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Case Report

Bilateral Orbital-cranio NF Type I: 2 Rare Cases Presentation

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Abstract

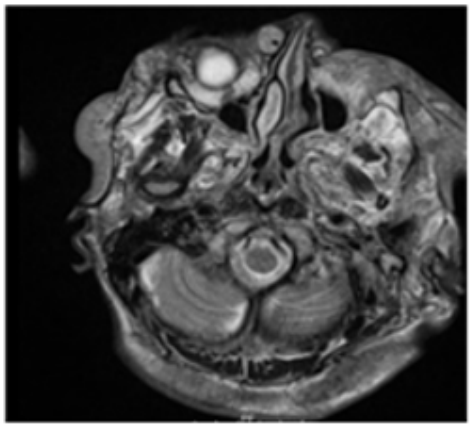
Introduction: Neurofibromatosis type I (NFI) is a fairly frequent (1 in 3000 live births), autosomal dominant, neurocutaneous disorder that has considerable clinical variability. In orbitocranio NFI (OCNF), which occurs in 1–22% of patients, NFs may cause progressive, disfiguring tumours of the orbital, facial, and temporal areas [1,2,4-8]. In this presentation, we report the experience with clinical observation and surgical management of rare bilateral OCNF, in two patients (2:162) seen at Chelsea and Westminster Hospital.

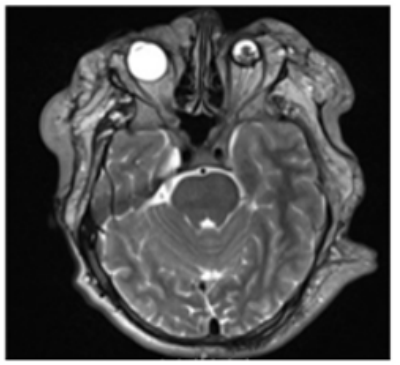
Method: Patients diagnosed with NFI were identified and medical records reviewed for demographic data, ophthalmologic examinations, surgical interventions and procedure outcome to create a retrospective, non-comparative case series of patients with bilateral OCNF seen at one medical centre over a 10-year period.

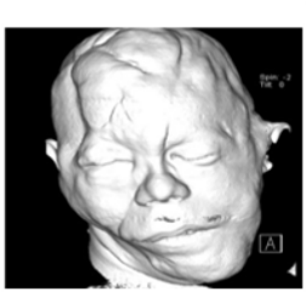
Result: Two patients with bilateral OCNF have been identified in a total of 162 cases with NFI. A 33yo man with extensive soft tissue infiltration (mass lesions) involving the face and anterior-superior scalp and neck, (Figure 1) ovoid mass posterior to the spinous process of C2, infiltration of the para pharyngeal spaces partially compressing the oropharynx, bilateral orbital involvement with small calcified globe on the left side and sphenoid wing dysplasia, asymmetry of the skull vault with relative thinning on the left hemisphere (Figure 2). He had several previous surgeries on both his eyelids, as far as cataract surgery and NF debulking from forehead and orbits. His current visual acuity is 6/18 in the right eye and no perception of light in the left.

Keywords: OCNF; Neurofibromatosis Type I (NFI); Tumour

Figure 1







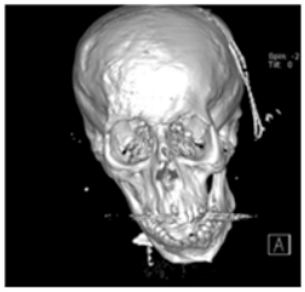


Figure 2

A 15 yo female with generalized NFI, positive family history of NFI (father) and NFI mutation positive (nonsense mutation exon 27a). Severe bilateral plexiform neurofibromatous changes involving orbits, forehead and periorbital regions more marked on the right (Figure 3). Right sided extension in pterygopalatine fossa, infraorbital canal and foramen ovale. Plexiform neurofibroma left hemi-tongue, right sphenoid wing dysplasia, multiple foci of abnormal signal intensity involving right cerebellar hemisphere, brain stem globus palladi and thalami. Long term swallowing problems with high arched palate, small stature, longstanding cervicothoracic scollosis. Severe visual impairment with glaucoma and bilateral cataracts (left removed). She had previous extensive orbital surgery (metal construction of the right orbital wall and roof), face and NF debulking.

Figure 3

Conclusion

Unilateral sphenoid wing dysplasia is one of the features of NF-I, but severe bilateral plexiform neurofibromatous changes involving orbits, forehead and periorbital regions in the same patient are very rare. (1:35 in the largest series to date). Patients with bilateral OCNF often require multiple procedures to preserve vision, prevent additional disfigurement and achieve cosmetic rehabilitation. Patients need regular ophthalmological monitoring given the potential for progressive visual and cosmetic consequences [3,9-12].

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