A Case of Segmental Neurofibromatosis in Confined to Posterior Tibial Nerve Dermatome

Posterior Tibial Sinir Dermatomuna Sınırlı Bir Segmental Nörofibromatozis Olgusu

Mutlu ÇAYIRLI,[a]
Mustafa TUNCA,[b]
Ahmet AKAR,[b]
Yavuz POYRAZOĞLU,[c]
Ali Fuat ÇİÇEK[d]

Clinics of
[a]Dermatology,
[b]General Surgery,
Mevki Military Hospital,
Departments of
[c]Dermatology,
[d]Pathology,
Gülhane Military Medical Academy,
Ankara

Geliş Tarihi/*Received:* 04.06.2014
Kabul Tarihi/*Accepted:* 09.07.2014

Yazışma Adresi/*Correspondence:*
Mutlu ÇAYIRLI
Mevki Military Hospital,
Clinics of Dermatology, Ankara,
TÜRKİYE/TURKEY
mutlu78tr@yahoo.com

ABSTRACT Neurofibromas are benign tumors arising from the peripheral nerve sheath. Neurofibromatosis type 1 is one of the most commonly seen autosomal dominant disorders which consists of multiple neurofibromas. Neurofibromas may rarely exist singly in people without neurofibromatosis type 1 at any location along the peripheral nervous system. Segmental neurofibromatosis is a rare form of neurofibromatosis in which skin lesions are limited to a region of the body. Here we report a patient of segmental neurofibromatosis who had multiple localized dermal neurofibromas on the lower limb with no other cutaneous or systemic findings. This is the first report of isolated segmental neurofibromatosis in confined to the posterior tibial nerve dermatome.

Key Words: Neurofibromatoses; neurofibroma

ÖZET Nörofibromlar periferal sinir kılıfından köken alan benin tümörlerdir. Nörofibromatozis tip-1 çok sayıda nörofibromdan oluşan ve en sık izlenen otozomal dominant kalıtımlı hastalıklardan biridir. Nörofibromlar nadiren nörofibramotozis tip- 1 olmayan insanlarda da periferik sinir sistemindeki herhangi bir bölgede izole olarak bulunabilirler. Segmental nörofibromatozis deri lezyonlarının vücudun bir bölgesinde sınırlı olduğu nadir izlenen bir nörofibromatozis formudur. Burada diğer kutanöz ya da sistemik bulgular olmaksızın alt ekstremitesinde lokalize çok sayıda dermal nörofibromu bulunan bir segmental nörofibromatozis olgusunu sunuyoruz. Bu olgu posterior tibial sinir dermatomunda sınırlı bildirilen ilk segmental nörofibromatozis olgusudur.

Anahtar Kelimeler: Nörofibromatozis; nörofibrom

Turkiye Klinikleri J Dermatol 2014;24(2):90-2

Neurofibromas are benign tumors arising from the peripheral nerve sheath. Existence of multiple dermal neurofibromas is a hallmark of neurofibromatosis type 1 (NF1), which is one of the most common autosomal dominant disorders. Neurofibromas may rarely exist singly in people without NF1 at any location along the peripheral nervous system.[1] Segmental neurofibromatosis (SN) is a rare form of disease in which skin lesions are limited to a region of the body. Here we report a patient of SN who had multiple localized dermal neurofibromas on the lower limb with no other cutaneous or systemic findings. This is the first report of isolated SN in a unique linear pattern confined to the posterior tibial nerve dermatome.

CASE REPORT

A 30-year-old female presented to dermatology outpatient department with asymptomatic swellings on her right leg of 15 years duration. The patient was complaining about an increase in the size and number of tumors in the last two years. Dermatologic examination revealed soft, multiple, flesh-colored, cutaneous nodules (ranging from 1-2 mm to 1 cm in diameter) over the right posterior tibial region in a dermatomal distribution (Figure-1). Clinically we considered these lesions as neurofibroma. However, there were no nodules or other cutaneous findings of NF1 at the rest of the body. Optic glioma or lisch nodules were not detected on ocular examination. The patient had no first degree relative with NF1. No other abnormalities were found in a detailed neurological examination. A clinical diagnosis of SN was considered. One of the nodular lesions was completely excised under local anesthesia for histopathological examination. Histopathology of the tumor revealed non-encapsulated but well-circumscribed lesion located in the dermis and subcutis (Figure 2). Proliferation of spindle cells with wavy nuclei located in fibrous stoma was seen. Atypical mitoses or nuclear pleomorphism were absent (Figure 3). These findings confirmed the clinical diagnosis of SN.

DISCUSSION

In contrast to NF1, SN is a rare disorder characterized by localized cutaneous neurofibromas and\or café-au-lait macules with no systemic involvement.[2] Distribution of tumors is commonly unilateral but can be bilateral. The most common affected site of the body is the thoraco-abdominal

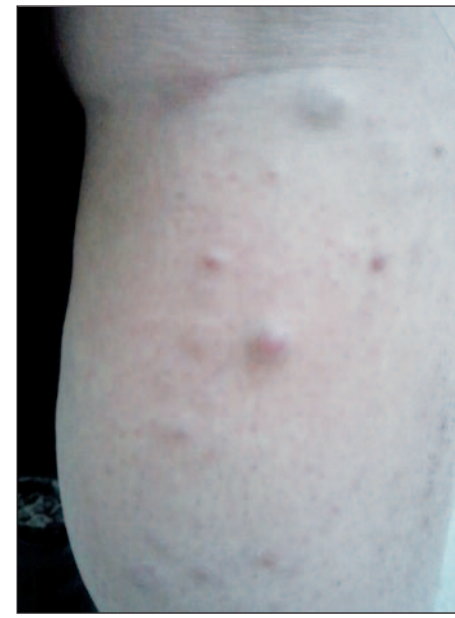

FIGURE 1: Cutaneous nodules over the right posterior tibial region in a dermatomal distribution.
(See color figure at http://www.turkiyeklinikleri.com/journal/dermatoloji-dergisi/1300-0330/)

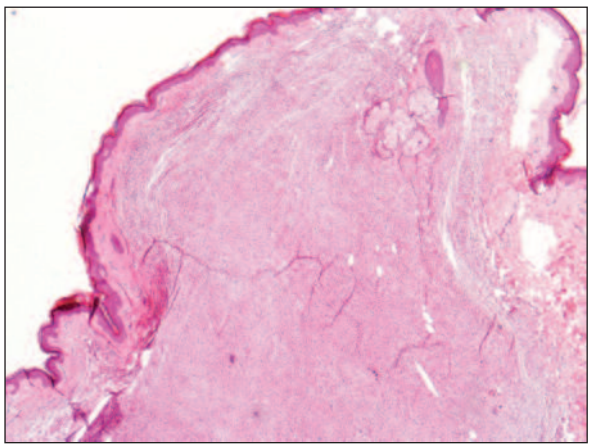

FIGURE 2: Non-encapsulated but well- circumscribed lesion located in the dermis and subcutis. (H&Ex20).
(See color figure at http://www.turkiyeklinikleri.com/journal/dermatoloji-dergisi/1300-0330/)

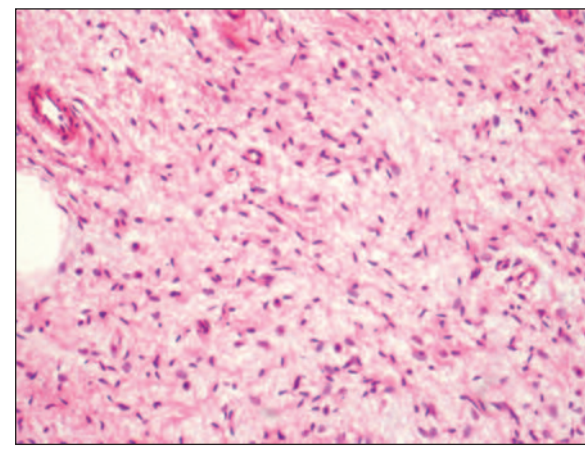

FIGURE 3: Proliferation of spindle cells with wavy nuclei located in fibrous stoma. Atypical mitoses or nuclear pleomorphism were absent. (H&Ex200).
(See color figure at http://www.turkiyeklinikleri.com/journal/dermatoloji-dergisi/1300-0330/)

region.[3] Axillary freckling and family history have been reported in limited number of cases. Cutaneous neurofibromas are usually asymptomatic but especially those deep seated on peripheral nerves and spinal roots can cause neurological dysfunction. The prevalence of complications including malignancies in SN is much lower than the NF1.[1]

The etiology of SN is not well-known but it is thought to arise from a postzygotic mutation in the NF1 gene of primitive neural crest cells leading to somatic mosaicism. Reguler NF-1, sarcoidosis, benign tumors (trichoepithelioma, leiomyoma), cutaneous lymphomas, xanthomas should be considered in the differential diagnosis of the disease.[4]

As in our case, cutaneous lesions can be observed in a limited dermatome and can be diagnosed incidentally while the patient is being examined for a different complaint. Therefore, a thorough physical, ophthalmologic and neurologic examination should be performed in patients with SN for rule out generalized disease that may present with systemic complications.

REFERENCES

1. Blakley P, Louis DN, Short MP, MacCollin M. A clinical study of patients with multiple isolated neurofibromas. J Med Genet 2001;38(7):485-8.
2. Gabhane SK, Kotwal MN, Bobhate SK. Segmental neurofibromatosis: a report of 3 cases. Indian J Dermatol 2010;55(1):105-8.
3. Jankovic I, Kovacevic P, Visnjic M, Jankovic D, Velickovic M. A unique case of hereditary bilateral segmental neurofibromatosis on the face. An Bras Dermatol 2012;87(6):895-8.
4. Morais P, Ferreira O, Bettencourt H, Azevedo F. Segmental neurofibromatosis: a rare variant of a common genodermatosis. Acta Dermatovenerol Alp Panonica Adriat 2010; 19(3):27-9.