Bilateral Segmental Neurofibromatosis Showing Different Dermatomal Distribution

Sang Hyun Cho, M.D., Eun Young Bae, M.D., Chang Nam Lee, M.D., Seung Hun Lee, M.D.*, Jeong Deuk Lee, M.D.

Departments of Dermatology and Internal Medicine, College of Medicine, The Catholic University of Korea, Seoul, Korea*

A 43-year-old woman presented with numerous cutaneous neurofibromas, limited to the left anterior chest(T2-3) and the right lower back(L1-2). These had been present for 10 years. Neither cafe-au-lait spot, intertriginous freckle, nor Lisch nodule was found. The family history was negative for neurofibromatosis. Biopsy specimens showed circumscribed, nonencapsulated neurofibromas. The present case was a rare form of bilateral segmental neurofibromatosis in that while most of the reported cases involved the same dermatome bilaterally, she had bilaterally different dermatomal neurofibromas. (Ann Dermatol 15(2) 71~74, 2003).

Key Words : Bilateral segmental neurofibromatosis

Bilateral segmental neurofibromatosis(NF) is a rare disorder characterized by cafe-au-lait spots and/or neurofibromas, limited to a region of the body, in a bilateral dermatomal distribution. We report an unusual form of bilateral segmental NF, involving the right lower back and the left anterior chest respectively. In other words, neurofibromas were presented on both the right and left sides of the trunk, but at separate dermatomes, not at the same level bilaterally.

CASE REPORT

A 43-year-old Korean woman presented with multiple, soft and non-tender papules and subcutaneous nodules on the left side of the chest and right side of the lower back. These had been present for 10 years. Since the beginning of the disease, the patient had noticed gradually increasing numbers of these lesions, limited to the two separate dermatomes. She denied associated pain and pruritus. There was no family history of neurofibromatosis.

Physical examination revealed soft, non-tender, flesh-colored papules and nodules, 0.1-2.0cm in diameter, in separate distributions involving the T2-3 on the left side of the chest and L1-2 on the right side of the back(Fig. 1). Many of the soft nodules were pushed down into the panniculus by light pressure with the finger("buttonholing") and sprung back when released. She had no cafe-au-lait spots, intertriginous freckles, or Lisch nodules. Complete blood cell count and chemical studies were normal. Chest X-ray and a computed tomography scan of the brain showed no abnormal findings.

Histologic examination of skin biopsies taken from the lesions on the chest and lower back respectively revealed well-circumscribed, plexiform arrangements of spindle cells within the dermis. The lesion was not encapsulated and consisted of spindle-shaped cells with bland, slightly wavy nuclei and slender elongated cytoplasmic processes in a myxoid background, in which eosinophilic wavy collagen bundles were apparent(Fig. 2). These findings were consistent with neurofibroma.

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Reprint request to : Jeong Deuk Lee, Department of Dermatology, Our Lady of Mercy Hospital, The Catholic University of Korea, #665 Bupyung-dong, Bupyung-gu, Inchon, 403-720, Korea
Tel. 82-32-510-5528, Fax: 82-32-510-5827
E-mail. leejd@olmh.cuk.ac.kr

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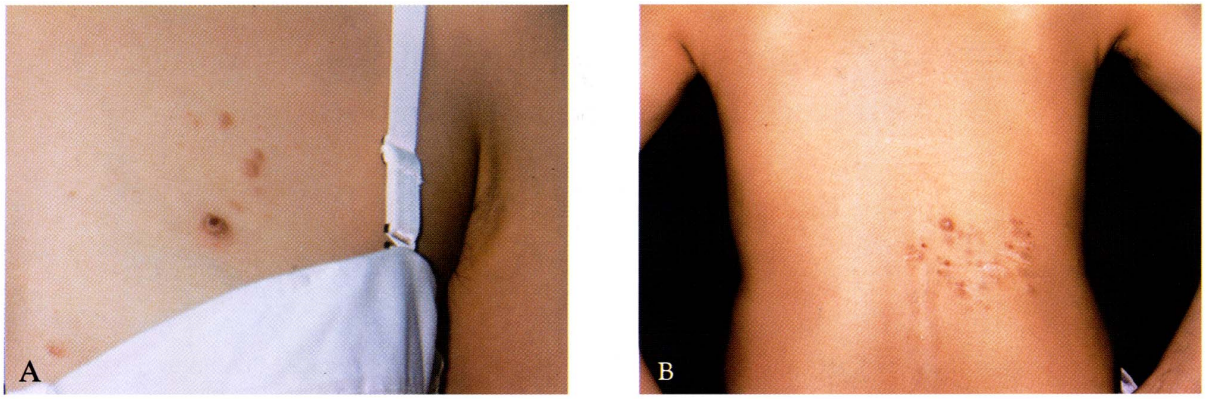

Fig. 1. Dermatomal distribution of neurofibromas on the left chest(A) and the right lower back(B).

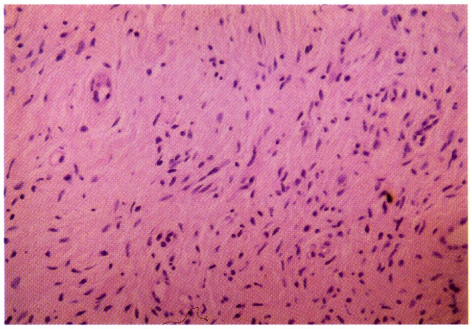

Fig. 2. Skin biopsy from subcutaneous mass on the left chest shows spindle-shaped cells and bundles of eosinophilic wavy fibrous tissue in myxoid stroma (hematoxylin and eosin; original magnification × 200).

Taking cosmetics into account, we excised four neurofibromas which were located on the chest and relatively large in size.

DISCUSSION

Neurofibromatosis is considered as a heterogeneous group of disorders manifested by developmental changes of the nervous system, bones and skin, and clinically defined by the presence of neurofibromas, multiple cafe´-au-lait(CAL) spots, intertriginous freckles and Lisch nodules.

Segmental neurofibromatosis is a rare form of neurofibromatosis that is characterized by neurofibromas and/or CAL spots in a dermatomal distribution. Segmental neurofibromatosis was first described by Gammel[1] in 1931. Crowe et al.[2] suggested the term sectional neurofibromatosis in 1956. In 1977, Miller and Sparkes[3] proposed the more precise term, segmental neurofibromatosis, which continues to be used in contemporary literature.

The neurofibromatosis is extremely variable and Riccardi[4] classified neurofibromatosis into eight categories in 1982. According to Riccardi's classification, type V neurofibromatosis includes only those cases of segmental neurofibromatosis which are unilateral, not associated with extracutaneous lesions and nonfamilial. So bilateral segmental cases are classified into type VIII neurofibromatosis. However, Roth et al[5]. were aware that many cases of segmental neurofibromatosis did not meet these stringent criteria, so they proposed a further classification of segmental neurofibromatosis into four subtypes in 1987(Table 1): Type I, true segmental form, no systemic involvement, nonfamilial; Type II, localized with deep involvement; Type III, hereditary segmental form; Type IV, bilateral segmental form. Our case would be classified as type VIII neurofibromatosis according to Riccardi's classification, and the fourth subtype of segmental neurofibromatosis according to the classification of Roth *et al.*

Bilateral segmental neurofibromatosis is a rare form of segmental neurofibromatosis. Wong searched 16 cases of bilateral segmental neurofibromatosis[6]. Since then, three more cases have been reported in the literatures[7-9]. While each of these cases shared the characteristic dermatomal distribution of neurofibromas, lentigenes and CAL spots, there are numerous differences in their clinical

Table 1. Classification of segmental neurofibromatosis by Roth *et al.*

Category	Description	Features
Type I	True segmental	Segmental CAL spots and/or neurofibromas, no systemic involvement, nonfamilial
Type II	Localized with deep involvement	Segmental with deep systemic involvement, Nonfamilial
Type III	Hereditary segmental	Segmental, no deep involvement, familial
Type IV	Bilateral segmental	Bilateral segmental CAL spots and/or neurofibromas, no deep involvement, nonfamilial

Table 2. Reported cases of bilateral neurofibromas at different dermatome without pigmented lesions

Author	Age/Sex	Neurofibromas	Pigmented lesions
Winkelmann and Johnson	47/M	Bilateral : right back, abdomen left knee	None
	70/M	Bilateral : right arm left infra-orbital	None
Present case	43/F	Bilateral : right lower back left chest	None

features. Wong classified bilateral segmental neurofibromatosis into three groups[6]. First, bilateral segmental neurofibromatosis had unilateral neurofibromas with contralateral pigmented lesions. Second, bilateral segmental neurofibromatosis had bilateral neurofibromas only, without pigmented lesions. Third, bilateral segmental neurofibromatosis had bilateral neurofibromas associated with unilateral pigmented lesions. The present case would be classified into the second group in Wong's classification.

While most of the reported cases of bilateral segmental neurofibromatosis involved same level of the body in a bilateral dermatomal distribution, our case differed from previously reported cases in that she had different dermatomal bilateral segmental neurofibromas(left: T2-3, right: L1-2). Only 2 previous cases of bilateral neurofibromas at different dermatome without pigmented lesion had been reported and the clinical features of these cases are summarized in Table 2[6,10].

Segmental neurofibromatosis is usually known as a non-inherited disease. Crowe *et al.*[11] suggested that segmental neurofibromatosis is a result of a somatic mutation in early embryonic development of the primary neural crest. Some authors have proposed that bilateral segmental neurofibromatosis may be due to two coincidental, independent mutations. As cases of familial segmental neurofibromatosis surfaced, Sloan et al.[12] suggested the possibility that some cases of segmental neurofibromatosis involved gonadal mosaicism as well as somatic mosaicism.

We report a case of bilateral segmental neurofibromatosis involving different dermatomes. Presentation of patients with cutaneous neurofibromas should prompt the physician to perform a thorough physical examination and to question the patient regarding family history. It is important for the physician to determine the type of neurofibromatosis the patient has so that, if indicated, an appropriate search for systemic disease and genetic counseling can be undertaken.

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