

CASE REPORT

NEUROFIBROMATOSIS TYPE I: CASE REPORT

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HOW TO CITE THIS ARTICLE:

Neha Gupta, Poonam Gupta, M. C. Baberwal, Hemant Mishra. "Neurofibromatosis Type I: Case Report". Journal of Evolution of Medical and Dental Sciences 2014; Vol. 3, Issue 68, December 08; Page: 14718-14722, DOI: 10.14260/jemds/2014/3977

ABSTRACT: Neurofibromatosis (NF) is a term that has been applied to a variety of related syndromes, characterized by neuro ectodermal tumors arising within multiple organs and autosomal-dominant inheritance. Neurofibromatosis type I(NF-1), known as well as Recklinghausen's disease, we have presented a case report of 10 year old boy with complain of scalp swelling on right postero-lateral aspect of scalp with multiple flat, hypo pigmented macule on back, neck. On radiology work up including MRI there were multiple plexiform neurofibromas, multiple non-neoplastic hamartomatous lesion suggestive of neurofibromatosis type-1.

KEYWORDS: Neurofibromatosis type-1, plexiform, recklinghausen's disease.

INTRODUCTION: Neurofibromatosis (NF) is a term that has been applied to a variety of related syndromes, characterized by neuro ectodermal tumors arising within multiple organs and autosomal-dominant inheritance. At least 8 different clinical phenotypes of neurofibromatosis have been identified and are linked to at least two genetic disorders. Neurofibromatosis type I (NF-1 is characterized by multiple café-au-lait spots and the occurrence of neurofibromas along peripheral nerves.

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History: A 10year old boy presented to neurology department of Mahatma Gandhi medical college Jaipur with complains of scalp swelling on right postero-lateral aspect. The swelling was soft, fusiform swelling with multiple flat, hypo pigmented macule on back and neck region.

Physical Examination: Dermatological status - On skin examination there were multiple flat, hypo pigmented macule present ranging from a few millimeters to several centimeters in diameter, some of them pedunculated; multiple café-au-lait spots with diameter > 1, 5 cm (fig1); axillary and inguinal freckling. The mucous membranes were not affected.

Ophthalmological status - Lisch's nodules on the iris of both eyes were without clinical visual involvement on ophthalmologic work up-On slit lamp examination there was evidence of lish nodules in iris (Fig. 3)

Lab and imaging studies, histologic findings and Consultations: The standard laboratory tests values were in the normal range.

Radiology work Up: Findings on MRI brain-(Fig. 4-8) diffusely infiltrating sheet like lobulated mass lesion was in scalp of right occipito-parietal region and extending inferiorly into the upper neck appearing hyper- intense on T2 and hypo intense on T1 weighted images. The lesion shows heterogeneous enhancement.

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No bony erosion or intracranial extension of the lesion is seen. Small similar ill-defined lesion was also seen in subcutaneous fat of left fronto-orbital region with extension into the superior extra conal fat of left orbit similar mass lesion was also noted in the pre vertebral space s/o multiple plexiform neurofibromas in right occipito -parietal scalp, left fronto- orbital region and pre vertebral space. There was an altered signal intensity lesion seen in right superior frontal cortex and sub cortical region in parasagittal location appearing hyper intense on T2 and FLAIR image and hypo intense on T1 images with no contrast enhancement s/o non neoplastic hamartomatous lesion.

DISCUSSION:

Diagnostic Criteria: According to the National Institute of Health Consensus Development Conference 1, at least two of the following criteria must be present to make the diagnosis of NF-1:

1. Five or more cafe-au-lait spots larger than 5 mm in diameter in pre pubertal patients; six or more cafe-au-lait spots larger than 15 mm 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 nodules.
6. A distinctive osseous lesion (pseudoarthrosis of the tibia or sphenoid wing dysplasia).
7. A first-degree relative diagnosed with NF-1 in accordance with the above criteria.

Plexiform neurofibromas of the orbit tend to originate from the orbital branches of the trigeminal nerve.¹ They often affect the upper eyelid, causing a characteristic sinusoidal deformity of the lid margin.² 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. Plexiform neuromas of the orbit are associated with congenital absence of the sphenoid or enlargement of the sellaturcica. Peripheral neurofibromas are benign tumors consisting predominately of Schwann’s cells and fibroblasts with endothelial, peri neural, and mast cells.³

There is evidence that they have a single cell origin despite multiple cell types within the tumors.⁴ Plexiform neurofibromas occur in about one third of NF-1 cases, most commonly on the trunk and less often on the limbs, head and neck. They are benign and rarely symptomatic, but they can cause significant cosmetic and visual problems if the orbit is involved. Café-au-lait spots are composed of epidermal melanocytes with giant pigment granules (macro melanosomes) within the cytoplasm and are of neural crest origin. They are not pathognomonic of neurofibromatosis, having been reported in association with several other conditions and in patients not affected by the condition.⁵

Hamartomas of the iris (melanocytic nevi) can be seen and are called Lisch nodules. They are variable in size and have a smooth, dome-shaped configuration.⁶

Although clinical findings are primarily neurocutaneous in nature, any organ system can be involved. The diagnosis requires six or more cafe au lait cafe-au-lait spots, each larger than 1, 5 cm in diameter. Axillary freckling is also highly

Suggestive of the diagnosis.^{7,8} Areas of hypo pigmentation or hyper pigmentation can also be seen.

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CONCLUSION: The patient described here is a very typical case of NF-1, which presents a considerable interest because of the high generalization of the skin lesion.⁹ In such cases, a detailed patient investigation is required, because of the possibility for generalized involvement of other organs.¹⁰ The proper clinical and genealogic analysis is important for the determination of the genetic risk and prognosis for their relatives of the proband.

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Fig. 1



Fig. 2

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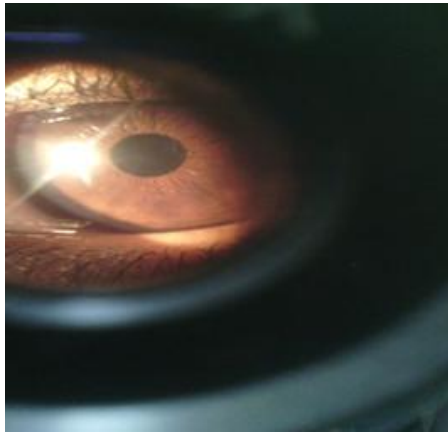


Fig. 3: On slit lamp examination there was evidence of lisch nodules in iris

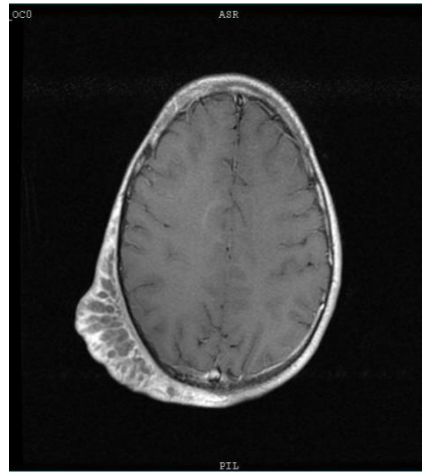


Fig. 4: Post Gd T1W image showing plexiform neurofibromas at right parieto-occipital region

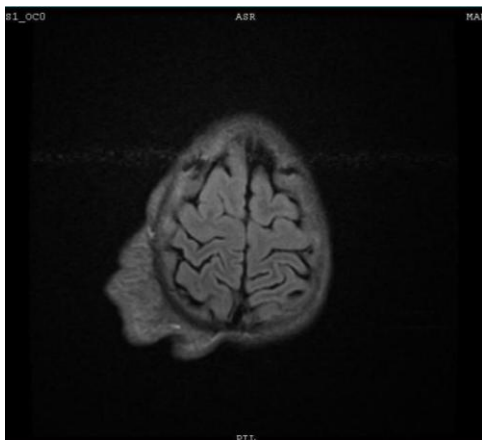


Fig. 5

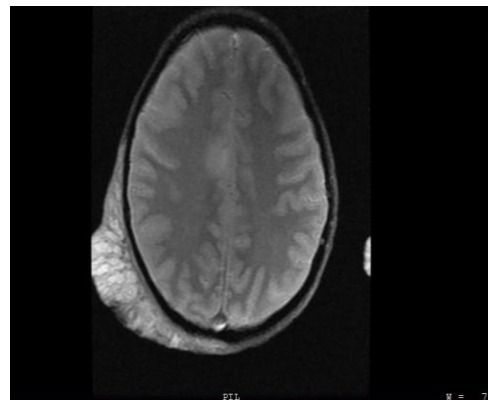


Fig. 6

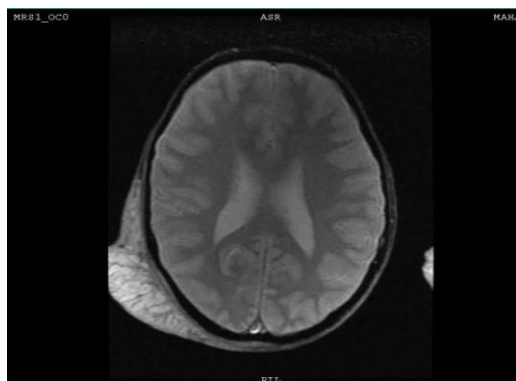


Fig. 7

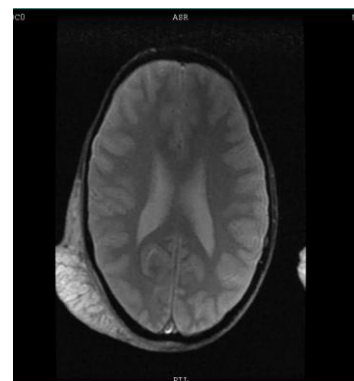


Fig. 8

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Date of Submission: 13/11/2014.
Date of Peer Review: 14/11/2014.
Date of Acceptance: 02/12/2014.
Date of Publishing: 08/12/2014.