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Orbital neurofibromatosis with enophthalmos

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SUMMARY. **Two patients with orbital neurofibromatosis associated with enophthalmos are presented. Assessment using 3D-CT scan shows an increase in the size of the orbital cavity and an enlargement of the inferior orbital fissure, which allows orbital fat to prolapse into the infratemporal fossa, causing enophthalmos.**

Neurofibromatosis is an inherited disorder, occurring in approximately one in 3000 live births. The disease may be manifested in any system or organ, including skin, viscera, nerve, muscle and skeleton; orbital involvement has been reported to take place in 6–14% of cases (Hunt and Pugh, 1961; Burrows, 1963; Jacoby *et al.*, 1980). Plexiform neurofibroma in the eyelids is the commonest abnormality in the orbit, resulting in ptosis. Exophthalmos is common and is caused by the presence of an intraorbital tumour mass or, more typically, absence of the greater wing of the sphenoid (Morax *et al.*, 1988). The latter lesion allows the temporal lobe to herniate into the orbit, causing pulsating exophthalmos; enlargement of bony orbital cavity, especially in an inferior direction, is commonly observed with this proptotic deformity.

Enophthalmos of the affected orbit, although rare, has been described as resulting from an overwhelming increase in size of the bony orbit compared to that of the orbital contents (Van der Meulen, 1987; Morax *et al.*, 1988; Jackson and Shaw, 1990). Two patients with orbital neurofibromatosis associated with enophthalmos are presented here with their 3D-CT imaging (Fukuta *et al.*, 1990).

Case reports

Case 1

A 41-year-old male presented with a neurofibromatosis involving the left forehead, eyelid, temporal region and cheek (Fig. 1). The left eye showed 5 mm of enophthalmos and 3 mm of downward dystopia.

The preoperative 3D-CT scan revealed an increase in the size of the left orbit. The inferior orbital rim on the left was 3 mm lower than the normal side. The left superior orbital fissure was enlarged but not large enough to cause temporal lobe herniation into the orbit. The inferior orbital fissure was increased in size, resulting in prolapse of orbital fat into the infra-temporal fossa (Fig. 2). The orbital bony cavity volume was 31.1 cc on the left and 25.7 cc on the right. Both eyes were the same size. The neurofibroma infiltrated the subcutaneous tissue of the left middle and upper face, and the muscles in the left infratemporal fossa were also involved.

Case 2

A 34-year-old female presented with neurofibromatosis limited to the right palpebral region (Fig. 3). She had previously undergone a procedure on the upper eyelid for ptosis. The right eye showed 3 mm inferior displacement and 4 mm enophthalmos. Her preoperative 3D-CT showed a slightly enlarged superior orbital fissure, inferior displacement of the inferior orbital rim by 3 mm and an expanded inferior orbital fissure, through which the orbital fat escaped into the infratemporal fossa (Fig. 4). The volume of the right orbit was 33.6 cc and the left was 27.5 cc.

Discussion

The skeletal abnormality of the orbit in neurofibromatosis was first documented radiographically by Moore in 1931. Thereafter, the defect of the posterior wall of the orbit was described by several investigators using plain X-rays (LeWald, 1933; Holt and Wright, 1948). Hunt and Pugh (1961) stated that this deficiency should be considered characteristic of neurofibromatosis since its incidence was fairly high (13 of the 192 cases in their series). Recently computer tomography has improved the understanding of anatomical disorders in orbital neurofibromatosis (Van der Meulen *et al.*, 1982; Ziemmerman *et al.*, 1983; Jackson *et al.*, 1983; Grenier *et al.*, 1984; Marchac, 1984; Bite *et al.*, 1987; Van der Meulen, 1987; Morax *et al.*, 1988; Poole, 1989; Harkens and Dolan, 1990). The CT findings are enlargement of the orbit and middle cranial fossa, hypoplasia of the ethmoid, maxilla, zygoma and mandible, and depression of the orbital floor. The enlargement of the superior orbital fissure, which is sometimes referred to as a defect of posterior wall, and the herniation of temporal lobe into the orbit through this defect have been well demonstrated by CT scan.

The enlargement of the inferior orbital fissure has never been addressed. This is not limited to orbital neurofibromatosis with enophthalmos; a 3D-CT study of six patients in our series with pulsating exophthalmos showed an increase in size of both superior and inferior orbital fissure in all cases.

Whether the enlargement of the superior orbital fissure is the result of erosion by neurofibromatous tissue or congenital mesodermal dysplasia has caused much discussion. Hunt and Pugh (1961) emphasised the latter aetiology, based on the evidence that the

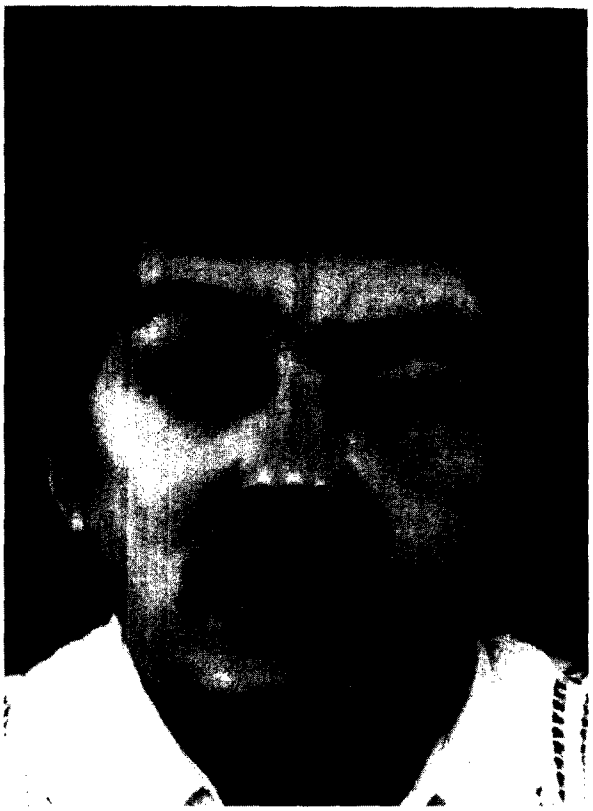

Fig. 1

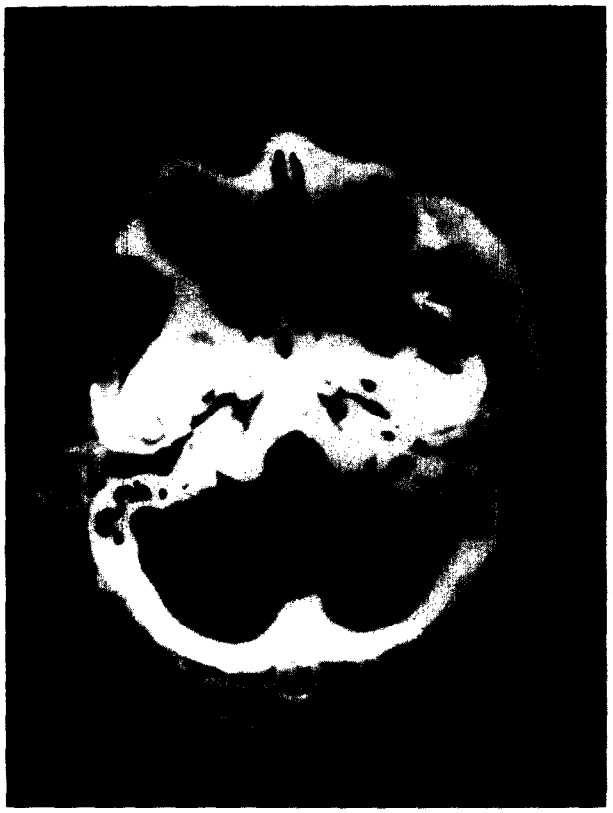

Fig. 2

Figure 1—Preoperative photograph of Case 1. A front view showing the redundant soft tissue in the forehead, eyelid and cheek of the left face.
Figure 2—A CT section of Case 1 through the inferior orbital fissure showing the prolapse of orbital fat (arrow) into the infratemporal fossa through the enlarged inferior orbital fissure.

Fig. 3

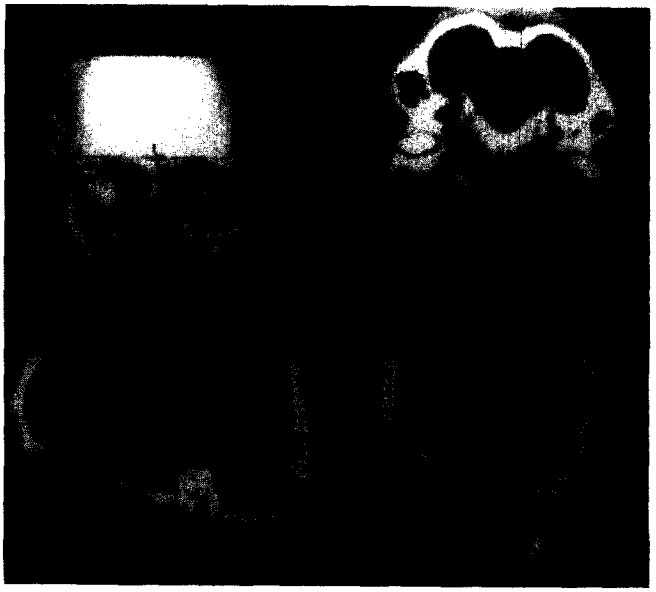

Fig. 4

Figure 3—Preoperative photograph of Case 2. A front view showing neurofibromatous involvement of the right temple and upper eyelid.
Figure 4—Preoperative 3D-CT scan of Case 2. Upper left: A bony surface image showing enlargement of the inferior orbital fissure and inferior displacement of the inferior orbital rim of the right orbit. Upper right: axial CT scan. Lower left: parasagittal scan. Lower right: coronal scan. All showing orbital fat herniation through the enlarged inferior orbital fissure.

pathological study of specimens taken from the posterior portion of the orbit frequently showed no tumour tissue. In the human embryo, the bones of the skull and face develop from the mesenchyme surrounding the cerebral vesicle or in the branchial arches. The lesser wing and root of the greater wing of the sphenoid are preformed in cartilage, followed by ossification. The lateral part of the greater sphenoidal wing and the maxilla are formed by direct ossification in the mesenchyme. In the third month, the large open space between the lesser wing and maxilla is separated into two areas by the triangular segment of the developing greater wing (Bollobás, 1984). As the ossification of each bony element spreads, these two bony gaps become narrowed and form the superior and inferior orbital fissures, respectively. If the mesenchymal element related to the sphenoid and maxilla is involved in the developmental defect, impairment of formation of those bones is considered to result in the enlargement of the superior and inferior orbital fissures. However, reports of progression of pulsatile exophthalmos over years (Poole, 1989; Harkens and Dolan, 1990) suggest that local pressure from the tumour may also play some role in the skeletal abnormalities.

Temporal lobe herniation into the orbit, or an intraorbital tumour mass such as neurofibroma, glioma and glaucomatous buphthalmos are associated with exophthalmos. The brain hernia characteristically produces the pulsating proptosis. The enlarged inferior orbital fissure which allows the orbital contents to prolapse into the infratemporal fossa, as well as the increase in the size of the orbital cavity, are considered to be causative elements of the enophthalmic deformity presented here (Van der Meulen, 1987; Morax *et al.*, 1988; Jackson and Shaw, 1990). Which lesion predominates is the decisive factor in orbital globe position in the A-P direction.

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