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Case report

The surgical and orthodontic management of cherubism in a growing child☆

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ABSTRACT

Cherubism is a rare non-neoplastic disease that affects the jaws causing significant destruction and disfigurement. The disease also affects the normal eruption, occlusion and function of the dento-alveolar complex. Cherubism may radiographically and histologically resemble central giant cell granuloma, fibrous dysplasia and other giant cell lesions. The case of a 15-year 4-month-old girl with no obvious facial swelling or signs of cherubism is described in this report. The patient presented with a dental malocclusion that included the ectopic eruption and displacement of teeth caused by the lesion. The radiographic findings derived from Cone Beam Computed Tomography (CBCT) technology are described. This case report details the orthodontic and surgical diagnosis and management of a teenager during the post growth period.

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1. Introduction

Cherubism is a rare non-neoplastic disease that affects the jaws causing significant destruction and disfigurement. It was first described by Jones (1933) as "familial multilocular cystic disease of the jaws with the eyes looking towards heaven appearance". Thus the term cherubism was used to describe the faces with bilateral swollen cheeks resembling the cherubs of Renaissance art. Cherubism is a rare hereditary benign bone disease with an autosomal dominant inheritance. Its familial distribution may affect different generations (Peters, 1979; Ongole et al., 2003), even though isolated non-familial cases have also been reported (Meng et al., 2005; Ozkan et al., 2003; Jain et al., 2007). Cherubism is often characterized by bilateral expansion of the mandible and/or the maxilla which becomes noticeable in early childhood and grows progressively until puberty, after which it shows partial or complete spontaneous remission (Jones et al., 1952; Peters, 1979, Valiathan and Prashanth, 1997; Lannon and Earley, 2001). Cherubism may range from undetectable clinical cases to aggressive ones that extend to the eyes, having significant orbital manifestations that may cause proptosis, looking towards heaven appearance and loss of vision (Hawes, 1989; Raposo-Amaral et al., 2007). One extreme reported case progressed rapidly resulting in death due to gastrointestinal and pulmonary infections (Silva et al., 2002). The severity of cherubism is graded using the Motamedi classification system (Kalantar Motamedi, 1998) as it incorporates both anatomic involvement and aggressiveness of the disease. Symptoms range from no clinical or radiographic detectable features to gross deformity of the mandible and maxilla, airway obstruction or impairment, impaired vision and hearing (Hawes, 1989).

1.1. Facial and oral manifestations

In classic cherubism the normal bone is replaced by fibrous tissue causing expansion of the affected site and its replacement with immature bone. The mandible is most commonly affected bilaterally at the body and the ramus region (Yamaguchi et al., 1999, Kozakiewicz et al., 2001; Schultze-Mosgau, 2003; Beaman et al., 2004; Jain et al., 2007, Penarrocha et al., 2006; Silva et al., 2007), however unilateral involvement has also been described (Reade et al., 1984; Meng et al., 2005; Ozkan et al., 2003). The condyles are usually spared but condylar involvement has been reported (Beaman et al., 2004; Jain et al., 2007), maxillary involvement is less frequent and usually less extensive. Dental complications of cherubism include arch and dental abnormalities, delayed eruption, narrow V-shaped palatal arch, root resorption, malalignment, displacement and impaction of teeth (Yamaguchi et al., 1999). Frequently, ectopic unerupted teeth are involved by the lesion (Pontes et al., 2007; Kalantar Motamedi, 1998). Thus the occlusion is affected functionally and esthetically.

1.2. Diagnostic tests

Cherubism has a characteristic radiographic pattern of expansile bone remodelling, with a diffuse bilateral multilocular nature

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(Yamaguchi et al., 1999; Ozkan et al., 2003; Beaman et al., 2004; Jain et al., 2007, Penarrocha et al., 2006). Modern imaging has made it easier to visualize the true extent of the lesion. Computed Tomography (CT) provides a clear delineation of the disease extent in cherubism, which is often not possible on plain films due to overlapping of facial bones. Cherubism appears as multilocular cystic lesions bilaterally affecting the maxilla and mandible on CT scans and plain radiographs (Carvalho Silva et al., 2007). Magnetic Resonance Imaging (MRI) is used to assess the orbital involvement of the lesion when the patient is developing progressive proptosis (Jain et al., 2007). Cone Beam CT (CBCT) has been recently become available as a low radiation imaging modality for the head and neck region. It has been widely used in maxillofacial surgery and orthodontics (Kau et al., 2005; Palomo et al., 2006). Cone Beam CT has the advantage of showing fine detail and the extent of the lesion accurately along with the affected adjacent structures. Three dimensional models can be constructed to visualize the degree of expansion and destruction of bone. CBCT could also be used to monitor the disease process, since it delivers lower radiation doses and superior images compared to traditional CT.

Cherubism was previously considered as a subtype of fibrous dysplasia because of the common radiographic and histological resemblance. Histologically, cherubism resembles central giant cell granluloma, hyperparathyroidism and giant cell tumour with numerous randomly distributed multinucleated giant cells and vascular spaces within a fibrous connective tissue stroma. However, it was found that cherubism results from mutations on the SH3BP2 gene on chromosome 4p16.3 (Hyckel et al., 2005; Tiziani et al., 1999), therefore it is now classified as an independent entity. Cherubism has also been associated with Noonan and other syndromes (Wolvius et al., 2006; Dunlap et al., 1989). Bone markers such as phosphorus, serum calcium, and alkaline phosphatase are usually within the normal levels with respect to age. Therefore, blood chemistry analysis is performed to determine the level of calcium and phosphate to help exclude other bone diseases.

1.3. Management

The management of the disease varies considerably according to the extent, aggressiveness and clinical behaviour. Conservative treatment is the most common form of treatment of cherubism unless the lesion is causing serious disfigurement and expansion that necessitates some intervention. Surgical or medical treatment with calcitonin or interferon has been suggested (Lannon and Earley, 2001; de Lange et al., 2007).

Until now the use of Cone Beam CT has not been described as an imaging modality for cherubism, and orthodontic treatment in a “cherubic” child has not been described in detail in the literature (Hyckel et al., 2005; Morley and Stoneman 1984).

2. Case report

In this case report, we discuss the use of CBCT in defining the extent of the disease in a patient with a unique presentation of cherubism, not showing any obvious facial deformity or swelling. Interestingly, the typical features of cherubism were visible on the Cone Beam CT. The displacement, ectopic eruption and misalignment of teeth were also seen. The patient underwent orthodontic treatment during the disease process to correct the malocclusion and reposition an impacted molar.

2.1. Facial and oral manifestations

A white15-year 4-month-old girl was referred to the department of Oral and Maxillofacial Surgery University of Texas Houston Science Centre in November 2006 with a lesion affecting the mandible bilaterally. The patient was asymptomatic with no obvious facial swelling or expansion in the mandible, no history of trauma, infections, numbness, or altered sensation to the face. There was no family history of hereditary disease. Intra-oral examination revealed missing 2nd permanent molars bilaterally with crowding of the dentition evident in the upper and lower arches. The patient was concerned with the appearance and alignment of her dentition and wanted to improve her smile and appearance (Fig. 1).

2.2. Diagnostic tests

A panorex radiograph was taken and showed two large bilateral multilocular lesions in the mandible extending from the body to the ramus associated with an impacted 3rd molar on the right side and impacted 2nd and 3rd molars on the left side. The lower left 2nd molar (LL7) was displaced by the lesion to the lower border of the mandible, and the impacted lower left 3rd molar (LL8) was undergoing ectopic eruption above the LL7. The patient also had a CT scan to better visualize the extent of the lesion. A differential diagnosis of cherubism, hyperthyroidism, and Gorlin’s syndrome was suggested. Arteriovenous malformation was ruled out by CT with contrast. Serum calcium and parathyroid hormone levels were within normal range. A further CBCT was taken with the Sirona Galileos machine (Sirona Dental Systems), demonstrating the precise extent of the lesion and the involved anatomic structures along with the degree of destruction. It also showed the exact relationship of the lesion to the impacted teeth and made it possible to evaluate and plan the surgical extraction and assess the possible treatment options for the patient (26) (Fig. 2). The relationship of the impacted teeth to the occlusal plane and lower border of the mandible was assessed in all three planes with the aid and manipulation of the Galileos Galaxis 3D Software (Fig. 3). Incisional biopsy was taken under general anaesthesia. The pathology report was of a central giant cell granuloma with prevascular eosinophilic cuffing, consistent with cherubism. Hyperparathyroidism was excluded by the blood chemistry analysis performed showing calcium within its normal limits. Fibrous dysplasia was also ruled out by the presence of lamina dura around teeth and absence of the classical “ground glass” appearance radiographically. The bilateral distribution of the lesion, radiographic findings with histological support are consistent with a diagnosis of cherubism. The subject’s brother was screened using a panorex as a precaution to exclude familial cherubism. The panorex showed no abnormalities.

2.3. Management

Interferon treatment was started at M.D Anderson Cancer Centre to abort the giant cell process and possibly reduce the deformation of the mandible. An orthodontic consultation was requested regarding the lower left molars and their position relative to the dental arch.

Orthodontic examination revealed that the patient had dental Class II Division 1 malocclusion with irregularities to alignment of the dentition on a mild skeletal 2 base. The orthodontist (C.H.K.) and oral-maxillofacial surgeon (M.E.W.) planned for the extraction of the mandibular left third molar (LL8) and the alignment of the mandibular left second molar (LL7) using a gold chain attached to a bracket. Surgical removal of all impacted third molars with a secondary biopsy from the lesion to confirm the diagnosis and monitor progress was planned and carried out under general anaesthesia. The LL7 was surgically exposed by the