

Early Presentation of Cherubism

Cherubism is a rare genetic disorder with approximately 300 cases reported worldwide. The disorder typically begins in children at ages of 2-7 years affecting males and females with equal frequency [1]. The lesions usually first appear symmetrically in the angle of mandible; rarely involvement of condyles and zygomatic arches has been reported. Lesions are limited to the jaws, and in most cases begin to regress with the onset of puberty. Respiratory problems due to backward displacement of tongue or obliteration of the nasal airway may manifest as upper airway obstruction. Extracranial involvement is extremely rare. Biochemistry is usually normal in these patients [2].

A 9-month-old girl presented with progressive enlargement of the facial bones first noticed at 3 months of age. The enlargement was gradual, involving the maxilla and the mandible bilaterally initially (**Fig. 1**); followed by development of palpable firm to hard lesions over affected bones without any pressure symptoms. She was referred with a probable diagnosis of fibrous dysplasia. CT scan revealed symmetrical enlargement of mandibles involving the body, ramus, coronoid and condylar processes with loss of normal trabecular pattern and ground glass opacity in involved bones (**Fig. 1**). Maxilla, sphenoid wings, body and pterygoid plates showed similar changes. A bone scan revealed overgrowth of mandible with increased uptake and no other significant bony abnormality. A biopsy was advised but the parents refused for the same.

Grading systems for cherubism have been suggested to describe location and severity of lesions. There are no distinguishable histological lesions specific for cherubism. The disease usually occurs due to dominant mutations on *SH3BP2* gene located on chromosome 4p16.3 [3,4]. The differential diagnoses include brown tumor of hyperparathyroidism, giant cell lesions, fibrous dysplasia, aneurysmal bone cyst and the hyperparathyroidism-jaw tumor syndrome.

Follow-up every 2 to 5 years is advisable after the disease becomes quiescent. Surgical intervention is indicated when aesthetic or functional concerns arise.

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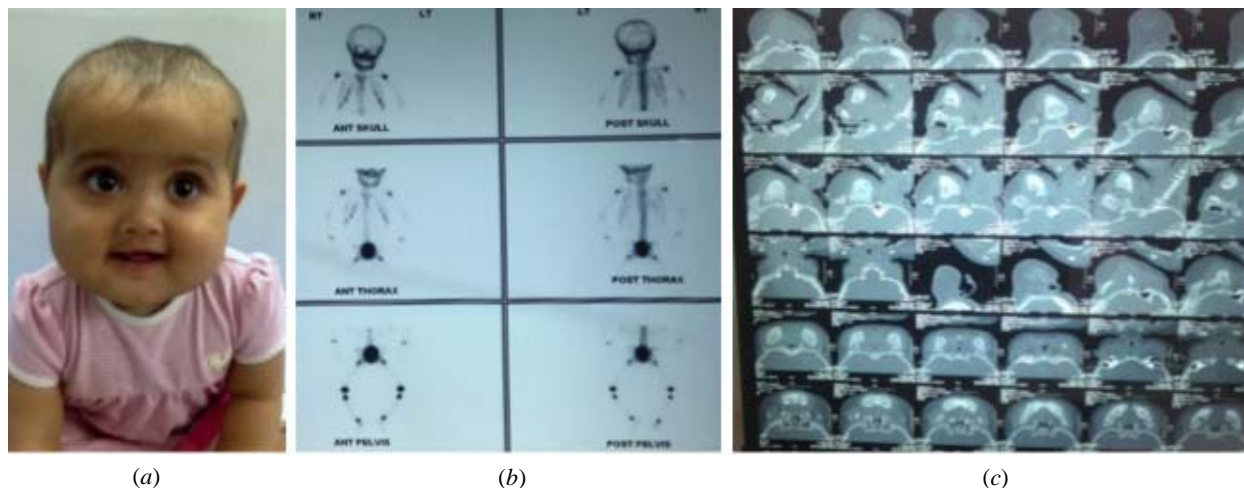


FIG.1 (a) Typical faces of child showing fullness of maxilla and mandible with upward slanting of eyes and the sclera visible below the irises; (b) Bone scan demonstrating increased uptake in the mandible and maxilla with no other abnormality; (c) CT scan of the child showing symmetrical enlargement of the mandibles.