



## SEGMENTAL NEUROFIBROMA WITH MULTIPLE LENTIGENES – A RARE CASE REPORT

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<p><b>Article Info</b></p> <p>Received 15/12/2015 Revised 27/12/2015 Accepted 02/02/2016</p> <p><b>Key words:</b> Segmental Neurofibromatosis, lentigenes, café au lait macules</p>	<p><b>ABSTRACT</b></p> <p>Neurofibromatosis is a genetic disorder characterized by benign growths of peripheral nerve sheaths, neurofibroma and café au lait macules (CALM) associated with various cutaneous and systemic manifestations. It is an autosomal dominant disease with complete penetrance by the age of 5 years. They are soft, violaceous, sessile or pedunculated tumor mass of varying size distributed mostly over the trunk and extremities. We report a case of segmental neurofibromatosis in a 45 years old female who presented with complaints of raised skin lesions in right side of the body and multiple pigmented lesions all over the body and axilla for the past 30 years.</p>
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### INTRODUCTION

Crowe et al [1] was the first person to describe Segmental neurofibromatosis (SNF) in 1956 and the authors termed it *Sectorial neurofibromatosis*. Miller and Sparkes [2] in 1977 renamed this term as SNF. Riccardi [3] included SNF as type-V in his classification of neurofibromatosis. The clinical features of SNF are: Café-au-lait spots and/or neurofibroma in a solitary unilateral segment of the body, no family history, with no crossing of the median line, and no systemic involvement. Roth *et al* [5] further subdivided the SNFs into four subtypes: hereditary, bilateral true segmental and localized with deep involvement.

### CASE REPORT:

A 45 years old female presented to our OPD with complaints of raised skin lesions in right side of body for the past 30 years. History of itching was present. She also gave history of multiple pigmented lesions all over body

and axilla since childhood. There was no history of visual disturbances, seizures, bone pain, difficulty in breathing, palpitation, and headache. There was no history of similar complaints in the family.

Dermatological examination revealed multiple hyperpigmented soft nodules of size varying from few millimeter upto 2 cm over the right shoulder, right side of abdomen, back and right leg. Button hole sign was positive. No pedunculated or plexiform lesions were noted. Multiple hyperpigmented macules and patches present in right half of body with few spill over lesions in left. Intertriginous and palmar freckling was present. Ophthalmological examination was done to rule out Lisch nodule and optic glioma which proved to be normal. Orthopaedic evaluation did not reveal any abnormalities. Neurological examination done was also normal. Scalp, oral mucosa and nail were normal. Routine investigations done were normal.



**Figure 1. Clinical picture shows multiple pigmented nodules of varying size of 1 x 1mm to 3 x 2mm present over the back.**



**Figure 2. Clinical picture showing multiple pigmented macules and a single well defined patch of size 1 x 1cm are seen over the abdomen.**



## DISCUSSION AND CONCLUSION

Phacomatosis is a group of disorders with cutaneous as well as central and peripheral nerve tumours. Neurofibromatosis is one such disorder with various skin features which may be detected prenatally, at birth or even thereafter. The first characterization of the disorder was made by von Recklinghausen in 1882. Several clinical syndromes have been identified such as neurofibromatosis type 1 (NF1) also known as von Recklinghausen disease, neurofibromatosis type 2, segmental or mosaic NF-1, familial café au lait spots, schwannomatosis. Neurofibromatosis type 1 being the most common of these disorders occurs at a frequency of 1 in 3500 births and with nearly 100% penetrance of the disease by the age of 5 years [4,5]. Neurofibromatosis is transmitted as an autosomal dominant trait. SNF is a rare disorder with its prevalence rate of 0.0014 and 0.002% [6]. The NF1 gene in Chromosome 17 encodes neurofibromin which downregulates Ras activity by means of alteration or premature stop codon. Almost 50% of cases are sporadic

As per Riccardi's classification NF 5 relates to segmental or unilateral neurofibroma or Café au lait macules in one or more dermatomes which is non familial. Post zygotic mutation especially of gonads due to germline mosaicism is implicated as a cause for this condition. Clinically, patients may be divided into four groups: (i) with only pigmentary changes, (ii) with isolated plexiform neurofibromas (iii) with both pigmentary changes and neurofibromas and (iv) with only neurofibromas. The neurofibromas are mostly asymptomatic and range from

0.1 cm to several centimeters in diameter. They tend to arise in dermatomal distribution, most commonly cervical followed by lumbar, thoracic, and sacral region [8]. Pigmentary changes constitute café-au-lait macules and axillary freckling, with the former being more common. Some patients of SNF have had complications of NF1 which includes optic pathway gliomas, learning difficulties, and pseudoarthrosis. Those with learning difficulties tend to have large areas of cutaneous involvement. Lisch nodules are seen rarely in segmental NF. The importance in recognizing this specific type, lies on genetic anticipation where a full blown neurofibromatosis is predictable in the forthcoming generation.

This case is being reported because of its rarity and rare associated features (lentigenes).

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## CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

## STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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