Case Report

A rare case of neurofibromatosis type I involving the right upper eyelid

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Abstract

Neurofibromatosis is a rare genetic disorder primarily affecting the tissues developed from the neural crest. It has two distinct types, Neurofibromatosis type I and Neurofibromatosis type II (NF1 and NF2). Bilateral Lisch nodules (iris hamartomas), cafe au lait spots over the extremities and a history of first degree relative with the same disease are some of the characteristics of NF1. Secondary glaucomas along with ectropion uvea is also common. NF type II is characterized by bilateral acoustic neuromas and cataracts. Here we report a case of a 25 yr old female having neurofibromatosis type I involving the right upper eyelid, the extremeties and the iris of both eyes.
Keywords: Neurofibromatosis, Lischnodules, cafe' au lait spots.

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INTRODUCTION

Neurofibromatosis is a genetic disorder which primarily affects the growth of tissues developed from neural crest. Its two main types are NF type I (von Recklinghausen disease) which has an autosomal dominant inheritance affecting the long arm of chromosome 17. It is characterised by cafe au lait spots over the extremities, axiallary or inguinal freckling (non exposed parts), optic nerve gliomas, Lisch nodules (iris hamartomas) and a history of a first degree relative with the same disease[1]. There may be a presence of congenital glaucoma associated with ectropion uvea. Rarely a retinal astrocytoma may be seen which may give a mulberry appearance if the nodules are extremely large and cause retinal detatchment. The type II variant of NF is rarer than type I and is caused due to a loss of alleles on chromosome 22. It has a presence bilateral acoustic neuromas. The patients may have a posterior subcapsular, capsular or a cortical cataract during their presenile age. Ocular motor defects are also commonly seen. Rarely Combined hamartomas of the RPE are seen along with an epi retinal membrane. Unilateral Lisch's nodules have also been reported in some cases.

CASE STUDY

A 25 year old female had come to the ophthalmology department with complaints of mass over right upper eyelid since childhood. She was apparently alright till the age of 5 years when she noticed a mass over over her right eyelid (Fig. 1). It was painless and gradually increasing in size, having reached its current size by the age of 7 years. It was associated with drooping of eyelids (ptosis), but no diminuition of vision. Patient had no other ocular complaints. Patient had no history of systemic illnesses like diabetes mellitus or hypertension. Her father is a known case of NF I having mass over left upper eyelid with similar characteristics as the patient, cafe au lait spots over the extremities and Lisch's nodules on the iris of both eyes. On local examination, over the face there was a presence of multiple macules having a size of 0.2cm X 0.2cm. On examination of the upper and lower extremities there was a presence of multiple, well defined, skin colored nodules of sizes ranging from 2cm X 0.5cm to 3cm X 2cm (Fig. 1 and 2). There was a presence of similar nodular distribution over the back and

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abdomen. There was a presence of brownish macules (cafe au lait spots) interspersed in between the nodules of variable sizes. Head position and facial symmetry was normal. Extra ocular muscle movements were normal in all directions without any over action. On ocular examination of the right eye, the visual acuity of the patient was 6/6 and that for near vision was N6. Dimensions of the mass over the eyelid were 3cm X 2cm. Its extent was from lateral orbital wall to 2mm medial to the root of nose and from inferior edge of eyebrow extending on to the lid. On palpation, it was non tender, non reducible, non pulsatile and freely mobile, not attached to the bone. Skin over the swelling was normal, showing no signs of inflammation. There was a presence of ptosis but no diminution of vision as the upper lid did not cover the pupillary area. On Slit lamp examination of right eye, Lisch nodules were seen over the iris (Fig. 4). They were brown in color, seven in number, gelatinous and 1mm – 2mm in diameter. Mechanical ptosis due to the lid mass is evident. There was no evidence of proptosis. Intra ocular pressure was checked with a schiotz indentation tonometer which was 17.3mm of Hg. On dilated fundus examination (by instilling tropico plus eyedrops) of the right eye, optic disc had a slight temporal pallor, rest of the fundus examination showed no evident abnormalities. On examination of the left eye, visual acuity was 6/6 for distant vision and N6 for near vision. On slit lamp examination, iris showed the presence of Lisch nodules which were nine in number with characteristics similar to that in the right eye (Fig. 5). Intra ocular pressure was 17.3mm of Hg. Dilated fundus examination was normal. The diagnosis of neurofibromatosis type I was made from the clinical findings as per its diagnostic criteria.

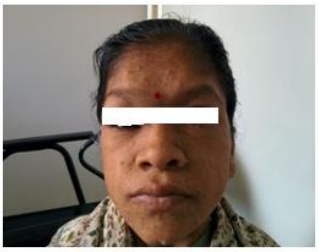

Figure 1:

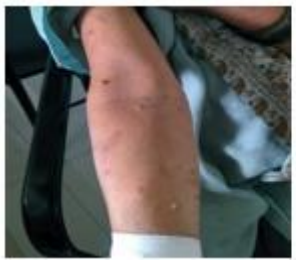

Figure 2:

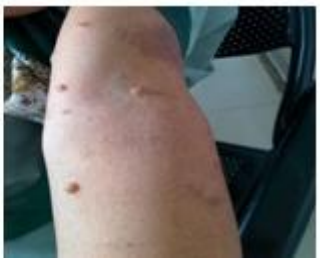

Figure 3:

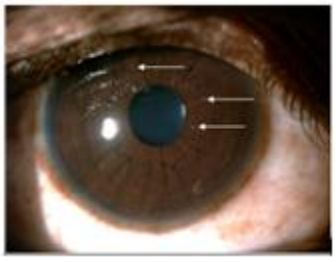

Figure 4:

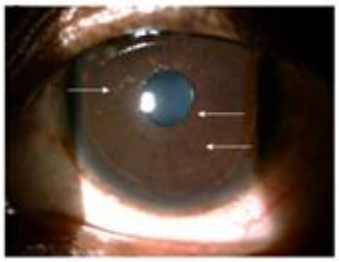

Figure 5:

DISCUSSION AND CONCLUSION

Neurofibromatosis is a rare hereditary disorder which has an autosomal dominant inheritance affecting the long arm of chromosome 17. Involving the tissues developed from neural crest, particularly the nerves of sensation, schwann's cells (responsible for formation of myelin) and melanocytes[2]. The description is given by a german pathologist, Friedrich Daniel von Recklinghausen, who accurately described this condition, hence neurofibromatosis is often referred to as von Recklinghausen's disease (NF1). According to The National Institute of Health Consensus Development

Conference[3], at least two of the following criterias must be present to make the diagnosis of NF1:

1. Five or more cafe-au-lait spots larger than 15mm in diameter in post pubertal patients.
2. Two or more neurofibromas of any type, or one plexiform neurofibroma.
3. Axillary or inguinal freckling
4. Optic glioma
5. Two or more Lisch's nodules
6. A distinctive osseous lesion (pseudoarthrosis of the tibia or sphenoid wing dysplasia)
7. A first degree relative diagnosed with NF1 in accordance with the above criteria.

Our case fulfils 3 out of the the 7 above mentioned criteria, namely , the presence of five or more cafe-au-lait spots, two or more Lisch's nodules and a first degree relative of the patient diagnosed with NF-1. Plexiform neurofibromas of the orbit tend to originate from the orbital branches of the trigeminal nerve. They often affect the upper eyelid, causing a characteristic sinusoidal deformity of the lid margin[4]. The tumor is soft and feels like a 'bag of worms'. The resultant displacement of the globe or ptosis can result in amblyopia in children. In this case however the visual acuity of the patient was 6/6, N6 in both the right as well as the left eye. Lisch's nodules (Hamartomas of the iris) are variable in size and have a smooth dome shaped configuration. They may also be seen in the trabecular meshwork which may eventually lead to a secondary glaucoma. Father of the patient is a case of NF1 according to the above mentioned diagnostic criteria. Surgical amelioration of the lid neurofibroma in this patient is possible after assessing the extent of involvement but the tumour has a tendency to recur. This patient has slight ptosis on the right. S shaped eyelid margin results due to a plexiform neurofibroma. Detection and management of associated glaucoma or optic nerve gliomas or meningiomas are essential. Patients with optic nerve gliomas should be monitored regularly. Intra ocular pressure of both the eyes should be monitored periodically along with a dilated fundus examination to note any glaucomatous changes of the optic nerve. Visual field testing and neuroimaging studies done in this patient were normal. On follow up marriage counseling of the patient should be done. Periodic CT scans should be done to rule out optic gliomas. Intra ocular pressure should be monitored regularly along with visual fields studies to rule out any presence of secondary glaucomas.

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