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Nonfamilial cherubism in a 6-month-old infant: a case report

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Abstract

Background Cherubism is known as a very rare autosomal dominant familial disorder of childhood caused by a mutation in the SH3BP2 gene on 4p16.3. It has not yet been observed at birth and is usually diagnosed in children aged 2–7. Here, we present a non-hereditary case of cherubism at a very early age.

Case presentation A 6-month-old girl presented with bilateral progressive jaw enlargement. On physical examination, bilateral asymmetrical jaw enlargement, predominantly on the left side, and some enlarged, non-tender, mobile submandibular lymph nodes were detected. No other abnormality was observed. Further investigations with radiology suggested cherubism and Burkitt's lymphoma as differential diagnoses. Later on, histopathologic evaluations were suggestive of cherubism. No surgical interventions were indicated, and the child is on regular follow-ups.

Conclusion Non-hereditary Cherubism, despite scarcity, can present in children below two years of age, even as early as the beginning of primary dentition. Accurate and swift diagnosis is essential to avert physical and psychological complications. Our case report shows the importance of keeping cherubism in mind as a differential diagnosis of bone disease, even in children under a year old, and the value of interdisciplinary collaboration in dealing with rare genetic disorders.

Keywords Bone disease, Cherubism, Familial benign giant-cell tumor of the jaws, Familial multiloculated cystic disease of the jaw, Case report

Introduction

Cherubism is a familial disorder of childhood, first described by William A. Jones in 1933. He named the disorder *Cherubism* due to the cherubic-like appearance, the full round cheeks, and the upward gaze of the eyes in his three patients [1–3]. With only 300 cases reported by 2012 and variable estimated prevalence reported in the literature from 1 per 10,000 to 180,000, overall cherubism is known as a very rare disease with no definite prevalence determined up to date [4–6].

The hallmark of the disease is symmetrical multilocular radiolucent lesions expanding in the mandible and maxilla [4]. This, besides the swelling of the submandibular

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case, except for an elevated LDH and PLT, other laboratory findings were in the normal range. However, Calcium, phosphorus, alkaline phosphatase, and TNF-α were not assessed in our patients, which could be another limitation of this study.

While generally, no treatment is necessary as the mass tends to disappear with the child reaching puberty, in severe cases with functional disability, invasive treatment like surgery or radiotherapy has been suggested. Likewise, our patient had mild involvement and is under longitudinal observation [21, 22]. Surgery is not recommended in mild cases due to the possibility of recurrence. Although Steroids, TNF-α inhibitors, calcitonin, antineoplastic agents, immunosuppressants, and monoclonal antibodies have been reported as an option for the pharmacological treatment of cherubism, none have yet been confirmed by clinical trials as a possibility for effective treatment [22–24].

Conclusion

To our knowledge, this is the first reported case of cherubism in a 6-month-old infant.

Non-hereditary Cherubism, despite scarcity, can present in children below two years of age, even as early as the beginning of primary dentition.

Although treatment options for cherubism remain limited, understanding this condition's clinical and histological features can help clinicians provide appropriate care and counseling to affected patients and their families, thus preventing false diagnoses and unnecessary investigations, especially in young patients like ours. Further research and long-term follow-up are needed to understand cherubism's pathogenesis better and develop new therapeutic approaches. Overall, our case report indicates the importance of having cherubism in mind as a differential diagnosis of bone disease, even in children under a year old, and highlights the value of interdisciplinary collaboration in managing rare genetic disorders.

Abbreviations

CXR	Chest X-ray
CT	Computed Tomography
SWS	Sturge-Weber syndrome
MRI	Magnetic resonance imaging
TNF-α	Tumor necrosis factor-α
LDH	Lactate dehydrogenase
PLT	Platelet
WBC	White blood cell
RBC	Red blood cell
ESR	Erythrocyte sedimentation rate
CRP	C-reactive protein
PT	Prothrombin time
PTT	Partial thromboplastin time
BUN	Blood urea nitrogen
Cr	Creatinine
INR	International normalized ratio

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Author contributions

SSH diagnosed the disorder, wrote the first draft of the manuscript, and supervised the project. AA was involved in data gathering and gaining ethical approval. Our pathologist, MM, made a diagnosis and reported the case based on pathology evaluations and prepared Fig. 3. ASD helped diagnose, reviewed, and approved the final draft FY contributed by reporting the patient's radiologic images and prepared Fig. 2. RB wrote, edited, and reviewed the manuscript. SR reviewed and approved the final version of the manuscript. All authors approved the final draft.

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Data availability

The datasets regarding our participant's laboratory, radiologic, and clinical findings during the current study are available from S.hamzavi55@yahoo.com upon reasonable request.

Declarations

Ethics approval and consent to participate

This study's ethical principles align with Helsinki's declarations. The patient's parents agreed to participate with their child in this project and provided written informed consent. The patient's identity was protected. This research was approved by the Shiraz University of Medical Sciences ethics committee. Approval ID: IR.SUMS.REC.1402.025.

Consent for publication

Written informed consent for publication was obtained from the child's parents. All radiologic, laboratory, and pathologic findings and images are anonymized, and no patient identification is reported.

Conflict of interests

The authors declare no conflict of interests.

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