A Case of Segmental Neurofibromatosis

Bir Segmental Nörofibromatozis Olgusu

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ABSTRACT Segmental neurofibromatosis is a rare variant of neurofibromatosis (NF) type 1. It is characterized with cafe-au-lait macules, freckling and/or neurofibromas limited to one region of the body. Females are affected more than males and the lesions are usually localized on the servicothoracic region. The prevalence of the disease associated systemic involvement and complications in segmental neurofibromatosis is much lower than in NF type 1. Segmental neurofibromatosis is believed to be a result from a postzigotic mutation of NF type 1 gene. We report the case of 37-year-old man presented with neurofibromas on the left side of the lumbar region.

Key Words: Neurofibromatosis 1; neurofibromatosis type 5

ÖZET Segmental nörofibromatozis, nörofibromatozis (NF)'in nadir görülen bir varyantıdır. Vücudun bir bölgesine sınırlı kafeola makülleri, çillenme ve/veya nörofibromlarla karakterizedir. Kadınlarda daha sık saptanır. Lezyonlar en sık serviko-torasik bölge yerleşimlidir. Hastalıkla ilişkili sistemik tutulum ve komplikasyonlara NF tip 1'e kıyasla daha nadir rastlanır. Segmental nörofibromatozisin somatik mozaisizm sonucu NF tip 1 geninin postzigotik mutasyonuna bağlı geliştiği düşünülmektedir. Bu yazıda sol lumbal bölgede nörofibromlar ile tanı koyduğumuz 37 yaşındaki erkek hastamızı sunmayı uygun bulduk.

Anahtar Kelimeler: Nörofibromatozis 1; nörofibromatözis tip 5

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Neurofibromatosis (NF) type 1 is an autosomal dominant disorder affecting one in 3500 people. It was first described by von Recklinghausen in 1882.[1] The cafe-au-lait macules, neurofibromas, axillary freckling and Lisch nodules of the iris are the most frequent features of the disease. Minor features include macrocephaly, hypertelorism, learning disabilities, thorax abnormalities and orthopedic problems such as scoliosis and pseudoarthrosis. Malignancies associated with the disease are optic glioma, peripheral nerve tumors, rhabdomyosarcoma, pheocromocytoma, duodenal carcinoma, colon cancer and leukemia.[2-4]

Segmental neurofibromatosis is a rare variant of NF type 1 with a prevalence range between 0.0014 and 0.002 percent.[5] It was first described by Gammel in 1931; and then Crowe et al. suggested the nomenclature 'sectorial neurofibromatosis' in 1956.[6] In 1977, Miller and Sparkes proposed the

term 'segmental neurofibromatosis'.[6] In 1982, Richardi classified neurofibromatosis into eight groups and termed segmental neurofibromatosis as NF type 5. According to his classification, segmental neurofibromatosis was characterized with cafe-au-lait macules and/or axillary freckling and/or neurofibromas in a single unilateral region of the body with no family history and systemic involvement.[5,6] Then, in 1987 Roth et al. divided the disease into four subtypes: true segmental type (Richardi type 5), localized type with deep involvement, hereditary type and bilateral type.[6]

CASE REPORT

A 37-year-old man presented with skin colored, nontender papules and nodules on the left lumbar region of the body since the age of 32. The number of the lesions had increased over time. On physical examination, four skin colored, soft papules and three nodules in a dermatomal distribution were observed on the left side of the lumbar region (Figure 1). Three nodules are excised by plastic surgeons. Histopathological examination showed a dermal well-circumscribed spindle-cell neoplasm (Figure 2). Immunhistochemically, S-100 protein staining was positive in tumor cells (Figure 3). Histopathological findings were consistent with neurofibroma. Axillary freckling and cafe-au-lait macules were absent. Lisch nodules were not detected by split lamb examination. He was in good health and his family history was unremarkable. His two daughter had no stigmata of neurofibromatosis. Routine laboratory examination, abdominopelvic ultrasound and craniospinal magnetic resonance imaging were normal. The clinical, histopathological and radiological findings were consistent with segmental neurofibromatosis.

DISCUSSION

Segmental neurofibromatosis may be divided into four categories according to different clinical features: patients only with pigmentary abnormalities, only with neurofibromas, both with pigmentary changes and neurofibromas and with isolated plexiform neurofibromas.[5] Neurofibromas in a dermatomal distribution is the most common presentation

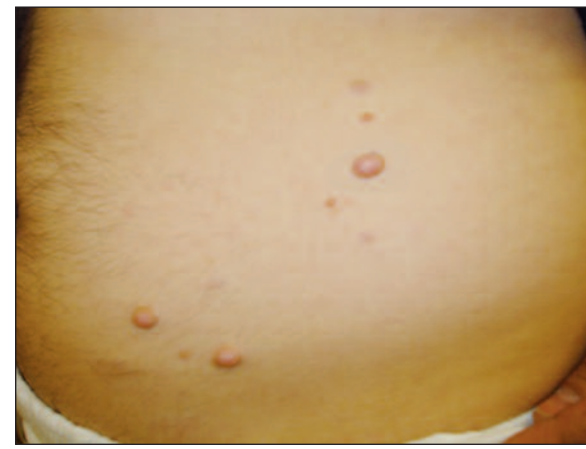

FIGURE 1: Skin colored, soft papules and nodules in a dermatomal distribution on the left side of the lumbar region.

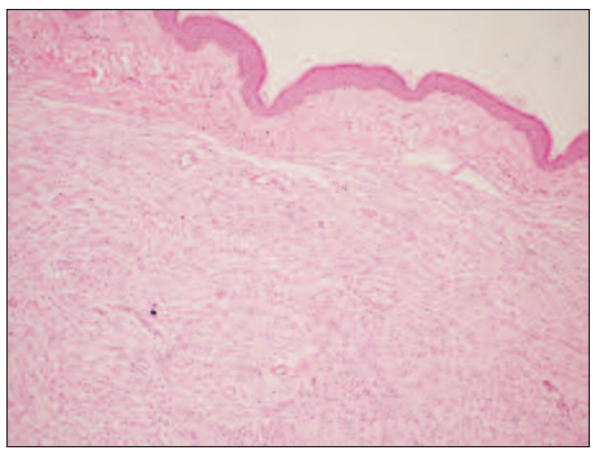

FIGURE 2: Dermal well-circumscribed spindle-cell neoplasm (H&Ex20).

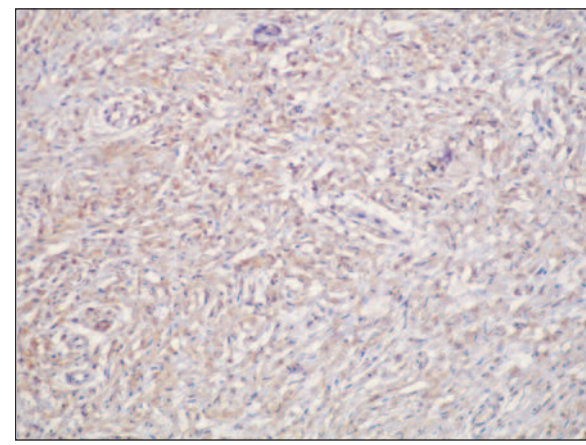

FIGURE 3: S-100 protein staining was positive in tumor cells (S100x40)

of the disease. The most common localization is the cervical segment followed by thoracic, lumbar and sacral regions of the body. In the literature patients with neurofibromas on face and extremities have been reported.[6,7] Segmental neurofibromatosis is mostly seen in Caucasian women in twenties.[8] It is usually unilateral but some cases with bilateral lesions have been reported.[9]

Segmental neurofibromatosis is thought to be a result of postzigotic NF type 1 gene mutation. If the somatic mutation occurs early in embryonic development, generalize disease (NF type 1) appears; whereas mutations that occurs late, results as a localized disease (segmental neurofibromatosis).[10] Gonadal mosaicism is thought to be the main factor for people with segmental neurofibromatosis having children with NF type 1.[11]

Skin disorders are the major manifestations of segmental neurofibromatosis. Systemic involvement and complications are much lower than the NF type 1. In the literature, segmental neurofibromatosis associated with skeletal abnormatilies, soft tissue hypertrophy, visceral neurofibromas, unilateral renal agenesis, renal angiomyolipoma and malignancies such as Hodgkin's lymphoma, colon carcinoma, gastric carcinoma, lung carcinoma, and peripheral nerve sheath tumour have been reported.[3,7,8,12-15]

The prevalence of segmental neurofibromatosis has been estimated to range between 0,0014% to 0,002%. We think that most of the cases are underdiagnosed because of the absence of symptoms. It is usually diagnosed while the patient is being examined for a different concern or cosmetic problems. However the risk for systemic involvement is low, all patients must be evaluted for complications. We report this case of segmental neurofibromatosis because of the rarity of the disease.

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