OPHTHALMOLOGICAL MANIFESTATIONS OF NEUROFIBROMATOSIS - A CLINICAL EVALUATION OF FIVE CASES

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Introduction

The neurofibromatosis consists of genetically distinct hereditary disorders that primarily affect cell growth of neural tissues. Inheritance is autosomal dominant with irregular penetrance and variable expressivity. The mutation rate is high. The 2 main types are: type 1(NF1) and type2 (NF2)

The other variants include segmental (NF3) and familial café-au-lait spots (NF4)

Neurofibromatosis type 1

Affects 1 in 4,000 individuals, present in childhood. The cardinal features are more than 6 café-au-lait spots and multiple cutaneous neurofibromas. There neural tumors in brain, spinal cord and nerves(cranial, peripheral and sympathetic). The skin lesions are café-au-lait spots, axillary freckles, fibroma molluscum, plexiform neuro fibromas.

The ocular features are optic nerve glioma, other neural tumors like neurilemmoma, plexiform neurofibroma, or meningioma.there may be spino-orbital encephalocele.

Eye lid plexiform neurofibroma when involves the upper lid may give rise to mechanical ptosis

Anterior segment lesions include lisch nodules prominent corneal nerves, congenital ectropion uveae. There may be glaucoma, choroidal hamartomas

Neurofibromatosis type 2

Affects 1 in 50,000 indivisuals. Systemic features include acoustic neuromas. Ocular features are juvenile posterior sub capsular cataract, and retinal hamartoma

Aim

To study the clinical presentation and management of neurofibromatosis.

Period of study-may 2003 to may 2005

Observation -case-1-

2yrs old child presented with hyphaema.there was large cornea,digitaly raised iop,loss of vision,and proptosis. Following trivial injury hyphaema developed.medical treatment with anti glaucoma drugs (timolole/d,glucomole/d), local steroid application caused reduction of hyphaema.There was axillary freckles, café -au -lait spots over the trunk. Under GA trabeculectomy was done. Hyphaema and raised iop was controlled.there was improvement in VA.

Case -2-

3yrs old child presented with loss of vision. There was digitally raised iop, large cornea. Axillary freckle and café- au- lait spots were present. Under GA trabeculectomy was done.iop was controlled and vision improved.

Case-3

29 yrs old female presented with red eye, and loss of vision.There was raised iop and mild uveitis.there was fibroma mollusum all over body including face. Local medical treatment with glucomol e/d and timolol e/d with local and systemic steroids caused reduction of iop from 30 mm of hg to 15 mm of hg.uveitis was controlled and vision improved.

Case-4

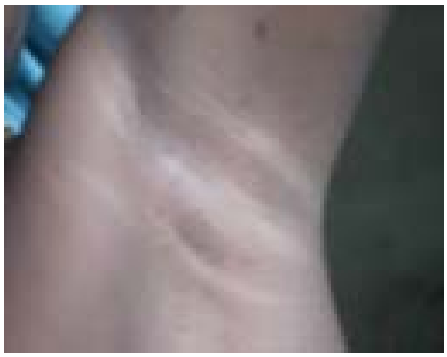
55Yrs old male presented with eyelid plexiform neurofibromatosis and secondary glaucoma.Following trabeculectomy iop reduced from 29mm of hg to17 mmof hg.Medical treatment continued with steroid e/d and antiglaucoma medication.

Case-5

40yrs old male presented with fibroma molluscum and eyelid plexiform neurofibroma.

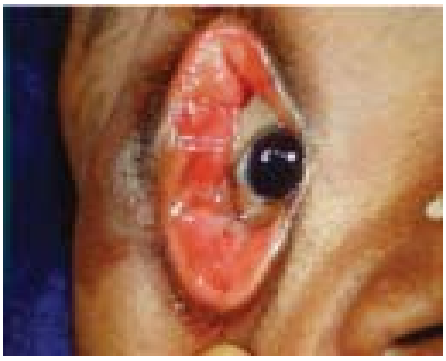
Sex incidence

Out of the 5 cases, 3 cases were male and 2 were female.

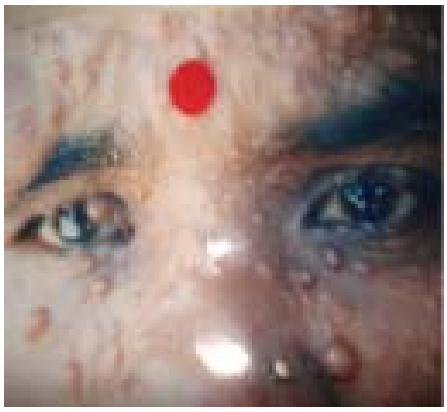


Axillary freckles

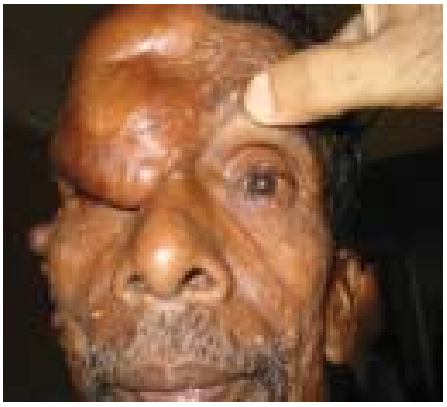
Café-au-lait spots



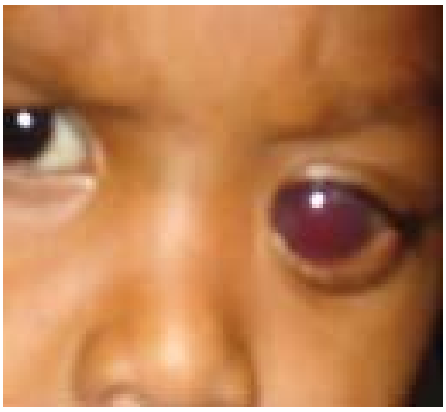
inside of plexiform neurofibromatosis



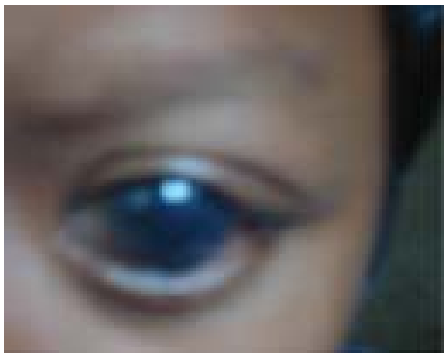
Presented with secondary glaucoma



Presented with secondary glaucoma



A two year old patient of neurofibromatosis presented with hyphaema



After absorption of hyphaema

Age incidence-

Upto 5 yrs-2
6 to30yrs-1
35 to 50yrs-1
51to65yrs-1

Mode of presentation-

2 patients presented with congenital glaucoma out of which 1 case had hyphaema and proptosis. 2 cases had eye lid plexiform, neurofibroma. And 1 case had chronic, simple glaucoma with fibroma molluscum.

Management

Difficulty was observed in treating these patients. Trabeculectomy was done in 3 cases.Good iop control was obtained following trabeculectomy. Medical treatment with steroid e/d and systemic steroid could control uveitis.

Discussion and conclusion

Neurofibromatosis is due to genetic mutation of chromosome 17 and 22 in type 1 and type 2 respectively.It occurs in connection with branches of fifth cranial nerve. It may occur in extremities.the affected nerves become enormously thickened as a result of mixofibromatous degeneration of endoneurium.If it occurs in scalp the underlying skull may be eroded. The involved skin sometimes hangs down in pendulous foldswith grotesque effects.

Fibroma molluscum consist of pedunculated, flabby, pigmented nodules which are frequently widely distributed

all over the body. They begin to appear at about puberty and increase in number throughout life.The number of lesions vary from few to several hundred. Histologically they consist of neurofibromasor schwannomas of cutaneous nerves.

Plexiform neurofibromas are larger and less well defined and tends to merge with the surrounding tissues.It may be present at birth or appear during childhood. Sometimes it involves face and cause disfigurement (The elephant man)

In this study there is slight male preponderance. Glaucoma was controlled by trabeculectomy. No surgical treatment required in 2 cases. Ophthalmological manifestations are common in neurofibromatosis and the treatment is challenging. Each case needs independent evaluation and the treatment is tailormade .

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