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Case Report
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A Rare Case of Grade III Cherubism

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Abstract

Cherubism is a benign condition affecting the maxilla and mandible; it is an autosomal dominant fibro-osseous disorder. The characteristic feature of cherubism is the "cherub" like appearance of the patients. This occurs due to multiple cystic lesions in the maxilla and mandible that gives a rounded face appearance. Here we describe a rare case of Grade III Cherubism affecting an eight year old girl. The patient was diagnosed and is currently undergoing conservative treatment.

Introduction

"Cherubism" was first coined and documented in 1933 by Dr. WA Jones of Kingston, Ontario, describing a case of three siblings of the same family of Jewish Russian heritage [1]. It is a self limiting disease and rarely apparent before the age of two years. It occurs in children and predominantly in boys. It is a non-neoplastic disease of bone characterized by clinical bilateral swelling of cheeks due to bony enlargement of jaws that give the patient a typical 'cherubic' look. Beginning in early childhood, both the lower jaw (the mandible) and the upper jaw (the maxilla) become enlarged as bone is replaced with painless, cyst-like growths. These growths give the cheeks a swollen, rounded appearance and often interfere with normal tooth development. In some people the condition is so mild that it may not be noticeable, while other cases are severe enough to cause problems with vision, breathing, speech, and swallowing. Enlargement of the jaw usually continues throughout childhood and stabilizes during puberty. The abnormal growths are gradually replaced with normal bone in early adulthood. As a result, many affected adults have a normal facial appearance.

Case Presentation

An 8-year old girl presented to us in the maxillofacial department of Bangabandhu Sheikh Mujib Medical University with the complaints of bilateral facial swelling for the past 5 years. The patient's parents stated that they first noticed the swelling when she was 3 years of age. It presented as a small swelling of the mid-face, and gradually began to grow. Concerned, they went to a local doctor, who could not give a definitive diagnosis or treatment. Gradually over the course of 5 years the swelling involved most of the mid- face particularly medially and also lateral portions of the lower third of the face. On further query no familial history of such lesions could be determined.

On examination large bilateral symmetrical expansion is evident affecting the mid-face extending from the malar prominence to the angle of the lip. There is obliteration of the naso-labial fold and an appearance of flattening of the nasal bridge and flaring of the nostrils. Profile of the patient revealed bilateral swelling of the lateral aspects of the lower third of the face. The pathologies were firm to hard on palpation, with no discernable outline. They were not tender. Intra-oral examination showed complete obliteration of the palatal arch as all of the premaxilla was involved with the swelling; anterior dentitions were missing. The vestibule was discernible but grossly diminished. Upper posterior dentition was intact though the vault of the palate was also involved with the swelling. In the lower arch both the anterior and posterior dentition appeared normal though there was gross expansion of both buccal and lingiual cortex. The patient had no difficulties in eating swallowing or speech and ocular examinations revealed no abnormality. For the purpose of this academic paper consent was taken from the patient.

An orthopentamogram and a contrast CT scan of the maxillofacial region was advised to assess the extent of the involvement. CT scan revealed mixed density osteolytic lesions involving the maxilla and mandible. Maxillary involvement included both premaxilla and the hard palate, both maxillary sinus and infraorbital rim. Floor of the orbit was involved as well. Upper anterior teeth appeared embedded within the lesion. The entire body and both rami of the mandible also showed gross destruction though a thin rim of alveolar bone including dentition remained intact.

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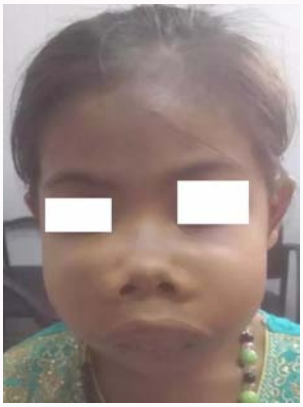

Figure 1: Frontal view of patient.

Figure 2: Lateral view of patient.

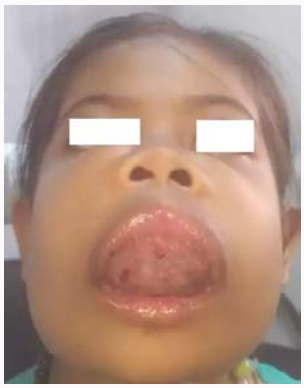

Figure 3: Intra-oral view.

The posterior and lower border of the mandible and both condoyle were also intact.

As there was no familial history of cherubism some routine investigations were advised to establish a definitive diagnosis. To rule out any metabolic abnormality we advised serum calcium, parathyroid hormone and alkaline phosphatase assay. All were found to be within normal range with regards to the age of the patient. An incisional biopsy followed by histopathology was also advised. Histopathology revealed the following report; sections showed pieces of soft tissue with bony tissue at one side. Few osteoclast-like giant cells were seen in a fibrous stroma. The fibrous stroma was highly vascular containing spindle fibroblasts arranged compactly. The histopathological diagnosis was a giant cell lesion consistent with a

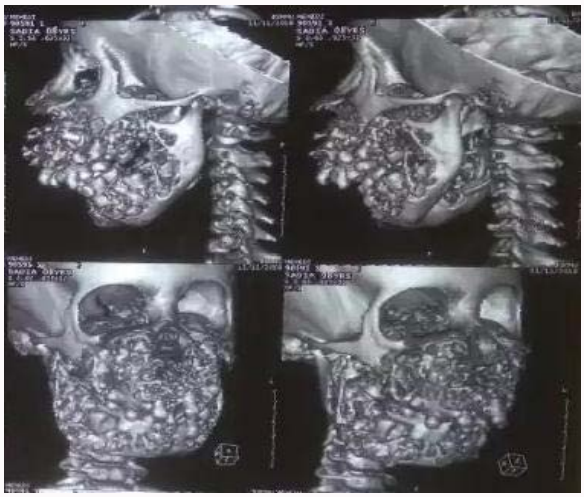

Figure 4: CT Scan of patient.

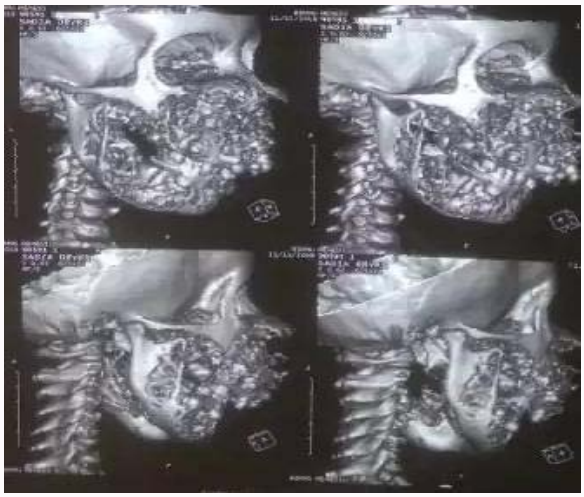

Figure 5: CT Scan of patient.

giant cell granuloma. Finally a definitive diagnosis of cherubism was established factoring in clinical, radiographic and histopathology findings.

Discussion

Cherubism belongs to a group of non-neoplastic bone lesion that affects only the jaws according to World Health Organization (WHO) [2-4]. It is also considered member of the family of fibrous osseous diseases and some authors refer this disorder as familial fibrous dysplasia. Other names for this disease like “familial benign giant cell tumor of the jaw” and “familial multi-locular cystic disease of the jaw” have also been used.

Dr. Jones first described Cherubism in 1933, as “familial multi-locular cystic disease involving mandible and maxilla” [1]. Later on the term cherubism was used this was in particular due to the cherub like appearance of the patient. The cherub like appearance was due to the symmetrical swelling of both jaws causing a rounded face appearance in addition sometimes patients were found to be looking upwards (eyes raised towards heaven) due to retrusion of the lower eyelid that causes a visible sclera beneath the iris [5-16].

Cherubism has been defined as a genetically determined alteration

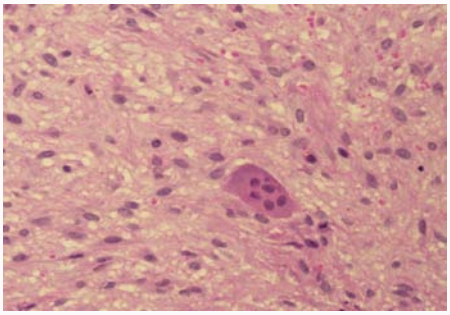

Figure 6: Histological section.

of tooth germ development with a mutation in gene SH3BP2 [3,17-19]. It is an inherited autosomal dominant trait which appears to have 100% penetrance in males and 50% to 70% penetrance in females [12]. Being a hereditary condition cherubism has also been described with some other hereditary syndromes like Ramon Syndrome, Noonan Syndrome and fragile X syndrome [12,20,21]. Though rare, non- familial manifestations of cherubism similar to our case have also been described [13-15].

Clinical course of the condition is usually predictable with the child presenting as healthy at birth, oral symptoms begin to arise during the 2nd or 3rd year of life. Classic bilateral symmetrical deformity of the face due to multiple cystic lesions involving both jaw bones is the main diagnostic criteria. The disease progresses until puberty and shows gradual regression in adulthood. Solitary lesions or some degree of deformity may persist well into adulthood, and a few aggressive versions of cherubism have also been described. Fibrous tissues when invade the floor of the orbits, causes the floor to lift upward irregularly which may push the eyes in different directions [22-24]. Due to the orbital floor involvement the inferior, lateral recti muscles and the intraconal space of the orbits may also involved [25,26].

- In 1957 Seward and Hawkey suggested a grading system for CherubismGrade I: Involvement of bilateral mandibular molar region and ascending rami, mandible body or mentis.
- Grade II: Involvement of bilateral maxillary tuberosities as well as the lesion of grade I diffused whole mandible.
- Grade III: Massive involvement of the entire maxilla and mandible except the condyles.
- Grade IV: Involvement of both jaws with condyles [21].

Our case falls under Grade III Cherubism. A similar grading system was also described by Arnott in 1978 [3].

Histopathologically cherubism is similar to other fibrous dysplastic lesions and cannot be differentiated. Most of the orbital lesions consist of osteoblast cellular proliferations, surrounded by giant cells and fibroblast cells. Presence of vascular channels have been reported [23,24]. Radiographic presentation usually shows multiple cystic lesions involving both jaws bilaterally. Features like "floating tooth syndrome" and "ground glass appearance" have been used to describe the radiographs [16]. CT scan is the primary imaging technique essential for establishing a diagnosis and for adequate course of action. It helps to identify the extent of bony involvement.

In general, the prognosis for cherubism is good. Being a self-limiting disease the "wait and watch" policy has been the main course of action recommended. Surgery is not a treatment of choice. But in case of expansion of tissue resulting in difficulty with airway or chewing capacity, biopsy and surgical intervention can be done. Medical attention for aesthetic and functional concern is required.

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