

Cranio-Orbital Anomalies in von Recklinghausen Neurofibromatosis Simulating an Intraorbital Space-Occupying Lesion

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Neurofibromatosis is a hereditary autosomal dominant disorder which is classified into two main types, referred to as von Recklinghausen neurofibromatosis (VRN) and bilateral acoustic schwannoma.¹⁻³ VRN, which is characterized by a variety of clinical features, is the most commonly encountered neurofibromatosis, affecting one in 3000 persons.⁴ To increase awareness of clinicians about the various pathology that can be associated with neurofibromatosis, the authors report a unique case of a VRN patient who had cranio-orbital anomalies which simulated a tumor.

Full endocrinological assessment was normal. The patient is being followed up.

Case Report

A 49-year-old Sri Lankan male, a known case of VRN, presented with a five-month history of diplopia and recurrent episodes of occipital headache associated with blurring of vision. He was investigated elsewhere and referred as a case of an intraorbital tumor. On examination, the patient had multiple cafe-au-lait spots all over the trunk and multiple skin nodules. He had a normal visual acuity bilaterally; however, there was limitation of lateral movement in the right eye. Fundoscopy was normal and there were no Lisch nodules detected in the iris.

The visual evoked response showed a P100 latency of 105 mSec in the right eye and 96 mSec in the left eye. Computed tomography (CT) scan showed an enlarged empty sella (Figure 1). The right sphenoid wing was absent (Figure 2). There was also evidence of brain tissue occupying the posterior aspects of the right orbit and the intracavernous portion of the right carotid artery appeared ectatic (Figure 3). Carotid angiography demonstrated the tortuous and ectatic right internal carotid artery (Figure 4).

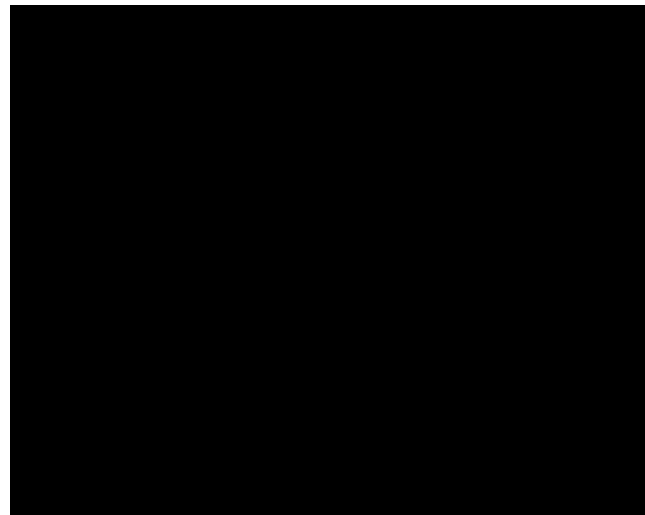


FIGURE 1. CT scan (+ IV contrast) coronal view showing an empty sella turcica.

TABLE 1. Cranio-orbital anomalies associated with VRN.⁶⁻⁸

Area affected	Anomalies affected
Skull	erosion of parietal bone macrocephaly congenital skull defect
Sphenoid bone	absence (hypoplasia) of the greater wing of sphenoid absence of the lesser wing of sphenoid sellar deformity sphenoid body lateral deformity
Orbit	erosion of the orbital wall - enlargement of the orbit - herniation of the temporal lobe into orbit erosion of the floor of the orbit
Petrous temporal bone and paranasal sinuses	enlargement of the middle cranial fossa posterior ethmoids lateral deformity poor pneumatization of the mastoids erosion of the internal acoustic meatus ethmoidal and maxillary bone hypoplasia

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Discussion

Since Fredric Daniel von Recklinghausen described neurofibromatosis more than a decade ago, a wide variety of manifestations related to this pathology have been recognized. The various cranio-orbital features encountered in VRN are summarized in Table 1.⁵⁻⁸

Orbital wall defects are reported to occur in 5% to 15% of VRN cases.^{6,9} On radiography, the orbital outline may

be enlarged with absence of the normal bony landmarks. Partial or complete absence of the greater or lesser wings of the sphenoid may be found (usually the greater wing is absent and the lesser wing is elevated and displaced medially).¹⁰ It is difficult to explain the numerous bone anomalies in VRN on the basis of involvement of the nervous system (ectodermal origin). It is believed that there is an associated mesodermal dysplasia and that the defect consists of absence or failure of development of that portion of the membranous bone separating the cranial contents from those of the orbit.¹¹ The resultant gap in the posterosuperior portion of the orbit allows direct contact of the temporal lobe with the orbital soft tissue, simulating an intraorbital space-occupying lesion. The continuous vascular pulsation of the brain may cause pulsating exophthalmos and can lead to grotesque facial deformity and loss of vision in the affected eye.¹⁰

In neurofibromatosis, the sella turcica may be enlarged and deepened, suggesting an intrasellar expanding lesion. There may be a downward tilting of the floor of the sella.¹² Erosion of the anterior clinoid process with a J-sign appearance to the floor and posterior clinoids have been described.¹² In our patient, empty sella occurred without previous exposure to surgery or radiotherapy. It is recognized that in neurofibromatosis patients, the sellar changes (and possibly the empty sella) can result from excavation and enlargement of the chiasmatic sulcus by neurofibromatosis tissue.^{7,12}

CT evidence of enlargement of the cavernous sinus is recognized in up to 71% of VRN patients.⁸ This may reflect a plexiform neurofibromatous involvement of either

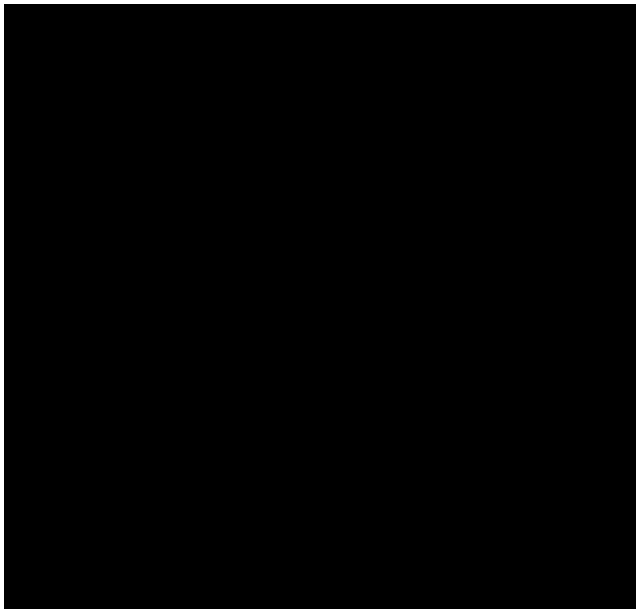


FIGURE 2. CT scan (bone window) showing an absent right sphenoid wing.

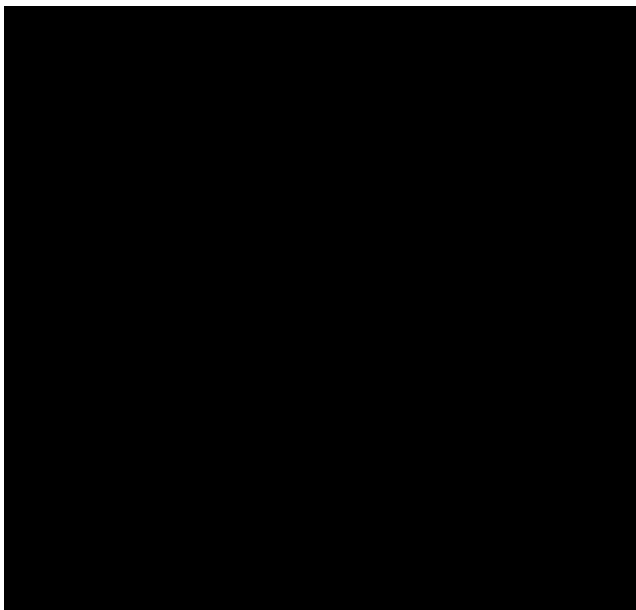


FIGURE 3. CT scan (+ IV contrast) showing brain tissue occupying the posterior aspects of the right orbit and ectatic intracavernous portion of the right internal carotid artery.

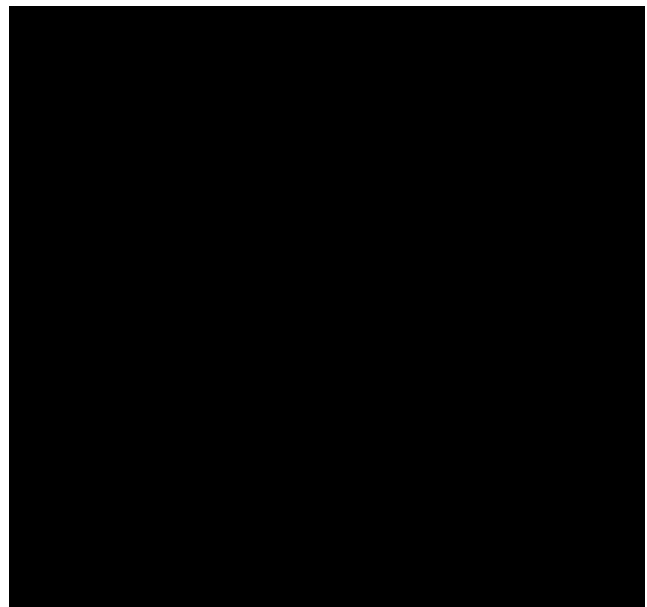


FIGURE 4. Right lateral common carotid angiogram showing a tortuous ectatic internal carotid artery.

the 3rd, 4th, 5th or 6th cranial nerves. In our case, there was no CT evidence of intracavernous plexiform neurofibroma. The enlargement was found to be related to the ectasia of the right carotid artery. The latter is a pathology very rarely associated with neurofibromatosis and its pathogenesis remains uncertain. The most commonly described pathological change has been abnormal cell proliferation within the arterial wall.¹³ In summary, an increasing awareness of the various malformations associated with neurofibromatosis is essential for the optimal management of these patients.

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