Cyst Turned out to be A Neurofibroma without any Associated Features of Neurofibromatosis

Dinesh G Patil*, Hemkala L Trivedi**, Anil D Dudhabhate*

Abstract
53 year old hindu male, farmer by occupation presented with painful swelling in inner aspect of left eye since 8 years

**Introduction**

Two histologically as well as biologically distinct lesions have been termed as neurofibroma. The most common form occur in skin (cutaneous neurofibroma or in peripheral nerves (solitary neurofibroma). They arise sporadically or in association with neurofibromatosis I. The skin lesions evident as nodules which may grow to be large sometimes with overlying hyperpigmentation.

The second type is plexiform neurofibroma which is considered to occur only in patients with neurofibromatosis I. The major concern in these patients is difficult in surgical removal of these plexiform tumours when they involve major trunk since they have significant potential for malignant transformation.

**Case Report**

53 year old hindu male farmer by occupation complaining of painful swelling in inner aspect of left eye since 8 years. The swelling was not increasing in size. No h/o any trauma, redness or diminution of vision. No h/o brown macules over the body or any swelling over the body. There is no similiar family history. Patient has no systemic illness.

O/E:- Visual acuity in RE was 6/6 and LE 6/12.

RE examination was normal externally.
LE examination showed a 3 x 3 x 3 mm firm to hard mobile tender mass in the area of left lacrimal sac, not adherent to skin or underlying structures. Conjunctiva, Cornea AC, Iris appeared normal. Pupil was circular reacting to light. Lens was clear.

EOM was full range. Sac syringing showed patency. Systemic examination doesnot reveal any abnormality.

Neurosurgery opinion taken to r/o neurofibromatosis was normal.

Orthopaedic opinion taken to r/o any bony abnormality (to r/o Neurofibromatosis) was normal.

Skin reference was also made to r/o any cutaneous lesions which was also normal.

**Investigations**

Hb- 15.1 gm% CBC-8300/cumm
P 72/L 27/E 1
FBS- 80 mg% PLBS- 100 mg%
BUN- 15 mg% Serum Creatinine- 1.2 mg%
Total Proteins-7.1 mg% Albumin- 4.1 mg%
SGOT- 34 IU/L SGPT- 35 IU/L
Serum Na- 138 meq/l Serum Cl- 107 meq/l
Bleeding time- 3 min Clotting time- 5 min 25 sec 20 sec
ECG- Within normal limit X-ray orbit- Within normal limit
CT Scan orbit- Within normal limit

MRI Orbit- showed ill defined nodular soft tissue mass in medial canthus of left eye. In view of long duration of symptoms, possibility of neoplastic aetiology is less likely.
Excision biopsy of the lesion confirmed the diagnosis of neurofibroma.

**Pathophysiology**

Eye involvement in neurofibromatosis is more common than the clinical examination indicates. Every part of the eye including lid and adnexae may be involved in case of generalised neurofibromatosis.

Neurofibroma in the eye can have various presentations. There may be asymmetry of face, cornea may be larger, lids may show ptosis and café au lait spots. Orbit may show bony destruction or hypertrophy, intracranial pulsations can be transmitted to the eye. The pulsation are synchronous with pulse, unaccompanied by bruit but there is no discomfort. Exophthalmos of the eye may occur as a result of:

1. Neurofibromatous involvement of the orbit.
2. Herniation of brain due to orbital bone erosion.
3. Gliomas or meningiomas of the optic nerve which may be associated.

Conjunctiva usually not involved. Uvea may be involved in diffuse manner involving choroid, ciliary body and iris. Buphthalmos might occur when glaucoma occurs.

Generalised neurofibromatosis comprises two autosomal dominant disorders. Neurofibromatosis I (von Recklinghausen disease) and Neurofibromatosis II (Acoustic Schwannoma).

In neurofibromatosis I 50% have definite family history. The remainder appears to represent a new mutation.

Neurofibromatosis I has 3 major features:

1. Multiple neural tumours (Neurofibroma) disperse anywhere on or in the body.
2. Second feature of neurofibromatosis is Café Au Lait spots present in 90% patients located over nerve trunks 6 or more spots > 1.5 cm in diameter if present in adult are likely to be neurofibromatosis I.
3. Pigmented hamartoma of the iris (Leisch nodules) are present in 94% of patients of age 6 years or older. They are helpful in establishing the diagnosis.

Wide range of associated abnormalities have been reported in these patients.

1. Erosive defects owing to contiguity of neurofibromatosis of bone
2. Scoliosis.
3. Intraosseous cystic lesions.
4. Subperiosteal bone cyst.
5. Pseudoarthrosis.

Patients with neurofibromatosis I have 2-4 fold increased risk of developing Wilm's tumour, rhabdomyosarcoma, meningioma, optic glioma and phaeochromocytoma.¹

**Histology**

Neurofibroma present in dermis and subcutaneous fat is well delineated but unencapsulated, composing of spindle cells. Adnexal structures sometimes get entrapped by the edge of the lesion. The stroma of this tumour is highly collagenised and contain little myxoid material. Lesions within peripheral nerve are of identical histologic appearance.²

**Treatment**

Surgical excision/biopsy of the tumour is done because of cosmetic concern. Risk of malignant transformation from solitary neurofibroma is extremely small.³

**Discussion**

The patient had solitary neurofibroma without any features of neurofibromatosis I which on clinical examination resembled cyst. Patient did not have neurofibromas on or in the body anywhere else. Patient did not have Café au lait spots or Leisch nodules on iris. Patient did not have any hypertrophy or destruction of the orbital bone or exophthalmos.

Since the patient had an atypical presentation without any associated features of neurofibromatosis I we thought the case to be interesting and reporting.

**References**


---

**CERVICAL CANCER IN INDIA**

Screening for cervical cancer is not a component of health care in rural India. This article reports on a trial health care in rural India. This article reports on a trial of cervical-cancer screening by human papillomavirus (HPV) testing, cytologic analysis, or visual inspection of the cervix with acetic acid in rural villages. The results, as compared with those in a group that received no screening, showed that a single round of HPV testing significantly reduced the incidence of invasive cervical cancer and mortality in rural Indian villages.