RESEARCH
www.commprac.com
ISSN 1462 2815

RADIOGRAPHIC UNVEILING OF CHERUBISM: A MULTIDISCIPLINARY JOURNEY IN DIAGNOSIS AND PRECISION TREATMENT PLANNING: A CASE REPORT

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Abstract

Cherubism, an autosomal dominant disorder characterized by painless and symmetrical jaw enlargement due to fibrous tissue replacement, presents challenges in diagnosis and management. This report details the case of a female patient with right eye ptosis and facial swelling, utilizing a CT Orbit without Contrast to unveil a complex pathology. Imaging revealed an expanded mandible, cystic spaces in bilateral sphenoid sinuses, and maxillary sinus enlargement with orbital floor encroachment. Despite orbital changes, eyeball integrity was preserved. The radiological impression suggested features indicative of Cherubism, emphasizing the need for clinical correlation. Surgical interventions, guided by CT scans, were discussed for severe cases. Orthodontic management and long-term follow-up, incorporating advanced imaging modalities, were underscored for comprehensive care. This case underscores the multidisciplinary approach required for Cherubism, integrating clinical, radiological, and genetic assessments for optimal patient outcomes.

Keywords: Cherubism, CT Scans, Radiological Diagnosis, Autosomal Dominant.

INTRODUCTION

Cherubism is a rare disease of autosomal dominant inheritance characterized by painless, frequently symmetrical, enlargement of the jaws because of the replacement of bone with fibrous tissue [1]. Cherubism was first described by Jones—is a benign fibro osseous disorder of childhood involving the lower two thirds of the face.
The true incidence is unknown, but the age of onset is between 2 and 10 years [2]. Cherubism is a rare, hereditary, non-neoplastic bone disease. It is characterized by

clinically evident bilateral, painless enlargements of the jaws that are said to give the patient a "chubby" appearance [3].

The disease is also called familial fibrous dysplasia of the jaws, but recent genetic investigation has shown it to be a separate entity at the molecular level [4]. Furthermore, Lannon et al. [5] mentioned the necessity to distinguish cherubism from central giant cell granuloma and giant cell tumor of the jaws, with which it holds a false synonymity. Usually, it presents as painless bilateral swelling of the jaws giving the patient a cherubic look with an eyes-to-heaven appearance [4,6].

Cherubism is characterized by bilateral expansion of the mandible and/or the maxilla that becomes noticeable within the first several years of life, becomes progressively larger until puberty, and gradually resolves by middle age [7]. The dentition is often abnormal, and patients have tooth agenesis, non-eruption, displacement, root resorption, and malocclusion [8]. Cherubism is an autosomal-dominant disease with variable penetrance and expressivity.

It has been associated with mutations in the SH3BP2 gene, which has been mapped to locus 4p16.3 [9,10]. The radiographic appearance is unique because of its diffuse, bilateral, multilocular nature, which retards the eruption of permanent teeth and may even prevent the formation of permanent molars [11]. Histologically, the lesions contain numerous multinucleated giant cells scattered throughout a fibrous connective tissue stroma [12].

CASE PRESENTATION

An Asian female patient reported to the department presenting with right eye ptosis persisting for 2 years and facial swelling ongoing for 4 years (FIGURE 1), underwent a CT Orbit without Contrast. The imaging revealed a complex pathology characterized by a grossly expanded mandible, abnormal bone texture with cystic spaces in the walls of bilateral sphenoid sinuses, and bilateral maxillary sinus enlargement.

Notably, the posterolateral wall of the maxillary sinuses protruded into the respective extraconal spaces of the bilateral orbits, causing superomedial displacement. Despite these orbital changes, the shape and integrity of both eyeballs appeared preserved, with no evidence of lens dislocation, vitreous hemorrhage, or foreign bodies.

Optic nerve, extraocular muscles, and retrobulbar fat were normal. The findings suggest a chronic process with potential compression of ocular structures, warranting further clinical correlation and interdisciplinary collaboration for a comprehensive diagnostic and management approach.

The radiological impression strongly suggests features indicative of cherubism, with fibrous dysplasia considered as an alternative diagnosis. Given the inherent limitations of solitary pathological and radiological investigations, the report underscores the crucial need for clinical correlation.

It emphasizes that these investigations serve as adjuncts rather than conclusive tools in isolation, urging a comprehensive interpretation in tandem with clinical symptoms, patient history, and additional tests.

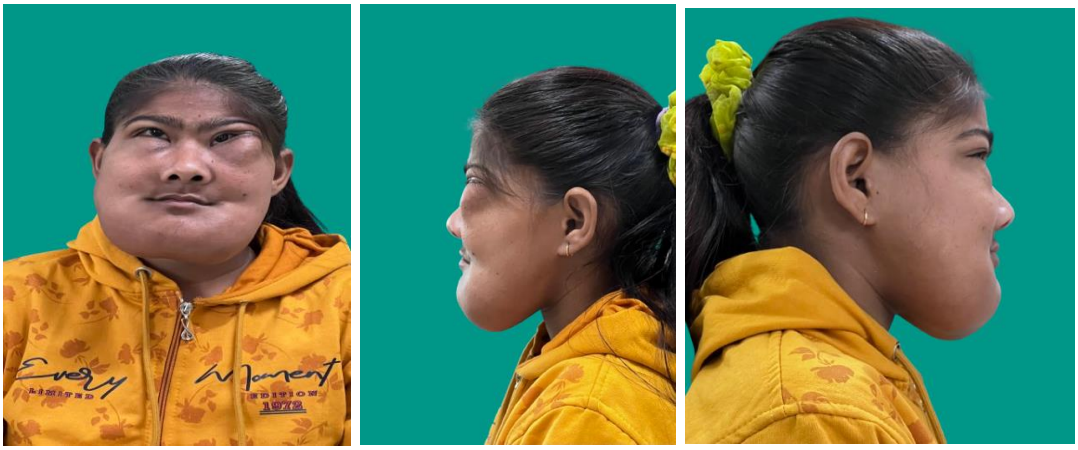

Figure 1 : Facıal Profıle Of The Patient

The condition initiates as a painless bilateral expansion of the affected bone, leading to the stretching of the skin in the upper face. This expansion involves the infraorbital rim and orbital floor, resulting in a distinctive appearance where a rim of sclera is visible beneath the iris, creating a classic "eyes upturned to heaven" look. The upward tilt of the eyeball is compounded by the stretching of the upper facial skin, pulling the lower lid downwards. As the bone involvement progresses, there is widening, causing displacement and failure to erupt developing teeth. Various dental abnormalities manifest, including agenesis of the 3rd mandibular molars, tooth displacement, premature exfoliation of primary teeth, delayed eruption of permanent teeth, and transposition and rotation of teeth. The permanent dentition often exhibits defects, and severe cases may involve root resorption.

Radiographically, the condition presents as an expansile, multilocular radiolucency. The presence of numerous unerupted teeth and destruction of alveolar bone can result in a phenomenon known as "Floating Tooth Syndrome." In adulthood, the cystic areas in the jaws undergo re-ossification, leading to irregular patchy sclerosis. The characteristic ground glass appearance is observed due to the small, tightly compressed trabecular pattern, although it is non-specific. This condition has been linked to Noonan's Syndrome.

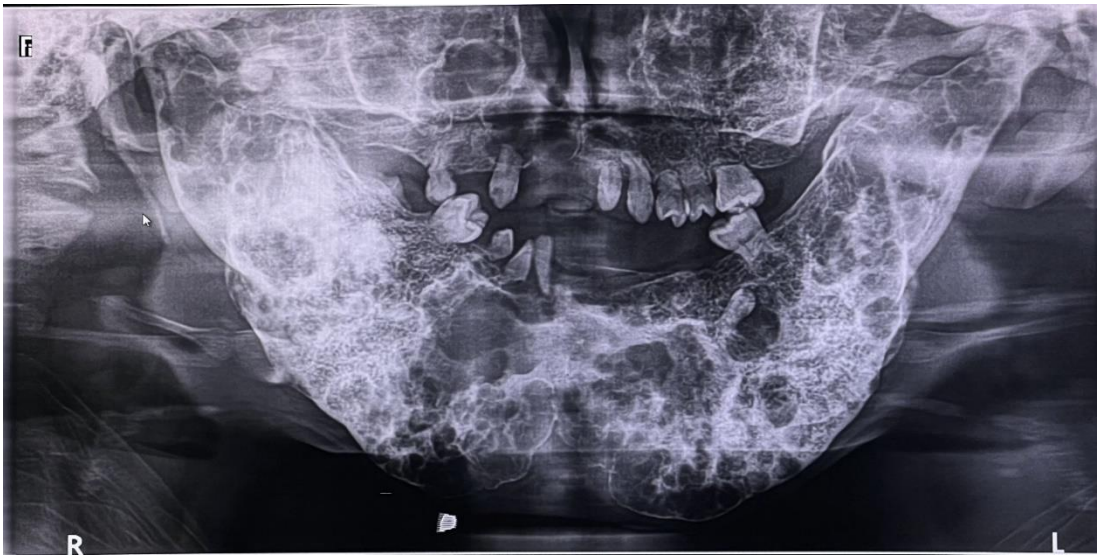

Figure 2: Orthopantogram Of The Patıent

The CT scan revealed significant remodeling of both the maxilla and mandible, displaying extensive expansion with internal trabeculations and a mildly sclerotic matrix, as illustrated in Figure 3. This remodeling led to the obliteration of the maxillary antrum and encroachment onto the orbital floor, as depicted in Figure 3, resulting in bilateral proptosis, which was clinically observed.

Notably, there were no signs of cortical break, fracture, or periosteal reaction within the jaw bones. The absence of bilateral condylar extension was a characteristic feature. Additionally, the CT scan clearly indicated the lack of extraosseous soft tissue extension.

CT scans play a crucial role in the detailed evaluation of Cherubism. Authors have consistently utilized CT imaging to visualize the extent of expansile remodeling in the maxilla and mandible. These scans reveal multilocular radiolucencies, internal trabeculations, and a mildly sclerotic matrix within the affected bones.

The characteristic findings on CT include obliteration of the maxillary antrum, encroachment onto the orbital floor, and the absence of cortical breaks or fractures. Bilateral proptosis, a clinical manifestation, is often correlated with the extent of orbital involvement observed on CT scans. The absence of extraosseous soft tissue extension is a notable feature in CT imaging.

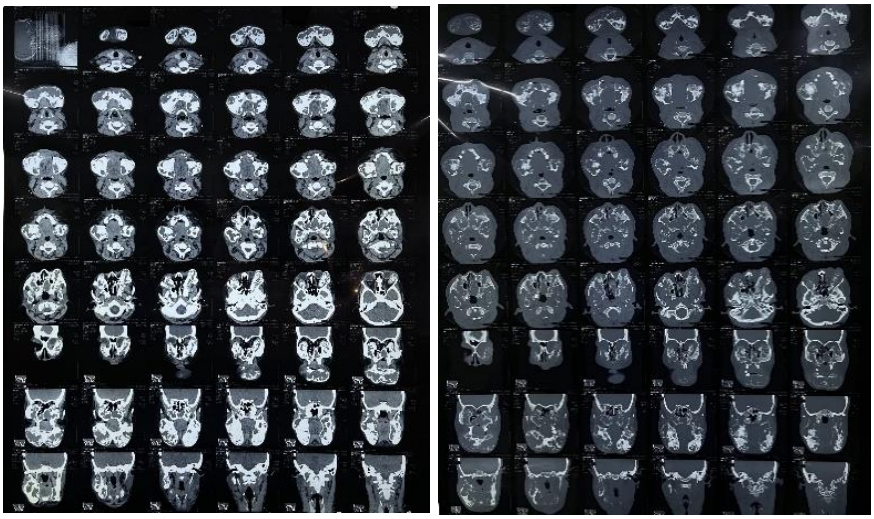

Figure 3 : CT Scan Of The Patıent

DISCUSSION

Cherubism stands as a rare and painless osseous disorder observed primarily in children and adolescents. While the radiologic features of cherubism are not pathognomonic, the diagnosis is strongly indicated by bilateral and relatively symmetric jaw involvement, specifically limited to the maxilla and mandible [13].

Imaging studies typically reveal extensive remodeling of the affected bones, accompanied by cortical thinning, multilocular radiolucencies displaying a coarse trabecular pattern, and a notable absence of periosteal reaction. These characteristic radiographic findings contribute to the identification and differentiation of cherubism from other jaw conditions [8,10].

This genetic disease follows an autosomal dominant pattern of inheritance, exhibiting significant variability in its clinical expression. The penetrance of the trait is noted to be as high as 100% in men and ranges from 50% to 70% in women.

Originally perceived as exclusively familial, there have been reported instances of sporadic cases. These sporadic occurrences may be attributed to factors such as incomplete penetrance or the emergence of new mutations within the affected genetic locus. The diverse ways in which the genetic trait manifests underscore the complexity and variability associated with this condition [4,5].

In 1992, Marck and Kudryk proposed a grading system for cherubism depending on the location of the lesions. Accordingly, grade 1 concerns bilateral involvement of mandibular rami, grade 2 corresponds to the involvement of both mandibular rami and maxillary tuberosities, grade 3 includes massive involvement of the entire maxilla and mandible except for the condyles, while grade 4 relates to the involvement of the orbits with ocular disturbances as well as the grade 3 lesions [14]. In our patient, the lesions were classified as grade 2, according to the grading system mentioned above.

Surgery is indicated only in more aggressive cases characterized by functional impairment such as speech, chewing or swallowing, and ocular disturbances, or with the presence of major deformities that may cause psychological problems for the patient. Surgical removal of the fibrotic lesions does not modify the natural course of the disease and gives temporary relief but may provoke rapid recurrence or exacerbate the disease if the patient is operated on during the active phase [15].

Calcitonin therapy has demonstrated efficacy in inducing regression in central giant-cell granuloma of the jaws, primarily through its inhibitory effects on bone resorption by multi-nucleate cells, specifically osteoclasts.

Given the presence of similar osteoclastic activity in cherubic lesions, calcitonin, functioning as an antiresorptive agent, appears to be effective in reducing cystic lesions within the jaws in individuals with cherubism. This therapeutic approach holds promise in addressing the underlying bone remodeling characteristic of cherubism, potentially contributing to the management and regression of the condition [15].

Radiotherapy is contraindicated in cherubism due to the risk of osteoradionecrosis, induction of malignancy and growth disturbances [16].

Cherubism has been identified to potentially co-occur with other genetic disorders, including Ramon's syndrome, Noonan's syndrome (also known as pseudo-Turner syndrome), neurofibromatosis type 1, Jaffe-Campanacci syndrome, and fragile X syndrome.

This association highlights the complex interplay and potential overlap of genetic factors contributing to various syndromic manifestations [17]. Clinicians should remain vigilant to the possibility of concurrent genetic conditions when diagnosing and managing individuals with cherubism. The recognition of these associations can contribute to a more comprehensive understanding of the genetic landscape and potential clinical implications for affected individuals [18,19].

The metaverse, which refers to a collective virtual shared space, is a concept that has gained attention for its potential applications across various domains, including healthcare.

While virtual reality (VR) technologies can be used for educational purposes, simulation, and even certain therapeutic interventions, there isn't currently a specific application of the metaverse, or VR specifically tailored for the treatment of cherubism.

However, advancements in technology may lead to innovative approaches in the future. For instance, virtual reality simulations could potentially be used for pre-operative planning or patient education in the context of surgical interventions for cherubism.

Additionally, virtual support communities within the metaverse could offer social and emotional support to individuals affected by cherubism. It's essential to consult with healthcare professionals, including specialists in genetics, dentistry, and maxillofacial surgery, for personalized management and treatment recommendations for cherubism [20].

CONCLUSION

Severe cases of Cherubism, characterized by significant functional or cosmetic concerns, may prompt surgical intervention. CT scans are instrumental in this regard, facilitating the planning and execution of surgical procedures. Surgical options encompass debulking of affected bone, contouring procedures, and the resection of excessively remodeled bone. The precision provided by CT imaging in identifying abnormal bone boundaries ensures targeted and effective surgical interventions, addressing both functional and aesthetic aspects. Orthodontic management is another integral facet of Cherubism treatment, specifically focusing on addressing dental abnormalities associated with the condition.

This includes the management of tooth displacement, alignment issues, and other dental anomalies. Panoramic radiographs (OPGs) are commonly utilized to assess dental conditions, aiding in the planning of orthodontic treatments. This comprehensive approach ensures that both the skeletal and dental components of Cherubism are considered in the overall treatment plan. Long-term follow-up is emphasized by authors to monitor disease progression, assess treatment outcomes, and identify potential recurrence.

CT scans play a crucial role in this extended monitoring, providing detailed information on bone re-ossification and the stability of surgical interventions. The integration of advanced imaging modalities, particularly CT scans and OPGs, in both diagnosis and treatment planning underscores the importance of a thorough and personalized approach to address the unique challenges posed by Cherubism. Regular follow-up with imaging remains essential to evaluate the effectiveness of interventions and ensure optimal long-term outcomes for patients.

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