

# Segmental Neurofibromatosis - A Rare Case Report and Review of Literature

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## Abstract

Segmental neurofibromatosis (SNF) (also known as NF Type V) is a rare disorder characterized by neurofibromas or café-au-lait macules in combination with neurofibromas, which are limited to one segment or region of the body. We report a case of a 56-year-old male patient who presented with multiple, firm, non-tender, skin-colored papules over the trunk grouped along the T10 dermatome bilaterally. Histopathology examination showed wavy, buckled nuclei of Schwann cells thereby suggesting the diagnosis of neurofibroma. A patient of SNF should be evaluated further to rule out any systemic features consistent with generalized NF. Although the risk of transmission to offspring is small, genetic counseling and evaluation for skin lesions and cognitive impairment can be advised.

**Key words:** Café-au-lait spots, Neurofibroma, Neurofibromatosis Type 1, Segmental neurofibromatosis

## INTRODUCTION

Segmental neurofibromatosis (SNF) (also known as NF Type V) is a rare disorder characterized by neurofibromas or café-au-lait macules in combination with neurofibromas, which are limited to one segment or region of the body. <sup>1</sup>NF can be divided into 3 broad categories, classified presently on the basis of their molecular aspects: NF Type 1, first described by von Recklinghausen in 1882, which is the most common form; NF Type 2, with the hallmark feature of bilateral vestibular neuromas; and finally, all other types of NF, which include atypical or variant forms of the disease. <sup>2</sup> Crowe *et al.* proposed the term sectorial NF for the localized form of NF was called sectorial NF by Crowe *et al.*, and later, Miller and Sparkes modified the nomenclature to 'SNF' - which happens to be the current term for neurofibromas having segmental distribution. <sup>3</sup> Thorax and abdomen (55%) is the most

common site for SNF, followed by upper extremities (20%), and lower limb and face (10% each). SNF on the face is very rare, and only about 10 cases have been encountered. <sup>4</sup>

## CASE REPORT

A 56-year-old male patient presented to our hospital with complaints of multiple swellings over the abdomen and back for 20 years. The patient had come to seek medical advice for these swellings which were completely asymptomatic according to him. There was no history suggestive of any cognitive impairment or visual disturbances. Furthermore, there was no history of NF in the family members.

On examination, there were multiple, firm, non-tender, skin-colored papules over the abdomen and back grouped along the T10 dermatome bilaterally (Figures 1 and 2). There were no ocular or bony features of NF.

Based on the history and clinical appearance of the lesions, a provisional diagnosis of multiple lipomas or neurofibromas was made.

Fine-needle aspiration cytology revealed a smear with moderate cellularity and spindle-shaped cells scattered

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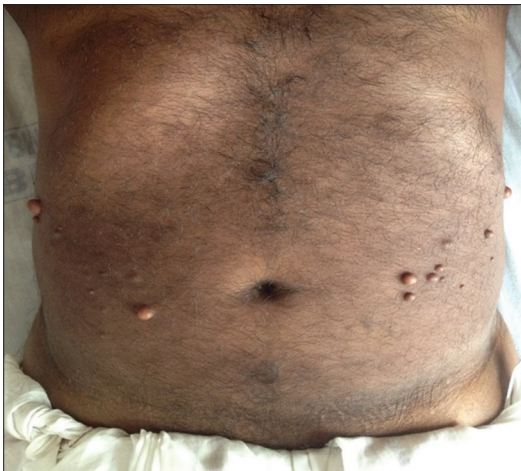
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**Figure 1: (a and b) Segmental distribution of neurofibromas (lateral view)**



**Figure 2: Segmental distribution of neurofibromas (anterior view)**

singly. The cells show round ovoid nuclei with regular nuclear membrane and moderate amount of cytoplasm. One of the papules was excised and subjected for histopathology examination which showed wavy, buckled nuclei of Schwann cells (Figure 3. H and E stain,  $\times 200$ ) and further confirmed the diagnosis of neurofibroma.

Since there were multiple neurofibromas grouped along the T10 dermatome and the distribution was consistent with the description of localized NF in the literature, we labeled the case as SNF.

## DISCUSSION

SNF is considered as a rare disorder with the unique characteristic of café-au-lait macules and/or neurofibromas following a regional distribution, and its prevalence has been estimated at about 0.0027%. It is proposed that SNF is related to post-zygotic mutation of the NF type 1 gene,

which leads to somatic mosaicism. Reports of patients who have localized disease but have children with generalized NF have been explained on the basis of genetic mosaicism.<sup>5</sup> The clinical features of SNF were established by Riccardi and classified this different presentation first as NF Type 5.<sup>6</sup>

Riccardi defined SNF as café-au-lait macules or neurofibromas in a single, unilateral segment of the body, without crossing of midline, no systemic involvement, and no family history. Cases that could not be explained and accommodated in according to Riccardi classification prompted Roth *et al.* to propose a further subclassification (Table 1): True segmental, localized with deep involvement, hereditary, and bilateral.<sup>7</sup>

In a group of 56,183 young male observed by Ingordo *et al.* where the subjects were between the age of 17 and 18 years and represented a population homogeneous with reference to age, sex, race, and country of origin, only 11 cases of NF with relative frequency of 0.020% were found. They observed only one case of SNF during the study (relative frequency 0.0018%). The authors thereby came to a conclusion that SNFs are probably not only underdiagnosed but are 10 times more uncommon than other forms of NF.<sup>8</sup>

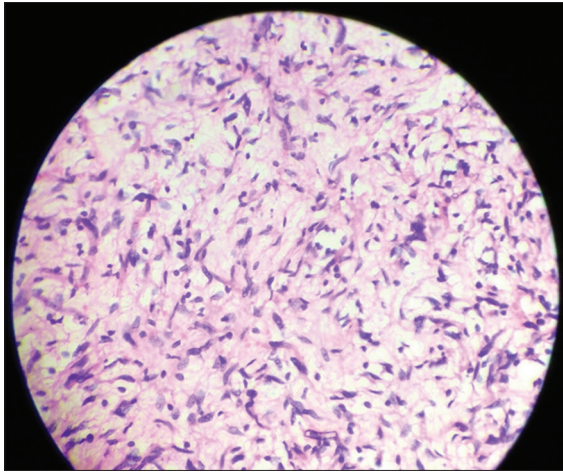
Clinically, patients can be categorized into four groups: (i) those with only pigmentary changes, (ii) with only neurofibromas, (iii) with both pigmentary changes and neurofibromas, and (iv) with isolated plexiform neurofibromas. Although most of the lesions are unilateral, there are reports of bilateral SNF.<sup>9</sup>

Counselling the patients of segmental neurofibromatosis about the clinical differences and some basic similarities with the classic NF Type 1 should form a part of routine management. However, it is significant to remember that SNF1 should be approached as a localized phenotype of NF1, not a separate form of NF, so that crucial screening is not overlooked.<sup>10</sup> The presence of optic gliomas and Lisch nodules has been reported in cases of SNF.<sup>11</sup>

There is a 5-15% lifetime risk of developing a malignancy with classical NF1, and this is about 2.5 times the risk seen in the general population.<sup>12</sup> In patients with SNF, the incidence of malignancy has been approximated at 5.3%.<sup>13</sup>

## CONCLUSION

A patient of SNF should be evaluated further to rule out any systemic features consistent with generalized NF. Although the risk of transmission to offspring is small, genetic counseling and evaluation for skin lesions and cognitive impairment can be advised. The patient should be followed up to monitor disease progression



**Figure 3: Photomicrograph showing wavy, buckled nuclei of Schwann cells (H and E stain, x200)**

**Table 1: Subclassification of SNF (Roth *et al.*)**

Subcategory	Description	Phenotypic features
I	True segmental	Unilateral, localized without family history of neurofibromatosis
II	Localized with deep involvement	Underlying deep systemic involvement
III	Hereditary	Localized with genetic transmission of neurofibromatosis
IV	Bilateral	Bilateral cutaneous manifestations

SNF: Segmental neurofibromatosis

and development of any systemic features of NF and any malignant change.

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