**Cherubism**

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# Cherubism

## Overview.

The term cherubism refers to the ‘‘chubby-cheeked’’ facial resemblance of angels, in renaissance paintings. Cherubism is an autoinflammatory bone disease where the onset occurs during childhood. It is characterized by bilateral and symmetric proliferative fibroosseuous lesions limited to the mandible and maxilla. It is a rare autosomal dominant bone disorder characterized by symmetrical expansion of the jaws where giant cell lesions replace bone. This disease sows carriable expressivity and the clinical presentation may range from asymptomatic bilateral mandibular/maxillary swelling to deforming life-threatening bone lesions.

## Etiology.

Cherubism is inherited in an autosomal dominant manner. Mutations in the SH3BP2 gene have been identified in about 80% of people with cherubism. In most of the remaining cases, the genetic cause of the condition is unknown. The SH3BP2 protein is particularly important for the function of cells involved in the replacement of old bone tissue with new bone and certain immune system cells. Too much SH3BP2 protein likely increases signaling in certain cells, causing an immune reaction, which is inflammation in the mandible and maxilla, and also triggers the production of osteoclasts, which are cells that break down bone. A combination of bone loss and inflammation likely underlies the cyst-like growth characteristic of cherubism.

**Clinical Presentation.**

The most common symptoms of cherubism are round, swollen-looking cheeks, wide jaw, loose, misplaced, or missing teeth, hypertelorism, maxillary enlargement and lower face swelling. Cherubism occurs between age two and seven years, it is a bilateral, symmetric enlargement of the mandible and/or maxilla including coronoids but usually sparing the condyles. Other cranial bones are usually unaffected. Swelling of mandibular and cervical lymph nodes, slow progression of the jaw lesions up to adolescence, and spontaneous regression typically starting after puberty and extending into the twenties. Upturned tilting of the eyeballs and rim of sclera visible beneath iris. There are certain dental abnormalities such as congenitally missing teeth, and displacement of permanent teeth secondary to the jaw lesions, hence malocclusion.

**Demographic.**

The incidence of cherubism is unknown. Research suggests that among people who have a mutated gene that causes cherubism, 100% of males and 50 to 70% of females display the physical signs of the condition. This means females can be carriers of the mutation but are unaware that they have it. Males cannot be considered carriers because they always show the physical signs of cherubism. A typical form of cherubism is during childhood.

**Biopsy / Histology / Radiographs.**

Some experts consider an incision biopsy sufficient to confirm the diagnosis. However, since cherubic phenotypes can be mimicked by other jaw tumors requiring different therapeutic strategies, thoughtful consideration of histologic analysis is warranted. Biopsy examination of the

lesions disclosed loose cellular fibrous connective tissue containing numerous multinucleated giant cells, fibroblasts, and vesicular nuclei.

Radiographic manifestations include bilateral, multilocular, soap bubbles-like, radiolucent areas within the mandible, usually located at the angles and rami. The coronoid processes are commonly involved, whereas the condyles are rarely affected. Lesions in the mandible are usually symmetric, whereas those in the maxilla may be asymmetric.

**Differential diagnosis.**

Observed radiographic appearance might be confused with other lesions containing giant cells such as hyperparathyroidism, and/or osteomalacia. Regarding the differential diagnosis, it should be emphasized that, whereas central giant cell lesions affect the central portion of the mandibular body and giant cell tumors rarely involve bone of the maxillomandibular complex, in cherubism the lesions are generally bilateral and involve both the maxilla and the mandible.

**Treatment.**

Treatment should be tailored to the individual's presentation and the evolution of the disease. For some people, surgery to remove the tissue is an option. It can help restore the mandible and the maxilla and cheeks to their previous size and shape. For misplaced teeth, orthodontics is an option and for missing or extracted teeth, implants are also an option. Children with cherubism should see their doctor and dentist regularly to monitor symptoms and the progress of this condition.

**Prognosis.**

In general, cherubism has a good prognosis. Cherubism does not progress after puberty, and as the patient grows to adulthood, the entire jawbone lesion tends to develop a more normal configuration. Surgery is not a treatment of choice. But in case of expansion of tissue resulting in difficulty with airway or chewing capacity, biopsy, and surgical intervention can be done. Medical attention for aesthetic and functional concerns is required.

**Professional Relevance.**

It is relevant to have a good knowledge of this condition because as dental hygienists typically we are the first ones of the dental team to see this patient clinically and radiographically. We can refer this patient to a doctor to diagnose this inflammatory bone disease. Because cherubism presents with some kind of malocclusion, it may be more difficult for the patient to reach certain areas in order to have proper dental hygiene, these malocclusions can affect the oral hygiene of the patient. The dental hygienist is an educator as well and besides cleaning those difficult areas, the dental hygienist should advise good techniques fitted to these patients. One of the treatments that cherubism includes could be orthodontic treatment and for a good orthodontic treatment oral hygiene is key.

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