

Isolated nonpulsatile enophthalmos in neurofibromatosis: An uncommon entity

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Isolated enophthalmos is a rarely observed entity in neurofibromatosis (NF). A 12-year-old male presented with right lower eyelid fat prolapse and enophthalmos for the past 7 years. There was no history of antecedent trauma/surgery. Computed tomography of orbit revealed an ill-defined intraconal hyperdense lesion located between lateral and inferior rectus along with an enlarged inferior orbital fissure (IOF). Superior orbital fissure was minimally widened without prolapse of any intracranial contents. Excision biopsy along with repair of widened IOF was performed through inferior transconjunctival route. Histopathology was suggestive of plexiform neurofibroma with positivity for S-100 and epithelial membrane antigen. No associated cutaneous lesions were present. Nonpulsatile enophthalmos with eyelid fat prolapse can be a presenting sign of NF.

Key words: Bony orbit, enophthalmos, neurofibromatosis

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Neurofibromatosis (NF) is a heredo-familial/sporadic phakomatosis with the variable phenotypic presentation.^[1] Phenotypic variation exists in the form of varying degrees of eyelid, orbital, and middle cranial fossa involvement. Orbital involvement presents most commonly with exophthalmos caused by either enlarged orbital cavity with prolapse of temporal lobe or presence of an optic nerve tumor.^[1] Pulsatile enophthalmos has been reported in 8 cases so far in literature and is attributed to enlarged superior and inferior orbital fissure (IOF) with orbital fat prolapse into infratemporal fossa.^[2-7]

Herein, we describe a rare presentation of NF as nonpulsatile unilateral enophthalmos with lower eyelid fat prolapse.

Case Report

A 12-year-old male presented with swelling in the right lower eyelid with smaller appearance of the same eye for the past 7 years. There was no history of trauma or surgical intervention. His best-corrected visual acuity was 20/20 in both eyes. Hertel's exophthalmometry reading was 13 and 16 mm in right and left eye, respectively. Right lower eyelid preseptal area had a central bulge with no palpable mass lesion [Fig. 1a and b]. There was increased bulge prominence while looking upward. Valsalva manoeuvre was negative. Extra ocular motility was full and free in all directions of gaze. Anterior and posterior segment was unremarkable. Computed tomography (CT) of orbit demonstrated deficient lateral wall (zygoma) with enlarged IOF causing prolapse of orbital contents into infratemporal fossa [Fig. 1c and d]. CT orbits showed a hyperdense irregular intraconal soft tissue lesion in inferior orbit located between lateral and inferior rectus. Greater wing of sphenoid was dysplastic with enlarged superior orbital fissure [Fig. 2]. There were no cutaneous lesions. The possibility of an old unnoticed orbital fracture or sclerosing orbital tumor was thought of as provisional diagnosis. Parents had cosmetic concern regarding the lower eyelid

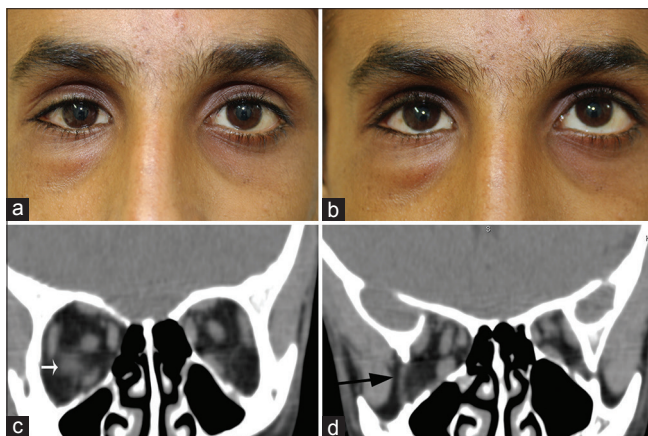


Figure 1: (a and b) External face photograph showing right enophthalmos with a prominent bulge in lower eyelid which increased on upgaze; (c) posterior coronal computed tomography cuts showing an ill-defined hyperdensity in right intraconal space between lateral and inferior rectus (marked with white arrow); (d) bulging periosteum across enlarged right inferior orbital fissure and prolapsing intraconal fat inferotemporally (marked with black arrow)

swelling. Inferior transconjunctival orbitotomy was carried out. Intraoperatively, a localized inferotemporal firm mass lesion was noted along the anteroinferior orbit, which had an appearance similar to tubular structures seen in eyelid neurofibroma. Lesion was extending into postseptal eyelid tissue. Excised lesion was sent for histopathology. Widened IOF with prolapsing orbital contents was noted. Prolapsed contents were repositioned back into the orbital cavity, and 1 mm thick porous polyethylene sheet was placed along the defect trimmed to the size of the defect. Postoperative course was uneventful. Microscopy showed a tumor characterized by tortuous proliferation of all components of peripheral nerves: the axons, Schwann cells, and perineural cells. Nerve sheath proliferation was seen within the perineurium of numerous dilated and disorganized nerve fascicles [Fig. 3a]. The nerve fibers were arranged in a haphazard to vaguely concentric manner with intervening fibromyxoid stroma. S-100 was positive in the Schwann cells and epithelial membrane antigen in the perineural cells. These features were consistent with a plexiform neurofibroma.

The presence of bony dysplasia (sphenoid) with a plexiform neurofibroma fulfilled the criteria for NF. At last follow-up of 6 months, there was residual enophthalmos of 2 mm with no prominent bulge in lower eyelid [Fig. 3b].

Discussion

Unilateral isolated enophthalmos in the presence of enlarged IOF without antecedent history of trauma should raise suspicion of NF. Etiologies of enophthalmos include orbital floor fracture, fat atrophy (posttrauma), facial hemiatrophy, silent sinus syndrome, orbital metastasis, sclerosing orbital fibrosis, and microphthalmos. NF rarely presents as enophthalmos. Clinical review of enophthalmos by Cline *et al.* reported only one case attributed to NF out of 26 cases.^[8]

Isolated orbital NF without eyelid/facial involvement is rare. Eyelid involvement with drooping of upper eyelid is the most commonly seen anomaly in orbitotemporal NF. The current case had involvement of inferior orbit presumably along the distribution of neural segment V2. The proposed reason for localized bulge in lower eyelid was anterior orbital neurofibroma, which resolved completely postoperatively. In the past, pulsatile enophthalmos has been reported in association with the enlarged orbital cavity and absent greater wing of sphenoid. Pulsatility depends on the size of the defect and intervening tissue thickness between transmitted brain pulsations and globe. Our case did not have sphenoid wing dysplasia to the extent of allowing intracranial pulsation transmittance to the globe. Here, enlarged orbital cavity with prolapse of contents through IOF lead to enophthalmos. In 1960, Burrows categorized bony changes seen in NF radiologically into enlarged optic canal, absent greater wing of sphenoid, enlarged bony orbit, bulging temporal fossa, and enlarged pituitary fossa in the order of decreasing prevalence.^[7] Hunt and Pugh emphasized that congenital mesodermal dysplasia for lack of ossification of maxilla and sphenoid bones responsible for it rather than erosion induced by NF tissue. In our case, NF lesion was not sizeable enough to produce any bony changes, thus supporting Hunt's hypothesis. Fukuta and

Jackson reported two cases of NF with enophthalmos. Both cases had eyelid and infratemporal fossa involvement with globe dystopia.^[6] Sachdeva *et al.* published a case report

with pulsatile enophthalmos in NF-1 along with herniation of temporal lobe into the orbit, causing esotropia, and visual loss.^[9] Our case had only orbital involvement in the form of an isolated neurofibroma arising in inferior orbit and enlarged IOF. Inferior orbital involvement in NF was also reported by Gurland *et al.* where multiple orbital masses were noted eroding through orbital floor and mistaken for fracture.^[10] Articles published on enophthalmos in orbital NF are summarized in Table 1.^[2-7,10,11]

Conclusion

Our case of NF is rare in its presentation and should be considered in the differential diagnosis of nonpulsatile enophthalmos with an orbital mass in association with bony changes typified by enlarged superior and IOFs.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

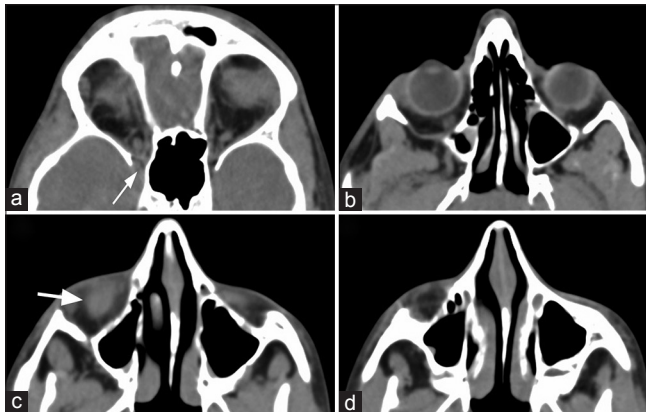


Figure 2: (a) Mid-axial computed tomography orbit showing enlarged superior orbital fissure on the right side compared to left (marked with white arrow); (b) inferior-axial computed tomography orbit showing deficient lateral wall with periosteal bulge; (c and d) axial computed tomography image depicting the location of tumor in mid-anterior orbit (marked with white arrow) with visible fat prolapse in more anterior cuts

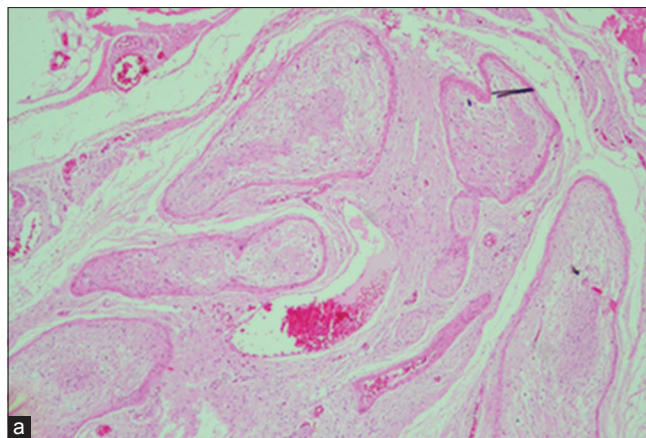


Figure 3: (a) Dilated and tortuous fascicles of proliferating nerve fibers with intervening fibromyxoid stroma (H and E, x200); (b) Residual enophthalmos with no bulge in lower eyelid at 6 weeks

Table 1: Summary of published articles on Enophthalmos in neurofibromatosis

Study (year)	Globe position (number of cases)	Eyelid involvement	Radiological findings
Burrows (1963)	Pulsatile enophthalmos and hypoglobus (1)	Present	Large “bare” orbit with defect of both sphenoidal wings
Lenshoek (1968)	Pulsatile enophthalmos/exophthalmos (1)	NA	Large “bare” orbit with defect of both sphenoidal wings
Savino (1977)	Pulsatile enophthalmos (1)	Present	Sphenoidal dysplasia with enlarged sella turcica
Fukuta (1993)	Enophthalmos with hypoglobus; Pulsatility not mentioned (2)	Present	Enlarged bony orbit with enlarged inferior orbital fissure
Rufa (2006)	Pulsatile enophthalmos (1)	Absent	Large “bare” orbit with defect of both sphenoidal wings
Lehn (2013)	Pulsatile enophthalmos (1)	Present	Absent greater wing of sphenoid
Sachdeva (2015)	Pulsatile enophthalmos (1)	Absent	Absent greater wing of sphenoid
Current case	Nonpulsatile enophthalmos	Absent	Enlarged inferior and superior orbital fissure

NA: Not available

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Conflicts of interest

There are no conflicts of interest.

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