Non-familial Cherubism

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Abstract
Cherubism is a rare, self-limiting disease that usually affects jaws of pediatric population and is characterized by diffuse, bilateral and multilocular bony enlargement of jaws with a typical radiographic and histopathological appearance. Here, we describe a case of an 8-year-old child without any family history of this genetic disorder.

Keywords: Bilateral jaw swelling, Cherubism, giant cell lesion

Introduction
Cherubism is rare inherited condition affecting the jaws characterized by the replacement of normal bone by a proliferation of fibrovascular tissue containing multinucleated giant cells.[1] Cherubism was first described by Jones in 1933. This osteolytic genetic disorder presents in childhood and tends to regress spontaneously after puberty. Its pathogenesis is unknown.[2] Cherubism is inherited as autosomal dominant disorders.[3] Some cases of Cherubism develop without family histories of the disorder. These presumably represent examples of spontaneous mutation.[1]

Case Report
An 8-year-old male child presented to our outpatient department with slowly enlarging painless, symmetrical swelling of the jaw. There was no significant family history [Figure 1]. Mouth opening was adequate, with “v”-shaped palate. Enlargement of the jaws was hard on palpation and whole mandible was involved. Submandibular lymph node was palpable bilaterally. Skin over the swelling was normal, intact, and freely movable. The tooth present was displaced and deviated from its normal position, with multiple missing tooth both in mandible and in maxilla. Expansion of both buccal and lingual cortical plate was observed, overlying mucosa was normal [Figure 2] Orthopantomograph revealed bilateral, multilocular, radiolucent lesion with thinning of cortical rims and displacement of unerupted teeth in mandible. Dental abnormality was observed more in mandible than in maxilla [Figure 3]. Haematological investigations were within normal limits. Histolopathological examination showed multiple multinucleated giant cells within cellular spindle cell stroma.

On the basis of clinical, radiological, and histolopathological examinations, the diagnosis of Cherubism was made. Due to expected tendency of the lesion to become static and may show regression at puberty, no surgical intervention was undertaken and the patient was kept on follow-up.
Cherubism is a disease of childhood[8] that usually presents before the age of 5, with male affected more commonly than female.[8] The disease is transmitted as an autosomal dominant trait with greater prevalence in males (100%) than in females (50-70%).[1,5] There was no history of the disease in any of the family members of our patient, thus it comes to be a non-familial case. Earlier, such case was reported in different literatures.[6-9] The disorder presents as gradually enlarging, non-painful bilateral swelling of the mandible and sometimes of the maxilla as well.[10] The radiographical appearance of Cherubism is characteristic with bilaterally symmetrical, well-defined multilocular radiolucencies in the mandible, with less frequent involvement of maxilla.[1,7,8] Non-involvement of mandibular condyle was earlier considered a hallmark of the condition.[15,6,8] The abnormalities of the dentition are in the form of incomplete development of teeth, root resorption, displacement, or loss of teeth. Histologically, the lesion contains numerous multinucleated giant cells scattered throughout a fibrous connective tissue.[9] This case showed similar, classical radiographical appearance of the disease and the mandibular condyle was not involved.

Usually, there is no need for active treatment of Cherubism. Since the lesion undergoes spontaneous regression, it is better if surgical intervention is delayed until after puberty. However, in patients with functional or cosmetic problems or emotional disturbances, surgical intervention can be considered.[9]

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