Original Research Paper

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Ophthalmology

LISCH NODULES AN OCCULAR MANIFESTATION IN A CASE OF NEUROFIBROMATOSIS

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ABSTRACT Neurofibromatosis (NF) is a multisystem disorder caused by a genetic mutation on chromosome 17. Neurofibromatosis 1 is the form with the most characteristic ocular manifestations. Lisch Nodules of the iris are the well-known diagnostic criteria for the disease. Lisch nodules are usually seen at in young age. Our patient is an 8 years old female who presented with pain and swelling over the left chest and who showed features of neurofibromatosis like cafe au lait spots and lisch nodules. Similar lesions were present in her mother. Lisch nodules are pathognomic of NF1 and should be differentiated from iris nevi observed in the general population. This disease is associated with increased morbidity and mortality. With an improved understanding of the disease and advancement in management, we can prevent complications.

KEYWORDS : NF1, neurofibromatosis, iris nodules, lisch nodules, Iris hamartomas, café-au-lait spots.

INTRODUCTION

In 1918 Waardenburg first described the pigmented iris hamartomas. Austrian ophthalmologist- Karl Lischin in 1937 reported the association of these iris hamartomas with neurofibromatosis type 1 (NF1). Riccardi in 1981 used term Lisch nodules in a formal publication. [3] The most common ophthalmologic manifestations of NF1 are Lisch nodules. Lisch nodules appear as yellow or brown coloured dome-shaped lesions on the iris which are best identified on slit-lamp examination. Multiple Lisch nodules are considered specific for NF1, which even precede the neurofibroma; but they lack any correlation with the disease course and severity. [1] Lisch nodules are melanocytic hamartomas that do not affect vision. The usual presentation is between 5 and 10 years of age. [2]

CASE REPORT

An eighth years old female was diagnosed with neurofibromatosis-1and was referred to ophthalmology OPD to look for ocular manifestations of NF1. The parameters at birth were within normal limits. She is born out of non-consanguineous marriage; Full term normal vaginal delivery and postnatal period was uneventful and is immunized up to date.

Her general physical examination showed two large hyperpigmented patches and multiple small hyperpigmented patches over her upper body suggestive of café au lait spots as shown in fig1,2,3. Similar kinds of lesions were present in her mother.

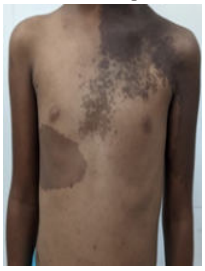

Fig. 1

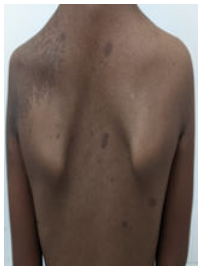

Fig.2

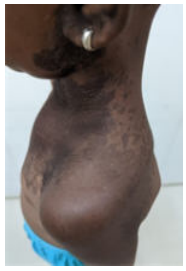

Fig.3

On Ocular examination; vision OD is 6/24 with pinhole 6/6 and OS is 6/18 with pinhole 6/6 on Snellen's chart. OU conjunctiva, cornea, and the anterior chamber was normal and pupils were reactive. OU iris shows 5-6 brown-coloured, dome-shaped lesions suggestive of Lisch nodules as shown in fig4,5. OU extraocular movements were normal in all gazes without pain or diplopia in any gaze. OU Dilated fundus examination was within normal limits.

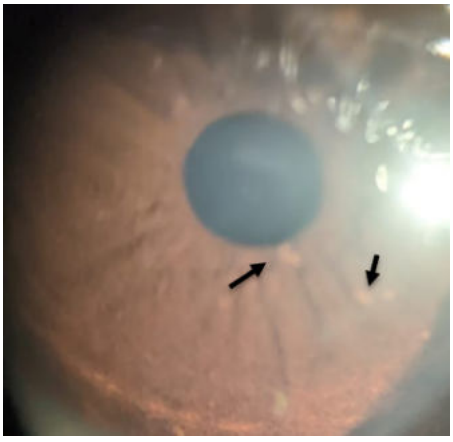

Fig.4 OD shows lisch nodules (black arrows)

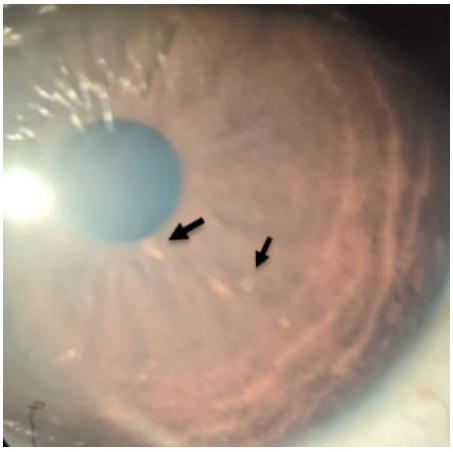

Fig.5 OS shows lisch nodules (black arrows)

As child had complaints of decreased hearing for which an ENT consultation was done and pure-tone audiometry showed bilateral minimal hearing loss.

MRI Brain showed ill-defined T2/FLAIR hyperintense signal intensity in the posterior limb of the left internal capsule, bilateral medial

thalami, midbrain, pons, medulla oblongata, upper cervical spinal cord, bilateral dentate nuclei and bilateral cerebellar deep white matter-likely focal areas of signal intensity. MRI Orbit (plain) showed no significant abnormality in both orbits.

DISCUSSION

Neurofibromatosis is one of the autosomal dominant neuro-oculo-cutaneous disorders which has three forms NF1, NF2, and schwannomatosis; of which neurofibromatosis type 1(NF1) is most common with a prevalence of 1 in 2500-3000 births. [1] Neurofibromatosis (NF) is due to a genetic mutation on chromosome 17-17q11.2 in NF type 1 (NF1) and on chromosome 22-22q12.2 in NF type 2.[4] According to the diagnostic criteria given by the National Institute of Health at least two of the seven features should be present, including six or more cafe´-au-lait spots (> 5 mm diameter in pre-pubertal and > 15 mm diameter in post-pubertal persons), axillary or inguinal freckles, at least two typical neurofibromas With at least two Lisch nodules (iris hamartomas), one plexiform neurofibroma, optic glioma, sphenoid dysplasia, long-bone pseudoarthrosis, and a first-degree relative with NF1.[1]

CONCLUSION

Neurofibromatosis is mainly a neurological and cutaneous disorder but can also affect various other organs such as bones and eyes requiring a collaborative and interdisciplinary approach. Ophthalmologists should be aware of the characteristics of this disease which can help in identifying it at an earlier stage and provide prompt treatment and save eyesight.

Acknowledgment:

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