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CHERUBISM: A RADIOLOGICAL AND CLINICAL PRESENTATION*

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SUMMARY: Yücel ÖT, Genç E, Kaya S. (Department of Otorhinolaryngology, Hacettepe University Faculty of Medicine, Ankara, Turkey). Cherubism: a radiological and clinical presentation. Turk J Pediatr 1998; 40: 453-459.

Cherubism is the hereditary form of the fibrous dysplasia of the jaws, but it may be seen sporadically as well. The disease has a self-limited nature and is rarely apparent before the age of two. There is no need to interfere surgically with these lesions of the mandible or the maxilla unless the child is severely affected, i.e. the disease deteriorates respiration, deglutition, vision, or the psychiatric makeup of the child due to cosmetic reasons. The clinical presentation and radiological evaluation of these children are so typical that the pediatrician and pediatric otolaryngologist need to be informed about this rarely seen disease. A case of a cherubic child, with his clinical appearance as well as his radiological evaluation, and discussion about the clinical outcome are presented. *Key words: familial fibrous dysplasia, cherubism, mandible, maxilla.*

Cherubism, the hereditary form of fibrous dysplasia, was first described by Jones[1] for in 1933, four out of five siblings of a Jewish family.

There is a proliferation of fibrous tissue and giant cells to the extent that the "bone-forming" mesenchyme is disturbed at the jaws, causing painless, symmetric swellings. Cherubism histologically appears as a giant cell reparative granuloma and is accepted to have an autosomal dominant inheritance[2].

Cherubism has been described as a hereditary illness, but we peresent a sporadic case with typical clinical and radiological presentation to inform physicians regarding this rarely seen disease.

Case Report

A seven-year-old boy was admitted to our hospital because of the symmetrical swelling of his malar areas and lower jaw. His father stated that the swelling on the face had begun at the age of four, simultaneously with that at the chin. Family history inquired of discretely was negative for such a disease in other siblings of the family and for the relatives as well.

The patient's eyes were rolled with an upward gaze, disclosing narrow bands of sclera beneath the iris. The palatal vault was found to have a "V" shape and there was a considerable bulging of the roof, mostly on the right side. (Figs. 1 and 2).

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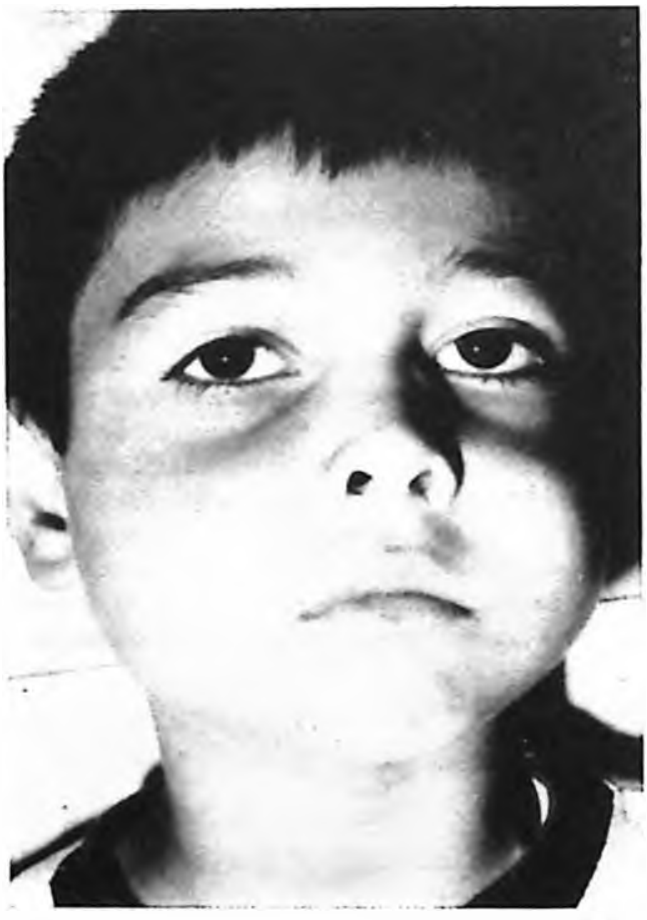

Fig. 1: General appearance of the cherubic child with upward gaze and porminent malar areas.

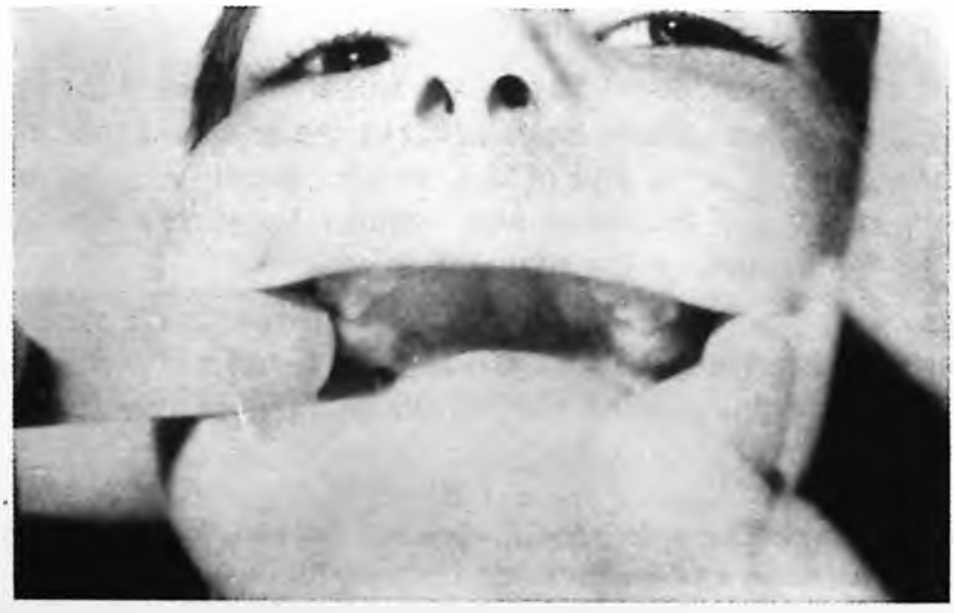

Fig. 2: The "V"-shaped palate.

X-ray studies showed bilateral symmetric, well-defined, multilocular areas of radiolucency, mostly at the molar areas. The thinning of the cortical bone is very typical with the expansion of the medullary part of the bone (Figs. 3 and 4). Computed tomography (CT) scan of the mandible and maxillary region showed these lesions more clearly as symmetric thinning of the cortical bone; multilocular hypodense areas in the core can be noted (Figs. 5 and 6).

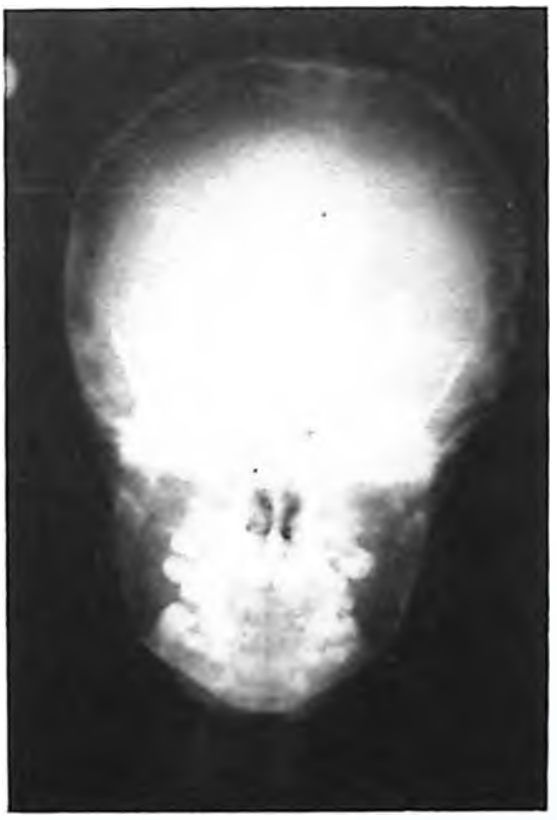

Fig. 3: The antero-posterior mandibular x-ray of the patient showing cortical thinning.

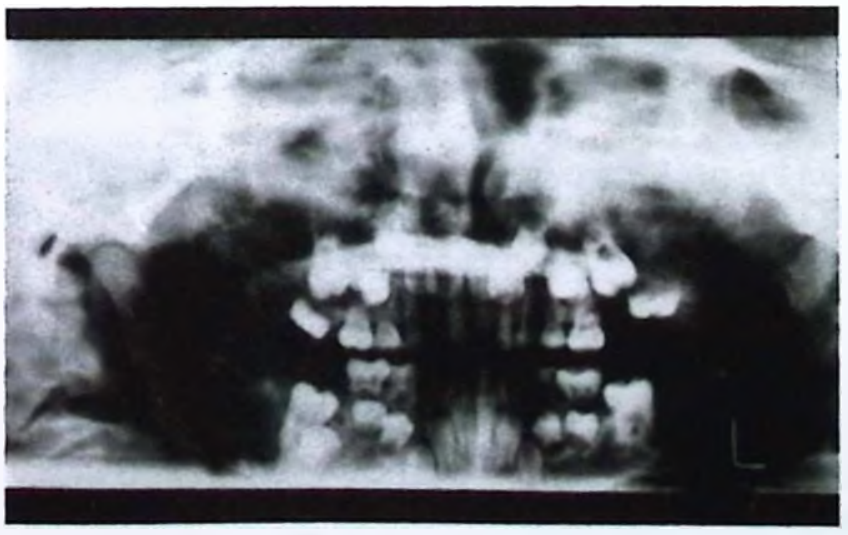

Fig. 4: The panoramic mandibular x-ray of the patient. The status of the teeth is typical.

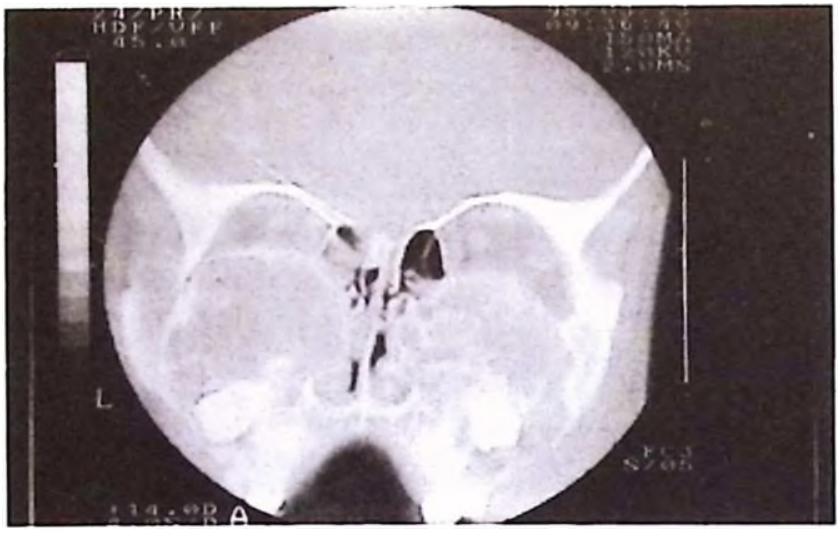

Fig. 5 The computed tomography (CT) scan of the maxilla showing the cortical thinning and medullary ballooning.

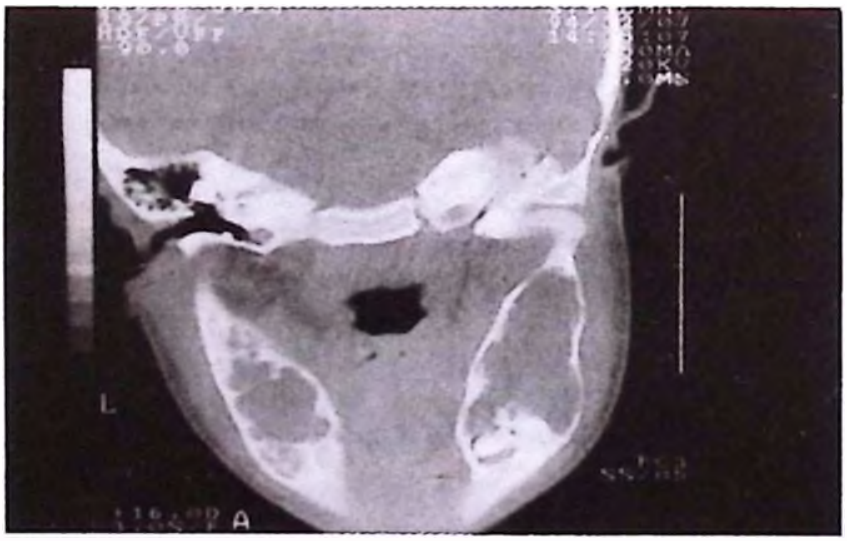

Fig. 6: The CT of the mandible with extreme cortical thinning and multilocularity in the medullary part.

On both sides of the neck there were enlarged lymph nodes, multiple in number, with the largest being two cm in diameter. A lymph node biopsy done previously in another center and confirmed in our pathology department revealed that the nodes had chronic inflammatory nonspecific changes. The bone marrow aspirate taken by the same center showed the hypercellularity of the bone marrow.

The systemic examination and laboratory work-up, except for a slight elevation of alkaline phosphatase, were normal.

In view of the history, the physical examination and the radiological evaluation of the patient, a diagnosis of "cherubism" was made.

Discussion

Since 1933 when cherubism was first reported many synonyms have been used to define the condition. Some of them are: the original name given by Jones of "familial multilocular cystic disease"[1], "bilateral giant cell tumors"[3], "familial fibrous swelling"[4], "familial intraosseous fibrous swelling"[5], disseminated juvenile fibrous dysplasia"[6], "familial osseous dysplasia"[7], and "hereditary fibrous dysplasia"[8].

The disease is generally not realized by the family at birth but rather by the age of two; it becomes overt especially at the region of the angulus of the mandible, the molar, retromolar regions and sometimes may involve the maxilla as well[8, 9]. In addition, a "V"-shaped palatal vault is frequently observed in these patients.

The maxilla, which can be affected up to 67 percent, showed the same morphological changes as seen in the mandible. The initial maxillary lesions occur in the region of the maxillary tuberosity and at the alveolar bone adjacent to it[8]. The maxillary sinuses are small or totally obliterated. According to Burland et al.[7], the cherubic look is seen in the patients in whom the disease involved the maxilla, especially the infraorbital ridge, the anterior antral wall and the floor of the orbita. The presented case is one in which the floor of the orbita and the infraorbital ridge were severely involved (Fig. 5).

The radiological appearance of cherubism is characterized by multilocular cysts that are separated by a thin bone and irregular shapes, with bilateral involvement, affecting the mandible and the maxilla. These cysts cause the expansion of the bone, but cortical bone is not involved and no periosteal bone formation is observed[4, 8, 9].

The laboratory work-up of the patients contain normal values except for the alkaline phosphatase levels[8]. Our case's laboratory work-up was consistent with these findings.

Cervical lymphadenopathy, which is characteristic of the disease, regresses synchronously with the facial lesions[8]. Our patient also had bilateral cervical lumps, and the histologic examination of these lumps revealed lymphatic hyperplasia. This reactive state of the lymph nodes is attributed to various things, including an inflammatory event secondary to stasis or possible dental sepsis[9].

Involution usually begins by the age of 10 for girls; boys reach that stage at about the age of 14[10]. With the regression of the disease, new bony septa are formed with an increase in bone density. The mandibular margins are more easily defined and the multilocular pattern is observed to have a sclerotic bony shape in the regressed patient[11].

Cherubism is considered a mutation of a non-sex linked gene affecting the bone development of the jaws. Although Quan et al.[12] related the genetic changes to the spontaneous deletion in the FMR1 gene, this still needs to be proven. Although the disease was accepted as a familial condition for many years, it is clear that our case is a sporadic one like some others reported in the literature[13, 14].

In the differential diagnosis of this disease, fibrous dysplasia, bilateral parotid swellings, masseter hypertrophy, gingival fibromatosis, infantile cortical hyperostosis, giant cell reparative granuloma, benign giant cell tumor, hyperparathyroidism, odontogenic cysts, histiocytosis X, and ameloblastoma should be considered[8, 9].

In the case of fibrous dysplasia, since the histopathological appearance is very similar to that of cherubism, a difficulty arises. As a definitive criteria, fibrous dysplasia is not hereditary, and more commonly affects the female. Sexual precocity is usually present[15]. When fibrous dysplasia is considered, maxillary involvement is more frequent than mandibular involvement and the lesions are not symmetrical. It has an onset in older ages as well. In fact, the two diseases are very close but not similar entities. As our case is a sporadic one, we had to depend on the clinical presentation and radiological evaluation in the diagnosis of our case.

The giant cell tumors are seen at an older age with no bilateral symmetric involvement and are "extremely rare in the jaw bones"[16]. Infantile hyperostosis is seen as early as six months of age, but the lesions appear as cortical thickening instead of thinning and the cystic changes are in the medullary part of the bone.

There are many controversies about the treatment modality of the disease. Jones[1] states that each patient should be evaluated individually according to the manifestations of the tumor. His guidelines, which still have a place among treatment modalities, include: a) no active treatment, b) extraction of the teeth in the involved area, c) contouring of the involved area, and finally d) complete curettage. But Lawrence et al.[9] points out that the extraction of the teeth makes no difference. The role of surgery in this disease is limited and sometimes may aggravate the disease[17]. Since spontaneous regression is reported for many of the cases, surgery is planned only when cosmetic, respiratory, vision or speech problems exist[15, 18].

The use of radiation therapy is universally condemned for the treatment of cherubism since its use may lead to sarcoma formation, osteoradionecrosis or interference in the bone growth and facial development of the child[8, 15].

Our main reason to operate on this patient would be for cosmetic purposes. But after two years of follow-up we did not see any progression of the lesions and, as the patient did not have any concern about his appearance, we did not perform any surgery. We think that if the child has problems about his appearance in the future, we can trim the mandible after regression of the lesions at the age of fourteen.

In conclusion, cherubism is a rarely seen disease of the maxilla and mandible with a clinical presentation of firm and painless, symmetric swelling of the jaws. The radiological appearance of these lesions is quite typical and must be distinguished from other lesions of the jaws. As this disease has a self-limiting nature, there is no need to interfere surgically unless a complication develops such as vision or psychiatric problems, or difficulty in breathing or swallowing.

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