OCULAR MANIFESTATIONS IN NEUROFIBROMATOSIS-1

Presenter
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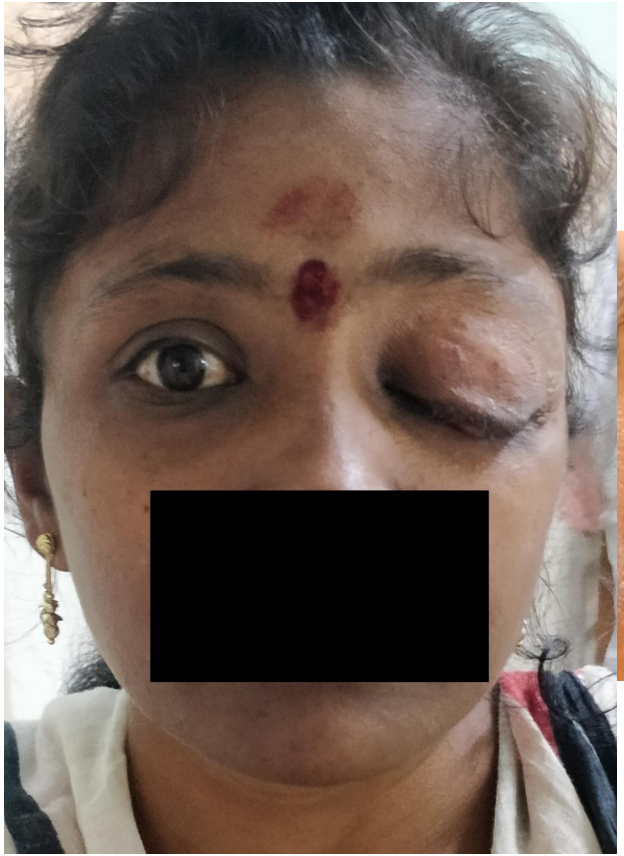
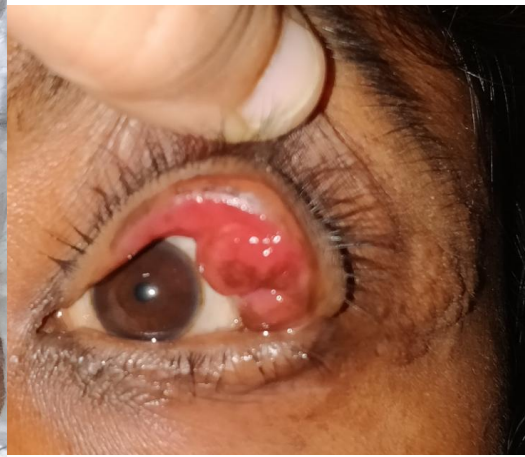
AIM&OBJECTIVES

To report a case of Neurofibromatosis 1 with eyelid neurofibroma and lisch nodules on the iris.

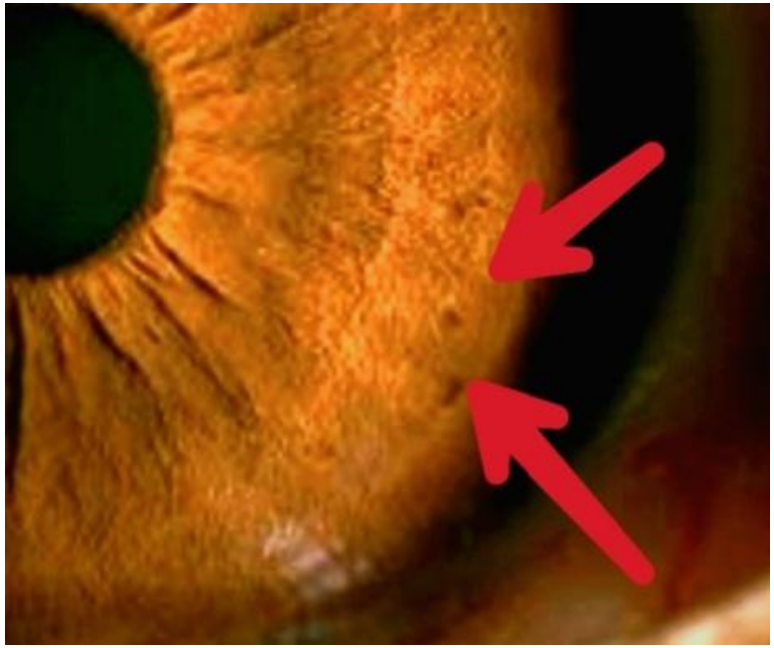
MATERIAL & METHODS:A 35 yrs old female presented to OPD with mechanical ptosis of left lid which was painless for the past 1 year.On evertion of the eyelid, there is a nodular mass temporally measuring 2*1 cms.





Anterior chamber assessment revealed lisch nodules on the iris which confirmed the diagnosis of neurofibromatosis-1.

Posterior chamber was normal.Optic disc and macula were normal.MRI showed no optic nerve glioma.She denied any other lesions on the body.The patient was referred to higher centre for blepharoplasty.



DISCUSSION

Neurofibromatosis is a genetic disorder observed with relative prevalence among neurocutaneous syndromes. Two forms of this genetic disorder may be delineated: neurofibromatosis type1, neurofibromatosis type2.

Neurofibromatosis type1 is also known as von Recklinghausen's disease, representing the overwhelming majority of NF cases and the focus of this report.NF1 carries with it a vast spectrum of clinical presentations, which mainly consist of neurofibromas, macules, cafe-au-lait spots, and optic gliomas.

Due to the incurable nature of neurofibromatosis type1 and 2, symptomatic treatment is currently the only effective management of this disease.Hence,ophthalmic intervention is much desired therapy to prevent facial disfigurement.Before procedures could be undertaken,diagnosis should be confirmed through appropriate evaluation