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NEUROFIBROMATOSIS TYPE 1 –A CASE REPORT

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ABSTRACT Neurofibromatosis is a group of disorder characterized by neuroectodermal tumors arising within multiple organs. It is broadly divided into two - Neurofibromatosis 1 (NF1) and neurofibromatosis 2 (NF2). Neurofibromatosis along with some of other similar disorders is collectively termed as Phakomatosis. NF1 type 1 is aka as Von Recklinghausen's disease /peripheral neurofibromatosis / classic neurofibromatosis whereas the other name for NF 2 is Schwannoma or central neurofibromatosis. It has autosomal dominant inheritance.

KEYWORDS : Lisch's nodules , café au lait spots , freckles

INTRODUCTION

NF1 is a multisystem disorder which require interdisciplinary management by physician/pediatrician , ophthalmologist and dermatologist . Neurofibromatosis type 1 (NF1) or Von Recklinghausen's disease is a rare genetic disorder characterized by the development of multiple noncancerous (benign) tumors of nerves and skin (neurofibromas) and areas of abnormally decreased or increased pigmentation of the skin (1).

NF-1 (peripheral neurofibromatosis, classic neurofibromatosis, Von -Recklinghausen's disease) have characteristic features which includes café au lait spots, axillary and inguinal freckles, Lisch nodules on the surface of iris, optic nerve gliomas and neurofibromas of the central nervous system and skin . The café au lait spots in this syndrome tend to be more numerous and larger than the lesions arising nonsyndromically in the general population (2). Lisch nodules are melanocytic hamartomas of the iris stroma and appear as tan to light brown nodules studding the surface of the iris. In NF-1 subcutaneous neurofibromas are commonly found on the eyelids either as pedunculated tumors or as plexiform neuromas which have the texture of a "bag of worms" on palpation.The optic nerve gliomas tend to cause axial proptosis and optic atrophy in early childhood . CNS neurofibromas has a prevalence between 1 per 3000 live births and can cause hemiparesis, hemiatrophy, and seizures in some of the affected individuals.

Here, we report a case of neurofibromatosis type 1 (NF 1) along with discussion on clinical features, importance of early diagnosis, and interdisciplinary approach for the management .

CASE REPORT

A 12-year-old male patient was referred to the out patient department of Ophthalmology from the Dept. of Pediatrics for detailed ocular assesment in the view of neurofibromatosis .

HISTORY

As informed by the parents, disease started in the early childhood as multiple hyper pigmented skin patches on the arm and chest which were present since birth which then increased in size gradually.Child did not complaint of any ocular symptoms.

EXAMINATION

Patient was well oriented and co-operative .On physical examination , multiple hyper pigmented skin macules(café-au-lait spots) on the chest, arms and back were seen(Fig 2a &b) size varied from 0.5 - 13cm, multiple lesions were >1.5 cm in diameter. Inguinal freckles were present. On local ophthalmic examination multiple Lisch's nodules (Fig 1a& b) were present on the surface of iris in both eyes, without any clinical visual involvement.

CNS involvement was ruled out by pediatrician beforehand. Child was also referred to dermatologist for further evaluation.

LAB AND RADIOLOGICAL INVESTIGATIONS

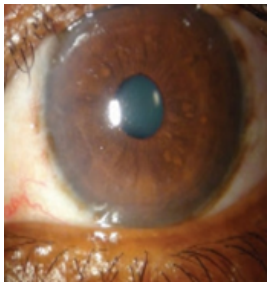
The standard laboratory tests as advised by the pediatric and dermatology dept. were in the normal range. X-ray photography was within the normal too.

DIAGNOSTIC CRITERIA OF NEUROFIBROMATOSIS TYPE 1

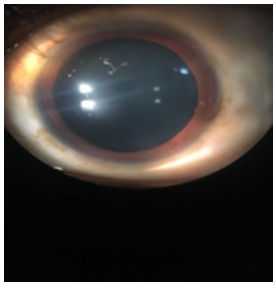
The diagnosis NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health Consensus Development Conference (3).

1. 6 or more café-au-lait spots = >5 mm prepubertal , >15 mm postpubertal
2. > 2 neurofibromas or 1 plexiform neurofibroma
3. Freckling axillary/inguinal
4. > 2 Lisch nodules (iris hamartomas)
5. Optic pathway glioma
6. Bone lesion (sphenoid dysplasia, thinning long bone cortex + pseudarthrosis)
7. First degree relative

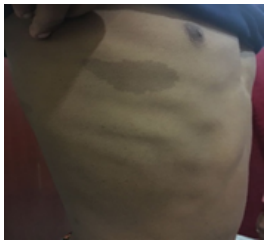
FIGURE



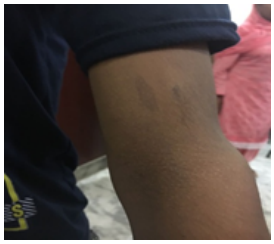
(Fig 1a)



(Fig 1b)



(Fig 1a)



(Fig 1b)

DISCUSSION

NF-1 is the most common type of phakomatoses and accounts for 90% of all the cases (2). Pigmentary changes are nearly present at birth , but neurofibromas are not common at that age. Café-au-lait spots are present in 99% of all the newborns, discoloration generally emerge before the development of neurofibromas and their amounts increase with age (4). Cutaneous neurofibromas are soft, fleshy-pink colored tumors, commonly seen on the trunk and limbs as sessile or dome-shaped masses.

Lisch's nodules (melanocytic pigmented iris hamartomas) are present in 94% cases. Hypertension may be present in some due renal vascular stenosis or pheochromocytoma .The incidence of pheochromocytoma , rhabdomyosarcoma, leukemia, and Wilms tumor is higher than in the general population.

Neurofibromatosis type 1 (NF1) represents a major risk factor for development of malignancy, mainly malignant peripheral nerve sheath tumors, optic gliomas and leukemias.

MANAGEMENT

The management is multi-focal as it consist of both the treatment of the disease and rehabilitation .

1. The skin tumors should be excised if they are cosmetically objectionable or show an increase in size suggesting malignant changes.
2. Cranial and spinal neurofibromas should be excised and the gliomas , meningiomas also demand surgical measures as well.
3. Bilateral optic nerve gliomas are usually treated with radiation, unilateral ones are excised.
4. Genetic counseling.
5. Most children with NF1 follow a regular education, one –third of patients may need additional therapy such as language therapy, extra-curricular educational support, or psychological support.

FOOTNOTES

Source of support: Nil
Conflict of interest: None

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