Case Report

Cherubism: A rare case diagnosed on fine-needle aspiration cytology

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ABSTRACT

Cherubism is a rare self-limiting nonneoplastic disease of the bone characterized by bilateral painless enlargement of the jaws giving a cherubic appearance to the patient. It is an autosomal-dominant disorder due to mutations in the SH3 domain-binding protein 2 gene on chromosome 4p16.3, but may occur sporadically. Here, we describe the clinical, radiological, and cytological features of cherubism in a 12-year-old girl.

Key words: Cherubism, jaw, mandible, painless enlargement

INTRODUCTION

Cherubism or multilocular cystic disease of the jaws was first recognized in 1933 by Jones.[1] He designated the descriptive name "cherubism" because the full round cheeks and the upward cast of the eyes give the children a cherubic appearance.[2] It is a very rare disorder with an estimated 300 cases reported in literature.[2] It is a benign, self-limiting fibro-osseous disorder characterized by bilateral symmetric enlargement of the mandible or maxilla.[3] The pattern of inheritance is autosomal-dominant[4] with tendency toward spontaneous remission. It is evident around 3rd or 4th year of life and it is one of the very few genetically determined osteoclastic lesions. Ramon and Engelberg[5] proposed a grading system for cherubism: Grade 1 (involvement of both mandibular ascending rami); Grade 2 (same as Grade 1 plus involvement of both axillary tuberosities); Grade 3 (massive involvement of whole maxilla and mandible, except the condylar processes); and Grade 4 (same as Grade 3 with involvement of the floor of the orbits causing orbital compression). We report a case of a girl presenting with diffuse painless enlargement of the lower jaw.

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CASE REPORT

A 12-year-old female child presented with the chief complaints of diffuse painless enlargement of both jaws. History revealed that it started as a small swelling that was progressively increasing in size for the past 4 months. Family history showed that the patient's elder sister had similar fullness of cheek in childhood, which spontaneously decreased substantially in size with increase in age. Clinical examination showed diffuse bilateral enlargement of the mandible [Figure 1]. On dental examination, disturbance in the arrangement of teeth was noted. X-ray revealed bilateral, multilocular, radiolucent lesions extending from the ascending ramus to the body of the mandible excluding the condyle region. Computerized tomography demonstrated multicystic bony lesions in the mandible with expansion and erosion in both cortices of the mandible. Laboratory investigations were performed, which showed an elevated serum alkaline phosphatase (ALP) level while serum calcium, parathormone, and other lab investigations were within normal limits.

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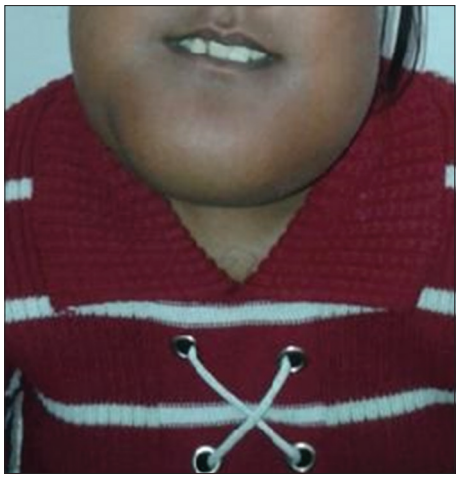

Figure 1: Clinical photograph of the patient showing diffuse enlargement involving both the jaws

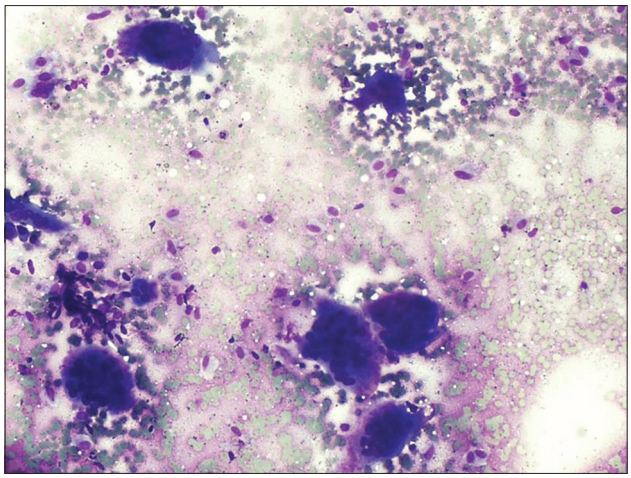

Figure 2: Fine-needle aspiration cytology smear showing stromal cells and multinucleated osteoclastic giant cells against a hemorrhagic background (Giemsa, ×100)

Thereafter, fine-needle aspiration cytology of the face swelling was done. Giemsa-stained smears were cellular showing many oval-to-spindle-shaped stromal cells having moderate amount of cytoplasm; along with these, large number of multinucleated osteoclastic giant cells against a background of hemorrhage were seen [Figure 2]. Thus, the possibility of giant cell-rich lesion was considered. Based on the characteristic clinical picture, typical family history, and cytology and radiology findings, a final diagnosis of cherubism was given. The patient and her family were thereafter counseled and were kept on regular follow-up examinations.

DISCUSSION

According to the WHO classification, cherubism belongs to the nonneoplastic bone lesion group involving the mandible. It was first described by Jones in 1933 as "familial multilocular cystic disease of jaws."[1] It is a very rare disorder with an estimated 300 cases reported in literature.[2] The pattern of inheritance is autosomal-dominant and its penetrance is 100% in males and 50–70% in females. However, the clinical expression of the disease is variable.[4] It usually begins between 2 and 5 years of age followed by a phase of rapid growth until 7 and 8 years, ultimately leading to a phase of slow growth until puberty and a phase of stabilization and remission at about 30 years.[6] The widely accepted theory for the pathogenesis of cherubism is the perivascular fibrosis leading to the mesenchymal disorder and decreased oxygenation.[7] Mutations in the axon 9 of the SH3 domain-binding protein 2 (SH3BP2) genes on Ch 4p band 16.3 have been identified in many families with cherubism.[3] There are indications that SH3BP2 plays a role in regulating the increased osteoblast and osteoclast activities in normal tooth eruption and so, point mutations in the gene could cause pathologic activation of osteoclasts. Mutation in gene encoding for fibroblast growth factor receptor III has also been seen in some cases of cherubism.[6] Radiographically, it appears as numerous well-defined multilocular radiolucencies of the jaws. Mineral metabolism is normal in these patients with serum levels of calcium, parathyroid hormone (PTH), PTH-related peptide, calcitonin, and ALP usually being within normal range.[2,7] Serum levels for ALP, however, may be increased during the active stages of cherubism.[2,7] The differential diagnosis of cherubism includes giant cell tumor, giant cell granuloma, and brown tumor of hyperparathyroidism, which may show similar cytological and histological picture.[8] However, these differential diagnoses can be ruled out based on the other findings such as giant cell granuloma is usually seen in adults, is unilateral, and develops in the anterior mandible;[7] giant cell tumor commonly involves epiphyses of long bones and normal serum levels of calcium, phosphorus, and ALP, which rules out hyperparathyroidism.[2] Thus, the limited and symmetrical distribution of the cherubism lesions along with familial history, clinical, and radiological findings can often facilitate the distinction of cherubism from these conditions.[2,8,9] Thus, there is no distinctive cytological feature that is specific for the diagnosis of cherubism, and clinical correlation is extremely important in distinguishing it from other differentials. The treatment ranges from close regular follow-up to surgical intervention for relieving complications and/or cosmetic purposes. Surgical procedures should be performed after puberty when the lesions are quiescent.[2] Therapy with calcitonin has been suggested by few authors to curtail the disease and reduce the need for surgery by inducing bone resorption.[7]

CONCLUSION

Cherubism is a rare osseous disorder of children and adolescents. Although the radiologic characteristics of the disease are not pathognomonic, it should be included in

the differential diagnosis of painless bony enlargement of jaw in children. Since it is a self-regressing condition, follow-up with minimally invasive treatment is only required.

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Conflicts of interest
There are no conflicts of interest.

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