Case Report

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Complex Unilateral Orbital Dysplasia with Plexiform Neurofibroma, Presented as Unilateral Pulsating Proptosis: Staged Surgery and our Learning

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Abstract

Presence of sphenoid wing dysplasia fulfills one of the diagnostic criterias of neurofibromatosis type 1 (NF1). This defect of the skull base can manifests as pulsating proptosis. Associated calvarial defects, especially away from the lambdoid suture are a much rarer presentation. This report describes the successful removal of plexiform neurofibroma followed by reconstruction of the dysplastic sphenoid wing in an NF1 patient. Later on, he underwent blepharoplasty for scarring of the upper eyelid. After successful management of the overall procedure in three stages, the patient achieved improved visual outcome along with acceptable cosmetic appearance.

Key words: Sphenoid wing dysplasia; neurofibromatosis; proptosis.

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Introduction

Neurofibromatosis type 1 (NF 1) is an autosomal dominant disorder, having high penetrance with variable phenotypic expression.[1–3] Sphenoid wing dysplasia of the skull base is a well recognized feature among the NF 1 population.[3] Calvarial defects are rare and are thought to occur primarily near the lambdoid suture. Sometimes, these skeletal abnormalities can be associated with an ipsilateral orbital plexiform neurofibroma. Through the bone defect, protrusion of the temporal lobe into the orbit can cause pulsating proptosis.[4–6] According to the previously reported literature, intracranial route is recommended for the reconstructive surgery because of the wider access to the deformed skull base in NF1-affected individuals.[7–12] Recently, lateral orbitotomy can be used as an alternative approach, allowing the implementation of a titanium mesh to separate the brain from the orbital contents.[13] In our reported case, the patient presented with complex orbital dysplasia along with a large calvarial defect, justifies our reconstructive surgery through intracranial route. Later on, we went for blepharoplasty in collaboration with Department of plastic surgery for improved cosmetic outcome. We also reviewed the relevant literature and focused on the pathogenesis of this complex orbital dysplasia.

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Case Report

History and physical examination:

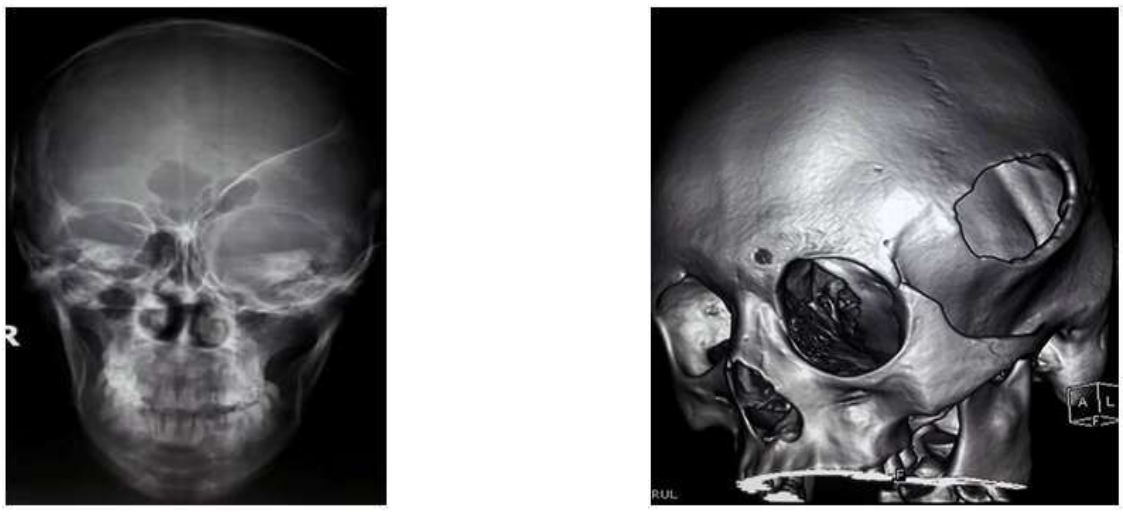
A 16-year-old man was presented to us with unilateral pulsating proptosis for 5 years and diffused swelling in the left temporal region since

his childhood. With this features, he applied herbal medications in his upper eyelid following which he developed significant scarring. Due to visual impairment and socially embarrassing cosmetic appearance, he had been referred to our facility for definitive management. On examination, the patient had more than six café-au-lait spots greater than 15 mm in maximum diameter, inguinal and axillary freckling and several cutaneous neurofibromas. The left orbit was enlarged and protruded in the cranio-caudal direction and the eyeball was displaced downward and medially (Fig. 1). The patient had impairment of vision of the affected eye in the form of only counting fingers from 3 feet with restriction of temporal field of vision. Jackson *et al.*[14] classified the orbito temporal manifestations of NF 1 into 3 Groups (Group 1, orbital soft-tissue involvement with intact vision; Group 2, both orbital soft-tissue and bone involvement with intact vision; and Group 3, both orbital soft-tissue and bone involvement with impaired vision or blind eye). According to their classification, the patient fitted into Group 3.

Fig. 1: Photograph showing extra-axial proptosis with scarring of upper eyelid.

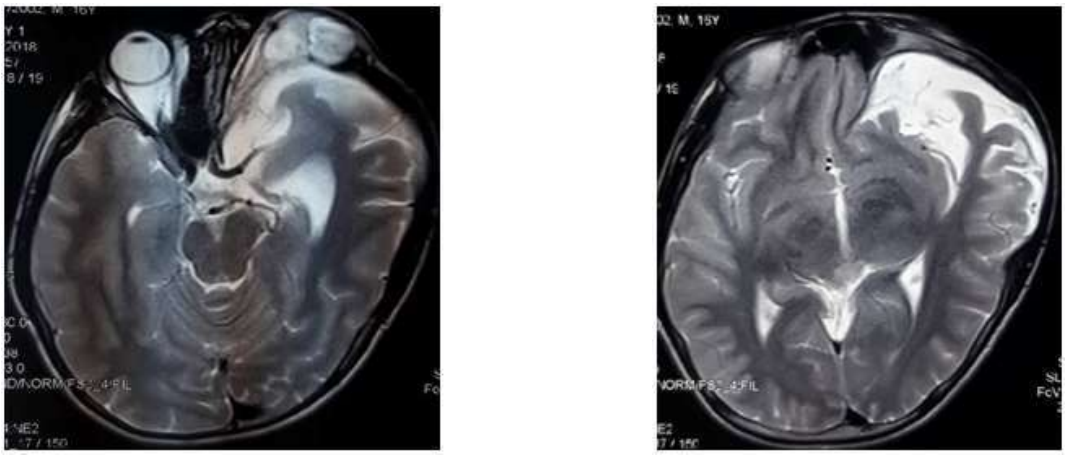
Investigations

Plain x-ray of the skull showed bare orbit sign with deviation of crista galli towards the left side (Figs. 2A, 2B). Computed tomography (CT) scan with 3D reconstruction of the skull were taken to assess the type and extent of the pathology. The scan depicted the loss of orbital roof and part of parietal and squamous temporal bone (Fig. 2B). The MRI of brain, axial view demonstrated the forward displacement of the orbital contents with the expansion of middle cranial fossa and dural ectasia. There was a hyper-intense well circumscribed extra-conal lesion present along the long axis of the optic nerve (Figs. 3A, 3B).



A B

Fig. 2: Plain X-ray of the skull showing bare orbit sign and deviation of crista galli towards the left side (A); CT scan with 3D reconstruction showing deficient orbital roof and a large oval calvarial defect (B).

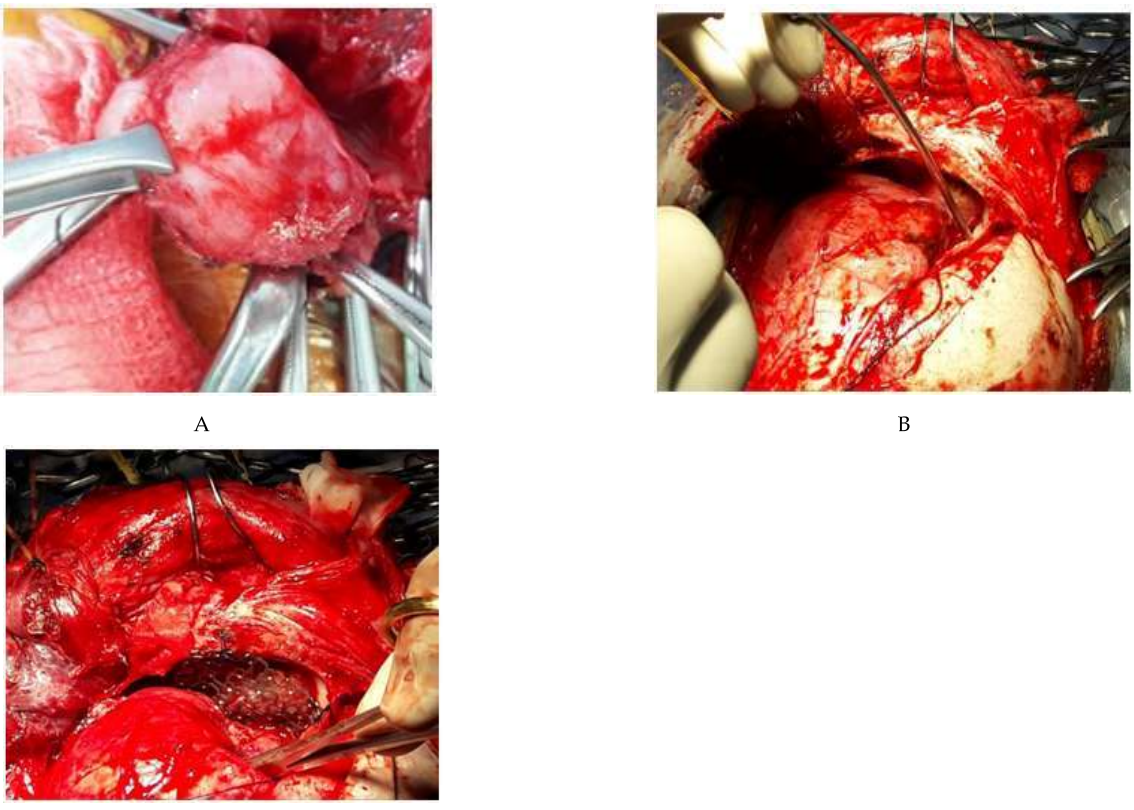


A B

Fig. 3: MRI of the brain showing expansion of middle cranial fossa contents with absence of lesser sphenoid wing, causing anterior displacement of the orbital contents (A) ; and presence of dural ectasia (B).

Operative procedure

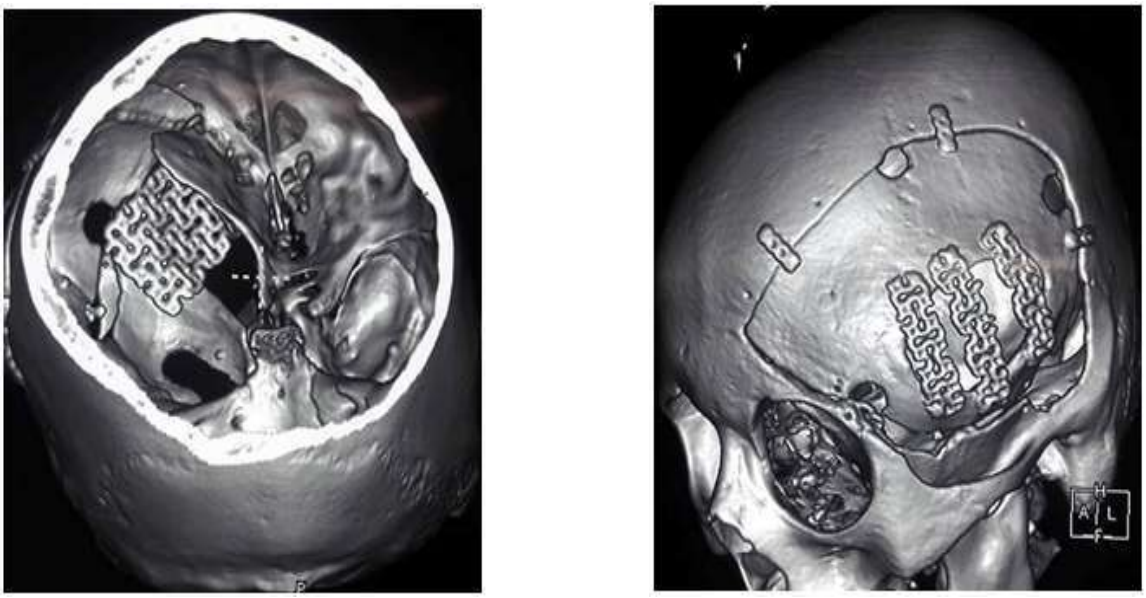
With all aseptic precaution, the patient underwent en bloc removal of the plexiform neurofibroma by subconjuctival dissection (Figs. 4A, 4B, 4C). 2 weeks later, he underwent left sided extended onto temporal craniotomy, excision of the deformed crista galli, reconstruction of the orbital roof and calvaria with titanium mesh (Figs. 5A, 5B). Later on, he underwent blepharoplasty to release the scarring in collaboration with the department of plastic surgery.



A B

C

Fig. 4: Subconjunctival En bloc excision of the plexiform neurofibroma (A) ; shifted crista galli and deficient orbital roof (B) ; Reconstruction of the roof with titanium mesh (C).



A B

Fig. 5: CT scan of brain 3D reconstruction showing, reconstruction of the orbital roof (A) and calvarial defect (B) with titanium mesh.

Postoperative period: Uneventful

Hospital course:

Vision improved from counting finger to 6/36. Patient could close his eyes with acceptable cosmetic appearance.

Discussion

Neurofibromatosis type 1 is a neurocutaneous disorder affecting approximately one in 3000 live births. The diagnostic clinical features are cutaneous neurofibromas, café au lait spots, Lisch nodules, axillary or inguinal freckling and nerve sheath tumors. It is an inherited autosomal dominant disorder caused by mutations in the NF 1 tumor suppressorgene, located on chromosome 17q11.2. The gene product, neurofibromin is a potent down-regulator of Ras proteins, which control cellular functions such as proliferation, differentiation and apoptosis. Loss of heterozygosity for the NF1 gene results in elevated levels of Ras guanosinetriphosphate in cells and the formation of neurofibromas. Osseous manifestations have been reported in approximately 50% of patients with NF1, manifests asidiopathic scoliosis (10–26% of patients), sphenoid wing dysplasia (3–11.3%), tibial pseudoarthrosis (1–4%), short stature, spinal meningocele, and macrocephaly.[15–20] In patients with NF1, there is a statistically significant association between sphenoid wing dysplasia and long bone defects.[21]

There is a close anatomical relationship between bony dysplasia and adjacent space-occupying lesion. It has been observed that plexiform neurofibromas are commonly present in patients with dystrophic scoliosis.[22–23] Prior case reports showed plexiform neurofibroma were found overlying the calvarial defect and/or sphenoid dysplasia.[24–26] Friedrich *et al*.[4] reported that orbital plexiform neurofibromas can be present in association with ipsilateral orbital dysplasia.[27] Other associated lesions have also been reported including meningioma, meningocele and lacunar skull.[28–30]

Another prominent space-occupying lesion associated with bony defects is a dural ectasia. Though this dural ectasia is most commonly associated with dystrophic scoliosis but can also be present in sphenoid dysplasia. In our reported case, dural ectasia was found in association with an adjacent sphenoid dysplasia along with a large calvarial defect. It has been suggested that the dural ectasia exerts a mechanical force that contributes to bony remodeling. Two emerging hypothesis suggests that the skull defects result either from a developmental malformation or from a destructive process. One hypothesis suggested that sphenoid dysplasia were the result of a malformation of the sphenoid bone arising from the mesoderm or neuroectoderm, resulting in unilateral defects that were thought to be of congenital origin.[31] Subsequently, with the revolution of radiology

and imaging techniques, there were several case reports of acquired skull defects, with and without adjacent tumors.[32]

Fibroblast growth factor signaling is important in both the endochondral and intramembranous ossification and thought to be involved in the secondary activation of the Ras pathway. It was thought that an adjacent plexiform neurofibroma and dural ectasia combined produces the pressure on the endocortical surface, which ultimately results in osteolysis. More recently, studies have evaluated the role of neurofibromin on bone metabolism. Vitro studies showed that loss of neurofibromin results in down regulation of osteoblastic activity and up regulation of osteoclastic activity.[33–35] In association, metabolic abnormalities also contribute to bony defects in NF1. Patients often exhibit low levels of 25—hydroxyvitamin D, which has been associated with osteopenia or fractures. It is postulated that vitamin D concentrations are inversely related to the number of café-au-lait macules.[36]

Regarding management, cranioplasty exerted a high incidence of resorption in the previously reported literature. Graft survival is improved with titanium mesh to reinforce the autologous graft and to promote stabilization of new bone.[37] In comparison with the previous study, our patient underwent reconstructive surgery through the same craniotomy, en bloc excision of neurofibroma as well as blepharoplasty. It is the author's recommendation that orbital neurofibroma should be explored early for optimum visual outcome. Later on, judicious reconstruction of the bone defects should be done which will vary among the patients.

Conclusion

Complex orbital dysplasia along with clavarial defects are rare complications of NF 1. Although plexiform neurofibromas and duralectasias are frequently observed, but not always present with skull defects. Our reported case supports the evolving theories of a dynamic skull defect pathophysiology. Despite having a rewarding overall outcome in short term follow up of our patient, progression may occur in more than half of the skull defects, reinforcing the need for long term follow-up.

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Ethics Approval: There are no ethical issues in this paper.

Abbreviations

CSF	:	Cerebrospinal fluid
CT	:	Computed tomography
FGF	:	Fibroblast growth factor
MRI	:	Magnetic resonance imaging
NF 1	:	Neurofibromatosis type I

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