Diagnostic Challenge

The Diagnostic Challenge is submitted by the Canadian Academy of Oral and Maxillofacial Radiology (CAOMR). The challenge consists of the presentation of a radiology case.

Since its inception in 1973, the CAOMR has been the official voice of oral and maxillofacial radiology in Canada. The Academy contributes to organized dentistry on broad issues related to dentistry in general and issues specifically related to radiology. Its members promote excellence in radiology through specialized clinical practice, education and research.

CAOMR Challenge No. 11

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A 19-year-old man presented with bilateral, painless enlargement of the body and angles of the mandible. He had a round face and broad cheeks. Upon examination, the submandibular lymph nodes were enlarged. His father gave a history of having a similar rounded facies himself with bilateral jaw enlargement, which had spontaneously regressed with age.

A pantomograph (**Fig. 1**) showed multilocular radiolucencies in the lower molar regions extending posteriorly into the coronoid processes of the mandible. The maxillary tuberosities were also affected. Multiple impacted and displaced teeth were seen.

Laboratory results showed an increase in the level of the serum alkaline phosphatase.

The patient was followed up for 3 years. At age 22, he presented with superomedial displacement of the right eye and an upward cast of the eyeball, with exposure of the sclera below the pupil. His other facial contours seemed to be normal at this time. A pantomograph (**Fig. 2**) showed signs of new bone formation (healing) in the areas previously occupied by the lesions.

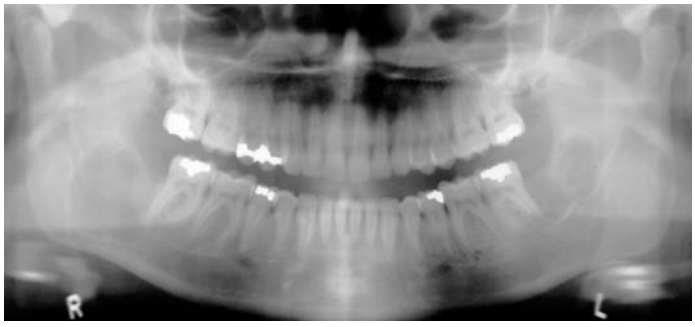

Figure 1: *Pantomograph of 19-year-old patient.*

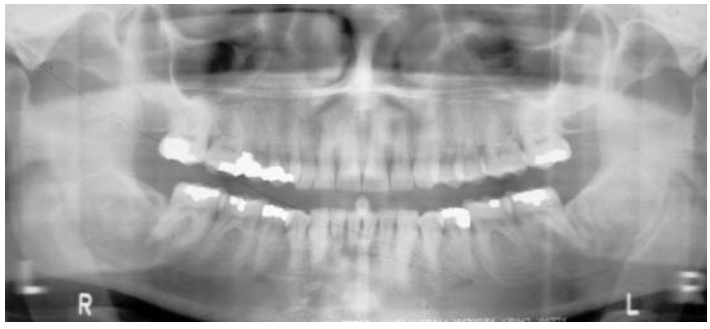

Figure 2: *Pantomograph made when the patient was 22 years old.*

What is your diagnosis?

(See page 670 for answer)

Answer to CAOMR Challenge No. 11

Cherubism is a rare, self-limiting, non-neoplastic bone lesion that primarily affects the jaws of children and young adults bilaterally. It is considered to be hereditary, with an autosomal dominant pattern.[1] The appearance of affected children is normal at birth but swellings of the jaws appear between 2 to 7 years of age. Males are affected twice as often as females. The penetrance is 100% in men and 50% to 70% in women.[2] The gene for the disease has been mapped to chromosome 4p16.3.[3]

This disease was first described by Jones[4] in 1933 in 3 children of a Jewish family. He made the analogy that the children looked similar to the renaissance cherubs and thus, suggested this clinically attractive name.

The mandible is always involved, whereas the involvement of the maxilla is variable. When the latter is involved, the palate may be V-shaped.[5] Displaced, malposed, impacted and unerupted teeth are common findings. Supernumerary and missing teeth can also occur. Premature loss of deciduous teeth and delayed eruption of the permanent teeth have also been reported.

The exposure of the sclera below the irides results in the apparent upward gaze, which has been attributed to elevation of the eye, retraction of the lower lid and loss of lower lid support. The orbital involvement in this disease usually appears late in affected individuals.

Radiographs show bilateral, multilocular, radiolucent areas within the jawbones. The coronoid processes are commonly involved, whereas the condyles are rarely affected.

Seward and Hanky[6] suggested a 3-tier classification for the disease:

- grade 1: bilateral lesions confined to the mandible extending up to the coronoid processes;
- grade 2: the same as grade 1, but with lesions in the maxillary tuberosities as well;
- grade 3: both jaws diffusely affected.

Although histopathologic investigation is not required in most cases to establish the diagnosis, when performed, it reveals osteoclast-like multinucleated giant cells in a moderately loose fibrous stroma with no evidence of neoplastic change.

Cherubism is reported to be associated with some well-described syndromes, including Noonan syndrome, Ramon syndrome, and Jaffe-Campanacci syndrome. The common features of Noonan syndrome include hypertelorism, "webbed" neck, mental retardation, cardiac defects, cryptorchidism and short stature. Ramon syndrome includes gingival fibromatosis, epilepsy, mental retardation and possibly insulin dependent diabetes. Jaffe-Campanacci syndrome is characterized by multiple nonossifying fibromata of the skeleton, café au lait spots, mental retardation, cryptorchidism, hypogonadism, and ocular and cardiovascular abnormalities.

The differential diagnoses for cherubism include giant cell tumour of the jaw, central giant cell lesion, brown tumour of hyperparathyroidism, fibrous dysplasia and aneurysmal bone cyst.[7–9]

The treatment protocol is primarily based on observation and follow-up. Since this disease often regresses spontaneously, surgical intervention may not be necessary other than for cosmetic and functional purposes. In recent years, experimental use of calcitonin in the treatment of cherubism has been described.[10] ◆

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