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Case Report

Comprehensive surgical management of cherubism with orbital involvement

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ABSTRACT

Cherubism is a hereditary childhood disease of autosomal dominant inheritance. It is more common in males. Its main clinical characteristic is a painless symmetrical swelling of the jaws with polycystic destruction of the bone structure and dental anomalies causing malocclusion. It is a self-limited disease, and some cases of spontaneous regression have been reported. Two clinical cases with remarkable orbital involvement are presented and the surgical management is described. Preprosthetic surgery and dental rehabilitation with osseointegrated implants was also carried out for the first patient. Surgery resulted in a significantly improved functional and aesthetic outcome for both patients, with no complications and a stable clinical situation on follow-up. Cherubism is a rare, non-neoplastic disease affecting the jaws but also the orbits. Facial deformity, malocclusion and visual impairment set the need for a multidisciplinary team approach in order to get successful outcomes in means of function and aesthetics.

1. Introduction

Cherubism is a rare hereditary childhood disease of autosomal dominant inheritance. It is more common in males. The penetrance lies at 80%, with variable expressivity. Its main clinical characteristic is a painless symmetrical swelling of the jaws with polycystic destruction of the bone structure. Complications include delayed dentition, dental root resorption, and malaligned and impacted teeth. It is a self-limited disease, and some cases of spontaneous regression have been reported. Approximately 250 cases of cherubism have been reported, but, to the best of our knowledge, only 11 papers describe an orbital involvement [1–11] and only 4 [12–15] on rehabilitation with dental implants.

2. Case reports

2.1. Case 1

An 18-year-old woman was initially examined in our department in 1986. At age 3, a progressive enlargement of her face with eye protrusion was noticed. Between ages 7 and 16 she underwent surgery 4 times in another institution. Physical examination revealed important exorbitism with superior displacement of the eyes (Fig. 1). The midface was enlarged transversally and vertically with malar hyperplasia. Several teeth were missing. The hard palate was flat and widened. CT scan showed two multilocular masses occupying the retroocular space bilaterally, causing exophthalmos (Fig. 2). The orthopantomography showed absence of many teeth, especially in the maxilla. Multilocular lytic lesions in the mandible were also apparent. Ophthalmological examination showed visual acuity of 0.2 OD, 0.4 OS. Ocular motility was normal except for mild restriction of supraduction and diplopia in that position. Two years later her visual acuity had worsened to 0.1 OD, 0.4 OS so she underwent surgery. In November 1988 a coronal and Weber–Ferguson–Lynch incision was made and a frontoparietal craniectomy was performed including the roof of the orbits bilaterally. The maxillary sinus was expanded and two

☆ Asian AOMS: Asian Association of Oral and Maxillofacial Surgeons; ASOMP: Asian Society of Oral and Maxillofacial Pathology; JSOP: Japanese Society of Oral Pathology; JSOMS: Japanese Society of Oral and Maxillofacial Surgeons; JSOM: Japanese Society of Oral Medicine; JAMI: Japanese Academy of Maxillofacial Implants.

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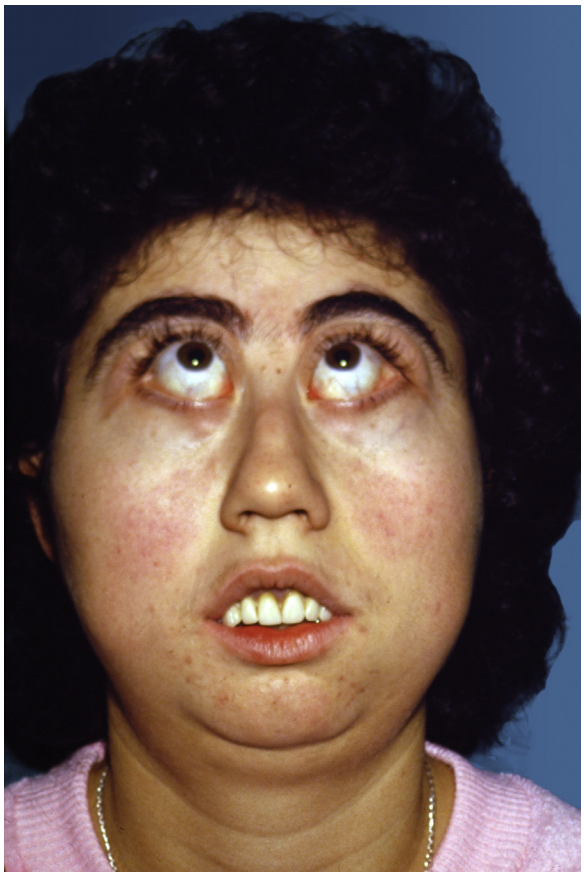

Fig. 1. Patient 1: preoperative aspect at 18 years of age.

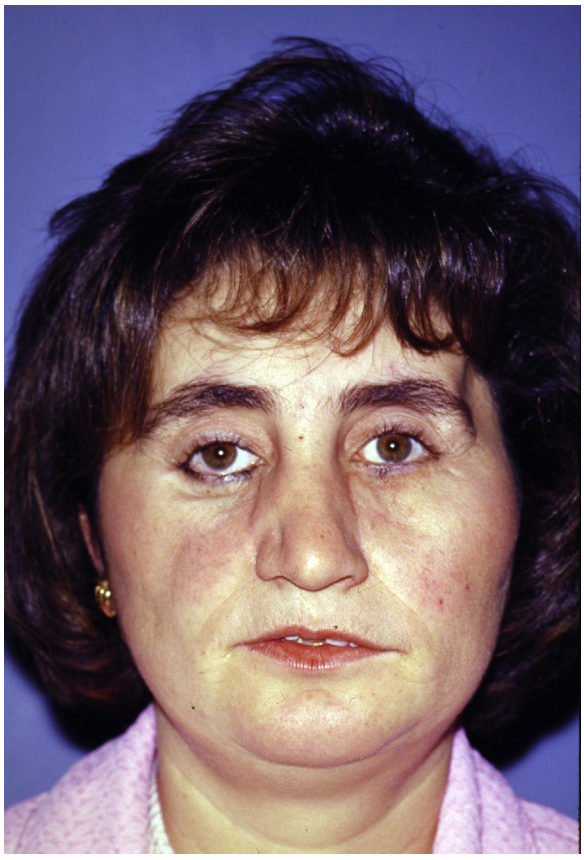

Fig. 3. Patient 1: postoperative clinical outcome.

fibroosseous masses were found in the infero-lateral walls of the orbits and pterigomaxilar and tuberositary regions. Osteotomies were performed on both malar bones medial to the masseter muscle insertion, the base of the nasal septum, from the internal infraorbital angle to the piriform notch, and the pterygomaxillary junction. The osteotomies on the external orbital rim and floor were performed via a subciliary approach; both infraorbital nerves were sectioned. The maxillomalar monoblock was removed. On the table, the 4 remaining premolars were extracted and the whole bone was remodelled. The reshaped monoblock was immediately reimplanted, and wire osteosynthesis was performed. A chondrocostal graft was used for reconstruction of both floors of the orbits (Fig. 3). A rhinoplasty was performed in a second stage. The pathology report showed fibrous proliferation with giant cells, which was diagnostic for cherubism.

The postoperative ophthalmological examination showed improvement of visual acuity (0.8 OD, 0.6 OS) with no diplopia.

The patient was referred to the genetics department for counselling.

The patient consulted 20 years later for cosmetic improvement, so a mentoplasty and lipofilling were performed and dental rehabilitation with bilateral sinus lift and osseointegrated implants was carried out (Figs. 4–6).

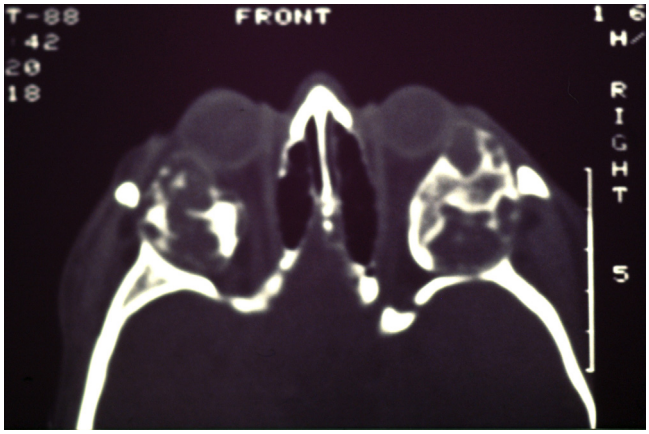

Fig. 2. Preoperative axial CT scan demonstrating orbital involvement and proptosis of the globes.

2.2. Case 2

A 28-year-old woman presented to our department in 2013. She had noticed mandibular enlargement since 2008 with progressive exophthalmos and decrease of visual acuity especially for the left eye. She had a luxation of the left globe a year ago. Examination showed a visual acuity of 0.6 OD, 0.1 OS, diplopia in dextroversion and exophthalmos of the left eye (Fig. 7). Two multilocular masses thinning, and expanding the cortical border of the maxilla, were found in the CT scan (Fig. 8). The retroocular space was occupied causing medial displacement of the vascular structures, muscles and optic nerves, and exophthalmos. Expanding multilocular lytic lesions remodelling and thinning the cortical border of the mandible were also found, located centrally in the mandible and extending to both bodies. In December 2013 she underwent surgery. A bilateral transconjuntival approach with canthotomy and cantholysis was performed, the infraorbital nerve was isolated and an osteotomy of the dysplastic bone in the orbital floor was made. A titanium mesh was used for reconstruction of the orbital floor. The dysplastic bone in the maxillary sinus was also excised.