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Case Report

Orbital involvement in cherubism

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Abstract

Cherubism is a rare, inherited condition characterized by fibro-osseous lesions of the maxilla and mandible. It has recently been localized to chromosome 4p16.3. The fullness of the lower half of the face and retraction of the lower lids gives the characteristic 'eyes raised to heaven' cherubic appearance. A case report of a 7-year-old girl with extensive orbital involvement of cherubism is presented. The patient underwent multidisciplinary surgery for the bony lesions, which extended from the maxillary antrum into adjacent structures, including extensive extraperiosteal orbital involvement. Cherubism may have orbital manifestations including lower lid retraction, proptosis, diplopia, globe displacement and visual loss due to optic atrophy. Ophthalmologists should be aware of the syndrome, its ophthalmic features and overall management.

Key words: bony, cherubism, fibro-osseous, orbit.

Case Report

A 7-year-old girl was referred for maxillofacial assessment after her dentist noted painless swelling of her jaws, dental anomalies and radiographic evidence of multilocular radiolucent lesions in the maxilla and mandible. Family history revealed her father had cherubism as a child and had undergone surgical treatment. She was then referred for ophthalmic assessment (Fig. 1).

Examination revealed the unaided Snellen visual acuity to be 6/6 in each eye. Extraocular movements were normal and cover testing showed she was orthophoric for distance and near. No relative afferent pupillary defect was detected and her fields were full to confrontation. Colour vision testing with Ishihara plates revealed 13/15 correct with her right eye and 14/15 correct with her left eye. Hertel exophthalmometry measured 15 mm (right) and 11 mm (left). Anterior segment examination was normal. Dilated fundus examination revealed normal discs with no evidence of pallor and healthy maculae. Retinoscopy showed 2.50 D of hypermetropia in each eye.

Computed tomography (CT) scan of the face showed multilocular radiolucent areas with intervening solid tissue involving the mandible and maxilla with marked extension of the lesion into the posterior right orbit (Fig. 2). The orbit had a reduced vertical dimension and the orbital contents were displaced superiorly. The lesion was closely related to the right optic nerve but did not appear to infiltrate the orbital contents.

Because of the extensive maxillary involvement with extension into the orbital cavity, the known expansile nature of the lesion and potential risk of compressive optic neuropathy, a multidisciplinary team performed surgery to debulk the orbital and maxillary lesions. Buccal vestibule maxillary incisions were made. Caldwell Luc antrostomy was performed. The anterior maxillary wall was partly removed for access. The lesion was removed from the right orbital floor subperiosteally.

Histopathology revealed fibrous proliferation with numerous multinuclear giant cells distributed irregularly throughout the lesion (Fig. 3). There was reactive new bone formation evident. The features were in keeping with the clinical diagnosis of cherubism.

One year postoperatively, the patient retained uncorrected visual acuity of 6/6 in both eyes. There was no proptosis nor evidence of any ocular problems (Figs 4,5). She subsequently had the lesions debulked from her mandible with no complications.

Discussion

Cherubism is a rare, painless, familial fibro-osseous disease involving the mandible and maxilla first described by Jones in 1933.[1] Approximately 175 cases of cherubism have been reported in the medical literature.[2] In three cases with orbital involvement, resection of the orbital lesions was performed for visual impairment,[3] visual impairment and pain,[3] and diplopia.[4] Cherubism is a familial disease, transmitted in

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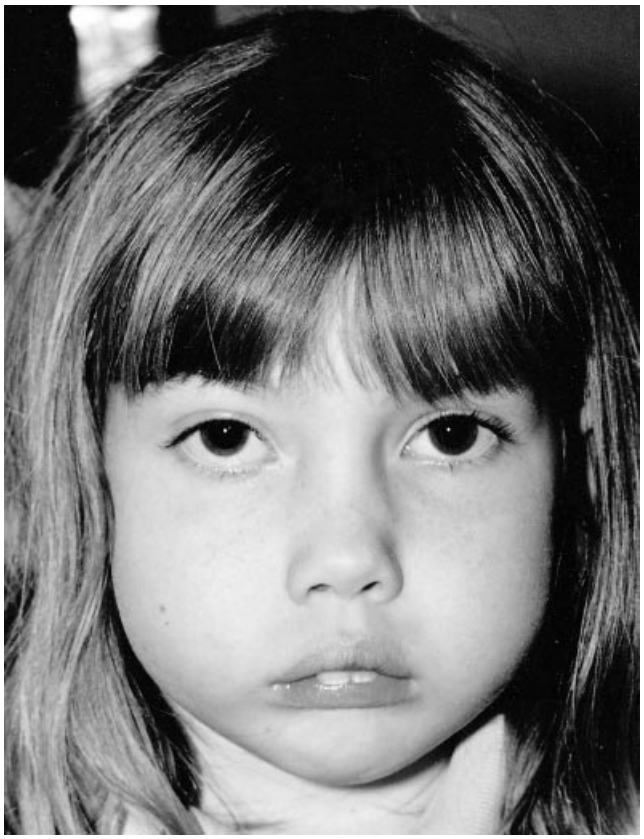

Figure 1. Preoperative photograph of 7-year-old girl with cherubism.

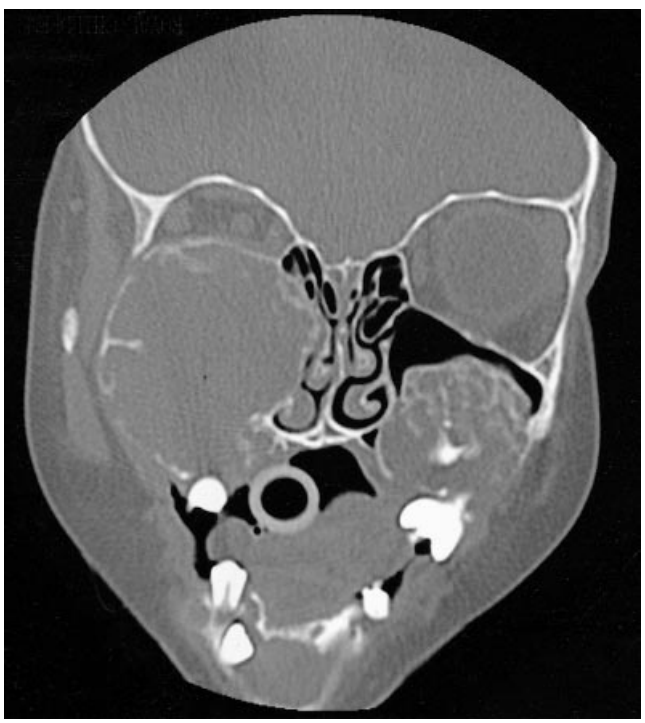

Figure 2. Bone window coronal computed tomography scan through the mid-orbits and maxilla demonstrating maxillary involvement and decreased right orbital volume.

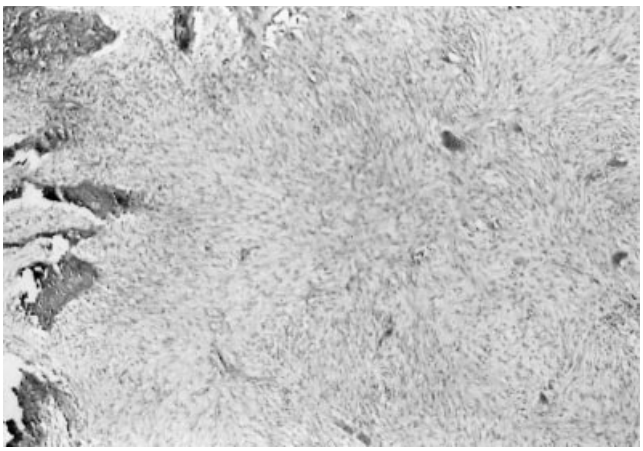

Figure 3. Histopathological appearance of the lesion showing mononuclear fibroblastic stroma with occasional multinucleate giant cells (H & E).

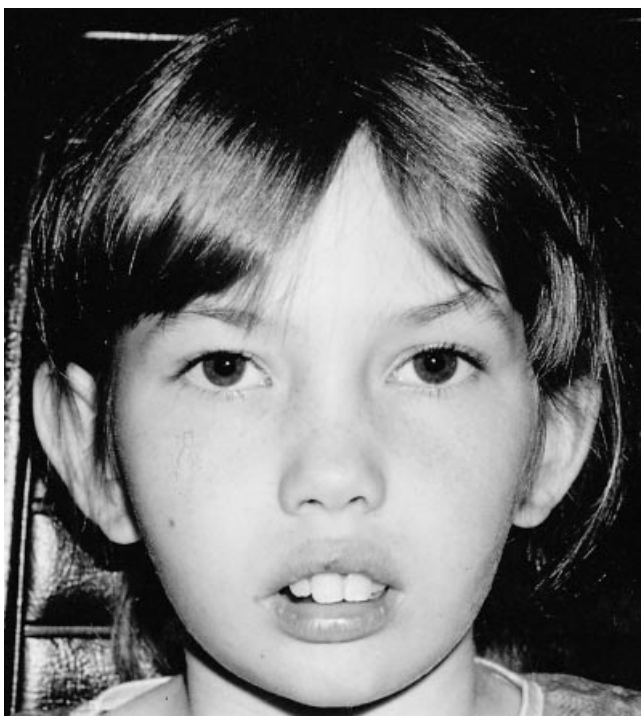

Figure 4. Postoperative photograph of the patient at 10 years of age.

an autosomal dominant fashion, with between 80 and 100% penetrance and variable expressivity.[5]

The maxilla and mandible are usually bilaterally enlarged giving a fullness of the cheeks and jaw. This also causes traction on the lower lids and with superior globe displacement, gives the eyes a 'raised to heaven' appearance that inspired Jones to coin the term cherubism after the cherubs of Renaissance art.[1,5,6] Proptosis, diplopia and visual loss due to optic atrophy have also been reported.[3,4]

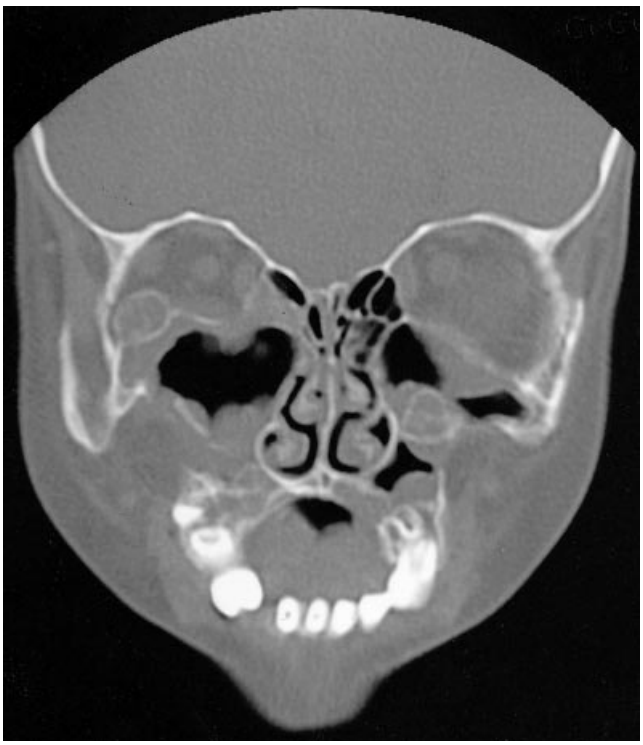

Figure 5. Postoperative coronal computed tomography scan showing debulked orbit and maxillary antrum.

The diagnosis of cherubism is usually made from the clinical picture, radiological findings, histopathological changes and family history,[6] but with the recent localization of the gene for cherubism to chromosome 4p16.3,[7,8] it is likely to become a genetic diagnosis. Classically, the affected child is normal at birth and develops the disease in the second or third year of life.[6] The mean age at presentation is 7 years and the disease tends to become more marked until puberty after which involution of the disease may occur.[5] With surgery there may be no residual deformity by adulthood.[5,6] Presentation is usually prompted by appearance and occasionally dental anomalies.[6] Radiographs reveal multiple multilocular radiolucent areas in the mandible and maxilla with expansion of the bony cortex.[1,6] The teeth are absent or displaced in the involved areas. The histology of the lesions reveals a mononuclear fibroblastic stroma, which contains multiple multinucleated giant cells, and there may also be irregular bone formation.[9] The histology is identical to that of giant cell granuloma of the jaw and must also be differentiated from fibrous dysplasia, hyperparathyroidism and osteoclastoma. Southgate *et al.* have shown the multinucleated giant cells to be osteoclasts.[9]

We have presented a case of cherubism with extraperiosteal orbital involvement in which ophthalmic input into the multidisciplinary management team was important. Although rare, ophthalmologists should be aware of this condition as an unusual cause of paediatric proptosis and globe displacement.

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