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CASE REPORT

Cherubism: early surgical approach and its effects on the quality of life in a pediatric patient

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Abstract:

Cherubism is a rare fibro-osseous osteodysplasia characterized by the symmetrical expansion of the jaws, often leading to facial deformities and functional impairment. This study presents the case of a pediatric patient diagnosed with cherubism who underwent early surgical intervention to improve quality of life and reduce the psychosocial impacts associated with the disease. The PedsQL™ 4.0 questionnaire was applied to assess quality of life before and after the surgical procedure. The results demonstrated a significant improvement in the social and school domains, reflecting a reduction in the psychosocial burden of cherubism. Six months post-procedure, no significant complications or signs of recurrence were observed, highlighting the efficacy of early intervention. These findings suggest that an early surgical approach is a valuable strategy for optimizing quality of life and minimizing the negative effects of cherubism, particularly in children undergoing social and academic development.

Keywords: Cherubism; Facial deformities; Quality of life.

INTRODUCTION

Cherubism is a bone disorder that exclusively affects the jaws, characterized by the replacement of medullary bone with fibro-osseous lesions[1,2]. It is an extremely rare condition, with approximately 600 cases reported worldwide[3]. Cherubism follows an autosomal dominant inheritance pattern and is associated with genetic mutations in the SH3BP2 gene, which plays a crucial role in the regulation of osteoclasts and osteoblasts[4].

The first description of the disease dates back to 1933, when it was initially referred to as "familial multilocular cystic disease of the jaws"[1,5,6]. Later, the term "cherubism" was adopted as the standard nomenclature due to the characteristic facial features observed in affected children, who present with rounded cheeks and upward-turned eyes, resembling the cherubs depicted in Renaissance paintings[1,6].

Cherubism typically manifests within the first decade of life as a painless, progressive, and approximately symmetrical swelling of the maxilla and/or mandible. In rare cases, orbital involvement may occur.

Statement of Clinical Significance
Early surgical intervention in cherubism provides significant functional and aesthetic benefits, promoting improved quality of life. This approach guides more effective clinical decision-making in Oral Pathology and Maxillofacial Surgery, focusing on the comprehensive rehabilitation of the patient.

The condition is frequently associated with dental eruption disturbances[2,4]. The phenotypic expression of the disease is highly variable, and in some cases, it may lead to severe, deforming bone lesions that are rarely life-threatening[3,5]. The fibro-osseous lesions and tissue expansion tend to progress until puberty, followed by spontaneous regression. Over time, the lesions are gradually replaced by woven bone, rendering facial abnormalities nearly imperceptible by the third decade of life[4,7].

The current recommended approach to cherubism is conservative management, with regular monitoring until the end of puberty due to the potential for spontaneous regression. However, in severe cases, early surgical intervention may be considered to improve both the patient's

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quality of life and facial appearance. Additionally, this approach can offer significant psychosocial benefits for children experiencing facial deformities during their formative years[2,8].

A significant concern for patients with cherubism and their families is that evident facial disfigurement may compromise quality of life[1,4,9]. Health-related quality of life (HRQoL) is a measure that reflects the value attributed to life while considering disabilities, functional status, perceptions, and opportunities, all of which are influenced by diseases, injuries, treatments, and policies[10]. Several studies report that facial deformities impact multiple aspects of HRQoL. However, few have specifically investigated the psychological impact and quality of life in individuals with cherubism[11,12].

This study aims to report the case of an seven-year-old girl diagnosed with cherubism, who underwent early surgical intervention due to the psychosocial distress caused by facial deformity. An analysis of health-related quality of life was conducted before and after surgery.

CASE REPORT

A 7-year-old female patient with a paternal history of cherubism was referred to the Oral and Maxillofacial Surgery Department at Governor Otávio Lage de Siqueira State Emergency Hospital (HUGOL), presenting with facial asymmetry associated with bilateral mandibular swelling.

During anamnesis, the patient's mother reported that facial growth had begun around the age of three and had become progressively more noticeable over time. According to the guardian, the child's physical and mental development was normal. During the consultation, the patient expressed dissatisfaction with her appearance, reported frequent teasing at school, and described difficulties performing typical activities for her age. Given this context, an evaluation of health-related quality of life (HRQoL) was conducted using the PedsQL™ 4.0 Generic Core Scales. The questionnaire was administered during a preoperative outpatient consultation by one of the attending oral and maxillofacial surgeons, in the presence of both the patient and her mother. The process was conducted in a calm and explanatory manner, ensuring that both the child and her caregiver clearly understood the questions. Both the child's self-assessment and the version with parental perception were completed in the same session, allowing for a comprehensive and contextualized assessment of the patient's baseline quality of life.

The PedsQL™ 4.0 Generic Core Scales is a validated instrument used to assess the quality of life of both healthy children and adolescents as well as those with acute or chronic conditions. It consists of 23 items distributed across four domains: physical (8 items), emotional (5 items), social (5 items), and school (5 items). In this case, the version designed for children aged 5 to 7 years was used[13].

The PedsQL™ 4.0 scores were calculated in accordance with the instrument's official guidelines. Each item was scored on a Likert-type scale (0 to 4), which was then reverse-scored and linearly transformed to a 0–100 metric scale: 0=100, 1=75, 2=50, 3=25, and 4=0. For the child's self-assessment, a visual facial scale with expressive icons was used, allowing intuitive responses corresponding to the same numerical values. The final score for each domain — physical functioning, emotional functioning, social functioning, and school functioning — was obtained by calculating the mean of the transformed item scores. This process was applied identically for the parent proxy version. The stratification of results followed reference parameters commonly used in pediatric HRQoL research: scores ≥80 were considered 'excellent', 60–79 'good', 40–59 'fair', and <40 'poor'[13].

Clinical examination

On extraoral examination, symmetric jaw expansion was observed, with increased volume in the mandibular body and ramus regions, without lymphadenopathy. Intraoral examination revealed alveolar bone enlargement and firm swelling in the posterior mandibular buccal vestibule. The area was asymptomatic upon palpation, but oral hygiene was deficient. Additionally, some deciduous teeth were missing, leading to wide edentulous spaces (Figure 1).

A computed tomography (CT) scan revealed bilateral multilocular osteolytic lesions affecting both the mandible and maxilla, associated with bone expansion and cortical thinning. Displaced tooth germs were observed within the lesions, which involved the posterior mandibular body and extended to the ramus. The mandibular head and condylar neck appeared normal (Figure 2). Laboratory tests showed normal serum calcium, alkaline phosphatase, and parathyroid hormone levels.

An incisional biopsy of the lesion was performed, and histopathological analysis demonstrated fibro-osseous proliferation, characterized by polygonal and elongated cells interspersed with multinucleated giant cells (Figure 3). The integrated analysis of clinical,

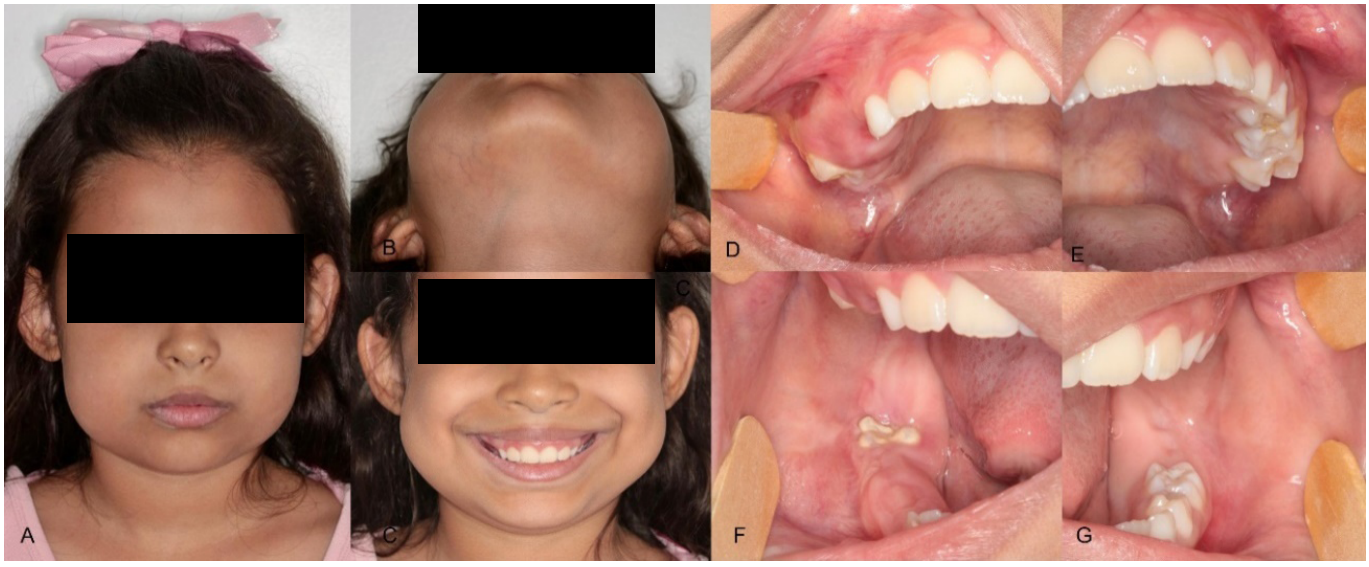

Figure 1. Preoperative extraoral and intraoral views showing increased bone volume in the posterior regions of the mandible and maxilla.

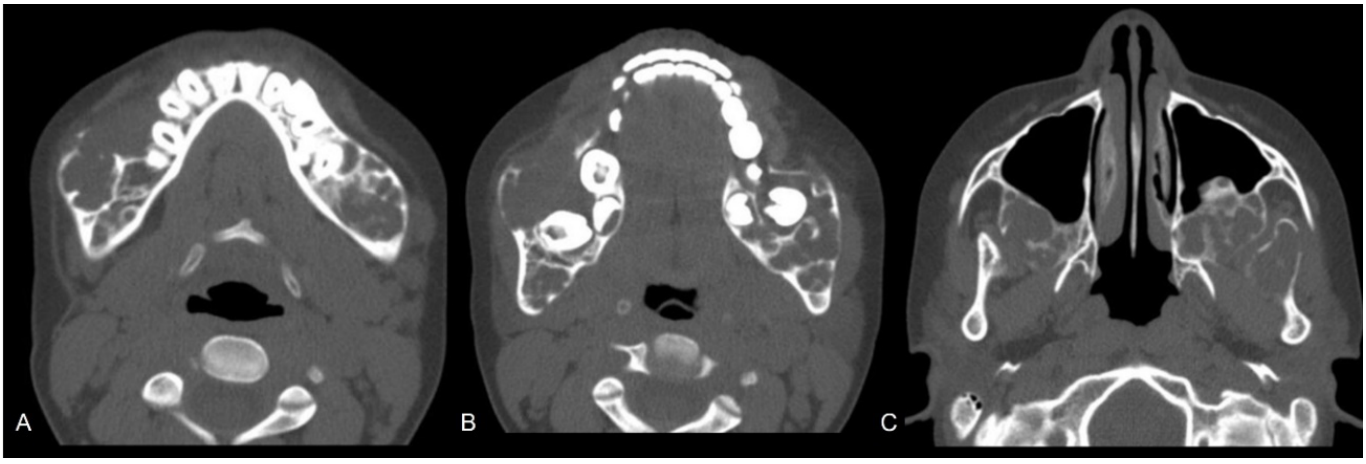

Figure 2. Axial computed tomography scan revealing bilateral multilocular osteolytic lesions in the mandible and maxilla, associated with bone expansion and cortical thinning.

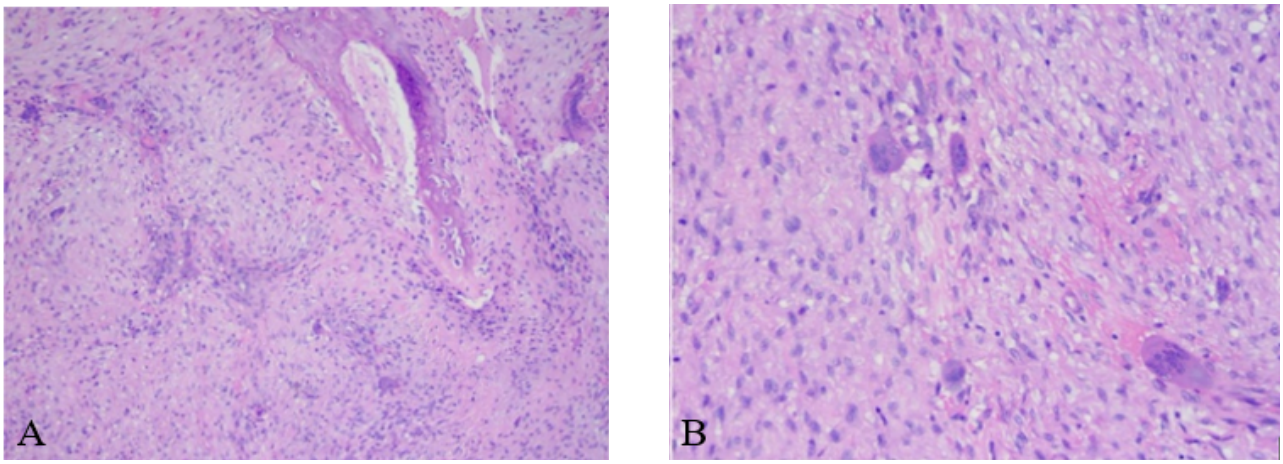

Figure 3. Histopathological examination of the H&E-stained sections revealed polygonal and elongated cells interspersed with multinucleated giant cells.

radiographic, laboratory, and histopathological findings confirmed the diagnosis of cherubism.

Surgical intervention

Surgical intervention was performed under general anesthesia. Bilateral intraoral incisions were made to access the mandible and maxilla. Upon lesion exposure, the friable and fibrous dysplastic tissue was carefully curetted, while preserving adjacent normal cortical bone. The inferior alveolar nerve was identified and preserved. Tooth germs located within the lesions were removed due to their intimate involvement in the fibro-osseous tissue, which made their preservation and spontaneous eruption unfeasible. Radiographic and intraoperative findings revealed that these developing teeth were completely embedded within dysplastic bone, with no favorable anatomical conditions for proper eruption or future functionality. Although the early removal of tooth germs may entail future dental rehabilitation needs, in this case, their unfavorable prognosis justified the decision. The patient will be able to undergo appropriate prosthetic or implant-supported rehabilitation at a suitable stage of craniofacial development. Hemostasis was maintained throughout the procedure, with no need for a blood transfusion (Figure 4).

Postoperative outcome

The postoperative period was uneventful, with no complications. Six months after surgery, the PedsQL™ 4.0 questionnaire was reassessed, revealing a significant improvement in both the patient's and her parents' quality of life (Table 1). The questionnaire was reapplied six months after surgery to allow sufficient time for surgical recovery, psychosocial adaptation, and reintegration into school and social environments. The six-month interval was chosen to assess more stable and clinically relevant improvements in quality of life, minimizing transient effects related to surgical recovery. Fifteen months postoperatively, no significant recurrence or long-term complications were observed (Figure 5).

DISCUSSION

Although cherubism is a benign condition, its lesions can progress to severe overgrowth, leading to multiple complications, such as speech, vision, breathing, dental development, and swallowing disorders[3,5]. These lesions significantly impact patients' lives and their families, compromising quality of life[4].

Based on this, the present study assessed the patient's quality of life using the PedsQL™ 4.0

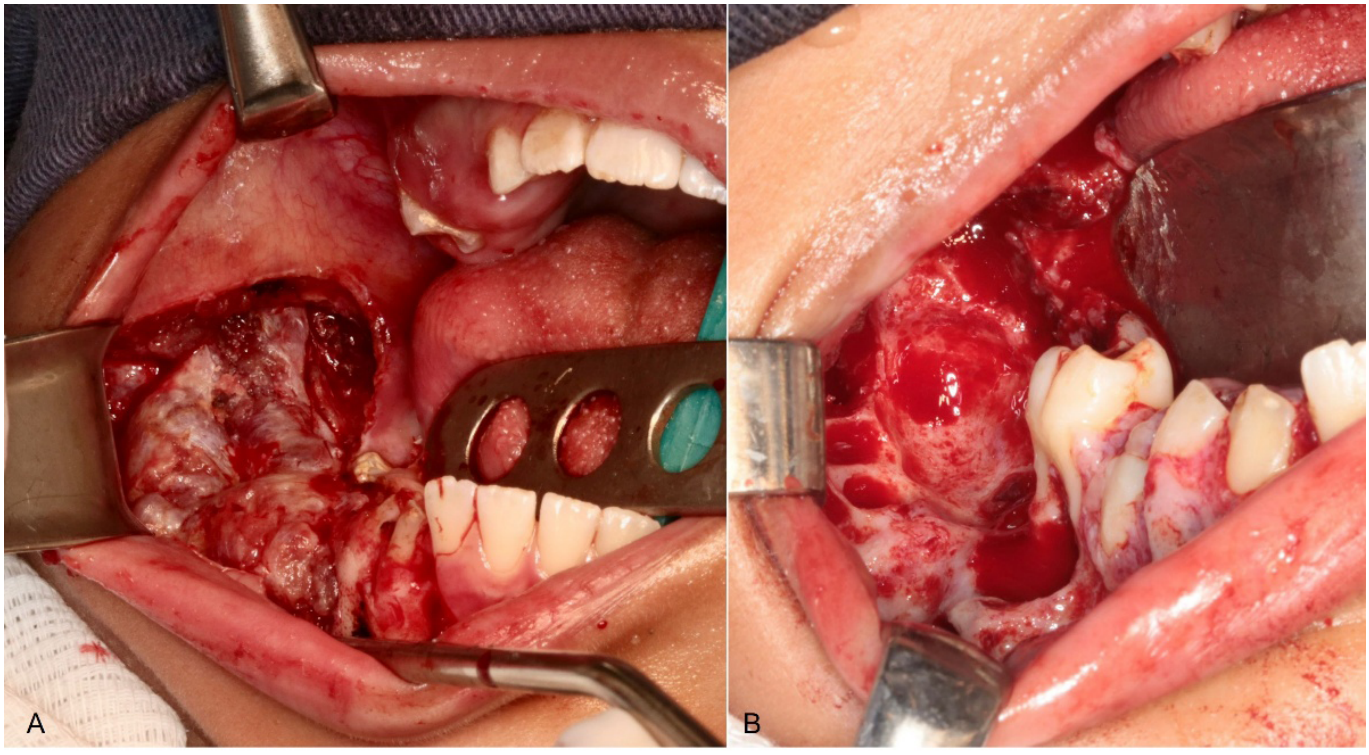

Figure 4. Intraoperative exposure showing friable, fibrous dysplastic tissue, along with the appearance of adjacent normal cortical bone after curettage.

questionnaire. The results demonstrated that the physical and functional changes associated with cherubism significantly affected quality of life, particularly in the social and school domains. These findings align with previous studies showing that children with disabilities often have fewer friends and experience social interaction difficulties[13].

The characteristic facial changes of cherubism frequently lead to social exclusion and the development of negative relationships, negatively impacting psychosocial development. Therefore, in addition to medical and dental follow-up, providing psychological support is crucial to mitigate long-term negative effects[14,15].

Cherubism lesions have often been described as self-limiting, with a reduction in size and number of lesions as puberty progresses, becoming almost imperceptible by the third decade of life[3-5]. However, studies indicate that not all patients exhibit spontaneous regression. In a study by Kueper et al., 84% of untreated patients showed no signs of regression, suggesting that the natural limitation of lesion growth should not be considered a rule[16].

The management of cherubism should be individualized, with therapeutic protocols that include curettage, surgical interventions, steroid injections, subcutaneous administration of interferon-α, and systemic calcitonin therapy[17]. Recently, the use of Imatinib, a tyrosine kinase inhibitor (TKI), has been reported as a potential therapeutic alternative for cases of cherubism[18,19]. Imatinib is traditionally used in the treatment of pediatric leukemias due to its action on the BCR-Abl tyrosine kinase[20]. In clinical case reports, the drug was administered on a compassionate-use basis, off-label, based on its potential inhibitory effect on osteoclast formation. The authors of these reports noted not only stabilization of the lesions but also signs of regression, along with improvement in patients' facial dysmorphology[18,19]. However, recent experimental evidence has questioned its efficacy, particularly in *in vivo* studies using *Sh3bp2* mutant mice — an established animal model of cherubism — in which imatinib administration did not lead to significant improvement in inflammatory or osteopenic phenotypes[20]. Furthermore, imatinib is a high-cost medication, which limits its broader applicability, especially in clinical settings with limited resources. Early surgical intervention is a viable option to enhance quality of life and facial appearance in cases where there is significant aesthetic or psychological impairment[2,21,22].

In the present case, surgical intervention was chosen to remove dysplastic tissue and impacted tooth germs within the lesions, aiming to halt disease progression, prevent complications, and improve the patient's quality of life. Son et al. reported positive outcomes with intensive surgical treatment, including curettage,

Table 1. Detailed PedsQL™ 4.0 scores (raw values, transformed scores, and qualitative interpretation) for the patient and her mother, before and after surgery.

Domain	Respondent	Raw Score (Sum)	Transformed Score (0–100)	Qualitative Interpretation
Physical functioning	SA – Pre	10	68.75	Good
	SA – Post	0	100	Excellent
	PP – Pre	125	15.63	Poor
	PP – Post	575	71.88	Good
Emotional functioning	SA – Pre	12	40.00	Fair
	SA – Post	2	90.00	Excellent
	PP – Pre	75	15.00	Poor
	PP – Post	225	45.00	Fair
Social functioning	SA – Pre	20	0.00	Poor
	SA – Post	0	100	Excellent
	PP – Pre	50	10.00	Poor
	PP – Post	500	100.00	Excellent
School functioning	SA – Pre	18	10.00	Poor
	SA – Post	6	70.00	Good
	PP – Pre	0	0.00	Poor
	PP – Post	150	30.00	Poor

resection, and jaw contouring, effectively correcting facial deformities and removing lesions[21].

Although the literature on the surgical management of cherubism remains limited, evidence suggests that early intervention may prevent severe complications, such as permanent tooth loss and significant facial deformities[2]. Moreover, considering that children with craniofacial anomalies have a higher incidence of anxiety and depression, particularly between 8 and 10 years of age, early surgical interventions can provide substantial psychosocial benefits.

CONCLUSION

The treatment choice for cherubism should be individually planned, considering factors such as patient age, disease characteristics, affected regions, progression rate, and clinical manifestations. Additionally, this study highlights the importance of assessing the psychosocial impact and quality of life of patients with cherubism, emphasizing the need for approaches that address not only clinical management but also overall well-being. We conclude that early surgical

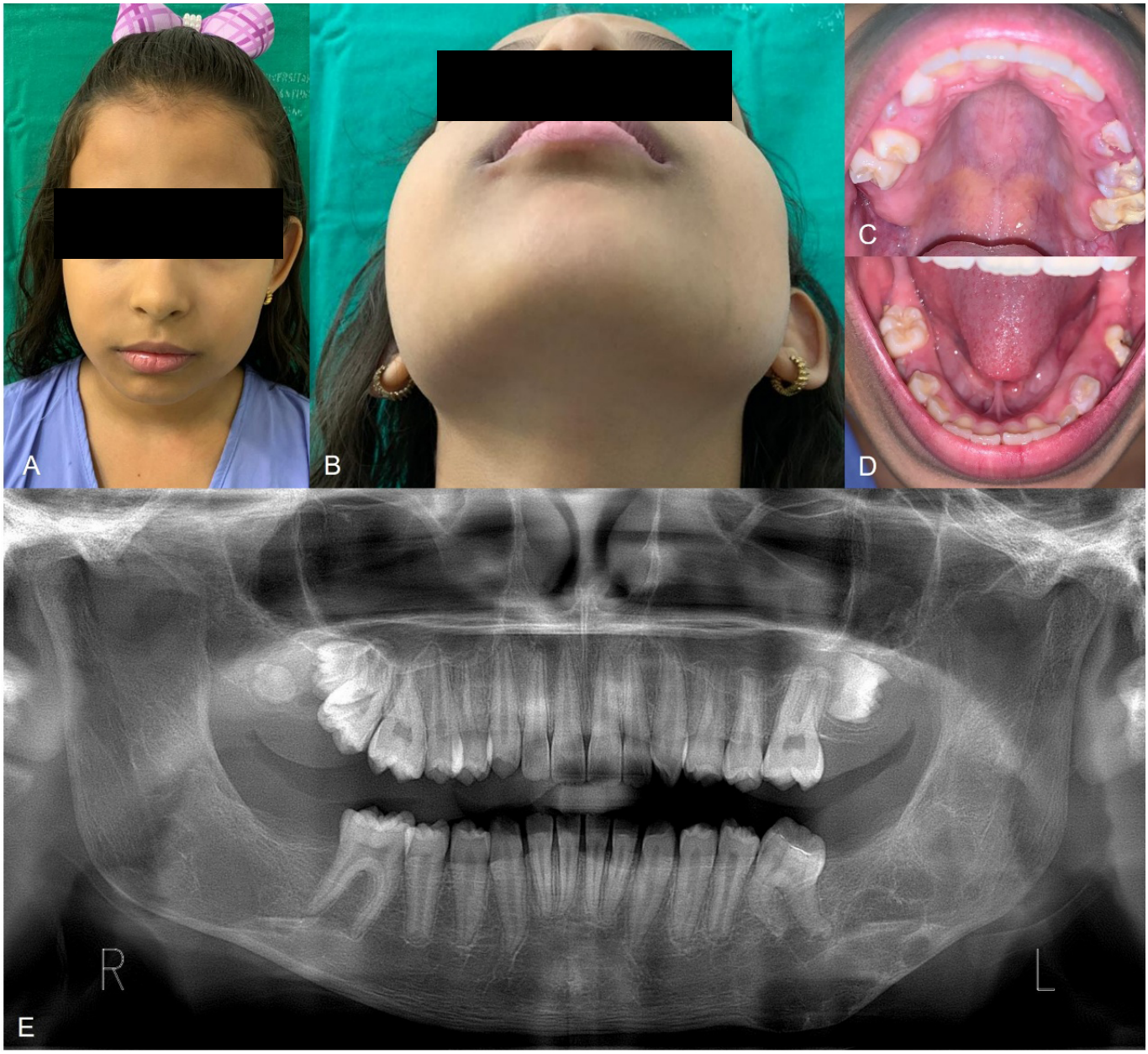

Figure 5. Postoperative extraoral and intraoral views demonstrating a reduction in bone volume in the maxillary and mandibular regions, as well as an improvement in mandibular contour.

intervention can significantly enhance quality of life, particularly by reducing its impact on the social and school domains.

Other information

Large language models

During the preparation of this work, the authors used ChatGPT (version GPT-4) only to improve readability and language. After using this tool, the authors reviewed and edited the content as needed and take full responsibility for the content of the publication.

AUTHORS' CONTRIBUTIONS

LTMQC: conceptualization, data curation, formal analysis, writing – original draft, writing – review & editing. AOP: data curation, formal analysis, writing – review & editing. RCS: data curation. formal analysis, methodology. EZ: investigation, writing – review & editing. SPA: investigation. VBF: data curation, investigation. RJS: conceptualization, investigation, project administration, writing – review & editing.

CONFLICT OF INTEREST STATEMENT

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Competing interests: The authors have no relevant financial or non-financial interests to disclose.

Ethics approval: The study was conducted in accordance with the ethical principles established by the Institutional Research Ethics Committee, ensuring compliance with current ethical and scientific guidelines. Approval protocol: 06856919.5.0000.5082.

REFERENCES

1. Tekin AF, Ünal ÖF, Göksel S, Özcan İ. Clinical and radiological evaluation of cherubism: a rare case report. Radiol Case Rep. 2020;15(4):416-9. https://doi.org/10.1016/j.radcr.2020.01.003
2. Moss W, Pires G, Zahid R, Tyrell R, Rhemtulla I, Gociman B. Severe cherubism treated with curettage, osteotomy, and bony repositioning: a case series of three patients. Plast Reconstr Surg Glob Open. 2022;10(1):e4079. https://doi.org/10.1097/GOX.0000000000004079
3. Liles SI, Hoppe IC, Arnold L. Denosumab therapy in cherubism. Cleft Palate Craniofacial J. 2023;60(12):1665-73. https://doi.org/10.1177/10556656221113891
4. Lahfidi A, Traore WM, Diallo ID, Lrhorfi N, Elhaddad S, Allali N, et al. Cherubism: a rare case report with literature review. Radiol Case Rep. 2022;17(10):3971-3. https://doi.org/10.1016/j.radcr.2022.07.063
5. Chrcanovic BR, Guimarães LM, Gomes CC, Gomez RS. Cherubism: a systematic literature review of clinical and molecular aspects. Int J Oral Maxillofac Surg. 2021;50(1):43-53. https://doi.org/10.1016/j.ijom.2020.05.021
6. Garlick JW, Willis RN, Donato DP, Gociman B. Cherubism in a 4-year-old boy managed with tumor curettage, mandibular osteotomies and repositioning. Plast Aesthet Res. 2018;5:29. https://doi.org/10.20517/2347-9264.2018.36
7. Holley TJ, Giannini PJ, Narayana N, Desa VP. Early detection of cherubism with eventual bilateral progression: a literature review and case report. Oral Surg Oral Med Oral Pathol Oral Radiol. 2019;127(3):e77-83. https://doi.org/10.1016/j.oooo.2018.08.011
8. Jung KW, Yun JM, Lee JM, Choi IS. Sinonasal manifestations of severe cherubism: a case with 11-year follow-up. Ear Nose Throat J. 2025;104(1_suppl):257S-62S. https://doi.org/10.1177/01455613221127588
9. Papadaki ME, Lietman SA, Levine MA, Olsen BR, Kaban LB, Reichenberger EJ. Cherubism: best clinical practice. Orphanet J Rare Dis. 2012;7(Suppl. 1):S6. https://doi.org/10.1186/1750-1172-7-S1-S6
10. Haraldstad K, Wahl A, Andenæs R, Andersen JR, Andersen MH, Beisland E, et al. A systematic review of quality of life research in medicine and health sciences. Qual Life Res. 2019;28(10):2641-50. https://doi.org/10.1007/s11136-019-02214-9
11. Geirdal AØ, Saltnes SS, Storhaug K, Åsten P, Nordgarden H, Jensen JL. Living with orofacial conditions: psychological distress and quality of life in adults affected with Treacher Collins syndrome, cherubism, or oligodontia/ectodermal dysplasia-a comparative study. Qual Life Res. 2015;24(4):927-35. https://doi.org/10.1007/s11136-014-0826-1
12. Tapia VJ, Epstein S, Tolmach OS, Hassan AS, Chung NN, Gosman AA. Health-related quality-of-life instruments for pediatric patients with diverse facial deformities: a systematic literature review. Plast Reconstr Surg. 2016;138(1):175-87. https://doi.org/10.1097/PRS.0000000000002285
13. Shapiro DN, Waljee J, Ranganathan K, Buchman S, Warschausky S. Using the patient reported outcomes measurement information system to evaluate psychosocial functioning among children with craniofacial anomalies. Plast Reconstr Surg. 2015;135(6):1673-9. https://doi.org/10.1097/PRS.0000000000001269
14. Riatto BM, Riatto SG, Andrade LP. Querubismo e qualidade de vida: impactos físicos e psicológicos. Revista Diálogos em Saúde. 2021;4(1):13-28.
15. Costabile C, Brunello MIB. Repercussões da inclusão escolar sobre o cotidiano de crianças com deficiência: um estudo a partir do relato das famílias. Rev Ter Ocup Univ São Paulo. 2005;16(3):124-30.
16. Kueper J, Tsimbal C, Olsen BR, Kaban L, Liao EC. SH3BP2-related fibro-osseous disorders of the maxilla and mandible: a systematic review. Int J Oral Maxillofac Surg. 2022;51(1):54-61. https://doi.org/10.1016/j.ijom.2021.04.001
17. Gupta S, Singh K, Garg A, Bhandari PS, Sah SK, Reichenberger E, et al. Clinicoradiologic follow up of cherubism with aggressive characteristics: a series of 3 cases. Oral Surg Oral Med Oral Pathol Oral Radiol. 2019;128(5):e191-201. https://doi.org/10.1016/j.oooo.2019.01.082
18. Eiden S, Lausch E, Meckel S. Involution von cherubismus im MRT unter therapie mit imatinib. RoFo. 2017;189(7):675-7. https://doi.org/10.1055/s-0043-105074

19. Ricalde P, Ahson I, Schaefer ST. A paradigm shift in the management of cherubism? A preliminary report using imatinib. J Oral Maxillofac Surg. 2019;77(6):1278.e1-1278.e7. https://doi.org/10.1016/j.joms.2019.02.021
20. Mukai T, Akagi T, Asano SH, Tosa I, Ono M, Kittaka M, et al. Imatinib has minimal effects on inflammatory and osteopenic phenotypes in a murine cherubism model. Oral Dis. 2023;29(3):1089-101. https://doi.org/10.1111/odi.14073
21. Son JH, Marshall DC, Valiathan M, Otteson T, Ferretti G, Grigorian PA, et al. Innovative surgical treatment of severe cherubism. Cleft Palate Craniofac J. 2019;56(1):90-3. https://doi.org/10.1177/1055665618774008
22. Holley TJ, Giannini PJ, Narayana N, Desa VP. Early detection of cherubism with eventual bilateral progression: a literature review and case report. Oral Surg Oral Med Oral Pathol Oral Radiol. 2019;127(3):e77-83. https://doi.org/10.1016/j.oooo.2018.08.011