

A Case of Segmental Neurofibromatosis

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Segmental neurofibromatosis is a rare disorder characterized by café-au-lait spots and neurofibromas, or only neurofibroma, limited to one region of the body. Disease-associated systemic involvement is uncommon. Most patients with segmental neurofibromatosis do not have a family history of neurofibromatosis. In Korea, there have been 3 reported cases of segmental neurofibromatosis. None of them had café-au-lait spots, systemic involvement, or family history. This report describes a case of segmental neurofibromatosis in a 25 -year-old woman who had Becker's nevus-like café-au-lait spots and this was linked to the presence of Fanconi's syndrome in a second degree relative of the patient. (*Ann Dermatol* 11(2) 109~111, 1999).

Key Words : Café-au-lait spot, Fanconi's syndrome, Segmental neurofibromatosis

Segmental neurofibromatosis(SNF, neurofibromatosis type V) is a rare disorder characterized by café-au-lait spots(CALS) and/or neurofibromas limited to a region of the body, often in a unilateral, dermatomal distribution^{1,2,3}. More than 82 patients of SNF have been reported, and they usually do not have a family history of neurofibromatosis⁴. We report the first Korean case of SNF who showed a Becker's nevus-like clinical appearance.

CASE REPORT

A 25-year-old female patient had dark-brown colored multiple patches on her right shoulder and arm. These had been present from birth. An examination revealed multiple soft non-tender subcutaneous masses in the pigmented patches(4,5th thoracic dermatome(Fig.1)). She had no axillary freckling, Lisch nodules or disease-associated systemic involvement. A family history showed that her father and two sisters had single CALS in a variable area without any associated diseases. A niece of the patient was diagnosed as having Fanconi's syn-

drome with cutaneous multiple CALS at the A. university hospital. Three punch biopsies were done on two CALS in our patient, and all of the specimens showed multiple, well-circumscribed aggregates of spindle-shaped cells and bundles of eosinophilic wavy fibrous tissue in the dermis(Fig. 2A & B). The cells were positive for S-100 protein and negative for smooth muscle actin.

DISCUSSION

SNF was originally described as sectorial neurofibromatosis(NF) by Crowe et al, in four patients⁵. Riccardi classified NF into eight types that included segmental NF as NF type V. Until now, SNF has been described in 86 patients⁴; 34 men and 52 women. The age of onset varied from birth to 83 years; the mean age of onset was 28 years. In the men, the lesions initially appeared between childhood and the age of 74(median, 28 years); in women, between birth and 83 years(median, 27 years).

Skin lesions that have been found in patients with SNF include neurofibromas(86 patients), CALS(23 patients), and axillary freckling(at least 9 patients). The typical distribution of SNF was unilateral, but in five patients the neurofibromas were bilateral⁶. The lesion usually occupied a single dermatome; cervical(34 patient), thoracic(34 pa-

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tient), lumbar(20 patients), and sacral(5 patients). In some patients nearby dermatomes were involved

and in 16 patients there was no mention about the involved dermatomes.

There are some dermatomal pigmented lesions that must be differentiated, such as Becker's nevus, nevus spilus, progressive cribriform and zosteriform hyperpigmentation/macular pigmented lesion, and reticulated hyperpigmentation distributed in a zosteriform fashion. In our case, the first clinical impression was Becker's nevus, but the impression was corrected by pathological findings.

SNF is usually known as a non-inherited disease. Most patients(93%) did not have a family history of NF. Crowe et al³ suggested that SNF is a result of a somatic mutation of a single cell early in embryonic development that through subsequent cell division involves a number of progeny cells, leading to the limited distribution of neurofibromas. However, six patients had a first-degree relative with neurofibromatosis^{7,8,9,10}. Only 2 cases of SNF showed one first-degree relative with SNF(one case was father and another was daughter)⁸. Moss et al¹¹ said that a mosaic form of neurofibromatosis type I genes(17th chromosome long arm band 11.2) may be responsible for SNF where there was a familial history.

Fig. 1. Dark brown colored multiple pigmented patches and a protruding mass underneath the patch on the right shoulder.

Fig. 2A. A biopsy specimen from the pigmented patch. A circumscribed non-capsulated tumor of the dermis is composed of loosely spaced spindle cells and wavy collagenous strands. (H&E, $\times 40$).

2B. Thin spindle cells are associated with thin, wavy collagen bundles. The cells and collagen bundles are loosely spaced in a matrix. (H&E, $\times 400$).

Table 1. Reported cases of Korean segmental neurofibromatosis

Authors	Onset age	Sex	Location	F.Hx	*CALS	Systemic Sx
Hur et al	28	F	Rt. 3,4th cervical dermatome	-	-	-
	25	F	Rt. 2nd cervical dermatome	-	-	-
Chung et al	58	F	Rt. 3,4th cervical dermatome	-	-	-
Our case	birth	F	Rt. 4,5th thoracic dermatome	Fanconi's SD (niece)	+	-

*CALS : café-au-lait spot

Sloan et al¹². said that somatic mosaicism accompanied by gonadal mosaicism is a genetic mechanism of familial SNF. The extremely scarce reported case of SNF with familial transmission is probably due to a significant percentage of patients having significant family histories who failed to give this information on the presence of NF in offsprings and siblings.

The association of Fanconi's syndrome in second degree relatives of SNF as in our case is not reported in the literature, and the significance of this association is unknown. Only one case of Fanconi's syndrome presenting in a patient with NF was reported¹³. In that case, hypophosphatemic osteomalacia was found to be associated with neurofibromatosis.

Systemic involvement in patients with SNF is uncommon. Many authors believe that SNFs are in reality, underdiagnosed, because it occurs with a clinical picture that, because of the absence of symptoms, can be neglected by the patient and pass unnoticed by the physician.

There have been 3 reported cases of segmental neurofibromatosis in Korea. On comparing these with our case, our patient showed multiple CALS, and a family history of Fanconi's syndrome in a second degree relative (Table 1).

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