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CASE REPORT

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Radiographic, CT and MRI features of cherubism

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Abstract Cherubism is a paediatric disease affecting the jaws. It is a close radiological mimic of giant cell lesions of the mandible. The radiographic features of this condition are well known; however, the MR imaging features have not been well described. We describe the radiographic, CT and MR imaging features of cherubism in a 5-year-old girl. We highlight the MR appearances of cherubic lesions and the ability of MRI to not only accurately depict the anatomical extent of the lesions, but also to reveal signal intensity changes in those areas that are apparently normal on radiographs and CT images.

Keywords Cherubism · Mandible · Radiography · CT · MRI · Child

Introduction

Cherubism, first described by Jones [1] in 1933, is a benign disease that usually manifests at 2–3 years of age with painless progressive swelling of the cheeks due to characteristic symmetrical involvement of the maxilla and mandible, with frequently associated dental malformations. It is usually progressive until puberty, after which it shows partial or complete spontaneous involution. The condition was initially thought to be familial [1–4], but many sporadic cases have now been reported. Previously the disease was considered as a familial form of fibrous dysplasia localized to the jaw [3, 4], but recent studies have shown it to be a genetically separate entity [5–8]. Mandibular involvement is typically bilateral; there has been only one report of unilateral involvement in an adolescent [9]. Maxillary involvement is variable and always accompanies mandibular involvement. Active treatment of the disease is generally not necessary, although limited surgical resection may be performed for cosmetic reasons or if function can be improved [10].

Radiographically, it resembles fibrous dysplasia and giant cell lesions, with multiple radiolucent areas with a ground-glass appearance in the jaws. The MRI appearance of the lesions, however, has not been well described [11]. We describe the radiographic, CT and MR appearances of a child with familial cherubism.

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Case report

A 5-year-old girl presented with a painless progressive swelling of both cheeks present from the age of 1 year, with progressive dental malformation and loss of teeth, and mild bilateral proptosis for the previous 3 months. On examination, the child had bilaterally symmetrical swelling of upper and lower jaws, with mild bilateral proptosis and an upward gaze. Many of the upper and lower teeth had been lost.

Radiographs revealed variably expansile, multiloculated lucent lesions distributed in the maxilla and mandible bilaterally with equivocal involvement of the right mandibular condyle. There was displacement as well as absence of many deciduous and permanent teeth (Fig. 1). The lateral view of the skull revealed a soft-tissue density mass in the posteroinferior part of the antra, causing a bulge in the posterior part of the orbital floor. The upper teeth were absent posteriorly, resulting in visualization of a large part of the hard palate ('hard palate' sign [4]) (Fig. 2).

CT clearly showed the presence of symmetrical, multiloculated cystic lesions affecting the mandible and maxilla. The mandibular condyles were seemingly not involved. The cysts had rounded scalloped margins and caused marked bony expansion, particularly of the outer cortex; they were filled with soft-tissue density material (Fig. 3).

MRI was performed to assess orbital involvement as the patient was developing progressive proptosis. The MR images clearly delineated the extent of mandibular and maxillary involvement and confirmed the lack of orbital involvement. Images in the sagittal and coronal planes

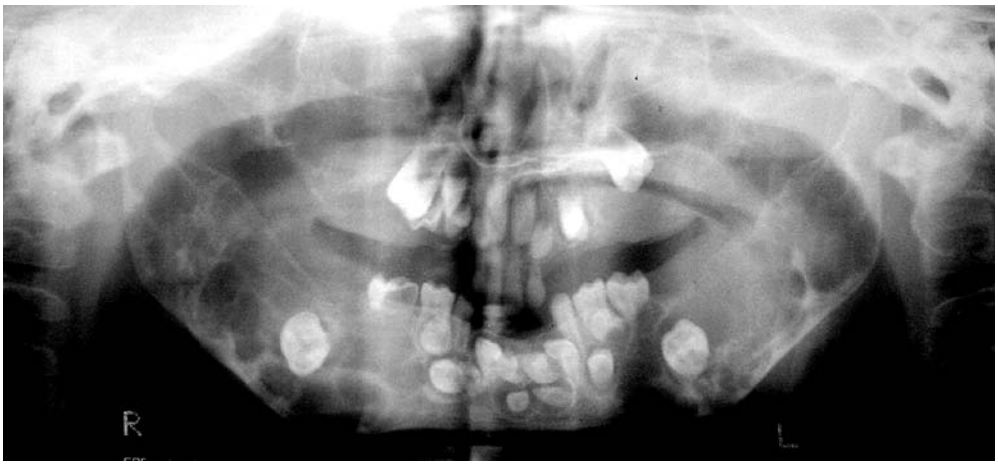

Fig. 1 Panorex view shows expansile, multiloculated, radiolucent lesions distributed symmetrically in the mandible, with apparent sparing of the mandibular condyles. There is extensive loss and displacement of teeth

clearly demonstrated the lesions in the roof of maxilla protruding into the floor of both orbits causing proptosis, as well as their relation to the optic nerves. The signal intensity of the lesions was heterogeneous with areas of isointensity to skeletal muscle on T1-weighted (T1-W) images that were correspondingly hyper- to isointense on T2-weighted (T2-W) images. Within these lesions, there were areas that were T1 hypointense and T2 hyperintense (Figs. 4 and 5). In addition, MRI also demonstrated similar signal intensity changes in the right mandibular condyle (Fig. 6).

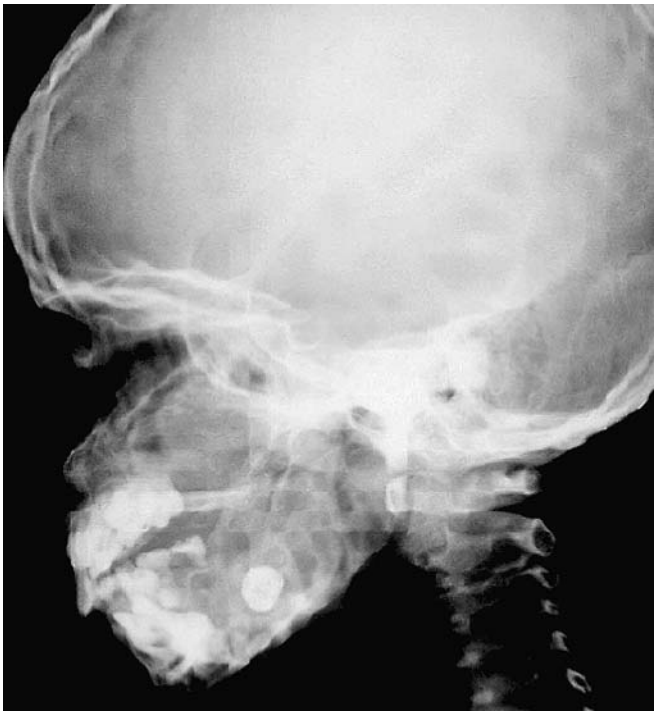

Fig. 2 Lateral view of the skull shows a soft-tissue density in the region of maxillary antra causing anterior displacement of the maxillary teeth with visualization of the posterior part of the hard palate (the 'hard palate' sign)

The child's mother also had similar painless swelling of both cheeks that slowly progressed until puberty and partially regressed in adulthood. The lesions had been partially excised for cosmetic purposes. Unfortunately, preoperative imaging was not available. Radiographs in adulthood showed multiloculated lesions with sclerotic margins distributed throughout the mandible, but without maxillary involvement (Fig. 7).

A diagnosis of cherubism was made based upon the characteristic clinical and radiological findings on a background of a positive family history. Histopathological confirmation was not obtained. As there was no significant compression of the airway or optic nerves and considering the young age of the subject and the tendency of these lesions to regress spontaneously, surgical intervention for the facial deformity was not contemplated and the patient was managed conservatively.

Discussion

Cherubism usually presents before the age of 5 years, most often between 12 and 36 months. Early studies found cherubism to be a familial disease [1–4]; however, many nonfamilial cases have now been reported [12, 13]. It has been described as a subtype of fibrous dysplasia, particularly the hereditary craniofacial variety, because of the close radiological resemblance of the two conditions [3, 4]. However, recent genetic analysis has shown that cherubism results from different mutations from those in craniofacial fibrous dysplasia, and the two conditions are distinct entities at the molecular level [5–8, 11].

The hallmark of cherubism is bilaterally symmetrical, well-defined, multiloculated radiolucent lesions in the mandible that usually extend from the region of the molar teeth towards the midline. Unilateral involvement of the disease has also been documented [3, 9]. Maxillary involvement is less frequent and usually less extensive, and is characterized by a soft-tissue density in the posteroinferior part of the maxillary antrum with forward displacement or absence of dental follicles in that region. This results in prominence of the posterior part of the hard palate

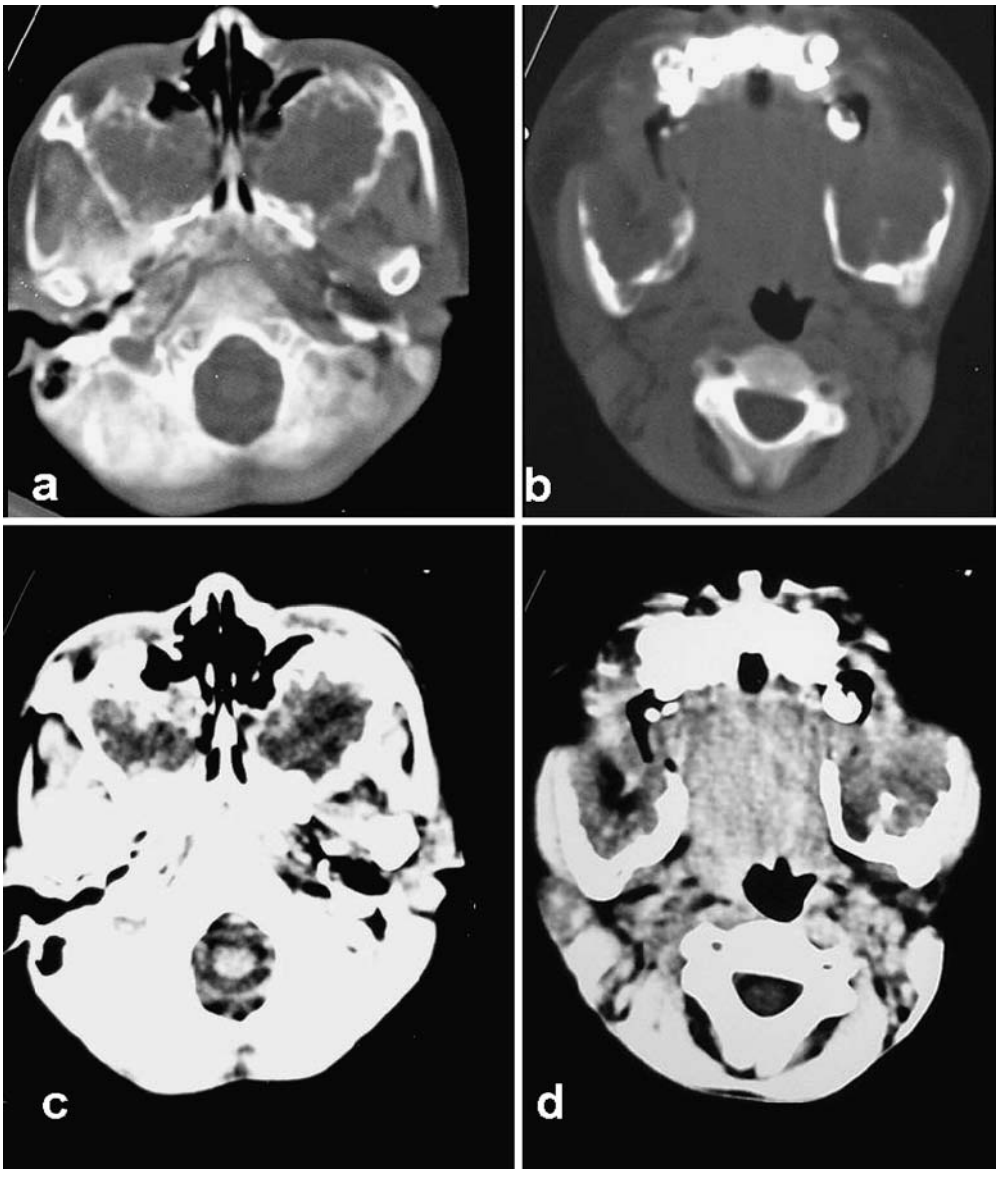

Fig. 3 Axial CT images at the level of the maxilla (**a**) and mandible (**b**) show grossly expansile cystic lesions involving both the bones with cortical scalloping. These cystic areas contain a low attenuation material within as seen on corresponding images at soft-tissue window settings (**c**, **d**). The mandibular condyles are normal

on a lateral skull radiograph ('hard palate' sign [4]). Dental anomalies such as nondevelopment or incomplete development of teeth, root resorption, and displacement, loss, or absence of teeth are frequently present. Our patient showed all these classic radiographic findings of cherubism.

The CT findings in our patient were also in agreement with those reported in the literature [11, 13], with multilocular lesions in the mandible and maxilla, having soft-tissue density within, with more expansion of the outer cortex of bone. 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.

MRI revealed the presence of heterogeneous signal intensity lesions, with areas of isointensity to skeletal muscle on T1-W images, which were correspondingly hyper- to isointense on T2-W images. Within these lesions, there were areas with hypointensity on T1-W and hyperintensity on T2-W images. These areas could possibly represent fluid-like signal intensity, but a FLAIR sequence was not available for confirmation. The MRI appearance of cherubism is not well described. Beaman et al. [11] noted nonspecific homogeneous isointensity to skeletal muscle on T1-W images and heterogeneous isointensity on fat-suppressed T2-W images. As noted by the authors, we also found MRI to be extremely useful in elucidating the exact anatomical location and extent of the cherubic lesions, particularly their relationship to the orbits and the optic nerves. The additional advantage of MRI is the lack of ionizing radiation and excellent soft-tissue contrast. In our case, MRI also demonstrated signal intensity changes involving the right condyle of the mandible. Involvement of these areas was not apparent on radiographs or CT images. Sparing of mandibular condyles was earlier considered a hallmark of this condi-

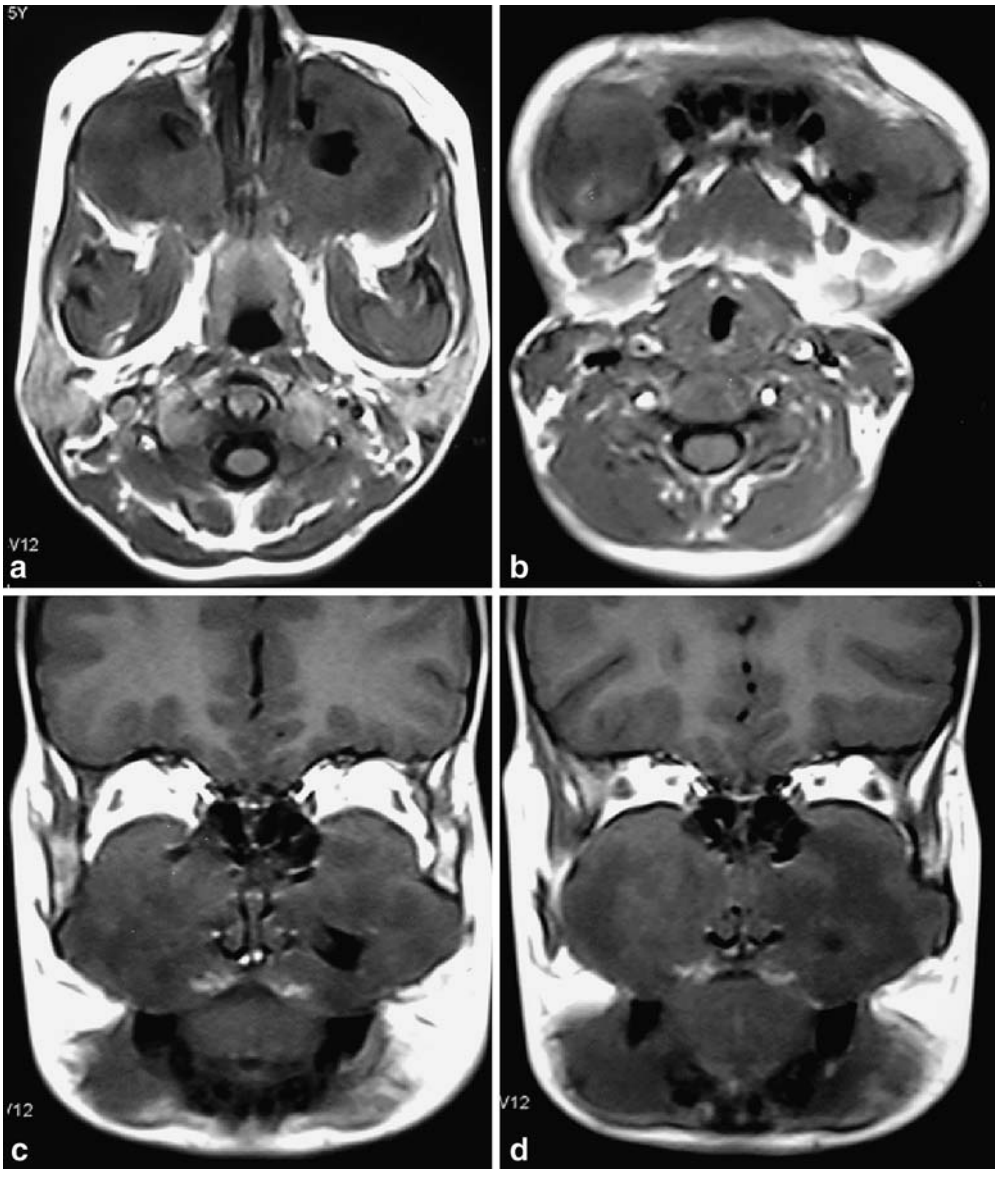

Fig. 4 T1-W axial (**a**, **b**) and coronal (**c**, **d**) images show heterogeneous signal intensity, expansile lesions with areas of isointensity to skeletal muscle in the maxilla and mandible, with areas of lower signal intensity within. Coronal images show the bulging of bilateral orbital floors by the maxillary lesions

tion [1–4, 12], but Beaman et al. [11] and Bianchi et al. [13] have also shown that condyles may rarely be involved. Therefore, the finding of condylar involvement should not necessitate an alternate diagnosis if characteristic cherubic lesions are seen involving the mandible and maxilla in a child.

The main differential diagnoses of cherubism include craniofacial variety of fibrous dysplasia, central giant-cell granuloma, brown tumours, and familial gigantiform cementoma. Fibrous dysplasia is not usually familial, presents slightly later between 10 and 30 years of age, does not show the typical "cherubic" look, and the lesions do not have a tendency to regress after puberty. In addition, the multiloculated, ground-glass lesions of fibrous dysplasia are rarely as symmetrical as the cherubic lesions. Central giant cell granuloma is a close mimic of cherubism but presents between 10 and 30 years of age. Radiologically, central giant-cell granuloma lesions have a predilection to involve the anterior mandible, they are rarely bilateral or symmetrical, and it is unusual for them to involve the posterior part of mandible or maxilla. Brown tumours of hyperparathyroidism are rare in the jaw region. Multiloculated radiolucent lesions may be seen with cortical bone thinning, but lesions are not symmetrical or bilateral. In addition, concomitant radiological signs of hyperparathyroidism, along with biochemical abnormalities make the distinction between the two, easy. Familial gigantiform cementoma is a rare disorder involving mandible and maxilla. The lesions are focal in distribution, predominate in the maxilla, and frequently extend to involve the orbits and nasal septum, the latter locations usually excluding the diagnosis of cherubism.

To conclude, cherubism is a rare, giant-cell-containing lesion of the jaw bones. CT and particularly MRI play a useful role in assessing the true extent of the lesions. MRI may also demonstrate signal intensity changes in areas that are apparently normal on radiographs or CT.

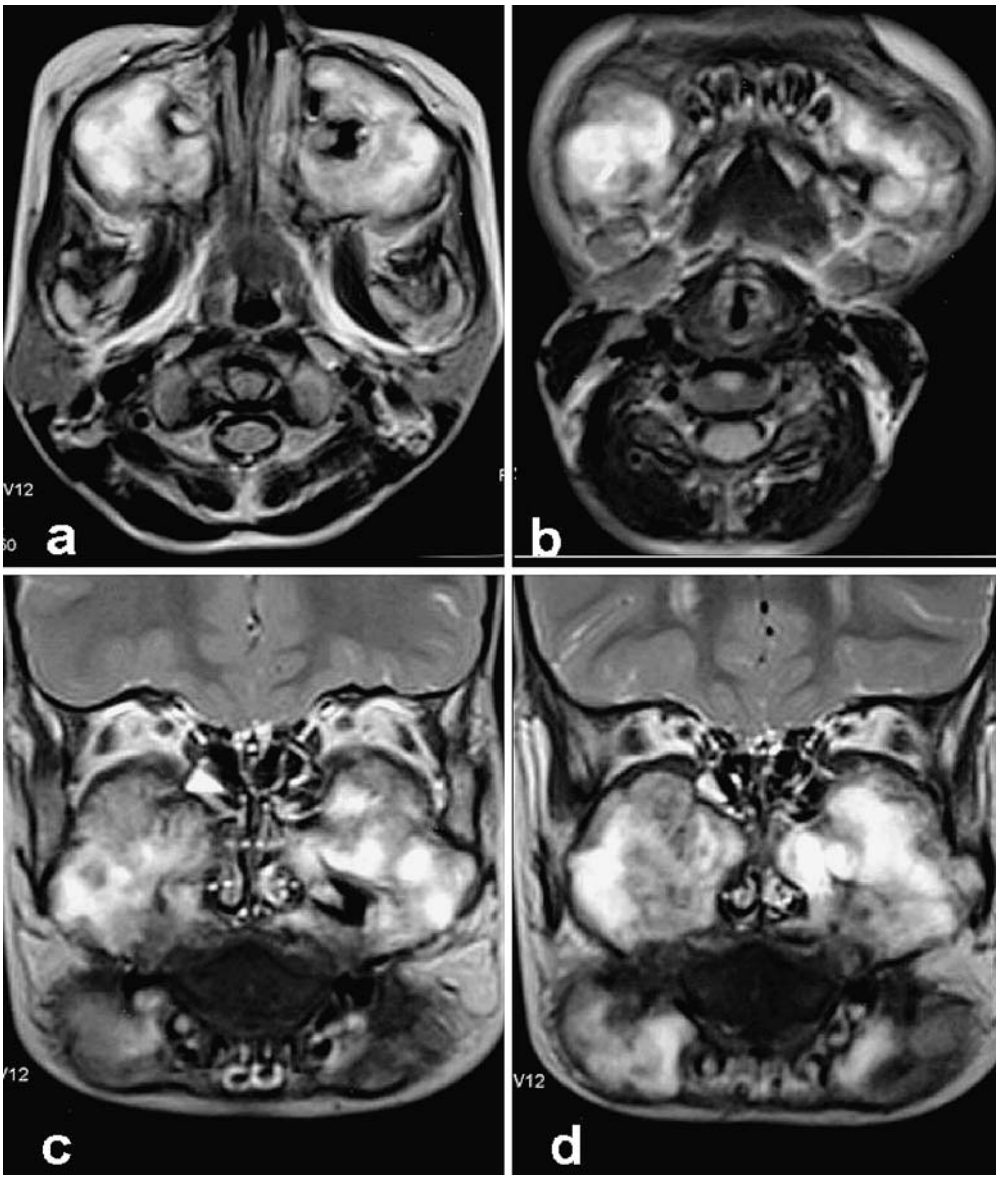

Fig. 5 Corresponding T2-W axial (**a**, **b**) and coronal (**c**, **d**) images show that the lesions have areas of hyper- to isointensity to skeletal muscle at the periphery and areas of increased signal intensity within

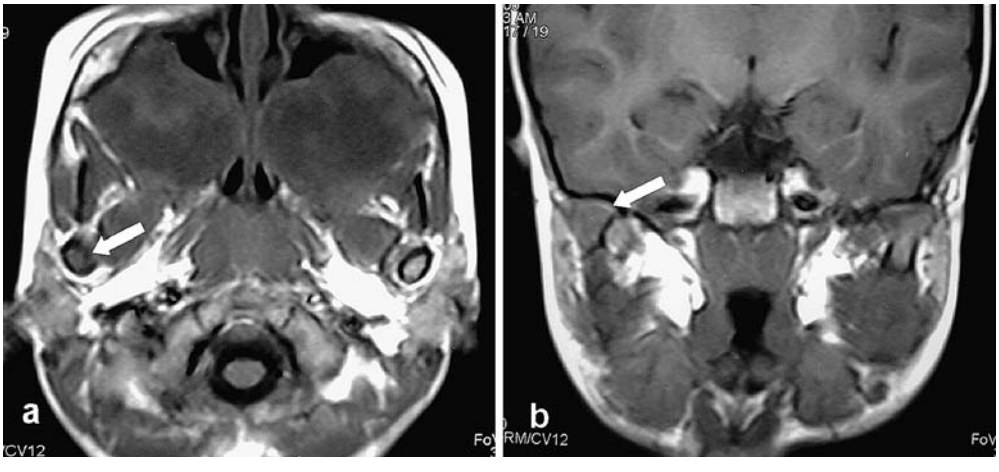

Fig. 6 T1-W axial (**a**) and coronal (**b**) images at the level of the mandibular condyles show the alteration of signal intensity involving the right condyle of the mandible (*arrows*)

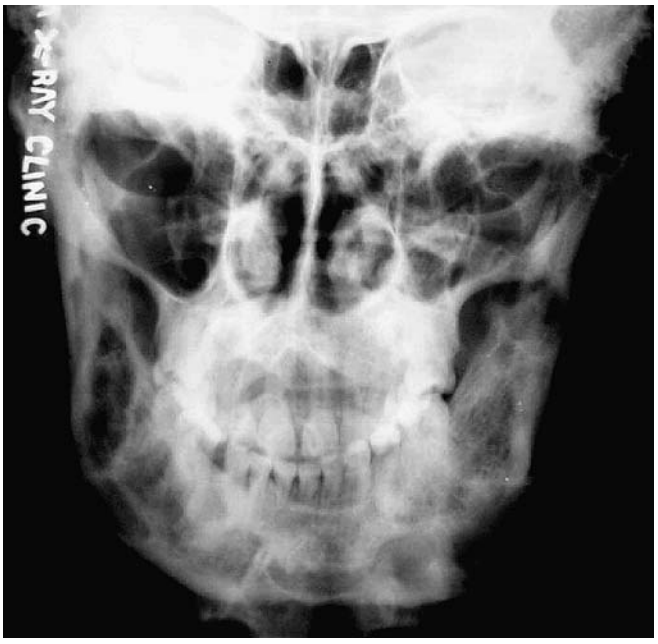

Fig. 7 Plain radiograph of the mother shows similar, expansile, multiloculated, radiolucent lesions with a sclerotic rim involving the mandible in a symmetrical distribution

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