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CLINICAL CASE REPORT

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Cherubism With Bilateral Mandible and Maxilla Involvement

A Case Report

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Abstract: Cherubism is a rare, nonneoplastic, self-limiting fibro-osseous that occurs in children. Affected children usually appear normal at birth. Lesions are characterized by the replacement of bone with fibrovascular tissue containing many multinucleated giant cells. Most studies have reported cherubism to be familial and with bilateral involvement of the mandibles. The authors describe a nonfamilial case of cherubism, involving both the mandible and the maxilla, in a 4-year-old female child with slowly enlarging, painless, symmetrical swelling of both cheeks.

Cherubism is a rare disease that is usually limited to the mandible, but the maxilla may be involved. Computed tomography scan and biopsy are helpful for early diagnosis.

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Abbreviation: CT = computed tomography.

INTRODUCTION

Cherubism is a familial, nonneoplastic, self-limiting fibro-osseous disease. Bilateral swelling of the jaws develops between 1 and 7 years of age.[1–5] It is commonly characterized by bilateral and symmetric fibro-osseous lesions limited to the mandible. In the vast majority of reported cases, the lesions are limited to the mandibles, with the condyles being spared.[3–9] In the case reported here of a 4-year-old girl, both mandibles and maxillae, however, were involved and no significant family history was found. To our knowledge, there are no reports of cherubism involving the condyles. Radiography and multislice computed tomography with three-dimensional reformation were performed. Biopsy yielded a pathologic diagnosis.

CASE REPORT

This case report was approved by the Review Board of Shandong Power Central Hospital and the patient's parents signed informed consent for publication of the case report and any accompanying images. A 4-year-old female child presented at the outpatients department with slowly enlarging, painless, symmetrical swelling of both cheeks. When the girl was approximately 2 years old, the parents noticed a change in her facial symmetry, which later became more obvious. 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 (Fig. 1). There was no history of any trauma, pain, pus discharge, blood discharge, fever, paresthesia, anorexia, or weight loss. No significant family history was reported.

CLINICAL FINDINGS

Radiography

Panoramic radiography revealed involvement of both the mandible and the maxilla, and showed a soap bubble-like multilocular radiolucency with thin and expanded cortices. Unerupted teeth included: 15, 25, 35, 37, 45, and 47 was impacted (Fig. 2). Multislice computed tomography showed the presence of multilocular cystic lesions filled with soft-tissue density material and dental anomalies affecting the body and the sides of the mandible. Similar lesions were also observed in the maxilla (Fig. 3).

Biopsy

A biopsy was obtained from the central area of the left rami. Histologic examination showed many large multinucleated giant cells in a loose, delicate fibrillar connective tissue stroma containing large numbers of fibroblasts and many small blood vessels (Fig. 4). As the child grows older, the lesions often become static and may show regression at puberty. In the current case, no surgical intervention was under taken and the patient was followed up at regular intervals.

DISCUSSION

Cherubism is a disease of childhood, first described by Jones in 1933.[1] It usually presents before the age of 5 years, and is more common in men. The exact etiology is unknown, but heredity plays an important role. Cherubism is typically limited to the craniofacial region. In the vast majority of patients, lesions occur only in the mandibles and the condyles are always spared. In the current case, both mandibles and the maxillae, however, were involved and the mandibular condyles were involved. To our knowledge, there is the first report of condylar involvement.

Cherubism is a rare inherited condition affecting the jaws and it is characterized by the replacement of normal bone by a proliferation of fibrovascular tissue containing multinucleated giant cells.[1] Its pathogenesis is unknown but heredity plays an important role.[2,3] Some studies have reported cherubism to be a

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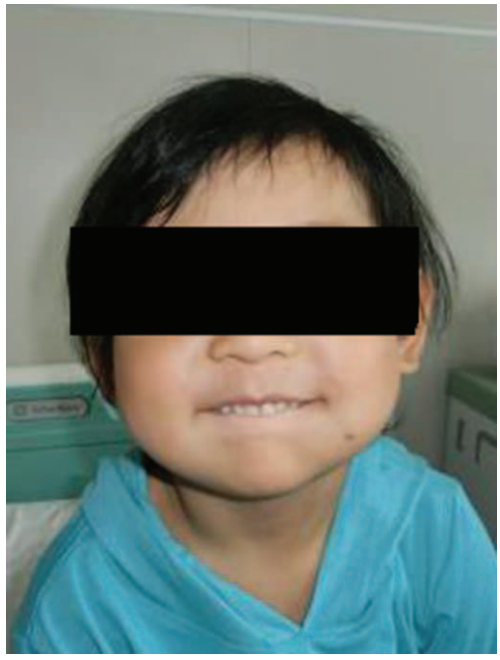

FIGURE 1. Photograph of the face of our case at 4 years of age. It shows symmetrical swelling of both cheeks. On palpation, the swellings were bony and hard in consistency. Skin over the swelling was smooth and freely movable.

family pathology; however, both hereditary and nonhereditary cases have been described.[4,5] In this patient, there was no history of the disease in any of the family members, and therefore, it was a nonfamilial case. In most patients, cherubism is because of dominant mutations in the *SH3BP2* gene on chromosome 4p16.3.[6,7,8,9] In addition to genetic factors, Caballero and Vinals[10] indicated other possible causes of cherubism, such as mesenchymal alterations during jaw development, an odontogenic origin or even hormonal and traumatic factors.

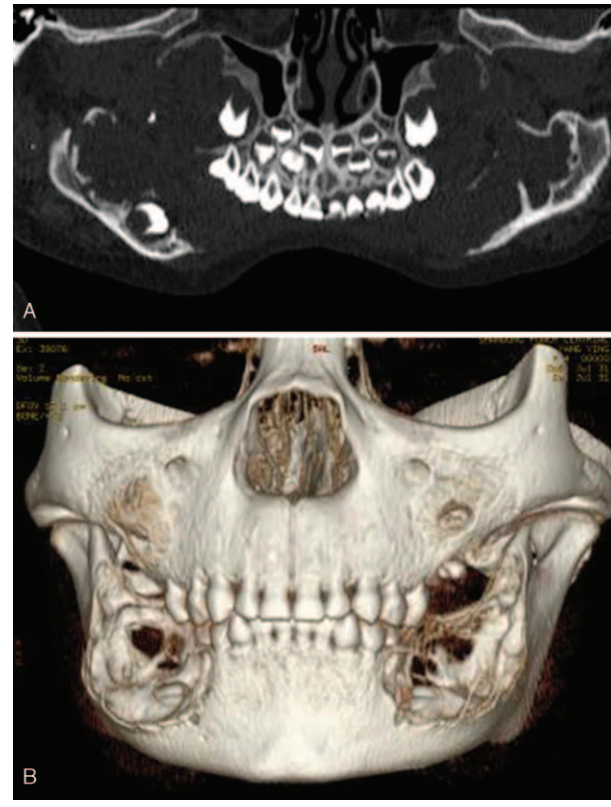

FIGURE 3. Computer tomography using curved-planar-reformation (A) and volume rendered images (B) shows the presence of multilocular cystic lesions filled with soft-tissue density material and dental anomalies affecting both the mandible and the maxilla.

Cherubism is characterized by bilateral and symmetric fibro-osseous lesions limited to the mandible, maxilla, or both.[5,6] Jones described that patients appeared to be ''looking towards heaven'' like a cherub angel, because of the round appearance of the cheeks, resulting from jaw hypertrophy.

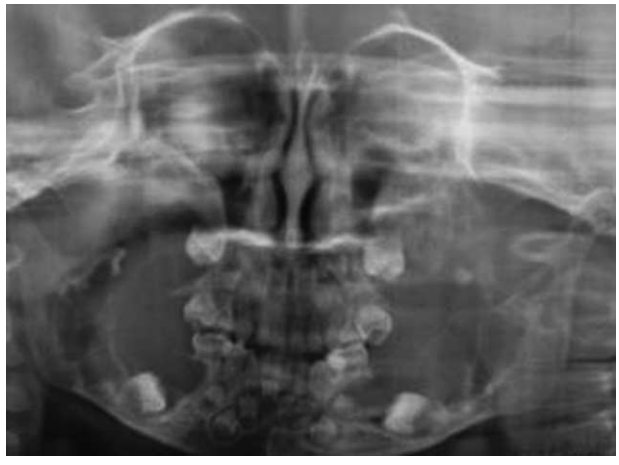

FIGURE 2. A panoramic radiograph of the patient. Both the mandible and the maxilla have soap bubble-like multilocular radiolucencies with thin and expanded cortices. Unerupted teeth included: 15, 25, 35, 37, 45, and 47 was impacted.

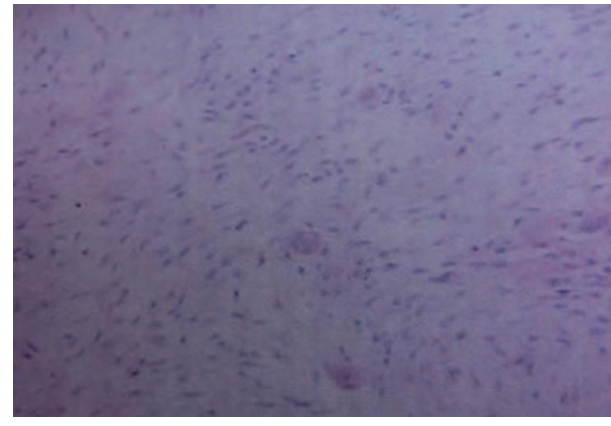

FIGURE 4. Hematoxylin–eosin staining at ×400 magnification. There are many large multinucleated giant cells in a loose, delicate fibrillar connective tissue stroma containing large numbers of fibroblasts and many small blood vessels.

Affected children usually present with painless progressive swelling of the cheeks, frequently associated with dental malformations.[11] This is consistent with the current case. The teeth are affected in the majority of patients with cherubism. The patient also had submandibular lymph node enlargement.[5,6]

Cherubism is a disease of childhood[12]; however, affected children tend to appear normal at birth. Swelling of the jaws usually occurs between 1 and 7 years of age, most often between 12 and 36 months,[5,11,13] with men affected more commonly than women.[7,14,15,16] In this case, the patient showed symptoms of the disorder at the age of 2 years. Lesions increase in number and size until puberty, when they typically begin to regress, fill with bone and remodel until approximately 30 years of age when they often become undetectable.[5,9] Our patient has been followed up for 2 years by radiography, and no changes were seen since the first examination.

Computed tomography is widely used in the diagnosis of bone lesions because it can provide clearer observation of lesions than orthopantomogram. The lesion and its surrounding structures can be analyzed by multiplanar reformation and three-dimensional reconstructed images.[17,18,19] Jain et al[4] reported CT showed the presence of multilocular cystic lesions affecting the maxilla, involving the maxillary sinuses associated with sinusitis and causing severe bone enlargement. Colombo et al[20,21] reported that CT was effective at providing an image of the orbital floor and lesions extending to the edge of the orbit.

Magnetic resonance imaging is also widely used to evaluate musculoskeletal lesions. Because of its higher soft tissue resolution, it is often superior to CT in histologic diagnosis, and provides more information about the lesions and the surrounding soft tissues. Magnetic resonance imaging is very sensitive in identifying bone marrow abnormalities and it is exquisitely able to demonstrate the extent of the tumor. Unfortunately, our patient was claustrophobic and magnetic resonance imaging was not obtained.

We hope that our case report leads to increased awareness and recognition of the clinical and pathologic features of this distinct but rare self-limiting fibro-osseous disease. Careful clinical and radiologic assessments are required. Although the lesions are usually limited to the mandible, our case demonstrates that the maxillae and mandibular condyles may be involved; biopsy may be helpful in making the diagnosis.

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