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CASE REPORT
Craniofacial/Pediatric

Severe Cherubism Treated with Curettage, Osteotomy, and Bony Repositioning: A Case Series of Three Patients

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Summary: Cherubism is a rare, autosomal dominant condition characterized by the replacement of medullary bone by fibro-osseous lesions, predominantly in the bilateral maxillae and/or mandibles. The clinical presentation of cherubism can vary widely, from clinically undetectable to severe facial disfigurement. Although there are no established management guidelines for this condition, conservative management with observation is typically favored in most cases due to the possibility of spontaneous regression following puberty. In this article, we present three cases of moderate to severe cherubism managed with early surgical intervention utilizing curettage and osteotomy followed by bony repositioning. We aimed to show the feasibility and safety of this minimally invasive surgical technique in the management of moderate to severe cases of cherubism to provide improvement in patient quality of life, aesthetics, and function while also possibly mitigating the need for later reconstructive surgery. *(Plast Reconstr Surg Glob Open 2022;10:e4079; doi: 10.1097/GOX.0000000000004079; Published online 27 January 2022.)*

Cherubism refers to a pathologic condition in which medullary bone is replaced by fibro-osseous lesions, predominantly in the bilateral maxillae and/or mandibles, and rarely the orbits.[1] The condition is most often inherited in an autosomal dominant fashion with variable penetrance and is attributed to genetic alterations in the SH3BP2 gene responsible for osteoclast and osteoblast regulation.[2] Onset of cherubism occurs in the first few years of life, but clinical presentation can vary in severity and speed of progression. Although the presentation is often alarming for a cancerous disease process, it is nonneoplastic in nature and most cases spontaneously regress following puberty.[1]

Diagnosis typically relies on clinical, genetic, radiographic, and histopathologic assessment. Orthopantomogram or CT scan usually show nonspecific bilateral multilocular radiolucent areas in the affected regions, and histopathology shows multinucleated osteoclast-like giant cells within the soft-tissue stroma.[3] Current recommended management predominantly centers on a conservative approach with observation through late puberty for hopeful spontaneous regression. Additionally, pharmacologic treatment using calcitonin, bisphosphonates, calcineurin inhibitors, and imatinib have been described with mixed success.[4–6] In severe cases, early surgical intervention can be considered with the aim of improving patient quality of life and appearance.[7] Given the emphasis on conservative treatment, there is a paucity of literature detailing operative management strategies. The aim of the study was to describe utilizing curettage and osteotomy followed by bony repositioning for the treatment of moderate to severe cherubism in a series of three patients.[8]

SURGICAL TECHNIQUE

In all cases, under general anesthesia, the mandible was approached through lower buccal sulcus incisions. The tumor was encountered, as it had eroded through the bony cortex at the level of the mandibular angle. The friable dysplastic bone was easily curetted from the surrounding normal bony cortex. The inferior alveolar nerve was visualized and preserved from iatrogenic injury, as

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well intermedullary molars. After tumor excision, the displaced buccal mandibular cortex was osteotomized with a piezoelectric bone saw (DePuy Synthes). Anterior, inferior, and posterior osteotomies were performed, protecting the surrounding soft tissue that remained attached to the buccal mandibular cortex through intact periosteum. The mobilized buccal mandibular cortex was then infractured and medialized, obliterating the enlarged intramedullary space. The procedure was replicated on the contralateral side.

The maxilla was approached through an upper buccal sulcus incision on each side. The tumor was identified, eroding through the anterior wall of the maxillary sinus. The tumor was removed in a similar piecemeal fashion by curettage. Orbital osteotomies were not necessary. The pliable orbital floor was allowed to reposition spontaneously under the weight of the orbital contents. No fixation devices were utilized.

CASES

Case 1

A healthy 3-year-old girl with a history of paternal cherubism presented with progressive enlargement of the mandible. On physical examination, the body and ramus of the mandible were involved, leading to significant facial deformity (Fig. 1A). CT scan showed bilateral mandibular and maxillary lucent bony lesions with expansion and thinning of the cortical bone (Fig. 1B). The most affected regions of the mandible included the ramus, angle, and the posterior body. Because of the severity of the clinical presentation and CT scan findings, operative correction was undertaken.

The patient had an uneventful postoperative course, and she was most recently seen in clinic 19 months postoperative with no significant recurrence or disease-associated tooth loss. Her incisions are well healed, and she is functionally and aesthetically much improved from her initial visit (Fig. 2A). Postoperative CT scan also showed retained structural improvement (Fig. 2B).

Case 2

A healthy 4-year-old boy with a family history of maternal cherubism presented with progressive enlargement of the maxilla and the mandible accompanied by worsening difficulty with chewing and speech. Physical examination revealed severe disfigurement from bilateral maxillary and mandibular expanding lesions with stark malocclusion. The patient also exhibited an upward gaze, demonstrating evidence of impingement on the orbital floors. The patient underwent a CT scan for further assessment and preoperative planning, which demonstrated expansile enlargement of the mandible and maxilla by innumerable thin-walled lucent cystic structures. (**See figure, Supplemental Digital Content 1**, which displays a preoperative CT scan showcasing a severe case of cherubism requiring surgical intervention. **http://links.lww.com/PRSGO/B905**.) There were also areas of cortical disruption without evidence of malignant features. Because of the functional impairments and rapidly progressive nature of his condition, the patient was taken to the operating room for curettage and osteotomy followed by bony repositioning. During the operation, he had one permanent floating tooth encased in tumor which had to be removed. (**See figure, Supplemental Digital Content 1**, **http://links.lww.com/PRSGO/B905**.)

The patient had an uneventful postoperative course. At his most recent 16-month postoperative follow-up, the patient continues to be aesthetically improved but not fully corrected from his initial presentation with improvement in postoperative CT scan findings. Overall, he has improved facial contour and continues to be free of gaze limitations.

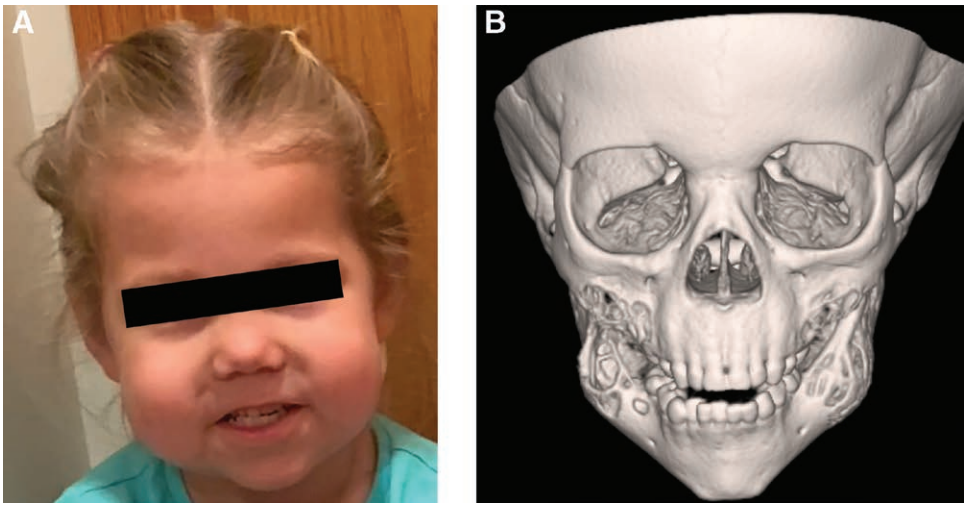

Fig. 1. Preoperative imaging. A, Preoperative appearance. B, Preoperative facial CT scan. The image shows bilateral mandibular and maxillary lucent bony lesions with expansion of the bone and thinning of the cortex.

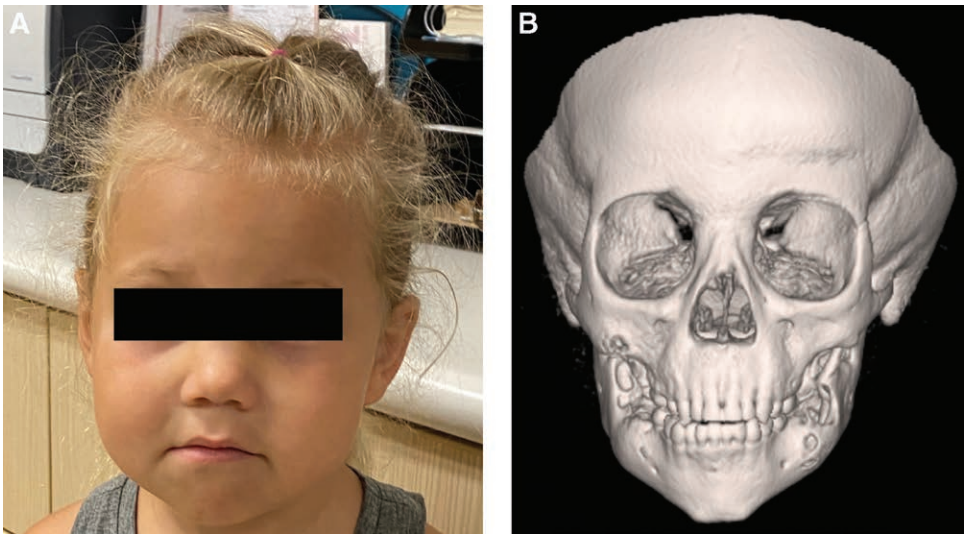

Fig. 2. Postoperative imaging. A, One-year postoperative appearance with improved cosmesis. B, Postoperative facial CT scan. The image shows improvement in bony contour.

Case 3

A 4-year-old patient with a family history of paternal cherubism presented with phonation difficulties and progressive cheek swelling. On clinical examination he had significant malocclusion and a narrowed palate. CT scan showed tumor burden displacing his mandibular cortexes and dentition. Because of his functional and psychosocial impairment, surgical management was pursued. Further details of this case have been published previously.[8]

He most recently followed up at 36 months postoperative. He continues to be recurrence-free without clinical features of his cherubism diagnosis with all his teeth retained.

DISCUSSION

There are many aspects to consider when pursuing surgical intervention in cases of cherubism.[9] Early surgical intervention may potentially prevent long-term sequalae. The removal of tumor may limit the overall disease process, which in turn may prevent permanent tooth loss and normalize facial aesthetics through their formative and consequent years. There are also reports of patients experiencing decreased ocular mobility after the regression phase, which in theory could be mitigated by early surgical intervention.[10] In our case series, all patients showed improvement in cosmesis at follow-up visits with decreased tumor burden.

A particular aspect that needs consideration when pursuing early surgical intervention is dental involvement. Dental involvement varies with the severity of the disease state, though it is common to encounter tooth roots with some absorption, underdevelopment of secondary dentition or agenesis in any case of cherubism. Even in more mild cases where medical therapy is pursued, tooth loss is almost universal.[7] In severe disease cases, even after the regression phase, extraction may become necessary in the cases of free-floating teeth. As such, a conservative approach may be detrimental for dental preservation.

Importantly, earlier intervention likely provides psychosocial benefits to children with facial deformities in their formative years. In one study, anxiety and depression were increased in children with craniofacial anomalies in the age range of 8–10 years when compared with children aged 14–17 years.[11] Early surgical intervention grants these children with disfiguring cherubism the possibility of normalcy.

From a technical standpoint, the piezoelectric bone cutter allows for a safer approach to the surrounding soft tissue when creating osteotomies. Our technique including curettage of the tumor, osteotomy of the cortical bone, and mandibular cortical repositioning restores the mandibular anatomy without damaging the deciduous teeth, permanent teeth, arteries, nerves, or buccal soft tissue.

CONCLUSION

Early surgical intervention with curettage and osteotomy followed by bony repositioning is a relatively minimally invasive and safe technique that can be considered for management of moderate to severe cases of cherubism.

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One patient case has been published in detail previously: Garlick JW, Willis RN, Donato DP, Gociman B. Cherubism in a 4-year-old boy managed with tumor curettage, mandibular osteotomies and repositioning. Plastic and Aesthetic Research. *2018;5(8):29. doi:10.20517/2347-9264.2018.36.*

PATIENT CONSENT

Parents or guardians provided written consent for the use of patient's images.

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