Segmental neurofibromatosis with deep schwannoma

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ABSTRACT

An elderly patient presented with two clusters of asymptomatic fleshy and pedunculated papules. Biopsy of the papules was consistent with neurofibromas. Decades prior she had undergone a surgery for the excision of a large schwannoma. Given her lack of other neurofibromatosis findings, the patient was diagnosed with multisegmental neurofibromatosis (multi-SN) with deep schwannoma, a possible new phenotype of SN. Because this entity may be associated with internal malignancy, it is important to screen and educate these patients as well as to provide regular follow-up.

Key words: Malignancy, neurofibroma, neurofibromatosis 1, schwannoma, segmental neurofibromatosis

INTRODUCTION

Segmental neurofibromatosis (SN) is a rare variant of a common autosomal dominant neurocutaneous disorder wherein a postzygotic mutation in the NF1 gene is thought to cause lesions distributed in one area of the body. Unilateral, bilateral, and late-onset cutaneous segmental neurofibromas have been described, as well as isolated deep-seated plexiform neurofibromas. There is also one case of multiple recurring schwannomas in deep and superficial locations, yet there have been no reported cases with deep schwannoma and clinically apparent multisegmental neurofibromas; increased knowledge and awareness of this phenotype is important.[1]

CASE REPORT

A 70-year-old woman presented to dermatology outpatient clinic for evaluation of two clusters of small, fleshy tumors that appeared over the previous three years. Twenty-five years prior, she had undergone a surgery for a deep nerve-based tumor lateral to her left knee. She had no other family members with similar lesions or a personal history of malignancy. On examination, several skin-colored to violaceous, fleshy, pedunculated papules and nodules were clustered over her left anterior shoulder and lateral left leg [Figure 1]. Biopsy confirmed that the cutaneous lesions were neurofibromas (Figure 2, showing non-encapsulated peripheral nerve elements and schwann cells with wire-like collagen fibrils). Given the clustered distribution of lesions and lack of other stereotypic findings of neurofibromatosis (café-au-lait macules, musculoskeletal deformities, axillary freckling, Lisch nodules of the iris), the patient was diagnosed with SN.

DISCUSSION

SN, classified by Riccardi as neurofibromatosis type V (NFV), was traditionally described as café-au-lait spots, freckling, and neurofibromas restricted to a discrete area.[2] However, Roth *et al*. realized that several patients with apparent SN did not meet these criteria and developed four subsets of NFV to better stratify and classify patients: True SN according to Riccardi are localized deep neurofibromas only, hereditary segmental, and bilateral segmental.[3] Recently, Hardin *et al*. reported that SN should not be

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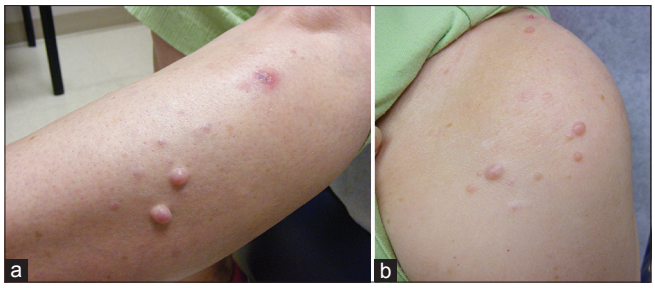

Figure 1: Cluster of fleshy, pedunculated papules on (a) left lateral leg and (b) left anterior shoulder

regarded as a distinct entity from neurofibromatosis I.[4] They believe cases previously referred to as unilateral or bilateral SN are now best referred to as mosaic generalized or mosaic localized neurofibromatosis.[3]

Tanito *et al*. studied characteristics of several NFV patients. They found 55% had pigmentary changes only, 8.6% had neurofibromas only, 22.4% had neurofibromas and pigmentary changes, and 13.8% had plexiform neurofibromas only.[5] The lesions often followed a dermatomal distribution, with the cervical areas being affected most commonly.

SN has been reported to be associated with malignant melanoma, breast, colon, gastric, and lung cancer, affecting 5.3% of patients.[6] As the current prevalence of cancer in the general population is lower, it seems that patients with SN are at an increased risk for certain malignancies. Most patients were diagnosed with cancers after their diagnosis of neurofibromatosis; therefore, a prudent review of systems and general examination for any neoplastic process could be of benefit in these patients.[6]

SN, or NFV, is a rare phakomatosis. To our knowledge, no report of late-onset multi-SN in two different dermatomes with associated deep schwannoma has been reported; this appears to be a new NF phenotype. Given the association of neurofibromatosis and plexiform lesions with various

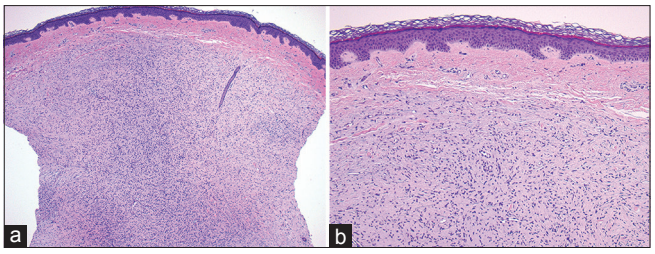

Figure 2: (a and b) H and E staining of tissue consistent with neurofibroma (description of histopath image required) (a: ×4 and b: ×10)

comorbidities, it is important to study the natural history of the disease to better counsel patients on risks, prevention, and family planning.

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Conflicts of interest

There are no conflicts of interest.

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