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Original Research Paper

Pathology

CHERUBISM: A RARE CASE REPORT WITH REVIEW OF LITERATURE

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ABSTRACT Cherubism is a rare, childhood, nonneoplastic , self limiting fibro-osseous lesion . Affected children usually appear normal at birth. It's a rare disorder and the precise incidence is unknown. It occurs between 1 to 7 years of age and regress spontaneously after puberty. Most studies have reported Cherubism to be familial and with bilateral involvement of the mandible,usually symmetrical. Here , we have described a familial case of Cherubism , involving both the mandibles and the maxilla in a 5 year old male child with a slowly enlarging, painless, asymmetrical swelling of both cheeks.

KEYWORDS : Cherubism; Childhood; nonneoplastic ; fibro-osseous lesion; self limiting.

INTRODUCTION:

Cherubism was first described by William Jones in 1933 . In 1938 the term "Cherubism"was used for the first time because the faces of the patients with more or less symmetrical swollen cheeks resemble the cheeks of renaissance art. It is also known as familial fibrous dysplasia of jaws, familial multilocular disease of the jaws and multilocular cystic disease of the jaw. Theexact etiology is unknown but hereditary plays an important role. In most patients cherubism is due to dominant mutation in the SH3BP6 gene on chromosome 4p16.3 . Until now, around 300 cases of Cherubism have been reported. It presents between 1 to 7 years of age andregress spontaneously after puberty, surgical intervention is needed only , if there are functional concerns. This inherited autosomal dominant disorder affecting jaw is characterized by replacement of normal bone by proliferation of fibrovascular tissue containing multinucleated giant cells.We are reporting this case because of its rarity. Computed tomography was performed and biopsy yielded a pathologic diagnosis 1-4 .

Case report:

A 5 year old male child presented at the outpatient department with a slowly enlarging, painless, bilaterally asymmetrical swelling of the mandible and maxilla. When the child was approximately 1.5 years old , the parents noticed a change in his facialsymmetry, which later became more obvious. He had a positive family history of Cherubism in his paternal uncle. There was no history of any trauma ,pain, pus discharge, blood discharge, fever, paresthesia, anorexia or weight loss. The patient did not have any complaints during respiration, phonation and swallowing.

On palpation the swellings were bony and hard in consistency.The skin over the swelling was smooth and freely movable, with no increase in temperature.Submandibular and cervical lymph nodes were not palpable. There was increased intercanthal distance and the left eye demonstrated inferior scleral show . Mouth opening was adequate. The patient had poor oral hygiene and teeth were displaced resulting into malocclusion and anterior cross bite. Patient even had posteriorly angulated ears.

Computed Tomography (CT) examination revealed involvement of both the mandible and the maxilla showing a soap bubble like multilocular radiolucency with expanded cortices,not involving the condyles. The fibro-osseous tissue was also extending into the left inferior orbital wall resulting in exposure of a rim of sclera beneath the iris – inferior scleral show.

According to Ramon and Engelberg grading , it was graded as Grade IV.

A bone biopsy was obtained from the right ramus and parasymphysis of the right mandible.

We received two bony bits each measuring 1x1x0.3cm.
On microscopy the tissue submitted showed marked proliferation of stromal spindle to oval cells interspersed with collagen bundles along with acellular cement like areas. At the periphery of lesions , shows mature bone trabeculae lined by osteoblast and osteoclasts. However no evidence of multinucleated giant cells were seen.

Hence,the final diagnosis of Cherubism was made.
As the child grows older, the lesion often becomes static and shows regression after puberty. In the current case as there were no functional concerns, hence no surgical intervention was undertaken and follow-up evaluation was advised with oral hygiene instructions.

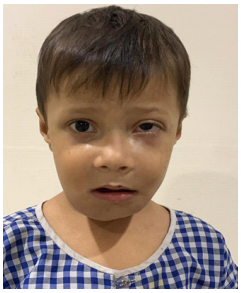


Figure 1: Photograph of the patient showing frontal view of face revealing prominent mandibular and maxillary contours,

increased intercanthal distance, inferior scleral show and posteriorly angulated ears.

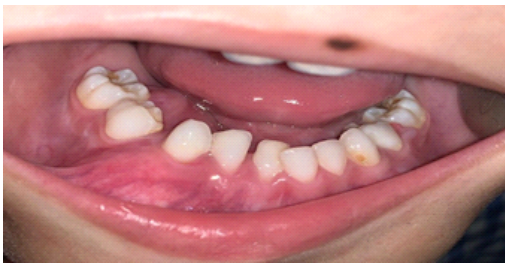


Figure 2: Photograph showing displaced teeth and poor oral hygiene.

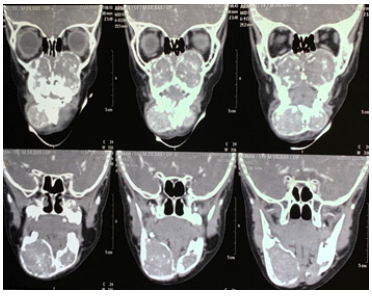
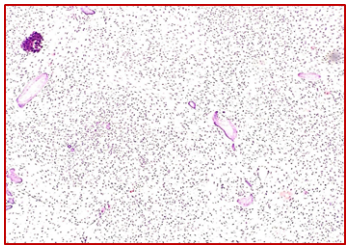
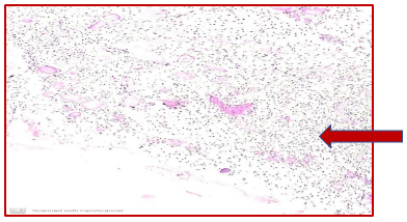


Figure 3: Photograph showing CT scan revealing involvement of both the mandible and maxilla showing soap bubble-like multilocular radiolucency with expanded cortices. The fibro-osseous tissue is extending into the left inferior orbital wall.



Microscopy – H&E 40x



Microscopy- H&E 100x showing showing spindle cells.

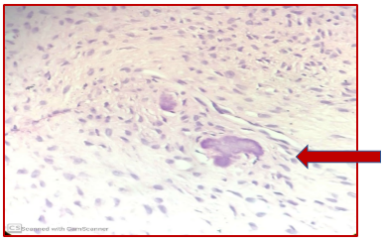


Figure 4,5,6 – showing acellular cement like areas.
Microscopy- H&E400x

DISCUSSION:

Cherubism has historically been considered a variant of fibrous dysplasia, but in reality is likely a distinct entity. A cherub is a toddler or baby angel, often portrayed in art to have chubby cheeks and an upward gaze. In fact, such a divine being is more accurately called a putto , but in modern English usage the terms have become blurred , and patients with cherubism are implied to have cherub-like faces.It's a rare disorder and the precise incidence is unknown . It is inherited in an autosomal dominant pattern and has variable penetrance , with onset in early childhood (typically in the 3-4 years of age) in our case the patient presented at 5 years of age. Interestingly inheritance is dependent on gender:100% in males and 50-70% in females, our patient was a male and had a positive family history of the same. Although the exact mechanism is unclear, an underlying SH3BP2 gene mutation is present in approximately 80% of cases.

Clinical presentation is due to characteristic cosmetic changes in the face, consisting of:

- bilateral, usually symmetric, jaw fullness with a slight upward turning of eyes. In our case though the swelling was present bilaterally , it was not symmetrical.
- bilateral expansile multiloculated cystic masses with symmetric involvement of mandible and maxilla

Additionally, submandibular lymph node enlargement may also be present . The teeth in the affected regions may be loose, and tooth eruption delayed 2-3.

Radiographic features consist of lucent expanded regions within the maxilla and mandible, with soap-bubble appearance.

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

- Grade I : Bilateral involvement of the ascending ramus of mandible.
- Grade II : Bilateral involvement of the ascending ramus of mandible and maxillary tuberosity
- 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 compression3

Histologically , Cherubism lesions resemble giant cell tumors because they contain many giant-cells and mononuclear or stromal cells. The fibrotic lesions are non-neoplastic. Cherubism cannot be diagnosed by histology alone because they are not distinguishable from other giant cell lesions of bone.

Histological features are indistinguishable from a giant cell granuloma.

Chomette and colleagues describe 3 histologically, immunohistochemically and ultrastructurally distinct stages in cherubism lesions [46]. In the first, osteolytic stage the authors found numerous round, fusiform and multinucleated giant-cells. The giant osteoclast-like cells are tartrate resistant acid phosphatase (TRAP) positive.

The tissue of the lesions is well vascularized. Fibroblastic cells with fewer giant-cells can be found in the periphery of the lesions. Hemosiderin, a breakdown product of hemoglobin and a sign of hemorrhage, is observed in endothelial cells and some surrounding fibroblasts. The second stage is characterized by proliferative Despite the pronounced changes, the disease stabilizes and often regresses without the need for treatment2

CONCLUSION:

Although rare, cherubism has a significant impact on affected children and their families . This is especially true in those cases where aggressive growth leads to facial deformity and functional problems. In the majority of cases , cherubism is self-limiting and no surgical treatment is necessary apart from

longitudinal clinical and radiographic observation. In cases of rapidly proliferating cherubism with significant functional consequences , resection may be indicated. Operative intervention does not change the disease progression but may improve function and appearance2

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Conflicts of Interest.
There are no conflicts of interest.

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