Clinical History:

A patient with known cherubism presented with facial deformity and bilateral exophthalmia.

Imaging Findings:

A 12-year-old boy with known cherubism was presented with facial deformity, and bilateral exophthalmia. There was no associated trauma, tooth extraction, constitutional disturbance or pain. The parents and other relatives, as far as could be investigated, did not have any facial deformity. Bilateral eyes were proptosed 6 mm with superior globe displacement and marked lower lid retraction. There was moderate restriction of upgaze in the left eye. In addition, there was scleral indentation due to orbital floor maxillary bone hypertrophy. The pupillary responses were normal and there was no optic atrophy but visual field examination showed generally reduced sensitivity in the left eye. A panoramic radiograph of the mandible revealed multiloculated osteolysis involving the entire mandible with dislocated teeth. Only the mandibular condyles were not involved. X-ray of the remaining skeleton showed no abnormality. CT scan showed soft tissue density masses occupying mandible, maxilla (Figure 1), and orbit with disrupted cortex (Figure 2a, b). Three-dimensional (3D) CT image exhibited a symmetrically expanded mandible and maxilla. In addition, 3D CT scan confirmed bilateral bony masses protruding along inferior orbital walls towards orbital apices (Figure 2).

Discussion:

Cherubism was first described by Jones in 1933 as familial multilocular cystic disease of the jaws. The disease is not present at birth. It appears to have 100% penetrance in male patients but only 50–70% penetrance in female patients. A genome-wide search has established linkage to chromosome 4p16 (1-3). The affected jaw begins to swell in early childhood and increases until puberty after which it does not progress further. The maxilla and mandible are usually bilaterally enlarged giving a fullness of the cheeks and jaw. Although, the mandible is more frequently affected but lesions affecting the maxilla are more aggressive, as observed in our patient. This also causes traction on the lower lids and with superior globe displacement (1, 2–5). Ramon and Engelberg (4) proposed a grading system for cherubism based on involvement; Grade 1: involvement of both mandibular ascending rami, Grade 2: grade 1 plus involvement of both maxillary tuberosities, Grade 3: massive involvement of the whole maxillae and mandible except the condylar processes Grade 4: grade 3 plus involvement the floor of the orbits, causing orbital compression. In our patient, the lesions were classified as grade 4, according to the grading system. The typical radiographic appearance of cherubism is that of bilateral, well-defined, multilocular radiolucencies that can affect the mandible and the maxilla. Expanding lesions often cause thinning of the cortex and, in the maxilla, may cause obliteration of the maxillary sinus. In our case, CT scanning helped to provide a clear delineation of the extent of disease, which was difficult on radiographs due to the overlap of the facial bones. In the present case, the CT
showed a multilocular appearance in the mandible created by the presence of bone septa and expansile remodeling and perforation in some places. Moreover, we noticed the involvement of both maxillae. The lesion expanded into the maxillary sinus and caused the osseous involvement of orbital floors bilaterally. In addition, virtual reconstructions using 3D-CT imaging were also performed to provide a better anatomical visualization of the extent of the lesions (2, 6–8). The differential diagnosis of cherubism consists of giant cell granuloma of the jaws, osteoclastoma, odontogenic cyst, ameloblastoma, odontogenic fibroma, myxoma, aneurysmal bone cyst, fibrous dysplasia and hyperparathyroidism (9, 10). Giant cell granuloma is usually unilateral and usually affects patients between the ages of 20 and 40 years, whereas cherubism is a symmetric lesion (9). Unlike cherubism, osteoclastoma rarely occurs in the jaws. Aneurysmal bone cyst may also exhibit giant cells, but its main feature is a cavity lined with tissue other than endothelium. Polyostotic fibrous dysplasia first presents in the second or third decade of life. Hyperparathyroidism rarely affects the jaws in an isolated manner. Bilateral odontogenic cysts are rare in the first 5 years of life (9, 10). In conclusion, cherubism can lead to various types of ophthalmologic complications. Exophthalmos and loss of visual acuity due to compression of the optic nerve are the most common. Clinicians must be aware of these complications for proper detection.

**Differential Diagnosis List:** Orbital involvement in cherubism

**Final Diagnosis:** Orbital involvement in cherubism

**References:**

Description: Figure 1. Bone window axial (a) and coronal (b) CT scan through the mid-orbits and maxilla demonstrating maxillary involvement and decreased bilateral orbital volume. Origin:
Description: Figure 1. Bone window axial (a) and coronal (b) CT scans through the mid-orbits and maxilla demonstrating maxillary involvement and decreased bilateral orbital volume. Origin:
Description: Figure 2. Anterior view of volume-rendering 3D CT scan showing gross bony hypertrophy of the orbital floors (L> R). Origin:
Description: Axial CT imaging demonstrating multicystic bone lesions with expansion and erosion in the mandible and the maxillae. Origin: