

CT Findings of Sporadic Cherubism in a 6-Year-Old Boy¹

6세 남아에서의 산발성 천사얼굴증 컴퓨터단층촬영 소견¹

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Cherubism is a rare hereditary disease that affects the jaws in children. This condition shows distinctive computed tomography (CT) imaging features of multilocular, expansile, cystic lesions limited to the maxilla and mandible bilaterally, which can play a key role in the diagnosis of cherubism. We report here a case of sporadic cherubism with characteristic radiologic findings in a 6-year-old Korean boy.

Index terms

Cherubism
Computed Tomography
Pediatrics

Received August 13, 2013; Accepted September 17, 2013

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INTRODUCTION

Cherubism is a rare hereditary disease of non-neoplastic origin that occurs during childhood, leading to the characteristic bony enlargement of the maxilla and mandible (1). The condition was initially thought to be hereditary, but a few sporadic cases have been described (2). We describe here the characteristic computed tomography (CT) features of this uncommon condition in a 6-year-old Korean boy.

CASE REPORT

A 6-year-old boy presented with progressive nasal obstruction and painless swelling of both cheeks which occurred over 4 months. He visited a dental clinic one year previously due to left gingival pain with swelling, and was diagnosed with secondary dental caries. On clinical examination, he showed bilateral symmetrical swelling of the cheeks with bony consistency. There was no local tenderness on palpation. He had no specific medical or family history.

The panoramic view showed expansile, multilocular, osteolytic

lesions involving the maxilla as well as the body and rami of the mandible (Fig. 1). Bilateral condylar extension was absent. CT images clearly revealed the presence of variably expansile, multilocular cystic lesions in the maxilla and mandible. The lesions showed expansile osseous remodeling with cortical thinning of the orbital walls and the nasal cavity, causing severe narrowing of the nasal airway (Fig. 2). There was no cortical breakdown, periosteal reaction, or associated soft-tissue mass. In the soft-tissue window setting, the lesions were shown to be filled with a low attenuation material containing multifocal sclerotic foci.

The patient was diagnosed with cherubism based on the characteristic clinical and radiologic findings. As the symptoms associated with airway obstruction were not severe, conservative management and regular follow-up were recommended.

DISCUSSION

The term 'cherubism' was first used in 1933 by Jones to describe clinical manifestations of full round cheeks, which resembled those of baby angels (cherubs) in Renaissance art (1, 3). The condition was initially thought to be hereditary with auto-

somal dominant inheritance. However, there have been a few sporadic forms of the disease without any definite familial history (2), and recent studies have identified the gene responsible for cherubism mapped to chromosome 4p16.3 (4).

Cherubism usually manifests before the age of five years with painless progressive swelling of the cheeks, causing upward turning of the eyes (“upward-to-heaven-looking eyes”) with exposure of the sclera inferior to the iris or protosis (1, 5). The clinical presentation is variable, and depends on the severity of the disease. Severe forms of the disease show not only massive deformity of the jaws but also respiratory difficulty with maxilla involvement. Dental problems such as incomplete or non-developed teeth, root resorption, and displacement or loss of teeth are also frequently present (6). Because the lesions usually progress until puberty and show spontaneous involution later, conservative management is preferred until puberty. However, surgical intervention may be indicated in patients with serious cosmetic or functional problems. If surgical intervention is needed, curettage or shaving for the

purposes of symptom relief have been considered as the treatments of choice (7).

The imaging features of cherubism are very distinctive on CT, which clearly depict the limited extent of the disease in the maxilla and mandible. The most representative imaging features are bilateral, well-defined, multilocular cystic lesions of the mandible with expansile remodeling of the bone and thinning of the cortex. The usual involvement of the disease begins at the angle of the mandible and extends into the ramus and body (6). Sparing of the mandibular condyles was considered before as one of the typical characteristics of the condition, but a few recent reports have described condylar involvement (3). Approximately 60-70% of lesions may show multifocal irregular patchy sclerosis within osteolytic lesions, which are thought to replace multilocular areas of diminished densities (6). Mandibular involvement is typically bilateral, but unilateral involvement has also been documented (8). Maxillary involvement is less common than mandibular involvement and shows a less extensive pattern. Maxillary lesions are always accompanied with mandibular involvement.

There have been a few studies on the MR imaging findings of cherubism (3), but they have been nonspecific and provided little help in evaluating this condition. In addition, several papers have reported that radiologic abnormalities could be observed during the screening of family members of patients with cherubism, although they showed no apparent clinical manifestations (9).

The radiologic differential diagnoses of cherubism are craniofacial fibrous dysplasia, giant cell lesions (including central giant cell granulomas and giant cell tumors), and brown tumors of hyperparathyroidism (3, 5). However, based on the clinical and

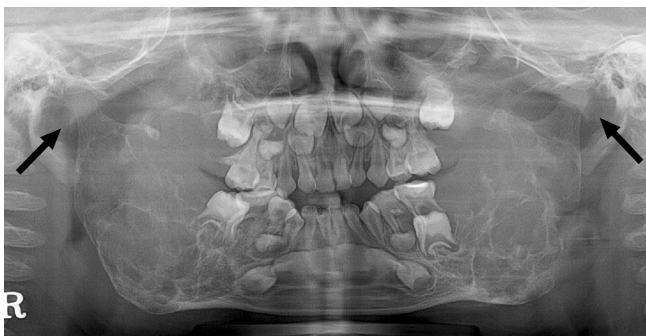


Fig. 1. A panoramic view shows expansile remodeling of the maxilla and mandible replaced by multilocular osteolytic lesions bilaterally. Sparing of both mandibular condyles (arrows) is also noted.

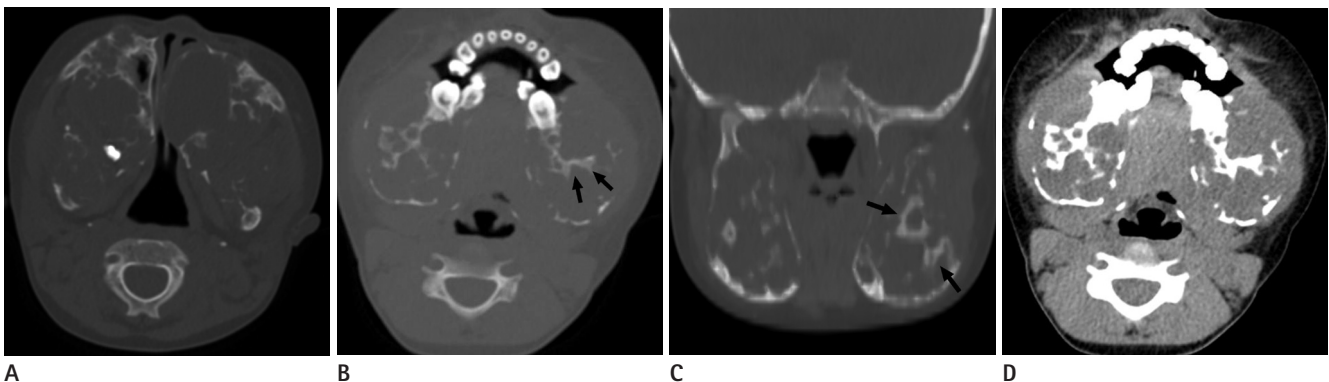


Fig. 2. Cherubism in a 6-year-old boy. **A-C.** Axial and coronal CT images reveal expansile, multilocular cystic lesions containing internal sclerotic matrix (arrows) in the maxilla and mandible. Obstruction of the nasal lumen and remodeling of the bony orbit by the expansile maxilla is noted. **D.** At soft-tissue window setting, these cystic areas contain low attenuation material.

radiologic manifestations, cherubism can be quite easily distinguished from other conditions involving facial bones. Various conditions such as fibrous dysplasia, giant cell lesions, and brown tumors are not usually familial, present at later ages, do not show the typical facial features of swollen cheeks and upward turning of the eyes, and do not involute during puberty. These conditions rarely show the symmetrical involvement seen in cherubism. In addition, a few odontogenic lesions including ameloblastomas and keratocystic odontogenic tumors should be included in the differential diagnosis. Among these, basal cell nevus syndrome presenting as multiple keratocystic odontogenic tumors in the posterior body of the mandible can mimic cherubism on imaging, but this syndrome usually also causes skin lesions or rib abnormalities without the characteristic facial swelling (10).

Cherubism is a very rare disease of the jaw in pediatric patients and shows the characteristic imaging findings of multilocular, expansile, radiolucent lesions involving the mandible and/or maxilla bilaterally. This condition can be diagnosed based on its clinical and radiologic findings, and, thus, it is important for radiologists and clinicians to recognize these typical manifestations when evaluating children with facial and dental problems. CT imaging plays an important role in diagnosing as well as assessing the extent of the lesions, may prevent unnecessary invasive procedures such as biopsies or surgeries, and can aid in family screening.

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6세 남아에서의 산발성 천사얼굴증 컴퓨터단층촬영 소견¹

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천사얼굴증은 드문 유전성 질환으로서 소아의 하악을 침범하는 질환이다. 이 질환은 컴퓨터단층촬영에서 다발성, 확장성의 낭성 병변이 상악과 하악을 양측으로 침범하는 특이한 소견을 보이며, 이는 천사얼굴증을 진단할 수 있는 중요한 소견이다. 우리는 한국 6세 남아에서 발견된 특징적인 영상학적 소견을 보이는 산발성 천사얼굴증 증례를 보고하고자 한다.

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