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DEN 2311- Oral Pathology

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Assignment: Cherubism

Etiology

Cherubism is a benign genetic disease where normal bone is replaced by fibrous granuloma containing giant cells and increased osteoclastic activity. It is an autosomal dominant syndrome caused by mutation of the gene SH3BP2 which encodes proteins involved in bone remodeling.

Clinical Presentation

Cherubism is characterized by symmetrical facial deformities due to expansive bone lesions in the maxilla and mandible. Depending on the extension and size of the expansion, these patients may present dysphagia, aesthetic, respiratory and speech dysfunction, as well as impaired vision and hearing. Cervical lymphadenopathy is usually present preceding the intraosseous lesions. Phenotypically, they present with a rounded face and enlarged mandible accompanied with an upward gaze due to elevation and distortion of the orbital socket, which gives them an appearance similar to “Cherubs” (chubby children depicted in renaissance art), hence the name of the condition. Intraoral clinical features include oligodontia, malocclusion, early tooth loss and failure of permanent tooth eruption.

Age and Sex

The condition starts between ages 2-5 and progresses through puberty. Some studies have reported a significant number of cases are expected to regress after puberty. No sex predilection has been reported.

Diagnosis/Lab Tests

Diagnosis of Cherubism consists in a combination of clinical exam, radiographic evidence and genetic testing. The Genetic testing is through analysis of DNA samples where mutation screening is performed, specifically studying the genetic sequence of the SH3BP2 gene through PCR procedures.

Radiographic Presentation

For Radiographic evaluation of patients with Cherubism, Panoramic imaging and CT scan are the best diagnostic tools. Radiographically, Cherubism patients present with uni or multilocular radiolucencies with multiple quadrant involvement primarily on the mandible but it also can be observed in the maxilla. These radiolucencies can present with or without expansion, thinning or perforation of the cortical bone. Some studies describe that in areas where there are no radiolucent lesions, other abnormalities like trabecular sclerosis, irregular outline and thickening of the bone have been observed.

Biopsy

The histopathology of cherubism is characterized by dense fibrous connective tissue with spindle shaped fibroblasts and randomly distributed multinucleated giant cells considered to be osteoclast-like.

Treatment

Because of the different presentation of Cherubism related to size of the lesion, expansion, and overall effect on the patient's quality of life, the treatment tends to be individualized case by case. For more moderate cases, usually the lesions are observed through puberty until they regress. For more advanced cases where deformity is the main concern, removal of the granulomas and surgical recontouring of the mandible is the approach of choice. The surgical management is preferred between the ages of 5-15. Medical management with drugs is still under research. The goal of the medical management is to reduce osteoclastic activity and improve bone remodeling. Drugs like Salmon Calcitonin intranasal and TNF-alpha antagonists have been tried with variable results. Additionally, a recent study from Oral Surgeons in Florida, has also experimented with Imatinib, a tyrosine kinase inhibitor (TKI) to manage the growth of the giant cell granulomas with positive results.

Prognosis

Due to this condition being very rare, there are no strong statistics about a definite prognosis. However, since several studies have reported that Cherubism does not progress past puberty, it has been determined that it does not have a poor prognosis. Individual prognosis will depend on size of granulomas, rate of expansion, deformities and effect on quality of life.

Dental Hygiene Care

The Dental management of patients with cherubism should be done with a multidisciplinary approach. Orthodontic treatment most likely will be required to avoid permanent damages in the dentition due to malocclusions, unerupted teeth and early tooth loss. The Dental Hygienist role would involve effective and efficient biofilm control to avoid periodontal problems which could be linked to the orthodontic implications. For those patients with bigger deformities, the dental hygienist should make sure that the patient and/or their primary caregiver is using the appropriate techniques and products for oral hygiene at home through education and motivation techniques. For patients with severe misalignment, malocclusions and fixed orthodontic appliances, interdental hygiene is crucial in order to avoid periodontal disease on a patient whose bone remodeling is already affected.

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