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*Case Report*

# Surgical Management of Oculotemporal Neurofibromatosis - A Case Report

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## ABSTRACT

**Introduction-** Neurofibromatosis is an autosomal dominant, multisystem disorder affecting cell growth of neural tissue, leading to tumour growths that impact skin, nervous system, eye and other organs. It is of two types- neurofibromatosis type 1 (NF1) and neurofibromatosis type 2 (NF 2). Patients with NF1 manifest variety of ophthalmic conditions including optic glioma, glaucoma, choroidal nodules, Lisch nodules and plexiform neurofibromas (causing mechanical ptosis), involving the eyelid, orbit, periorbital and facial structures. Plexiform neurofibromas in these areas are most appropriately labelled as OPPN (orbital periorbital plexiform neurofibroma) to encompass all locations where they occur. A frequent cause of visual loss is deprivational or anisometropic amblyopia. Equally important is the alteration in physical appearance, secondary to ptosis, dystopia and facial disfigurement leading to social embarrassment. A regular and long term monitoring is needed in all these cases Proper and timely management of these cases is a must to prevent complications.

**Case -** An 18 year old male presented with drooping of right upper lid associated with non progressive periorbital swelling and decreased vision since childhood. BCVA was finger count half metre OD and 6/9 OS. On examination right side mild dystopia was seen along with right upper lid severe ptosis which had a characteristic 'S' shaped, with a good LPS action (11mm). This was associated with periorbital swelling in the temporal fossa. The soft multilobulated swelling on the lateral half of the right upper eyelid on palpation felt like a "bag of worms". The patient was considered for removal of neurofibroma and cosmetic correction of right upper lid. The contralateral normal left upper eyelid and palpebral aperture was taken as the desired end result for full cosmetic correction. SURGERY- An en block full thickness resection of the excess of the right upper lid tissue (debulking ) with lid shortening , lateral canthopexy and the required LPS resection was performed.

**Conclusion-** Our patient underwent debulking with shortening of right upper lid along with lateral canthopexy and LPS resection, all done in one surgical procedure. At the end of 2 weeks post op follow up, a good cosmetic outcome with the desired lid height was obtained.

***Key words:*** Neurofibromatosis, oculotemporal, autosomal dominant disorder.

## INTRODUCTION

Neurofibromatosis is an autosomal dominant, multisystem disorder affecting cell growth of neural tissue, leading to tumour growths that impact skin, nervous system, eye and other organs. It is primarily divided into 2 sub-groups: neurofibromatosis type 1 (NF1), also known as von Recklinghausen or peripheral neurofibromatosis; and neurofibromatosis type 2 (NF 2), also known as bilateral acoustic

neurofibromatosis and central neurofibromatosis. [1]

Both NF1 and NF2 are acquired through an inherited autosomal dominant transmission or sporadic mutation, with presentation of NF1 more common than NF2. [2]

Patients with NF1 manifest variety of ophthalmic conditions including optic glioma, glaucoma, choroidal nodules, Lisch nodules and plexiform neurofibromas (causing mechanical ptosis) involving the eyelid, orbit, periorbital and facial structures. [3] All of these manifestations except Lisch nodules and choroidal nodules can result in visual loss, frequently during the age of visual maturation. A frequent cause of visual loss is deprivational or anisometropic amblyopia. Equally important is the alteration in physical appearance secondary to ptosis, dystopia and facial disfigurement leading to social embarrassment. Plexiform neurofibromas involving the eyelid, orbit and periorbital tissue have been described using a variety of names including orbito- temporal plexiform neurofibromatosis (PNs), orbito palpebral neurofibromatosis, oculotemporal neurofibromatosis, orbital neurofibromatosis and orbitofacial neurofibromatosis. Plexiform neurofibromas in these areas are most appropriately labelled as OPPN (orbital periorbital plexiform neurofibroma) to encompass all locations where they occur. [4,5]

Here we present a case of oculotemporal neurofibromatosis involving the upper eyelid and surrounding temporal and frontal regions which was managed satisfactorily with en block resection, debulking, lid shortening , lateral canthopexy and LPS resection , all done together in one surgical setting.

## CASE PRESENTATION

An 18 year old male presented with complaints of drooping of right upper lid associated with periorbital swelling and decreased vision since childhood. The loss of vision was gradual, painless and progressive. Right upper lid drooping was non progressive and present since birth. No history of trauma was present. Best corrected visual acuity was finger count half metre OD and 6/9 OS. Intraocular pressure was recorded to be 18 mmHg OD and 16 mmHg OS. On examination right side mild dystopia was seen along with right upper lid severe ptosis which had a characteristic 'S' shape. The levator palpebrae superioris action was good (11mm). This was associated with periorbital swelling in the temporal fossa and the soft multilobulated swelling on the lateral half of the right upper eyelid on palpation felt like a "bag of worms" Extra ocular movements were full and normal in all directions. Anterior segment examination revealed clear cornea, deep anterior chamber, normal papillary reaction and clear lens. Multiple Lisch nodules (>2) were found on iris on slit lamp examination in both eyes. Dilated fundus evaluation was within normal limits. General physical examination revealed multiple café au lait spots on skin of thoracic and lumbar region (>20 in number). The greatest measured spot was about 6x3 cm in size. Rest physical examination was within normal limits. The patient's history and clinical findings correlated with clinical diagnostic criteria of neurofibromatosis type 1. So a provisional diagnosis of NF1 was made.

Routine blood investigation, X ray of chest and long bones was conducted which was found to be normal. CT head and orbit was done and the findings suggested an ill defined soft tissue attenuating lesion in the scalp, right frontal region and subcutaneous tissue of lateral wall of right orbit and in pre septal region of right eyelid with mild fat stranding in the extra conal compartment of right orbit. No bony dysplasia or bony abnormality was seen in the orbit. Features were suggestive of plexiform neurofibroma.

**SURGERY-** The patient was considered for removal of neurofibroma and cosmetic correction of right upper lid. The measurement of the involved lateral aspect

of right upperlid was done for en block removal of full thickness involved right upper lid tissue. The measurement of the involved eyelid was done for both horizontal and vertical extent. The contralateral normal left upper eyelid and palpebral aperture was taken as the desired end result for full cosmetic correction. An en block full thickness resection of the excess of the right upperlid tissue (debulking) with lid shortening, lateral canthopexy (reconstruction of lateral canthus) and the required LPS resection was performed.

The patient was followed up post operatively and at the end of 2 weeks, a satisfactory cosmesis with the desired lid height was obtained.

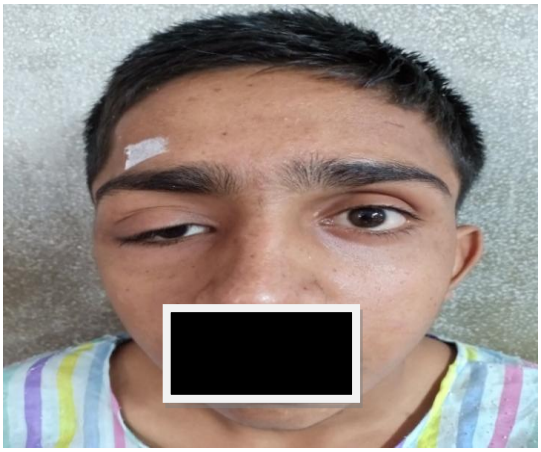

**Fig.1: Right upper eyelid 'S shaped' plexiform neurofibroma**

**Fig. 2: Day 1 –post operative**

**Fig.3: Day 7-post operative**

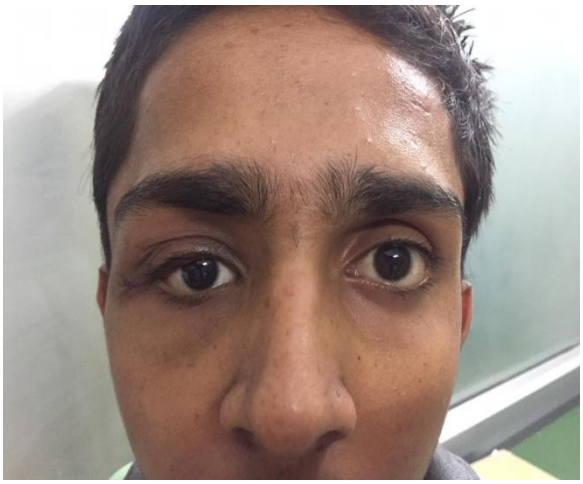

**Fig.4: 2 weeks post operative**

## DISCUSSION

Neurofibromatosis is a disease with an autosomal dominant trait with variable penetrance. Its frequency is 1 in 3000 live births. Skull and facial deformities occur in 22% of cases. [6] It is primarily divided into 2 sub-groups: neurofibromatosis type 1 (NF1); and neurofibromatosis type 2 (NF 2). NF-1 is caused by mutation in gene located on chromosome 17. In 50% cases, the individual inherits the mutated gene from a parent in an autosomal dominant pattern. In the other 50%, the chromosome 17 mutation is a new one. The NF 1 gene is large, making possibility of mutation relatively high. [7]

The severity and manifestations of NF1 vary widely. Most have distinctive café au lait spots which increase with age. The number and location of neurofibromas is variable implicating modifier genes.

Presence of >/= 2 of the following criteria are needed for diagnosis of NF1 developed by National Institute of Health:

- ✓ Six or more Café Au Lait Macules (CALM) ; >5 mm in greatest diameter at prepubertal age and >15 mm in greatest diameter in adults
- ✓ Axillary or inguinal freckling
- ✓ Two or more neurofibroma of any type or one plexiform neurofibroma.
- ✓ Optic glioma
- ✓ Two or more Lisch nodules
- ✓ A distinctive osseous lesion e.g. sphenoid dysplasia / thinning of cortex of long bones with or without pseudoarthrosis.
- ✓ First degree relative (parent, sibling, offspring) with NF1 by the above criteria. [8]

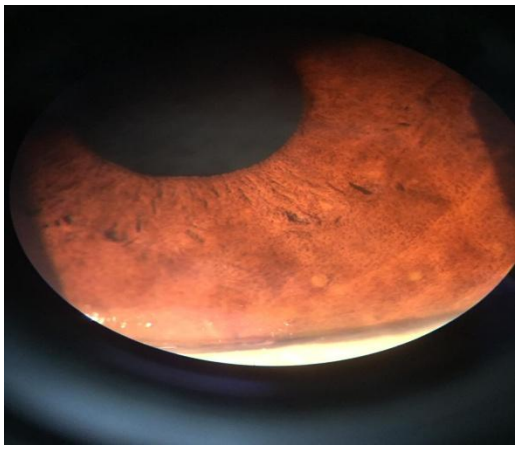

**Fig.5: Lisch nodules**

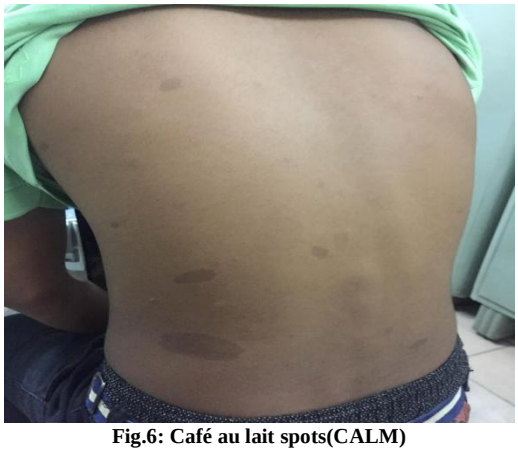

**Fig.6: Café au lait spots(CALM)**

Ophthalmic manifestations of NF 1 include Lisch nodules, plexiform neurofibromas, choroid hamartomas, retinal tumours, optic nerve gliomas, prominent corneal nerves etc. While assessing a patient with suspected ophthalmologic manifestation of Neurofibromatosis (NF) type 1, one should also consider iris nevi, glaucoma, juvenile glaucoma, tuberous sclerosis and NF2. [9]

Plexiform neurofibromas that infiltrate the orbit, temporal region or eyelid are potentially vision threatening. In young children, infiltration and odema of the orbit and eyelids may result into ptosis, proptosis, change in globe length, anisometropia and obscuration of visual axis causing amblyopia. Amblyopia secondary to the orbitotemporal plexiform neurofibroma is present in 62%, primarily from ptosis and anisometropia. [10]

The present case had multiple café au lait spots involving thoracic and lumbar region, plexiform neurofibroma involving the right upper eyelid and multiple lisch nodules in both the eyes; hence fulfilling the clinical diagnosis of NF1. Our case had the typical thickening of right upper lid with an S- shaped deformity and a 'bag of worms' sensation, characteristic of plexiform neurofibromatosis. No family history was present, hence, it is a case of sporadic mutation.

The decreased vision in present case was due to amblyopia which was caused by mechanical ptosis as the visual axis was obstructed by plexiform neurofibroma in right upper eyelid.

Our patient underwent debulking with shortening of right upperlid along with lateral canthopexy and LPS resection, all done in one surgical procedure. At the end of 2 weeks post op follow up, a satisfactory cosmetic outcome with the desired lid height was obtained.

No lab tests are pertinent in evaluating ophthalmic manifestation of NF1. However tissue biopsy of skin lesion is occasionally done to confirm diagnosis. The excised eyelid tissue in our patient was sent for histopathological examination and typical findings of neurofibroma, i.e., markedly enlarged nerves surrounded by thickened perineural structures, were found .

The treatment of oculotemporal neurofibromatosis is challenging. The surgical planning must include the evaluation of the exact amount of resection of the soft tissue and maintaining visual acuity as well as achieving a better aesthetic appearance. If the affected eye has limited eyesight, it is possible to pay more attention to cosmesis. Blepharoptosis surgery in neurofibromatosis is indicated if the visual axis is compromised or if there is a chance of amblyopia. [11]

The timing of surgery in neurofibromatosis including eyelid and orbit is still controversial. As a rapid growth phase of the neurofibromatosis is known to occur during childhood and puberty, most surgeons recommend undergoing surgery for oculotemporal neurofibromatosis during early childhood if it is associated with bony dysplasias /orbital neurofibroma, thus may prevent orbital deformity and preserve eye function. Early surgery should also be performed in cases of ptosis with pulsating exophthalmos. In contrast, correction of ptosis caused by an oculotemporal mass for cosmetic purposes is delayed until the age of 18, when the disease progression has stabilised. The recurrence rate of neurofibromatosis is relatively high. Ptosis of the upper eyelids may recur due to recurrence of the benign tumour. Recurrence may cause recurrence of exophthalmos as well. In addition, the possibility of malignant change should not be overlooked. Therefore, long term follow up is needed to observe the consequences of the surgical treatment. [12]

## CONCLUSION

Individual approach and planning is important in cases of neurofibromatosis. Surgical treatment of oculotemporal neurofibromatosis should aim at a good cosmetic and functional outcome. Regular follow up and monitoring is must in such cases, to look out for recurrence/ malignant transformation.

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