in relation to right side of mandible.