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Review Article

UNREVEALING THE MYSTERIES OF CHERUBISM- A CASE REPORT WITH BRIEF REVIEW

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ABSTRACT

Background: Cherubism is a benign fibro-osseous lesion involving jaw bones predominantly mandible. It appears to be a hereditary disease with an autosomal dominant pattern of inheritance. The lesion starts in childhood gradual in onset involving both sides causing asymmetry of facial profile. It regress spontaneously at the onset of puberty.

Aim: Revealing the unrevealed about cherubism: a fibro-osseous lesion affecting mainly jaw bones.

Case: we report a case of cherubism seen in the post pubertal age. We report a case for which panoramic and occlusal radiographs were taken, which are sufficient for diagnosis of this case. Clinical and radiological findings were discussed.

Conclusion: A rare disease but affected individuals have a significant impact on the health and general wellbeing. Affected bone is predominantly mandible causing facial asymmetry. Genetic test are used to diagnose the case. Surgical intervention, orthodontic treatment along with cosmetic /plastic surgery are a common line of management. At the higher end genetic therapy can also be regarded. To conclude we report a case of cherubism seen in a postpubertal individual with classic clinical features, sequence of progression, and radiographic features subtle to the diseases.

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INTRODUCTION

Fibro-osseous lesions are a group of genetically designated non neoplastic benign lesions of craniofacial and other skeletal system in the body. Cherubism is a hereditary form of fibrous dysplasia with a causative factor as transmission of autosomal dominant SH3BP2 gene mutation. An autosomal dominant disorder related to chromosome 4p16 that may be related to tooth development and eruption. The word cherubism means looking like angel, which resembles spherical facial appearance of angels painted in the Renaissance era.

This was first described in 1933 by Jones. WHO classified them into non- neoplastic bony lesions affecting only the jaws, bilaterally with age range of 2-7yrs. The lesion is characterized by immature bone formation and replacement of this bone with fibrous tissue. This is known by different names like familial or hereditary fibrous dysplasia, bilateral giant cell tumor and familial multilocular disease. With a more predilection for males rather than females. Starts mainly in childhood age, stay inactive for some time then progress slowly but regress after puberty without treatment.^{1,2,3,4,5,6} sporadic cases

are also reported due to mutation of the gene encoding for fibroblast growth factor receptor III (FGF-RIII)⁷. At molecular level the pathogenesis of cherubism has been described as due to a mutation in the gene encoding SH3 - binding protein 2 (SH3BP2) and possible degradation of the Msx-1 gene which is involved in the regulation of mesenchymal interaction during craniofacial morphogenesis⁸. Clinically the lesion appears as single bilateral painless swelling of spherical and symmetrical involving bilaterally the mandible predominantly, resulting in a fullness of the cheeks and retraction of the lower eyelids (maxilla), giving an upward turned appearance of the eyes-comparable to a cherub angel. Clinically it can also interfere with normal jaw function, speech and vision^{6,9}. Radiographically cherubism is characterized by bilateral, multilocular, radiolucent areas within the jawbones. The lesions usually appear around the mandibular angle and spread to the ascending rami and body of mandible⁷.

The syndromes associated are like Noonan's syndrome, Jaffe syndrome, Gardner syndrome and Ollier's disease. The routine radiograph can reveal the extent of bone involvement and the amount of bone destruction occurred⁹.

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The present case report describes this rare occurrence in a postpubertal girl.

A 18 yr. old female patient reported to the department of oral Medicine and Radiology with a complaint of bilateral swelling of face since 8 yrs.(fig 1) The patient reported that before the age of 5 she had normal facial features.



Profile Photo

As she grew she noticed slight variation of facial features, by the age 6yrs she noticed a facial asymmetry. By the age of 7 she noticed swelling on both sides of face. The swelling gradually increased in size. The parents took her to a paediatrician who after routine blood investigation put her on different medication. Since there was no regression of swelling they approached another paediatrician, who also gave medications. However the swelling kept gradually increasing and the asymmetry of face altered when compared with other children in locality. Even after six months to one year of medical consultation the swelling has not regressed and the patient stopped taking medication. They also visited a dentist in concern with the swelling and had undergone clinical and radiographic examination, but was referred to higher centers. The family history revealed none of her parents or grand parents or siblings had a history of such swelling on the face during their childhood. (fig1). General examination revealed no positive findings other than facial asymmetry. On extraoral examination the patient had bilateral nontender bony hard swelling involving the middle third of face, involving mainly the lower jaw. The swelling had an ill-defined boundary. The skin over the swelling was not stretched and no colour change. There was no discharge or visible pulsations. There were no other positive findings with respect to extraoral examination.

Intraoral examination revealed bilateral enlargement of mandibular bone, on both sides, the vestibular sulcus was reduced but not obliterated. The swelling was 4x 4, in size, nontender, extending from the angle of the mandible towards the midline with a smooth merging. No soft tissue lesions were observed. There were multiple decayed teeth in the upper and lower jaw. (fig 2&3) Radiographs taken were occlusal, OPG views. Panoramic radiographs showed multilocular lesions present bilaterally in the mandible, causing extensive obstruction of the body, involving the ramus, and extending anteriorly to the symphysis region, and up to the condylar head posteriorly (fig 6).



Intraoral view right and left quadrants

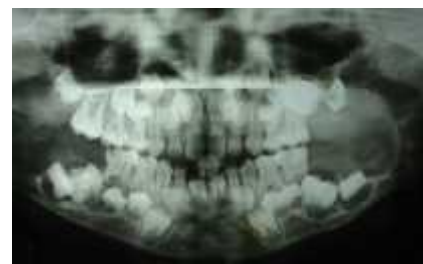


Intraoral view

The width of the mandible shows a considerable increase, the overall shape is altered, the trabeculations are altered, almost missing in the angle region, the mandibular canal path is altered, but interdental bone loss is considerably less.



First OPG



Second OPG

However, the maxillary bone was devoid of such changes. The mandibular occlusal radiographs showed a multilocular cyst-like appearance of the angle region with buccolingual expansion, predominantly buccally. The PA view showed a considerable expansion of the mandible angle region with loss of the normal skeletal boundary. (fig 8) The older OPG (fig 1&2) also shows considerable change, with much loss of trabeculations and change in the posterior region showing unilocular cystic appearance.



Final OPG



Mandibular Crosssectional occlusal view



PA view showing the expansion at the angle of mandible

DISCUSSION

Cherubism is a rare autosomal dominant fibro-osseous, non-neoplastic hereditary condition related to genetic mutation characterized by bilateral simultaneous expansion of jaw bones predominant involving mandible¹⁰. It also affects other skeletal system¹¹. This entity was first described in Ontario 1933 by William A. Jones in a family with several affected members. It is given the name because of 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 word "cherubic" originally designated member of the second order within the Christian celestial Chorus. These are creatures with specific physical features like starring eyes and wheel below the feet. It is a familial disease which runs in generation to generation^{2,5}.

This disease is characterized by bone degradation and fibrous tissue replacement at the angle of mandible and tuberosity of maxilla leading to the prominence of lower face. WHO put this disease in to a familial disorder involving predominantly mandible. This also can be referred to as hereditary fibrous dysplasia, bilateral giant cell tumor or familial multiloculated disease¹².

However the recent genetic mapping for these disease shown it to be a separate entity at the molecular level¹³. The condition is due to an autosomal dominant gene with 100% prevalence in male and 75% in female with variable expression and male to female ratio is 2:1⁵. The chromosome 4p16 has been established or has linkage in causing this diseases. This is also described as a disorder of age-related bone remodeling mostly involving the mandible, with loss of bone and replacement with large amounts of fibrous tissue³. Progression of disease usually stops after the onset of puberty or it regress slowly. The exact cause is unknown. The widely accepted theory for the disease is that, the perivascular fibrosis leads to a mesenchymal disorder and decreased oxygenation. The detected molecular pathogenesis is that with detection of a mutation in the gene encoding SH3-binding green fluorescent protein 2 (SH3BP2)¹². The associated syndromes with cherubism are neurofibromatosis type 1, Noonan-like/multiple giant cell lesion syndrome, Ramon syndrome, and Jaffe-Campanacci syndrome.

A grading system was developed by Ramon and Engelberg to describe the lesion:

- Grade 1:** involvement of both mandibular ascending rami.
- Grade 2:** grade 1 plus involvement of both maxillary tuberosities, and diffuse mandibular involvement.
- Grade 3:** massive involvement of the maxilla and mandible except for the condylar processes
- Grade 4:** grade 3 plus involvement of the floor of the orbits, causing orbital compression¹⁴.

Yet another system of grading was given by Motamedi based on the site and severity of occurrence:

- Grade 1:** fibro-osseous bilateral and symmetrical expansions in the rami of the mandible;
- Grade 2:** more severe involvement of the ramus and body of the mandible and the tuberosity region of the maxillae; and
- Grade 3:** involvement of maxilla and mandible in their entirety with considerable facial deformity.

But this was simplified by Raposo-Amaral by adding another grade to this and described the involvement of the orbits. The impact of this lesion on developments and eruption of deciduous and permanent teeth depends upon the time of onset and expansion of the lesion. The arrangement of deciduous teeth can be altered. When it affects the permanent dentition, there can be missing and malaligned teeth, rudimentary development of molars, abnormally shaped teeth, partially resorbed roots or delayed and ectopically erupting teeth. Extraction may be indicated in case of free floating or ectopically impacted. There are also situations when prosthesis and orthodontic treatment are needed¹¹.

Radiographic features are characterized by bilateral, multilocular, radiolucent areas within the jawbones. The lesions usually appear in the angle of mandible, less frequently in maxilla. The extent of the lesion involves massive areas of the jaw bone affected. The most prominent feature is an expansile lesion of the involved jaws with distinctive cortical thinning but no periosteal reactions, mandibular involvement is generally bilateral. Mandibular lesion often begin in the angle and gradually expand to involve the ramus and body resulting

in thing of the cortical plates. While in maxillary lesions, it may cause obliteration of maxillary sinus⁴.

We had a case with an swelling in the face involving the angle of mandible which according to the history showed gradual in onset. The clinical features were similar to a bony expansile swelling which the literature described as fibro-osseous lesion.

The radiographic features in OPG shows an expansile lesion involving the mandible, ramus, and body extending anteriorly. The older OPG (fig4) shows a unicystic lesion in the angle of mandible which increased in circumferential size in subsequent ones. The lesion matured in to multilocular one in the last OPG. In comparing the first and second OPG (fig4-6) the lesion shows a gradual increase in size involving and specific to the angle region

CONCLUSION

Although rare individuals affected with cherubism have a significant impact on the health and general wellbeing. Genetic test are used to diagnose the case. Surgical intervention, orthodontic treatment along with plastic surgery are a common line of management. At the higher end gene In conclusion we report a special case of cherubism seen in postpuberal age with classic clinical features, sequence of progression, and presentation of different radiographic features of the diseases.

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