Orthodontic management of a patient with cherubism: A case report

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Introduction: The aim of this case report was to present the successful orthodontic treatment of an adolescent girl with cherubism. **Methods:** The patient began treatment after puberty. Necessary extractions were performed, and she had full-arch treatment of the maxillary and mandibular anterior teeth. A series of archwires was used in a timely manner to create an adequate arch form and close the interdental spaces. Once treatment was completed, retention was ensured with a maxillary spring aligner and mandibular fixed retainer. **Results:** Maxillary alignment and an adequate arch form were achieved, and mandibular treatment was limited to the anterior segment. The patient finished treatment with an esthetically pleasing smile. **Conclusions:** There have been few reports of orthodontic treatment for a patient with cherubism. This case report provides evidence that complete or limited orthodontic treatment can be provided to these patients to improve facial esthetics and bolster their self-esteem. (Am J Orthod Dentofacial Orthop 2018;154:433-41)

Cherubism is a rare fibro-osseous disorder with about 300 reported cases in the literature.[1] Dr W. A. Jones described the disease in 1933 as a familial multilocular cystic disease of the jaws.[2] The name was given because of the facial characteristics, which are reminiscent of the cherubs portrayed in Renaissance art, with a chubby face and upward directed look.[3,4] Later studies found that cherubism is an autosomal dominant disease of the maxilla and mandible in which bone is replaced with excessive amounts of fibrous tissue, causing premature loss of deciduous teeth and impaction of succedaneous teeth.[2,5] There have been cases where the disease can be treated with local curettage of the lesions and surgical recontouring of the jaws. Little has been reported about the orthodontic treatment of patients with cherubism. The purpose of this case report was to present the diagnosis and orthodontic treatment of a patient with cherubism.

The general appearance of a patient with cherubism consists of bilateral mandibular expansion with enlargement of the cheeks.[6] This is a nonneoplastic, self-limiting, fibro-osseous disorder that affects the mandible and can also include the maxilla.[2,3] Lesions tend to occur twice as often in the mandible as in the maxilla. Symptoms of the disorder may not be apparent in infancy but begin to appear as early as 14 months to 3 or 4 years of age.[6,7] Enlargement of the mandible or maxilla can rapidly progress until middle childhood and then begin spontaneously to regress at puberty. Maxillary lesions are the first to regress, whereas mandibular lesions may remain active into adulthood.[8] The general appearance may return to almost normal with decreases in clinical swelling by the patient's third or fourth decade.[6,7] Cherubism also has an orbital component; the upward gaze with exposure of the sclera below the pupil is caused by elevation of the infraorbital rim and orbital floor and stretching of the facial skin.[3,5,9]

Radiographically, lesions appear as cystic multilocular radiolucencies that tend to begin near the mandibular angle, and spread to the ramus and body and the anterior ends of the ribs.[6,8] The bony expansion is painless, bilateral, and usually symmetrical. In severe cases, most of the mandible is involved. Maxillary lesions may occur at the same time but are more difficult to identify due to overlap with the maxillary sinus and nasal cavity. The extensive bone involvement causes a marked widening and distortion of the alveolar ridges. Destruction of the basal and alveolar bones causes ectopic, unerupted teeth to be involved with the lesions, giving a radiographic "floating tooth" appearance. In addition to the esthetic and psychological effects, the enlargements cause tooth

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displacement or failure of eruption, impair mastication, and create speech difficulties.

Generally, treatment of cherubism consists of biopsying the lesion, extracting any ectopic or unerupted teeth, and correcting the deformities surgically. Timing of surgical correction is difficult to determine. Excellent results have been obtained by early surgical intervention with curettage of the lesions. On the contrary, other studies have shown that early surgical interventions are followed by rapid regrowth of the lesions and worsening deformity.[2] Other treatment options consist of medical therapy with calcitonin or cosmetic osteoplasty after regression of the disease or in case of functional impairment.[10] More often than not, studies have proven that no treatment is also an option in which surgeons wait for stabilization and spontaneous remission, usually in the third decade of life. Orthodontic treatment is commonly required because the jaw distortion caused by the lesions leads to permanent dental abnormalities including premature loss of deciduous teeth and widely spaced, misplaced, unerupted, or absent succedaneous teeth, causing malocclusion.[11] The appearance of a patient with cherubism may be traumatic psychologically and socially; therefore, treatment should always be considered.

This rare developmental jaw condition is generally inherited as an autosomal dominant trait; several studies reported 70% penetrance in female patients and 90% in male patients.[7,8] Cherubism is caused by a mutation in gene *SH3Bp2*, from chromosome 4p16.3.[5,8] Ueki et al[12] found that all mutations reported were in exon 9 and affected 3 amino acids within a 6-amino acid sequence (RSPPDG) located in 31 to 36 amino acids. Although it is called an autosominal dominant inheritance with incompleted inheritance based on sex, it is interesting that haploinsufficiency of *SH3Bp2* in persons with that syndrome does not result in cherubism or cherubism-like characteristics. This finding and the clustering of amino acid missense mutations in *SH3Bp2* support the hypothesis that the mutations in *SH3Bp2* lead to a gain of function or act in a dominant-negative manner. The onset of the abnormalities of cherubism and their organ-restricted characteristics may be related to dental developmental processes in children, when signals unique to the mandible and maxilla are transmitted through the extracellular matrix, triggered by the eruption of secondary teeth.[6,12]

An accurate diagnosis of cherubism is based on clinical, histologic and radiographic findings.[4] Differential diagnosis of cherubism includes nevoid basal cell carcinoma syndrome, infantile cortical hyperostosis, hyperparathyroidism, odontogenic keratocyst, ameloblastoma, central giant cell granuloma, odontogenic myxoma, Noonan-like syndrome, and Ramon syndrome.[10,13]

Fig 1. Family portrait: patient in the center and her mother is second from the right (in hat).

DIAGNOSIS AND ETIOLOGY

A 7-year-old Hispanic girl came with her mother for an initial examination. The patient was initially diagnosed with cherubism at the age of two. Family history showed that the mother (Fig 1) has cherubism with the mandibular expansion resolved. A maternal first cousin (Figs 2 and 3) was also diagnosed with cherubism and showed clinical and radiographic characteristics of the disorder. The patient was placed on yearly recall at the pediatric dentistry division of Bronx-Lebanon Hospital Center and eventually referred for an orthodontic consultation and necessary treatment. At age 12, full orthodontic records were taken (Figs 4-6). Genetic testing was not completed.

At age 12, the patient had convex facial and Class II skeletal patterns with bilateral asymmetric mandibular expansion. The left mandibular ramus showed more expansion compared with the right side. The patient tested negative for pain upon palpation. An intraoral examination showed that she was in adult dentition with an unerupted maxillary right canine, lingual displacement of maxillary left second premolar, and buccal displacement of maxillary left first molar. The mandible showed ectopic eruption of the left first molar, severe rotation of the premolars, impaction of the canines, and an unerupted right first molar. Both arches had generalized spacing caused by many missing teeth. The incisor and molar relationships could not be determined. Overjet was 4 mm, and overbite was 90%. Overall, the patient's oral hygiene was fair.

The panoramic radiograph (Fig 5) showed significant multilocular radiolucienes throughout the entire mandible with multiple unerupted teeth giving a floating-tooth appearance. Supernumerary and impacted teeth were observed bilaterally in