Bilateral Optic Nerve Glioma in a Case of Neurofibromatosis

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Introduction

Neurofibromatosis Type 1 (NF-1) is a relatively common autosomal dominant disorder. In addition to multiple peripheral neurofibromas, NF-1 predisposes to central nervous system (CNS) tumors like optic nerve glioma, neurofibroma, ependymoma and meningioma. Bilateral optic nerve gliomas is relatively uncommon in NF-1 and optic nerve glioma usually presents as slowly progressive visual loss. Here we report a case of NF-1 with bilateral optic nerve glioma, presenting as sudden onset of defective vision in one eye.

Case report

22 year old female presented to our department with history of sudden onset of defective vision in right eye of 3 days duration, not associated with redness, pain, or photophobia.

There was no history of trauma to the eye. There is history of multiple swellings and hyperpigmented lesions all over the body since 10 years of age, progressively increasing in numbers. There is no history of similar illness in the family.

General examination showed multiple café au lait spots, cutaneous neurofibromas [fig. 1], molluscum fibrosum and axillary freckles. Neurological examination of motor, sensory, and cerebellar systems were normal.

Ocular examination showed 0.5 x 0.5mm sized firm swelling on right upper lid, decreased corneal sensation and grade 2 relative afferent pupillary defect in right eye. Vision in right eye was 6/60, with defective colour vision. Left eye vision was 6/6. Slit lamp examination showed prominent corneal nerves in both eyes and Lisch nodules at 5 O'clock in right eye, 2 and 4 O'clock in left eye. Fundus examination of right eye showed pale optic disc with well defined margin and normal vessels. Indirect ophthalmoscopy showed normal retinal periphery. Ophthalmoscopic examination of left eye was normal.

Investigation

MRI brain and orbit showed diffusely thickened non enhancing right optic nerve suggestive of optic nerve glioma [fig. 2]. Thickening was noted in the intra orbital portion of left optic nerve also. Non-enhancing patchy hyperintensities on T2 weighted images noted in globus pallidus, thalamus, brainstem, middle cerebellar peduncle [fig. 3] and supra tentorial white matter – likely to represent hamartomas in white matter and deep gray matter.
Patient was diagnosed as a case of Neurofibromatosis type 1 from the clinical picture.

As the patient presented with sudden onset of significant defective vision on right eye with RAPD and there was a delay in getting MRI, we started on systemic steroids in view of possible retrobulbar optic neuritis. Even though MRI showed optic nerve glioma, we continued on steroid as the patient’s vision was improving. As the patient was not willing for any intervention for glioma, she was discharged and advised follow up with tapering dose of steroids. On follow up at 2 weeks, patient’s vision was 6/9 which improved to 6/6 with -0.25 Dsph.

Discussion

Neurofibromatosis type 1 is a neurocutaneous syndrome characteristically associated with freckles in non-exposed areas, café au lait spots, Lisch nodules and cutaneous neurofibromas. Patients are at an increased risk of developing neoplasms of nervous system like optic nerve glioma, neurofibroma, ependymoma and meningioma. Optic nerve glioma develops in 15% of patients with NF-1, may be unilateral or bilateral and usually presents as slowly progressive painless loss of vision. Rarely it can present as sudden loss of vision due to hemorrhage into the glioma.

Our patient has got axillary freckles, café au lait spots, lisch nodules and cutaneous neurofibromas – fulfilling the criteria for diagnosing neurofibromatosis type 1. MRI showed bilateral optic nerve glioma. The patient presented with sudden loss of vision in right eye, which is unusual in optic nerve glioma - where the present action is as slowly progressive loss of vision. MRI did not show any evidence of haemorrhage into glioma to produce a sudden loss of vision. As the patient improved to normal vision, possible etiologies for loss of vision in our patient are unrelated optic neuritis or nonspecific inflammatory changes in the glioma, which might have improved with steroids.

So in conclusion, we present a case of bilateral optic nerve glioma in NF-1, which is relatively uncommon and sudden loss of vision in our case could be due to nonspecific inflammatory changes in the glioma or due to unrelated retrobulbar optic neuritis.

References