A Rare Case Report of Non-Familial Cherubism

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INTRODUCTION

Cherubism is a rare disease which affects jaws. It is seen in paediatric age group. It is an autosomal dominant disorder with male predominance. Many sporadic cases are also reported. This is a self-limiting disorder. The pathophysiology is replacement of normal bone by fibro-osseous tissue causing bilateral jaw enlargement. Increase in size of mandible on both sides results in spherical facial appearance.

Cherubism was first reported in 1933. Radiographically, the condition presents as bilateral, multiloculated, non-radio opaque foci which can involve bilateral maxilla and mandible with ground glass appearance. The lesions are epicentered in rami and tuberosities of mandible and maxilla respectively. The growth is directed anteriorly and pushes the teeth anteriorly. A scleral rim might be seen below the iris, giving the typical "eyes raised to heaven" sign.[1] Fibrous & osseous lesions, in cherubism are classified as quiescent, non-aggressive and aggressive depending on the symptoms and radiological appearances. Quiescent cherubism lesions were most commonly noted in elderly and shows no significant progression. It can regress with increase in age, surgical treatment indicated for cosmetic reasons or to reduce symptoms due to compression effects.

PRESENTATION OF CASE

An eleven years old, girl child came to hospital with complaints of headache and painless swelling involving the upper and lower jaw. The patient attendant gave history of progressive increase in size of the swelling. There was no family history of similar conditions.

On clinical examination, diffuse swelling involving bilateral cheek and jaw region was seen. CT facial bones study was requested further by the clinician. Radiological evaluation by computed tomography revealed diffuse bony expansion, osteolysis and ground glass appearance involving the bilateral maxilla and hemimandible. Areas of cortical breach are seen with associated soft tissue swelling at these regions. There is also involvement of the alveolar processes of maxilla and mandible with relative sparing of the symphysis menti.

CLINICAL DIAGNOSIS

The clinician diagnosed it as fibrous dysplasia / Cherubism.

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DIFFERENTIAL DIAGNOSIS

a) Fibrous dysplasia.
b) Aneurysmal bone cyst.
c) Ameloblastoma.
d) Giant cell lesions.
e) Multiple odontogenic keratocysts.

PATHOLOGICAL DISCUSSION

Histopathological examination revealed, multiple giant cells of varying sizes and variable number of cores in the background of mesenchymal stroma, characteristic of Cherubism.[2,3]

Marck and Kudryk in 1992 proposed a grading system.[4] This depends on the location of the lesions.
Type I: Lesions in rami bilateral mandibles.
Type II: Lesions in rami bilateral mandibles and tuberosity of maxilla.
Type III: Aggressive lesions in entire maxilla and mandible except for condyles.
Type IV: Lesions in the orbits with ocular disturbances and also the grade 3 lesions.

Motamedi (1998) and Raposo-Amaral (2007).[5, 6] classified and graded cherubism as follows:

Grade 1 - Lesions seen in mandible but there are no signs of root resorption.
- Sub type 1: Single lesion involving mandibular body.
- Sub type 2: More than one lesion involving mandibular body.
- Sub type 3: Single lesion involving ramus of mandible.
- Sub type 4: More than one lesion involving both rami.
- Sub type 5: More than one lesion involving the mandibular body and both rami.

Grade II - Multiple lesions involving the mandible and maxilla and there is no sign of root resorption
- Sub type 1: Involvement of both maxillary tuberosities and mandible.
- Sub type 2: Involvement of anterior maxilla and mandible.
- Sub type 3: involvement of mandible and entire maxilla.

Grade III - Invasive lesions of mandible coupled with signs of root resorption seen
- Sub type 1: Single lesion involving mandibular body.
- Sub type 2: More than one lesion involving mandibular body.
- Sub type 3: Single lesion involving ramus.
- Sub type 4: Multiple lesions of the mandibular rami.
- Sub type 5: Lesions involving the mandibular body and rami.

Grade IV - Lesions involving the mandible and maxilla and showing signs of root resorption.
- Sub type 1: Lesions of mandible and maxillary tuberosity.
- Sub type 2: Lesions of mandible and anterior maxilla.
- Sub type 3: Lesions of mandible and whole maxilla.

Grade V - Is Juvenile type, which is infrequent, fast growing, invasive and can disfigure. Lesions which are affecting the maxilla and mandible and can involve the coronoid and condyles.

Grade VI - It is most infrequent, enormously increasing in size, invasive and causes disfigurement.

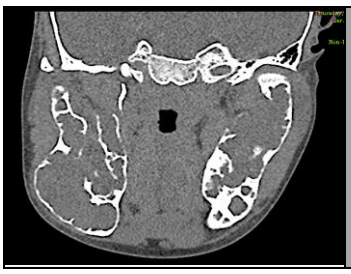

Figure 1.
CT Facial Bone Reconstructed Coronal Section Shows Osteolytic Expansile Lesion in Body Rami and Para Symphysis of Bilateral Hemi Mandible

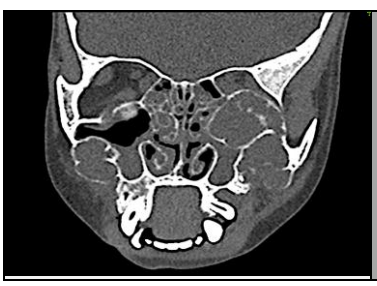

Figure 2.
CT Facial Bone Reconstructed Coronal Section Shows Osteolytic Expansile Lesion in Frontal and Zygomatic Processes of Bilateral Maxilla

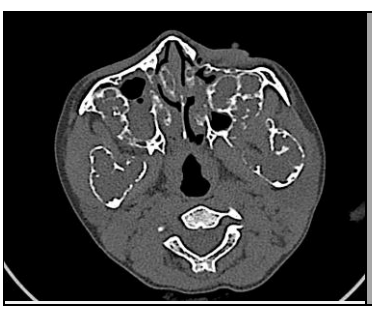

Figure 3.
CT Facial Bone Reconstructed Axial Section Shows Osteolytic Expansile Lesion with Mild Ground Glass Appearance in Bilateral Maxilla

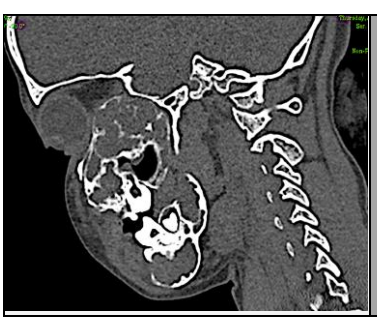

Figure 4.
CT Facial Bone Reconstructed Sagittal Section Shows Osteolytic Expansile Lesion with Mild Ground Glass Appearance in Right Maxilla and Mandible

In aggressive types of cherubism, there is extension of fibrous and osseous tissue into the floor of orbits. The eye ball can get inferiorly pushed causing exposure of scleral rim. Lesion can also involve the retro bulbar spaces and can lead to optic nerve indentation.

Cherubism will not show any manifestations in neonates and infants. Enlargement of the cheeks will be most commonly seen after 24 months of life. There will be swift enlargement of the affected regions of maxilla and mandible happening till the end of 1st decade of life. Following that, there won't be any significant changes until puberty. In the beginning of 2nd decade of life, there is gradual improvement in disease course. Nonetheless plain x ray may show abnormalities.

Chomette and colleagues described three stages in cherubism. Depending on histological, immune histochemical and structural.[7] First stage shows multiple multinucleated giant cells. These cells were found to be Tartrate Resistant Acid Phosphatase (TRAP) positive. These tissues are vascularized. Predominant fibroblastic cells seen in the periphery of the lesions. Haemosiderin is observed in endothelial cells.

Second stage, reparative stage shows proliferative spindle cells. Predominant fibroblastic nodules with central vessels are seen. Freshly synthesized bone matrix and osteoid are visualized.

Third stage which is osteoid forming stage seen along with mineralizing matrix. They contain more collagen and less cellularity. The multinucleated giant cells are called as osteoclasts. The elongated fibro blastoid cells are called as fibroblast cells or myofibroblast cells. The ovoid cells are metabolically active young fibroblasts. Ground glass appearance in bilateral maxilla.

DISCUSSION OF MANAGEMENT

Curettage is the surgery of choice as it gives better aesthetic appearance. Liposuction is also used to get good contour.[8,9,10] Radiotherapy is contraindicated. There is a fear of retardation of maxilla or mandibular growth or there can be radiotherapy induced bone necrosis or malignant degeneration. Medical therapy is also helpful; calcitonin is shown to be effective in literature but there are no proven results. Advanced treatment of Cherubim is the genetic therapy.

CONCLUSIONS

Cherubism is a rare osseous disorder involving maxilla and mandible. It is not diagnosed purely radiologically. Cherubism is diagnosed on the basis of age group, genetics, clinical presentation, radiological findings, laboratory and molecular findings.

Non familial cherubism is a rare entity, which causes prominent lower face. This condition can regress with increase in age. Radiological presentation of the disease most commonly begins in angle and ramus of mandible and continues to the mandibular body. Depending on the staging, bulk reduction can be performed.

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Disclosure forms provided by the authors are available with the full text of this article at jemds.com.

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