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Cherubism

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Cherubism or familial intraosseous fibrous swelling of the jaws is a disorder characterized by a proliferation of fibrous tissue and variable amounts of giant cells (Bruce *et al.*, 1953; Thoma, 1962).

Although the cause of cherubism is unknown, it is most probably a developmental disturbance of the bone-forming mesenchyme and has an autosomal dominant character with 100 per cent penetrance in males and 50-70 per cent in females.

The affected patient is usually normal at birth and starts to show the stigmata of the disease during the second, but occasionally the first, decade of life.

The mandibular angles are the usual sites of the lesions and their distribution is symmetrical. Maxillary involvement is noted in about two-thirds of cases, resulting in a V-shaped palatal vault. Premature loss of deciduous teeth and a failure of eruption of the permanent ones are common findings.

After a phase of rapid expansion, the jaw swellings tend to remain stationary or progress slowly for the next few years. Regression is said to occur at puberty (Batsakis, 1979).

Functional disability, pain and tenderness are not features of cherubism. Cervical adenopathy, hyperplastic and reactive in nature, is frequently reported and may even regress or disappear with the bony swellings at puberty, thus showing that they probably share a common underlying defect.

Roentgenograms show sharply-defined multilocular areas of radiolucency. The bone cortex is expanded and thinner. The condylar process of the mandible escapes. Developing teeth are often present within the lesions and are markedly displaced.

Case report

A 16-year-old patient (Fig. 1) presented with multiple facial swellings. The condition started at the age of nine years with the insidious appearance of bilateral submandibular swellings. These were followed by other swellings in the face and mandibular regions which gradually increased in size and led to disfigurement. There was no associated trauma, tooth extraction, constitutional disturbance or pain. A younger brother showed similar though less pronounced changes.

Examination revealed multiple bilateral hard swellings affecting the mandible (angle and ramus) and maxillae, especially near the nasal processes. The mandibular swellings were felt subcutaneously as well as intra-orally. The overlying skin and mucosa were normal, and freely mobile; there was no ulceration, discoloration or pigmentation, nor was there any tenderness. The palate was high-arched and there was a moderate degree of trismus.

The parents said that there was considerable delay in eruption of the teeth, which started when the patient was almost six years old, but other milestones such as sitting, walking and talking were normal. At the time of examination the patient had only two teeth in the upper and three in the lower jaw; these were also maldirected.

Bilateral palpable enlarged submandibular lymph nodes were found. The other groups were normal and there was no hepatomegaly or splenomegaly. The extremities and spine and the rest of the skeleton appeared normal. Mentality and sexual development were average.

Biochemical tests revealed an elevated serum calcium (13.5 mg. per cent) and normal serum creatinine, potassium, sodium and alkaline phosphatase.

Radiological studies (Fig. 2) showed multilocular cystic spaces affecting the greater part of the mandible, except for its condyles. The skeleton was normally calcified and multiple unerupted teeth were found in the upper and lower jaws.

Under general anesthesia one of the swellings was explored and its soft tissue contents examined histologically together with one of the submandibular lymph nodes.

Histologically the node showed a non-specific reation. The bone swelling revealed (Fig. 3) a great number of large multinucleated giant cells in a loose delicate fibrillar connective tissue stroma containing large numbers of fibroblasts and many small blood vessels.

Differential Diagnosis

Cherubism presents clinically as an expanding mandibular (and or maxillary) swelling and is characterised histologically by a proliferation of fibrous tissue with variable amounts of giant cell infiltration. Accordingly it should be differentiated

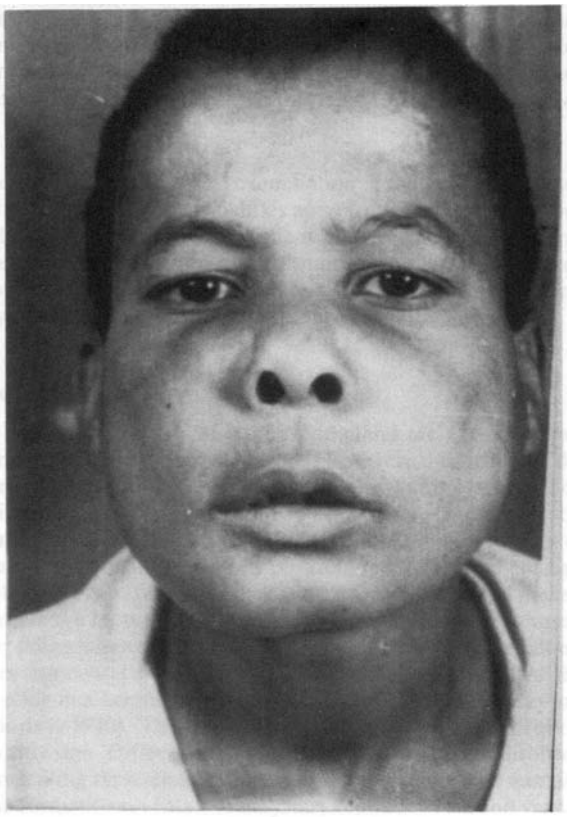


FIG. 1
Cherubism

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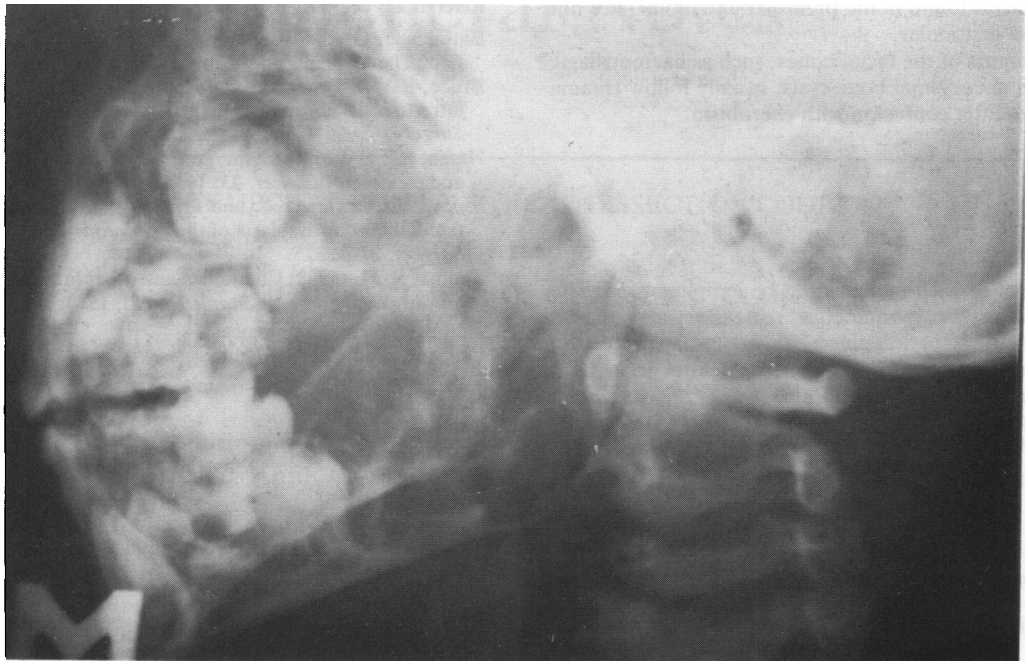


FIG. 2

Roentgenogram showing multiple cystic spaces and unerupted teeth.

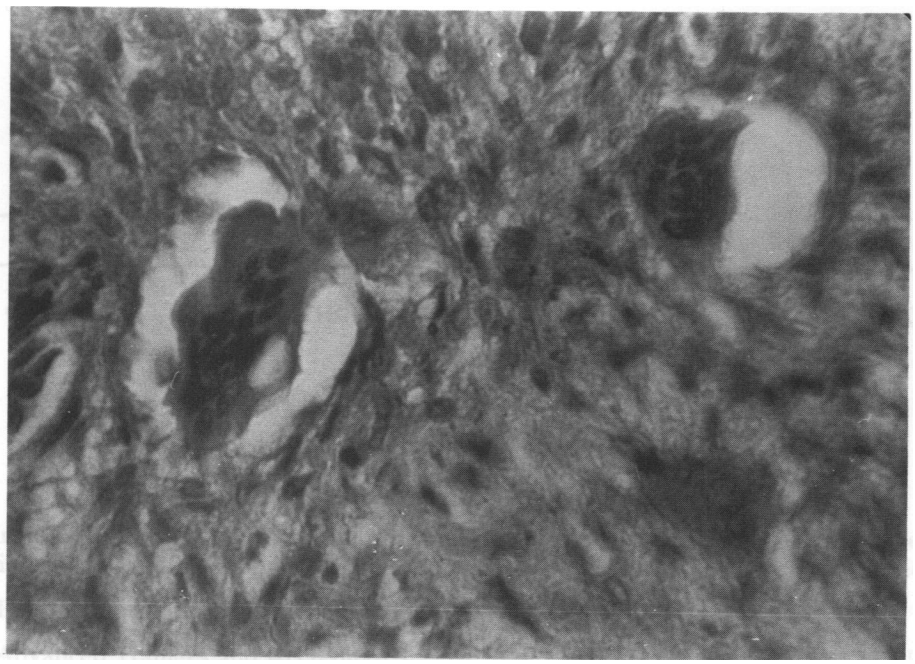


FIG. 3

Multinucleated giant-cells in loose, delicate fibroblastic stroma.

from other lesions which show either of these two features. The clinical, radiological and pathological picture should be evaluated in its entirety before a firm diagnosis is made, since cherubism may simulate one disease in one respect while differing entirely from it in others, e.g. it might be difficult or impossible to differentiate cherubism from giant cell reparative granuloma by microscopic examination alone.

Fibrous dysplasia has a clinical history and age incidence very similar to cherubism but the bone component so well illustrated in the former is entirely lacking in the latter.

The so-called 'brown tumour' of hyperparathyroidism may occur in the mandible or maxilla but the patient is usually older and the biochemical study (serum calcium, phosphorus and alkaline phosphatase) is significantly abnormal and radiographically there is loss of density of the skeleton as a whole. Histologically such brown tumours are composed of spindle cell connective tissue stroma containing numerous osteoclast-like multinucleated cells with free haemorrhages and phagocytosis.

True giant cell tumours of the jaws are extremely rare. The lesion is seldom seen below the age of twenty years; it affects the symphysis and bicuspid region of the mandible, and pain is a dominant feature. Histologically the lesion is dominated by giant cells with numerous nuclei and cellular stroma.

Giant cell reparative granuloma is not unlike cherubism microscopically, though clinically a history of trauma (usually a

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tooth extraction) is found in the premolar and molar sites not posterior to the first molar.

Vascular tumours of the facial bones, such as haemorrhagic bone cysts and aneurysmal bone cysts, usually follow trauma and should give little confusion with cherubism.

Management

The treatment of each patient must be individualized. The policy of waiting for spontaneous regression at puberty, already discussed, is permissible though not satisfactory in every case. If such promised improvement does not occur, as in our present case, curettage is helpful (Hamner and Ketcham, 1969). The use of radiation in any form is now universally condemned.

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