

Localized Multiple Pseudoatrophic Plaques: A Rare Clinical Form of Segmental Neurofibromatosis

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Sir,

Segmental neurofibromatosis (NF) is characterized by multiple neurofibromas or café-au-lait spots (1–4). It is unclear whether segmental NF is arranged within dermatomes or not. This disorder is considered to arise from a post-zygotic NF-1 gene mutation resulting in somatic mosaicism (5). We report here a case of segmental NF, in which multiple neurofibromas showed an unusual clinical presentation in the form of localized multiple pseudoatrophic plaques. The lesion histopathologically showed neoplastic proliferation in a dispersed pattern within the middle reticular dermis associated with only a small amount of collagenous stroma.

CASE REPORT

A 52-year-old Japanese woman presented with a 7-year history of multiple, plaque lesions on the right side of her neck. She sometimes felt itching at the area where the lesions were localized, and she scratched the area, making it 'red'. Physical examination revealed multiple, skin-coloured to slightly tan, roughly oval, slightly elevated plaque lesions, ranging from 0.6 to 1.8 cm in diameter, which were distributed on the right side of the neck in a band-like arrangement (Fig. 1). No herniation phenomenon was exhibited in these slightly soft plaque lesions on palpation, and some of the plaques had slight, fine wrinkling. The first clinical diagnosis was anetoderma (macular atrophy) or eruptive collagenoma. There were no café-au-lait spots, axillary freckles, Lisch nodules or other signs of NF. She had no family history of NF.

A biopsy specimen from one of the plaque lesions disclosed a proliferation of neoplastic cells with oval to spindle nuclei, dispersed within the reticular dermis and largely confined to the middle reticular dermis, with no involvement of the deep reticular dermis and

subcutaneous tissue (Fig. 2A). The epidermis and papillary dermis were normal. The neoplastic oval to spindle-shaped cells were associated with only a small amount of thin wavy collagenous stroma, reserving some normal collagen bundles (Fig. 2B). The hyperplastic peripheral nerves, intermingled with neoplastic cells and mucin, were also seen in the reticular dermis and subcutaneous tissue. Elastic tissue stain revealed the presence and no decrease of elastic fibres within the lesion of the middle reticular dermis as well as in the uninvolved, deeper reticular dermis. Immunohistochemical staining with S-100 protein labelled the neoplastic cells and highlighted their localization in the middle reticular dermis sparing the deep reticular dermis (Fig. 2C).

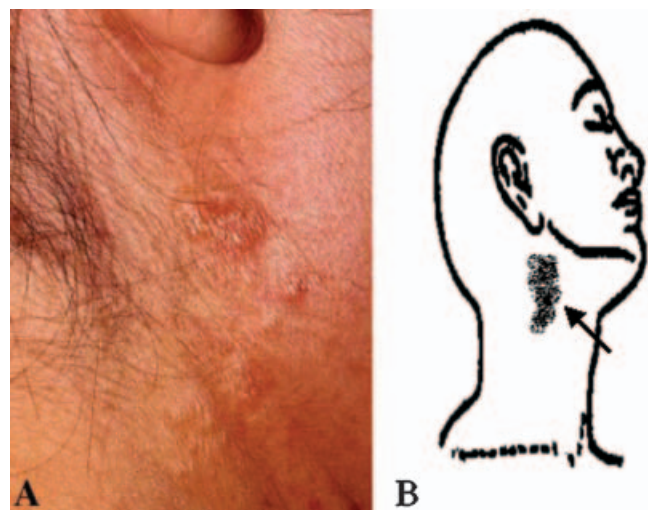


Fig. 1. (A) Multiple, skin-coloured to slightly tan, roughly oval, slightly elevated plaque lesions, on the right side of the neck in a band-like arrangement. (B) Schematic drawing of the distribution of the lesions (arrow).

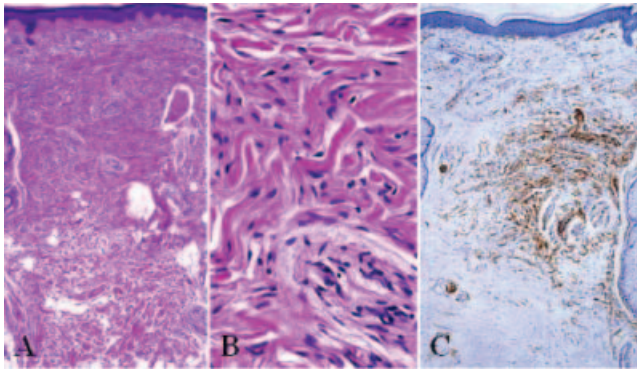


Fig. 2. (A) A proliferation of neoplastic neural cells, dispersed and largely confined to the middle reticular dermis. (B) Neoplastic neural cells with oval to spindle-shaped nuclei are associated with only a small number of stroma. A hyperplastic peripheral nerve with mucin can be seen. (C) Immunohistochemical staining with S-100 protein highlighting the localization of the neoplastic, neural cells in the middle reticular dermis (haematoxylin-eosin stain: A, $\times 14$; B, $\times 100$; immunostain: C, $\times 18.5$).

DISCUSSION

The usual clinical features of cutaneous neurofibromas seen both in NF-1 and in the sporadic type without stigmata of NF-1 are soft, skin-coloured or slightly tan, sessile or polypoid papules or nodules. Cutaneous neurofibromas in segmental NF also show those clinical features (1–3), although the features represent a wide range from eruptive small papules to multiple large nodules packed together with the appearance of a bunch of grapes (6, 7). Although it is well documented in the literature that the lesions usually occupy a single dermatome or a part of it (1–3), there is not yet enough evidence to confirm the relationship.

The present case of segmental NF showed an unusual clinical presentation in the form of localized multiple pseudoatrophic plaques. The histopathological features of cutaneous neurofibromas usually show a circumscribed lesion of neoplastic spindle cells associated with abundant, thin wavy collagenous stroma, which usually involves the whole reticular dermis, often affecting the subcutaneous tissue. By contrast, in the present case the neoplastic neural cells proliferated in a dispersed pattern within the reticular dermis and were largely localized in the middle reticular dermis, sparing the lower reticular dermis and subcutaneous tissue. In addition, they were associated with a lower amount of collagenous stroma. These histopathological features are considered to reflect the unusual clinical features of pseudoatrophic plaques.

Similar clinical features to those in the present case can be found in the reported cases, which described a rare clinical form of cutaneous neurofibroma, pseudoatrophic macules (8–10). The pseudoatrophic macules in the reported cases were all seen in the context of NF-1. Usually, the pseudoatrophic macules are slightly depressed from the surrounding skin as compared with

the present case of slightly elevated plaque lesions (8–10). One reported case demonstrated a sparse amount of neoplastic cells surrounding blood vessels in the reticular dermis without involving the deeper dermis and a reduction of normal collagen bundles present in the reticular dermis, which might be attributed to the clinical appearance of the pseudoatrophic macule (8). Another case of pseudoatrophic macule reported low-density collagenous stroma within the lesion, although the lesion involved the subcutaneous tissue as well as the whole reticular dermis (10). When neural neoplastic cells, dispersed in the dermis, proliferate mainly in surrounding fibrous, thick-walled capillaries and small veins with widened lumina, the lesions clinically become deep purple spots, called 'blue-red macules' (8, 11).

Pruritus observed in this case, first recognized as a feature of generalized neurofibromas in NF-1, has also been reported as a symptom in segmental NF (12). Pruritus may be helpful in the clinical diagnosis of segmental NF with unusual clinical features, such as the present case.

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