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Case Report

Histopathological insights into cherubism: a case presentation

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ABSTRACT

Cherubism is a rare hereditary fibro-osseous disorder of the jaws, typically presents as progressive, painless, bilateral jaw swelling in children between 2 to 5 years of age, with lesions showing maximum growth until puberty before regressing spontaneously. Radiographically, it is characterized by bilateral, multilocular radiolucencies in the posterior mandible and maxilla. Histopathologically, cherubism is marked by fibrotic stromal proliferation with multinucleated osteoclast-like giant cells. The presence of vascular channels and eosinophilic deposits further accentuates the unique histologic landscape of this condition. By documenting this histopathological perspective, we aim to reinforce its diagnostic significance and contribute to the growing repository of cherubism-related literature.

Keywords: Cherubism, Fibro-osseous lesion, Multinucleated giant cells, Perivascular cuffing

INTRODUCTION

Cherubism is a rare hereditary fibro-osseous disorder with characteristic clinical, radiographic, and histopathological features.[1] The presence of multinucleated giant cells within a highly vascular fibrocellular stroma remains a hallmark of the disease, resembling other giant cell lesions.[2,3] In our case, a notable histopathological finding was perivascular cuffing, which further supports the diagnosis. Recognizing these histopathological characteristics is crucial for differentiating cherubism from other giant cell-rich lesions, such as central giant cell granuloma (CGCG), brown tumor of hyperparathyroidism, and fibrous dysplasia. While cherubism is largely self-limiting, histopathological confirmation is essential for accurate diagnosis and management. A multidisciplinary approach, combining clinical, radiographic, and histopathological evaluation, ensures appropriate treatment planning and long-term follow-up to monitor disease progression and potential complications.[4-6]

CASE REPORT

An 8-year-old girl presented with bilateral swelling over the lower third of the face for 6 months. The swelling was gradually increasing in size and not associated with pain or limitation of mouth opening. There was no history of trauma or pus discharge. Medical, dental, and family history was non-contributory.

During the extraoral examination, diffuse swelling was observed over the angles of the mandible, extending to the malar bones on both sides. The skin covering the swelling appeared normal. Upon palpation, the swelling was non-painful and exhibited a hard consistency suggestive of bone involvement, with no localized increase in temperature. No lymphadenopathy was noted. Intraoral examination revealed erupting right mandibular first

molar. An OPG was suggested to confirm the diagnosis, assess the extent of bone involvement, and plan treatment. It showed expansile multilocular radiolucent lesions involving the mandible and maxilla symmetrically, particularly in the posterior regions giving the "soap bubble" or "ground-glass" appearance, typical of cherubism. Several unerupted, displaced, and malformed teeth were visible. The dental follicles appear widely separated due to the expansile nature of the lesion contributes to the characteristic cherubic facial appearance (Figure 1).

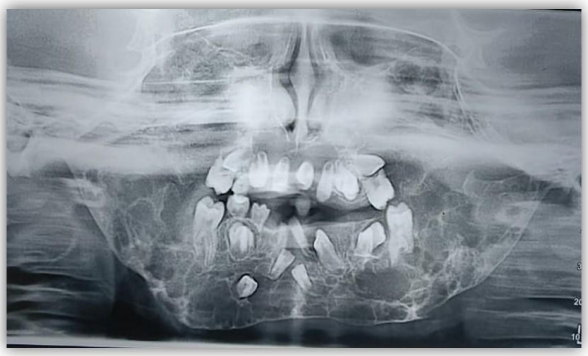

Figure 1: OPG showing expansile multilocular radiolucent lesions involving the mandible and maxilla symmetrically, particularly in the posterior regions giving the "soap bubble" or "ground-glass" appearance.

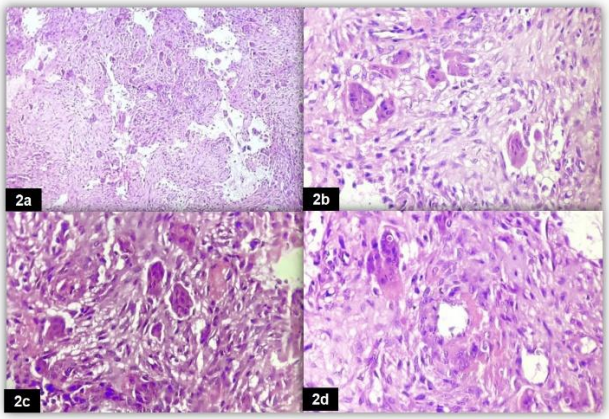

Figure 2 (A-D): Microscopic examination shows A-Highly cellular fibrovascular stroma interspersed with numerous multinucleated giant cells. B-Fibrovascular stroma with spindle-shaped fibroblasts. C-Multinucleated giant cells unevenly distributed throughout the stroma. D-Presence of perivascular cuffing, where eosinophilic deposits of collagen are seen surrounding blood vessels.

To rule out other metabolic bone diseases, serum markers were evaluated including alkaline phosphatase (ALP) and parathyroid hormone (PTH). ALP was slightly elevated, which was consistent with bone remodeling activity and PTH was found within normal limits, which helped to rule out hyperparathyroidism. Based on the clinical and radiographic features along with biochemical test, a provisional diagnosis of cherubism was given and an incisional biopsy was performed to confirm the diagnosis.

Incisional biopsy was done, and hematoxylin and eosin (H and E) stained section revealed a highly cellular fibrovascular stroma interspersed with numerous multinucleated giant cells (Figure 2 A). The fibrocellular stroma was composed of spindle-shaped fibroblasts (Figure 2 B). The multinucleated giant cells were distributed unevenly throughout the stroma and contained multiple nuclei, which were centrally or irregularly distributed within the cytoplasm (Figure 2 C). A notable feature in this case was the presence of perivascular cuffing, where eosinophilic deposits of collagen were seen surrounding blood vessels (Figure 2 D). There was no evidence of nuclear atypia, increased mitotic activity, or necrosis, ruling out aggressive neoplastic processes. A final diagnosis of cherubism was made based on the clinical, radiographic, and histopathological features.

DISCUSSION

Cherubism is classified by the world health organization (WHO) as a non-neoplastic, self-limiting fibro-osseous lesion exclusively involving the jaws. First described by Jones in 1933, it typically presents in early childhood and follows an autosomal dominant inheritance pattern with variable expressivity.[3,7] The condition is linked to mutations in the SH3BP2 gene, which plays a critical role in osteoclast regulation and bone remodeling.[3] The disorder usually manifests around 2 years of age, with progressive jaw expansion between 8 to 9 years and a tendency to stabilize or regress spontaneously after puberty. Complete regression is often observed by the third or fourth decade of life. The age at diagnosis is often dictated by the severity and extent of facial swelling. Males are generally more frequently and severely affected than females.[4]

Radiographically, cherubism typically appears as bilateral, multilocular radiolucencies involving the posterior mandible and maxilla, often described as having a “soap-bubble” or ground-glass appearance. The epicenter of these lesions is generally the mandibular rami and maxillary tuberosities, with progressive anterior expansion leading to anterior displacement of developing teeth. In some severe cases, maxillary involvement may result in infraorbital rim expansion, producing the characteristic upward gaze or "eyes raised to heaven" appearance due to scleral show.[7]

A grading system proposed for cherubism helps classify the condition based on the extent of jaw involvement: Grade I: Bilateral involvement of mandibular rami, grade II: Bilateral mandibular rami and maxillary tuberosities, grade III: Diffuse involvement of maxilla and mandible,

sparing the condyles and grade IV: Extensive involvement including the orbital floor with orbital compression.

Our case was classified as grade II cherubism according to this system.[4]

Histologically, Chomette et al described three progressive stages of cherubism: an initial osteolytic stage marked by numerous TRAP-positive multinucleated giant cells and a richly vascular fibroblastic stroma; a proliferative (reparative) stage dominated by spindle-shaped fibroblasts, central vascular nodules, and early osteoid deposition; and a final bone-forming stage characterized by increased collagen content, reduced cellularity, and the presence of alkaline phosphatase- and ATPase-positive osteoblastic activity.[6] In our case, histopathology revealed a fibrovascular stroma with unevenly distributed multinucleated giant cells, spindle-shaped fibroblasts, and perivascular collagen cuffing, consistent with the early to intermediate stage of cherubism.[8]

The differential diagnosis of cherubism includes other giant cell–rich and fibro-osseous lesions of the jaws. CGCG often presents unilaterally and lacks the bilateral symmetry characteristic of cherubism. Brown tumor of hyperparathyroidism may mimic cherubism both radiographically and histologically; however, normal PTH levels and absence of systemic signs in this case exclude this diagnosis. Fibrous dysplasia typically shows a unilateral ground-glass appearance and does not feature multinucleated giant cells. Juvenile ossifying fibroma presents with aggressive bone expansion but differs histologically, showing cementum-like calcifications without giant cells. The bilateral, symmetrical involvement combined with clinical, radiographic, biochemical, and histopathological findings supports the diagnosis of cherubism in this patient.

Surgical intervention is usually reserved for severe cases causing functional impairment or significant cosmetic concerns. In the present case, given the moderate grade and absence of complications, the patient is being followed up every six months to monitor lesion progression and dental development. Genetic testing was not performed due to resource constraints, but clinical and radiographic findings remain the mainstay for diagnosis and management decisions.[6]

CONCLUSION

Cherubism is a rare, non-neoplastic fibro-osseous condition characterized by bilateral, symmetrical jaw involvement predominantly in children. Accurate diagnosis relies on a combination of clinical presentation, characteristic radiographic features, biochemical markers, and histopathology. Early recognition and appropriate grading guide management, which is often conservative due to the self-limiting nature of the disease. Regular follow-up is essential to monitor lesion progression and address any functional or esthetic concerns. This case highlights the importance of a multidisciplinary approach for diagnosis and long-term care in cherubism.

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REFERENCES

1. Choi Y, Ji JM, KiM CH, Sung KP. Surgical management of severe cherubism persisting into early adulthood: a case report and literature review. Arch Craniofac Surg. 2024;25(1):38-43.
2. Jiao Y, Zhou M, Yang Y, Zhou J, Duan X. Cherubism misdiagnosed as giant cell tumor: a case report and review of literature. Int J Clin Experimental Med. 2015;8(3):4656.
3. Hamzavi SS, Askari A, Bahrololoom R, Mokhtari M, Sanaei Dashti A, Yarmahmoodi F, et al. Nonfamilial cherubism in a 6-month-old infant: a case report. BMC Pediatr. 2024;24(1):402.
4. Ram SG, Ajila V, Babu SG, Shetty P, Hegde S, Pillai DS. Cherubism: report of a case. Journal of Health and Allied Sciences NU. 2021;11(02):104-6.
5. Akolkar S, Hande A, Sonone AM, Chavhan A, Tehzeeb H. Cherubism Unmasked: A Case Report of Clinical and Histopathological Presentation. Cureus. 2024;16(3):e56456.
6. Pachva A, Narella K, Reddy AA, Kuar SP. Not All Chubby Cheeks Are Cute: A Case of Cherubism. Cureus. 2024;16(7):e65841.
7. Karthikeyan N, Varma S, Nagendran N, Moorthy P, Cheppala Rajan S. A rare case report of non-familial cherubism. J Evol Med Dent Sci. 2020;9:3503-5.
8. Papadaki ME, Lietman SA, Levine MA, Olsen BR, Kaban LB, Reichenberger EJ. Cherubism: best clinical practice. Orphanet J Rare Dis. 2012;7(1):1-4.

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