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## Segmental Neurofibromatosis: Atypical Localisation

Segmental Nörofibromatosis:  
Olağan Dışı Yerleşim

### Abstract

Neurofibromatosis (NF) is a genetic disease leading pathological findings in skin, soft tissue, bone and nervous system by affecting neural crest cells. Due to its heterogeneity neurofibromatosis was divided into eight different subgroups (NF-I NF-VIII) by Riccardi. Segmental neurofibromatosis (NF type V) is characterized by cutaneous neurofibromas and Café-au-lait spots limited with a segment of dermatome. Here we report this case with numerous, painless cutaneous nodules showing extension from the shoulder to the dorsal aspect of the right hand, since it a rare case.

**Key words:** Neurofibromatosis, segmental neurofibromatosis, NF-V, skin tumors, neural tumors, neurofibroma

**Conflicts of Interest:** The authors reported no conflict of interest related to this article.

### Özet

Nörofibromatozis (NF), nöral krest hücrelerini etkileyerek, deri, yumuşak doku, kemik ve sinir sisteminde patolojik bulgulara yol açan genetik bir hastalıktır. NF, heterojenitesinden dolayı Riccardi tarafından sekiz alt (NF-I NF-VIII) grupta sınıflandırılmıştır. Segmental nörofibromatozis (NF tip V) spesifik bir segment ya da dermatoma sınırlı, kutanöz nörofibromlar ve/veya Café-au-lait lekeleri ile karakterizedir. Biz, sağ üst ekstremitede proksimalden başlayarak el dorsumuna kadar lineer uzanım gösteren, çok sayıda ağrısız yumuşak nodüllerle prezente olan olgumuzu, nadir bir dermatomal dağılıma sahip olduğu için sunuyoruz.

**Anahtar kelimeler:** Nörofibromatozis, segmental nörofibromatozis, NF-V, deri tümörleri, nöral tümörler, nörofibrom

**Çıkar Çatışması:** Yazarlar bu makale ile ilgili olarak herhangi bir çıkar çatışması bildirmemiştir.

### Introduction

Neurofibromatosis (NF) is a genetic disorder which affects all neural crest cells (melanocytes, schwann cells and endoneurial fibroblasts) that causes pathological findings in the skin, soft tissue, bone and nervous system. The heterogeneity of the disease directed Riccardi (1) to classify NF into eight subtypes (NF-I to NF-VIII). One of these subtypes (NF-V) is segmental neurofibromatosis (SNF), which presents with cutaneous neurofibromas, and/or café-au-lait macules, that are limited to a specific segment or dermatoma of the body with no crossing of midline, no family history, and no

systemic involvement (1). Here we describe a case of SNF with a very rare dermatomal distribution.

### Case

A 65-year-old man was referred for evaluation of numerous, painless cutaneous nodules located on the dorsal aspect of the right hand, forearm and arm. About 10 years ago he noted the development of a small nodule on the extensor aspect of his right arm and all the other nodular lesions appeared 2 years ago after a pain on his arm. The lesions do not itch, burn, or bleed. No other members of his family or close relatives exhibited a similar condition. Past

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medical history was unremarkable except for a basal cell carcinoma.

Physical examination revealed multiple, soft, dome-shaped, skin colored or reddish nodules ranging in size from 3 mm to 1.8 cm in diameter, arranged linearly in a dermatomal distribution from the right shoulder to the dorsal aspect of the right hand (Figure 1). The nodules were manually invaginated into the skin with pressure (button-hole sign). Examination of the rest of the skin was normal. A biopsy specimen of the nodule showed typical features of neurofibroma (Figure 2). Cafe-au-lait macules, axillary freckling and lisch nodules on the iris were absent. Complete blood cell count, chemical studies, chest X-ray and nuclear magnetic resonance imaging of the head were normal. These features were consistent with our clinical diagnosis of SNF.

## Discussion

NF has an autosomal dominant inheritance and is a member of the neurocutaneous syndromes (phakomatoses). SNF was first described by Crowe et al. in 1956 (2). Ricardi incorporated SNF in his classification as type V. He thought that a postzygotic somatic mutation, in primitive neural crest cells, is the most likely cause for development and thus the lesion should be strictly non-inherited (1). Roth et al. also categorized this disease as true segmental, localized with deep involvement, hereditary, and bilateral type (3). The classification was manipulated by Weiss et al. a) "Classic" NF1 b) Whole gene deletion phenotype NF 1, and c) Alternate forms of NF1 (i.e. with incomplete/atypical features). SNF was included in the third category as a localized form of neurofibromatosis (4).

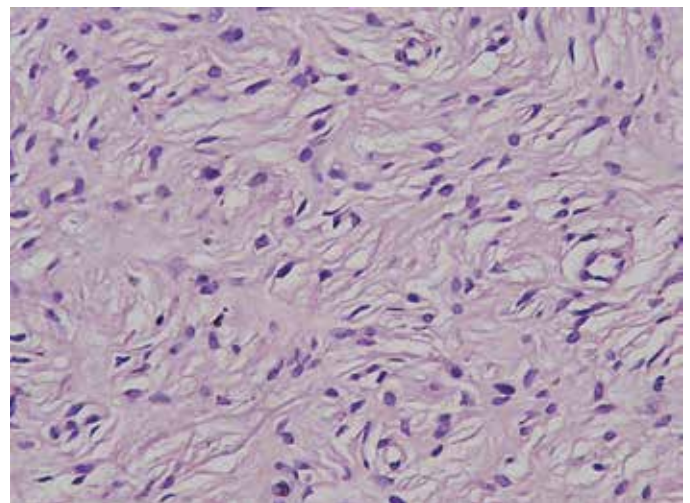
Incidence peaks in people aged 10-30 years and a second peak occurs between 50-70 years (5). Most commonly, patients present with only neurofibromas. The neurofibromas are mostly asymptomatic and range in size from 0.1 cm to several centimeters in diameter. They tend to arise in dermatomal distribution, most commonly cervical, followed by thoracic, lumbar, and sacral region. Pigmentary findings are common in children whereas, in adults neurofibromas are more common (6). In the majority of the cases that are characterized by pigmentary abnormalities, lesions tend to follow the lines of Blaschko (7).

In our case, neurofibromas were arranged linearly in a dermatomal distribution from the right shoulder to the dorsal aspect of the right hand. In literature only a few cases reported locating on the upper extremity (Table 1). As in our patient, both upper extremity and hand involvement, only one case has been reported (8). No other stigmata of NF was found, and no cases of NF were known in the patient's family.

Patients who present with only neurofibromas, need to be examined carefully for the criteria of generalized form of NF-1 and should be followed to detect any systemic complications that may occur.



**Figure 1. Dome-shaped, skin colored or reddish nodules arranged linearly in a dermatomal distribution from the right shoulder to the dorsal aspect of the right hand**



**Figure 2. 40x H&E Spindle-oval nucleated, S100 (+) and Vimentin (+) cell proliferation without necrosis, mitosis and atypia**

**Table 1. Characteristics of patients with segmental neurofibromatosis of the upper extremity\***

| Case No | Sex | Age of Onset | Site        | CI       | SI | FH | Reference No |
|---------|-----|--------------|-------------|----------|----|----|--------------|
| 1       | F   | 40           | Bi: A, H    | NF       | -  | -  | 7            |
| 2       | F   | NS           | R: H, FA    | CALM, NF | -  | -  | 8            |
| 3       | M   | 13           | L: FA, H    | CALM, NF | HL | -  | 9            |
| 4       | M   | 55           | R: A, FA, H | NF       | -  | -  | CR           |

A: arm, Bi: bilateral, CI: cutaneous involvement, CR: current report, F: female, FA: forearm, FH: family history, H: hand, HL: Hodgkin lymphoma, L: left, M: male, NS: not stated, R: right, SI: systemic involvement, present, -, absent

\*We only include patients who was diagnosed as having segmental neurofibromatosis of the arm and/or forearm and hand.

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