Case Report

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CHERUBISM: A CASE REPORT AND REVIEW OF LITERATURE

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ABSTRACT:

Cherubism, a pediatric disease, is a self limiting non-neoplastic autosomal dominant fibro-osseous disorder of jaws. 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 characterized by clinical bilateral swelling of cheeks due to bony enlargement of jaws that give the patient a typical 'cherubic' look. Regression occurs during puberty when the disease stabilizes after the growth period leaving some facial deformity and malocclusion. Cherubism may occur in solitary cases or in many members of the family, often in multiple generations. Since it was first described by Jones in 1933, many cases have been documented. Here a case of 5 years old cherubic child, with his clinical appearance as well as radiological evaluation and discussion about clinical outcome are presented. The patient was diagnosed but not treated.

Key words: Cherubism, multilocular radiolucencies, self limiting, fibro-osseous disorder, osteoclastic lesions, giant cells.

INTRODUCTION:

Cherubism is a benign, self-limiting fibro-osseous bone disease of childhood affecting only the jaws. It is evident around third or fourth year of life. In 1933, Jones first described the entity with its typical clinical features[(1)]. Typically the jaw lesions of cherubism are characterized by bilateral swelling of lower face. Because of this prominence of lower face, patient gives an appearance reminiscent of the 'cherubs' portrayed in Renaissance art, thus, this disease became known as Cherubism. It is one of the very few genetically determined osteoclastic lesions. Its gene2 is mapped on chromosome band 4p16.3 and is called SH3BP2 (for SH3 domain bind protein 2)[(3)]. The lesion tissue consists of vascular

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fibrous tissue containing varying numbers of multinucleated giant cells, which are diffused or focal. Radiologically, appearing as multilocular cystic lesion.

CASE DETAIL:

An 5 year-old female child (Fig. 1) reported by his father for sole complaint of painless, progressive and bilateral enlargement of lower face and jaws. The history revealed that the patient had been born as a full term normal delivery and showed no abnormalities until about the age of two year, but later symmetrical and bilateral swelling of lower face was seen. This enlargement had continued in gradually progressive fashion throughout the subsequent years. Family history showed that his father had a similar fullness of the cheeks in childhood that regressed after puberty. No other siblings gave such a history. On physical examination it was seen that patient was well built, active and mentally alert. No abnormality was found on clinical examination of the chest, abdomen, cardiovascular and central nervous system. No cutaneous pigmentation or other congenital abnormality was present, there was no evidence of endocrinal disturbance. Submandibular lymph nodes were bilaterally palpable , nontender and mobile.

On extraoral examination normal expression and color of the face was seen. There was no ophthalmic abnormality. There was symmetrical enlargement of both sides of mandible and minor involvement of maxilla. Enlargement was non tender and hard on palpation.

On intraoral evaluation few permanent teeth were seen (Fig. 2). Patient gave history of crowding in deciduous anterior teeth. Panoramic radiographs revealed extensive involvement of mandible. Multiple cystic areas were seen involving mandible on both sides extending up to the base of the condyle of the mandible (Fig. 3). There was marked thinning of cortex on both sides. Extension of lesion was also seen in maxilla. Posterior walls of maxillary sinus appeared more affected than anterior walls. Medial wall appeared normal on both sides. Laboratory investigations showed a hemoglobin level of 9.8 gm/dl (normal 13 to 18 gm/dl), an elevated alkaline phosphatase value of 623 IU/L (normal, 85 to 270 IU). Parathyroid hormone level and other laboratory investigations were within normal limits.

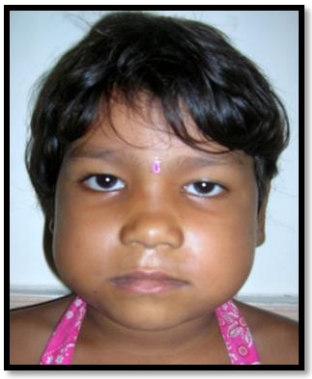

Fig. 1: Case showing bilateral swellings of face

Fig. 2: Intraoral View showing few missing permanent teeth

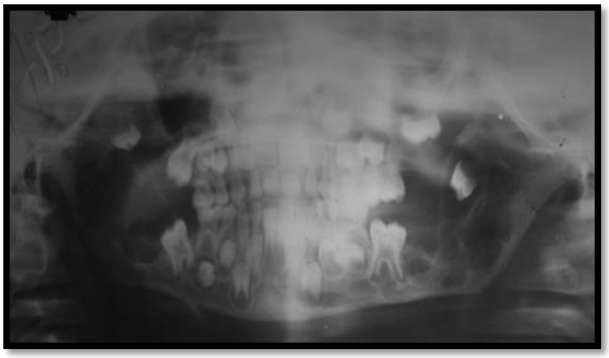

Fig 3: OPG reveals multiple cystic areas involving mandible on both sides extending upto the base of the condyle

DISCUSSION:

Cherubism is a rare hereditary autosomal dominant benign lesion of childhood. It appears as bilateral painless swellings of mandible and maxilla which progress until puberty, and then spontaneously abates. In 1933 Jones described the first case of cherubism in history of literature[1]. To date, many cases have been added to the literature without restriction to any one country or ethnic group. Cherubism appears to be uncommon in India compared with the incidence in other countries. According to World Health Organization (WHO) cherubism belongs to a group of non -neoplastic bone lesion that affect only the jaws[5] . It is also considered member of the family of fibrous osseous diseases and some authors refer this disorder as familial fibrous dysplasia. In 1978 Arnott suggested a grading system for the lesions of cherubism. Cherubism is divided into grades I, II, III and IV depending on location and the severity of involvement of jaws[4] .These classifications are based on extent of lesion at the time of evaluation. The grade often increases on follow-up examination. Our case falls under grade II of this classification, i.e. involvement of both maxillary tuberosities as well as the mandibular ascending rami. Clinical or radiographic findings of cherubism are not evident until the age of 14 months to three years of age. Typically, the earlier the lesion appears, the more rapidly it progresses. The progressive swelling of the face, with marked increase in fullness of cheeks and jaws, is common to all cases and is due to enlargement and expansion of the underlying bony structures, the skin and subcutaneous tissue being normal. The bilateral enlargement of maxilla when present, contributes to cherubic analogy by causing stretching of skin of the cheeks, thus exposing a thin line of sclera causing 'eyes raised to heaven' look. This was not reported in our case and is rarely encountered in other case reports. Frequently cherubism is accompanied by abnormalities in the configuration of dental arch and dental eruption. In severe cases tooth resorption occurs. The signs and symptoms of disease depend on the severity of the condition, range from clinically, radiologically undetectable features to grossly deformed jaws, upright palate, respiratory obstruction, impairment of vision and hearing. In few cases, cherubism has been described as being connected with other diseases and conditions such as Noonan's syndrome. Jaw and face lesions with displaced teeth are the only clinical abnormalities present in the child reported here.

Radiologically, it is characterized by bilateral multilocular cystic expansion of

the jaws. Cystic areas in the jaws become ossified resulting in irregular patchy sclerosis. The presence of numerous unerrupted teeth and the destruction of the alveolar bone may displace the teeth, producing an appearance referred as 'floating tooth syndrome'. Classic ground glass appearance because of compressed trabecular pattern is seen but is nonspecific as in our case. CT scan examination is useful in precisely demonstrating changes in lymphadenopathy and tissues surrounding cherubism. Plain radiographs and computed tomography scans are sufficient for diagnosis of cherubism. Magnetic resonance imaging, a non-invasive tomographic method is also useful to study the expansion of soft tissue, in particular in the aggressive forms and established preoperative vascular assessment. On bone scintigrams, low radioactivity (cold areas) was sometimes observed in patients with jaw bone diseases. These scintigrams also represented characteristic findings of cherubism. In general, cherubism has a good prognosis. Cherubism does not progress after puberty, and as the patient grows to adulthood, the entire jawbone lesion tends to develop a more normal configuration. 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.

Cherubism can be substantially differentiated from fibrous dysplasia by the bilateralism of the cystic bone defect. It should also be differentiated from ameloblastoma, multiple dentigerous cyst, central giant cell granuloma and Odontogenic myxoma. The only multifocal bone disease that could reasonably be expected to present as well-defined multiple jaw radiolucency and thus, diagnostic dilemmas is 'nevoid basal cell carcinoma syndrome'[(11)]. It does not however produce the facial swelling characteristic of cherubism.

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