Case Report

Rare Case of Ocular Manifestation of Neurofibromatosis

Parul Singh*, Shefali Pandey, Shaffi Chopra

Department of Ophthalmology, GSVM Medical College, Kanpur, Uttar Pradesh, India.

Abstract

Neurofibromatosis type 1 (NF-1) is a common autosomal dominant disorder affecting multiple systems. It presents with neurofibromas, skeletal deformities, and ophthalmic manifestations. This case highlights a rare occurrence of sphenoid wing agenesis, leading to orbital meningo-encephalocele and severe ocular displacement.

A 39-year-old male presented with a history of mass overhanging his right eyelid involving the right periorbital area since his childhood, which was gradually increasing in size and causing disfigurement of his face.

Keywords: Neurofibromatosis type 1, Sphenoid wing agenesis, Orbital meningo-encephalocele, Pulsatile proptosis.

Introduction

Neurofibromas benign tumor arising from peripheral nervous tissue. In this neoplasm, Schwann cells are admixed with perineural-like cells, fibroblasts, mast cells and CD 34+ spindle cells.[1]

In neurofibromas, Schwann cells show complete loss of the NF-1 gene. NF-1 gene is a tumor suppressor gene producing neurofibromin, which inhibits RAS activity.[1,2]

Neurofibromatosis is a hereditary autosomal dominant condition mainly of two categories: Type 1 and type 2.[1]

Type 1 is a systematic disease associated with non-neoplastic manifestations such as mental retardation, seizures, lisch nodules, café aulait spots and neurofibromas of all types, gliomas of the optic nerve, pheochromocytoma, malignant peripheral nerve sheath tumor and other hamartomas.[1]

Type 2 is most commonly associated with bilateral schwannomas and multiple meningiomas.[1]

Neurofibromatosis type 1 (NF-1) is a relatively common autosomal dominant disorder that occurs in one in 3,000 live births. Skeletal signs include scoliosis, sphenoid wing dysplasia, bony distortion and local cystic and erosive change. Although sphenoid wing dysplasia has been observed in between seven to 12.8% of patients with neurofibromatosis, the absence of a sphenoid wing is very rare.[1-5] We report here an unusual case of NF-1 with an associated absence of a sphenoid wing by using a non-contrast CT Head and orbit.

Case Report

A 39-year-old male presented with a history of mass overhanging his right eyelid involving the right periorbital area since his childhood, which was gradually increasing in size and causing disfigurement of his face. The version of the right eyelid revealed herniating Right orbital content with the right globe deviated down and having restriction of ocular movement in all gazes. On ophthalmological examination his Visual acuity is R/E-no perception of light; L/E - 4/60. Further examination revealed that his right eye was normotensive.

The mass is 7*8 cm in diameter. The mass had a bag of worm feel to it with a constant pulsation. The transillumination test is negative (Figures 1 and 2).

There was no pain and discharge associated with mass. The eyelid margin and adnexal structures are maintained. (Figures 3 and 4)

On physical examination, the patient's body weight is 67 kg, height 157 cm, Body temperature 36 degrees, heart rate 72 bpm. The patient gives a history of a benign mass in the center

Address for correspondence: Parul Singh
Department of Ophthalmology, GSVM Medical College, Kanpur, Uttar Pradesh, India.
E-mail: parulsingh1406@gmail.com

How to cite this article: Singh P, Pandey S, Chopra S. Rare Case of Ocular Manifestation of Neurofibromatosis. UP Journal of Ophthalmology. 2025;13(1): 38-41.
Received: 12-02-25, **Accepted:** 24-03-25, **Published:** 30-04-25

UP JOURNAL OF OPHTHALMOLOGY
An Official Journal of Uttar Pradesh State Ophthalmological Society, UPSOS (Northern Ophthalmological Society, NOS)
p-ISSN: 2319-2062 | DOI: 10.56692/upjo.2025130110

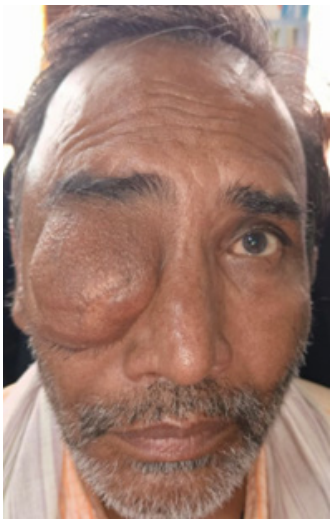

Figure 1: Mass overhanging Right eyelid

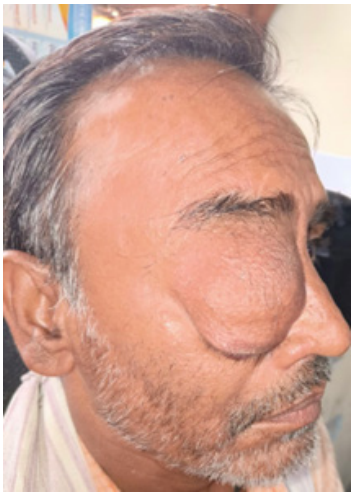

Figure 2: Lateral view of the patient representing overhanging mass

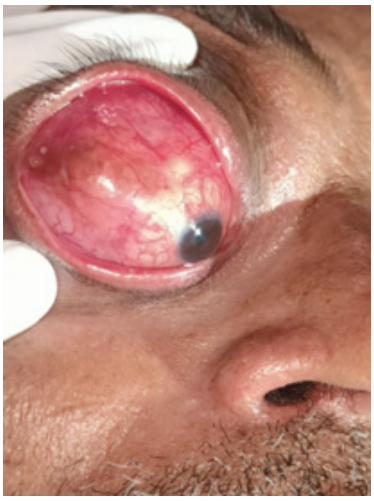

Figure 3: Herniating Right orbital content with the right globe deviated down

Figure 4: Protrusion of Right eyeball with dilated conjunctival vessels

of his chest, which was operated 20 years back. There were no other scar marks, nodules, or freckles anywhere in the body.

The patient gave a history of some surgery in his right temporal region when he was 7 years old but couldn't provide any documented verification for the same except for some scars present in the same region.

Laboratory investigations such as blood parameters, fasting blood glucose, coagulation profile, USG whole abdomen, chest X-ray, Mantoux test, rheumatoid factor and anti-nuclear antibody within normal limits.

Anterior Segment Examination

The lacrimal system, conjunctiva, cornea, anterior chamber, iris were within normal limits (Figure 5).

Fundus Examination

The patient's right eye fundus could not be assessed due to eyelid overgrowth and the eyeball deviated inward and downward. The left eye fundus examination was within normal limits with slightly hazy media due to cataracts and mild chorioretinal degeneration.

To determine the nature of the mass and cause of pulsation, CT head and orbit were advised to the patient.

Figure 5: Slit lamp examination of anterior segment of left eye

The CT scan revealed post-operative surgical clips are seen in situ in right temporal region. The large bone defect is seen in the right anterior and middle cranial fossa with herniation of CSF pockets into orbit. A small component of brain tissue is also seen in the right orbit. The meningo-encephalocele has displaced the right orbit inferiorly and laterally. Gliotic lesions are seen in the right frontal and temporal regions with the ipsilateral pulling of lateral ventricles. The right globe of orbit is normal in architecture. The lens is normal. No mass lesion is seen in the anterior or posterior compartment. Contents are homogeneous. The right optic nerve is stretched. Right orbital muscles are stretched. Right-sided proptosis is present. Third, fourth and lateral ventricles are normal in size and position. No midline shift. Basal Cisterns and cortical sulci are normal. The attenuation value of brain parenchyma is normal (Figures 6 and 7)

Discussion

Neurofibromatosis type 1 may affect all systems of the human body. It is a hamartomatous disorder that originates in the neural crest and secondarily affects the supporting mesenchyme. The disorder, which is characterized by pigmented cutaneous lesions and generalized tumors that originate in the neural crest (taking the form of a discrete fibroma nodule or a plexiform neurofibroma), manifests itself in various ways. Central nervous system indicators consist of an increased incidence of astrocytoma, meningiomas, schwannomas, and ependymomas. Other features of the disease include bony dysplasia, kyphoscoliosis, sphenoid wing hypoplasia and vascular dysplasia. Sphenoid dysplasia is one of the characteristics of NF-1, occurring in 5 to 10% of cases. Furthermore, abnormalities of the sphenoid wings are often considered pathognomonic. However, complete dysgenesis of the sphenoid wing is very rare.[6-9] Its radiological characteristics describe the area of defect in the greater sphenoid wing and enlargement of the middle cranial fossa. Many ophthalmic manifestations may occur in NF-1 patients. Lisch nodules are virtually all pathognomonic, and Huson *et al*.[10] found these iris hamartomas to be present in all of his NF-1 patients over 16 years of age. Other ophthalmic findings include choroidal hamartomas, plexiform neurofibromas, retinal phakomas and optic nerve gliomas. Exophthalmos and enophthalmos, potential sequelae, are associated with orbital tumors and/or sphenoid wing dysplasia.[6,11]

Unilateral enophthalmos, primarily viewed as a complication of an orbital fracture, may result from other causes such as microphthalmos, orbital fibrosis, and sclerosing orbital tumors.[6,8] Congenital microphthalmia occasionally accompanies sphenoid dysplasia or similar sphenoidal abnormalities. The neuro-ophthalmological findings of NF-1 are variable. Pulsating exophthalmos is a common one. However, in a few cases in which NF-1 patients have sphenoid dysplasia, enophthalmos was also seen as a result of microphthalmia or because the lateral wall of the orbit was deficient and allowed decompression of the neurofibromatosis

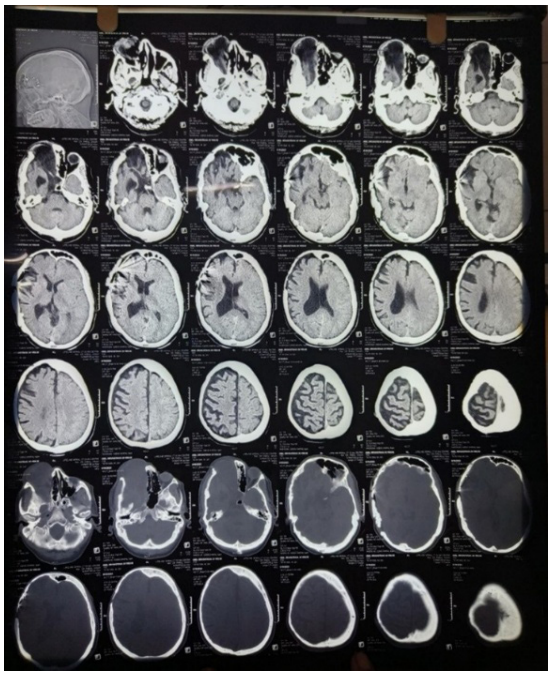

Figure 6: Coronal section CT Head and orbit showing remodelling of right orbit and sphenoid wing dysplasia

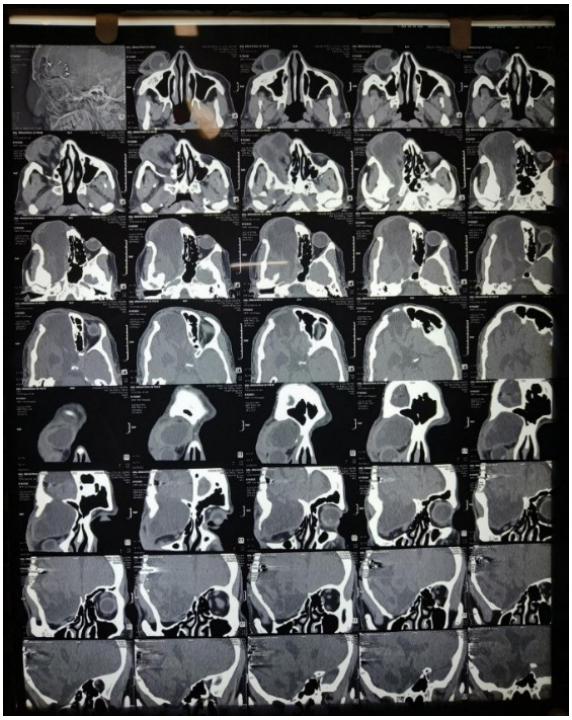

Figure 7: Coronal section CT Head and orbit at the level of paranasal sinuses representing significant proptosis R eye

tissue or retrobulbar fat into the temporal fossa.[12] The present case had NF-1 with sphenoid wing dysgenesis, resulting in meningo-encephalocele and herniation of CSF pockets into the orbit leading to proptosis.

In conclusion, sphenoid dysplasia is one of the features of NF-1, but the absence of a sphenoid wing is very rare.

References

1. Kumar V, Abbas AK, Aster JC. *Robbins and Cotran Pathologic Basis of Disease*. 9th ed. Philadelphia: Elsevier; p.1247-9, 1317.
2. Salmon JF. *Kanski's Clinical Ophthalmology: A Systematic Approach*. 9th ed. London: Elsevier; p.48-50.
3. Lee LR, Gigantelli JW, Kincaid MC. Localized neurofibroma of the orbit: a radiographic and histopathologic study. *Ophthal Plast Reconstr Surg*. 2000 May;16(3):241-6.
4. Santaolalla F, *et al*. Severe exophthalmos in trigeminal plexiform neurofibroma involving the orbit and the infratemporal fossa. *J Clin Neurosci*. 2009 Jul;16(7):970-2.
5. Kottler UB, *et al*. Isolated neurofibroma of the orbit with extensive myxoid changes: a clinicopathologic study including MRI and electron microscopic findings. *Orbit*. 2004 Mar;23.
6. Asil K, Gunduz Y, Yaldiz C, Aksoy YE. Intraorbital encephalocele presenting with exophthalmos and orbital dystopia: CT and MRI findings. *J Korean Neurosurg Soc*. 2015 Jan;57(1):58-60. PMID: 25674346 [PubMed] PMCID: PMC4323507.
7. Davidson RI, Kleinman PK. Anterior transorbital meningoencephaloceles: a defect in the pars orbitalis of the frontal bone. *AJNR Am J Neuroradiol*. 1980;1:579-82.
8. Khan N, Werke IV, Ismail F. Neurofibromatosis revisited: a pictorial review. *SA J Radiol*. 2010;16-18.
9. Erb MH, Uzcategui N, See RF, Burnstine MA. Orbitotemporal neurofibromatosis: classification and treatment. *Orbit*. 2007;26:223-8.
10. Jacquemin C, Bosley TM, Svedberg H. Deformities in craniofacial orbit deformities neurofibromatosis type 1. *AJNR Am J Neuroradiol*. 2003;24:1678-82
11. Dağıstan E, Canan A, Barut AY, Karagöz Y. Unilateral absence of sphenoid wing in a Neurofibromatosis type I patient: imaging findings. *J Clin Exp Investig*. 2013;4(3):364-6.
12. Onbas O, Aliagaoglu C, Calikoglu C, Kantarci M, Atasoy M, Alper F. Absence of a sphenoid wing in neurofibromatosis type 1 disease: imaging with multidetector computed tomography. *Korean J Radiol*. 2006 Jan;7(1):70-2. PMID: 16549958 PMCID: PMC2667581.