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Case Report

Managing a child with cherubism with functional consequences: a case report

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ABSTRACT

Cherubism is a rare begin, self-limiting autosomal dominant disorder characterized by bilateral and symmetric fibro-osseous lesions affecting the mandible and maxilla. The child with this disorder requires conservative management. Surgical interventions such as contouring, resection, or curettage are rarely required for disease and only in severe functional problems. This case study attempts to address the physical and psychological issues and challenges the child and his family members face.

Keywords: Cherubism, Fibro-osseous lesions, Familial fibrous dysplasia, Multilocular cysts

INTRODUCTION

Cherubism is a sporadic benign and self-limiting autosomal dominant disorder that occurs due to the mutation of the gene responsible for encoding SH3BP2 binding protein essential for bone metabolism.[1] Till now, only 300 cases have been reported worldwide. There is fibro-osseous bone deformity, rapid bone degradation followed by extensive bone remodelling with multilocular cysts filled with stromal and osteoclast-like cells, leading to bilateral mandible and maxillary size increment. The characteristic clinical feature closely resembles the "cherubs" from the portrait of renaissance art.[2] Hence, it is named "cherubism." Mr. William A. Jones documented the very first case in the year 1933, presented with bilateral painless expansion of the mandible. The genomic expression in males is 100%, compared to females, which is only 50-70 %.[3] However, many sporadic cases have also been reported.[4] The disorder occurs around the third or fourth year of life, with greater severity at the age of 2 to 5 years, gradually regressing after puberty. Depending upon the severity and location of the lesions, the disorder can be graded from 1 to 6, with lesion 1 affecting the mandible without signs of root resorption and lesion 6 affecting both the mandible and maxilla and the orbits aggressively.[5] In some rare case studies involvement of the zygomatic arches and condyles were also reported.[4-7]

CASE REPORT

Written informed consent from the family and assent have been obtained from the child for the case report.

A 14-year-old boy presented with the complaint of hard bilateral progressive swelling over the lower portion of the face since the age of 4 years, along with breathing difficulty and the development of mal-aligned teeth. There was tenderness and redness in the jaw, with mild pyrexia along with appetite loss, weight loss and decreased vision in the left eye. He frequently woke up at night due to coughs and colds and complained of mouth

breathing and snoring at night since last 6 years. Pre-morbidly he had normal facial features and no difficulty in swallowing, chewing, and speaking. The child was born full-term through normal vaginal delivery. The antenatal, perinatal, and postnatal periods were uneventful. Age-appropriate development milestones were attained (development quotient 100%). Family history revealed that the child's mother and elder sibling have bilateral bony growth in the maxilla. The second-order female sibling died during the cleft palate surgery. Earlier ENT referral for the ongoing complaint revealed bulged oral cavity and normal cribriform plate. Diagnostic nasal endoscopy (DNE) showed bilateral inferior turbinate hypertrophy along with obliteration of the floor of the nasal cavity with mucoid discharge. Nasopharynx could not be visualized. Currently his anthropometry was within normal limits. The vital parameters were within normal range for the age. The child had nasal obstruction and nasal intonation. The peculiar upward gaze of the child was present. Disruption of secondary dentition was observed in the child with rudimentary molars (Figure 1 and 2). Other systemic examinations were also normal. His complete blood count was suggestive of normocytic normochromic anemia. Liver function test showed raised ALP (519 U/L respectively). Parathyroid hormone level and other lab investigations were within normal limits. X-ray face (AP, lateral view) and plain and contrast CT scan of the face revealed unsymmetrical expansion lytic bony lesions with ground glass matrix which showed sclerotic changes involving mandible and maxilla along with associated bilateral narrowing of the nasal cavity and bilateral inferior and middle turbinates were obstructed by lytic lesions. The Biopsy specimens were composed of fibrous connective tissue containing multinucleated giant cells and diagnosed as grade II cherubism. The pathogenic mutation responsible for cherubism was not identified by Targeted mutation analysis. However, his clinical features are adequate to indicate Fibrous dysplasia which is considered as cherubism and the condition is self-resolving. Therefore, further genetic workup was not advised. The probable list of differential diagnosis shortlisted namely brown tumor of raised PTH, giant cell division, Noonan syndrome, aneurysmal bone cyst and hyperparathyroidism were ruled out.

He was managed by the health care team (HCT) consisting of a pediatrician, ENT surgeon, dental surgeon, plastic surgeon, nurse, dietician, and clinical psychologist interacted with the family. Steam inhalation was advised along with Xylometazoline nasal spray and normal saline drops for instillation. No surgical intervention was planned immediately but was kept as a standby intervention to debulk the fibrous tissue if airway obstruction worsens. Dental and plastic surgeons also advocated for conservative treatment and surgical correction was recommended after the attainment of skeletal maturity. The child was kept under regular follow-up. Parents were explained about the disease and the long-term management. The signs of respiratory distress were informed to them. The mother was encouraged to raise the child with her head for ease of breathing, keep the airway patent, and reduce pressure on the airway. On anticipation of impaired nutrition due to difficulty in chewing and swallowing due to jaw abnormalities, small, frequent, energy-dense meals containing a soft diet were advised. Self-imposed isolation and lack of socialization were the other problems due to altered facial appearance. The goal of the team approach to the family was to help them to understand, to alleviate anxiety and foster coping related to facial deformity and to help to develop positive self-esteem. The child was encouraged to participate in age-appropriate social activities in the school to build his confidence. The child continues to be visiting the facility regularly for the follow-up care.

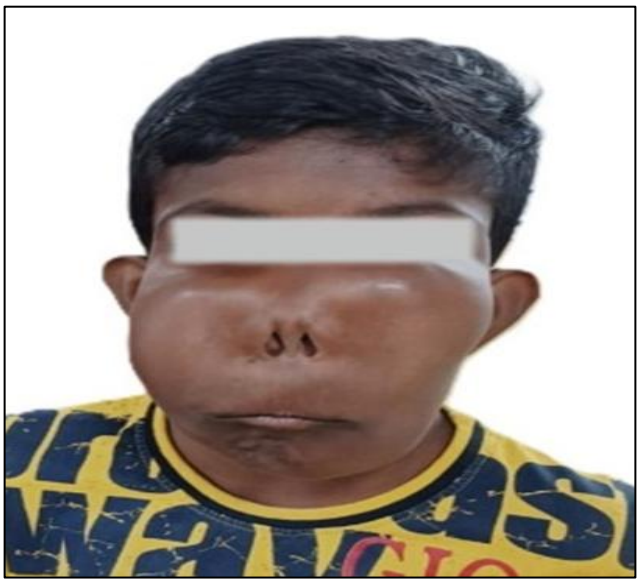

Figure 1: Bilateral swelling of the mandible and maxilla.

Figure 2: Intraoral view showing rudimentary molars.

DISCUSSION

Cherubism appears to be uncommon in India compared with the incidence in other countries.[7] Presentation is more common in males than females. Paradigmatically the cases are normal during infancy, inception is usually seen between the age of 14 months to 5 years, never the less in severe cases the presentation can occur at the time of the birth also.[5] Likewise in this case report, the case is male whose mother also had the same kind of presentation. Most of the cherubism cases show familial history, but some sporadic cases were also documented.[8,9] World health organization stated that cherubism is a cluster of non-neoplastic lesion in the bone which is limited only to the jaws. According to the literature this disorder is one such member of the family of fibrous osseous diseases group and hence it also called as Familial fibrous dysplasia.[7] Mostly there is mandibular involvement, however in 60% of the reported cases maxillary involvement was also observed. Rarely orbital involvement affecting the optic nerve leading to vision loss were documented.[2]

Maxillary enlargement on both the side of the jaw causes stretching of the skin on cheeks, which leads the sclera to look like a "eyes raised to heaven look". Similar finding was present in our patient. Frequently cherubism is accompanied by abnormalities in the configuration of dental arch and dental eruption. The presence of numerous unerupted teeth and the destruction of the alveolar bone may displace the teeth, producing an appearance referred as 'floating tooth syndrome'. Likewise seen in index case.[10]

Based on the findings of the radiological investigations there can be differential diagnosis like Noonan syndrome, fibrous dysplasia, giant cell granuloma, hyperparathyroidism.[11,12] Histologically differential diagnosis of primary hyperparathyroidism should be excluded.[13] The radiologic characteristic of cherubism is more diagnostic than histopathologic.[6,14-16] Cherubism has been graded by "Ramon and Engelberg" depending on lesion extension (Table 1). Our patient belonged to grade 2 cherubism in which the lesion involved both mandibular ascending rami and maxillary tuberosities. The laboratory studies of index case showed raised alkaline phosphatase (519 U/L), this finding is supported by the case report on cherubism where it was elevated to 611 U/L. The biopsy specimens were composed of fibrous connective tissue containing multinucleated giant cells and diagnosed as grade II cherubism. Depending on the severity of pathology, there's no effective treatment exists. Only surgical and medical treatment are there to provide some cure to this disorder.[17] For surgical modality, the 'wait and watch' is commonly followed. Surgery is most often planned before the puberty in cases of any grave deformity or severe psychological impact or worsening respiratory problems.[18] There is no evidence for an effective medical treatment till date. Among some trials, tacrolimus has shown some benefits.[19] Another study showed that calcitonin might also be useful since it showed inhibition of bone reabsorption.[20]

Table 1: Grading of cherubism (Ramon and Engelberg, 1986).

Grade	Clinical presentation
Grade 1	Involvement of both mandibular ascending rami
Grade 2	Involvement of both mandibular ascending rami and maxillary tuberosities
Grade 3	Massive involvement of whole maxilla and mandible, except the condylar processes
Grade 4	Massive involvement of whole maxilla and mandible, except the condylar processes with involvement of the floor of the orbits causing orbital compression; according to this grading system

CONCLUSION

Cherubism a rare, self-limiting autosomal dominant disorder has a significant impact on the physical and psychological wellbeing of the child and the family. The aggressive growth of fibro-osseous lesions leads to facial deformity and functional consequences. Operative interventions are generally reserved for improving the functions and the appearance of the child. Symptomatic interventions and psychological support and genetic counselling are the important interventions required for managing such case.

Informed consent and assent were obtained from all participants, and their privacy and confidentiality were safeguarded throughout the study.

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