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CASE REPORT

A 28 year-old male presented for evaluation and excision of multiple papules that had been appearing progressively over the previous 3 years. Physical examination revealed soft, flesh-colored papules and nodules, distributed in dermatomal fashion from the left chest wall anteriorly to the back posteriorly (Figure 1). The lesions did not cross the median line and no axillary freckles or café-au-lait macules were seen. There was no positive family history. The histopathological examination revealed a non-encapsulated, well-circumscribed spindle cell neoplasm, with cells arranged in fascicles (spindle to wavy), as well as buckled nuclei, indistinct cytoplasm and myxoid background, consistent with neurofibromas (Figure 2). The patient was diagnosed with segmental neurofibromatosis (SNF) and examined at the ophtalmology department, ruling out ocular disease. Magnetic resonance imaging showed no bone, central or peripheral nervous system abnormalities.

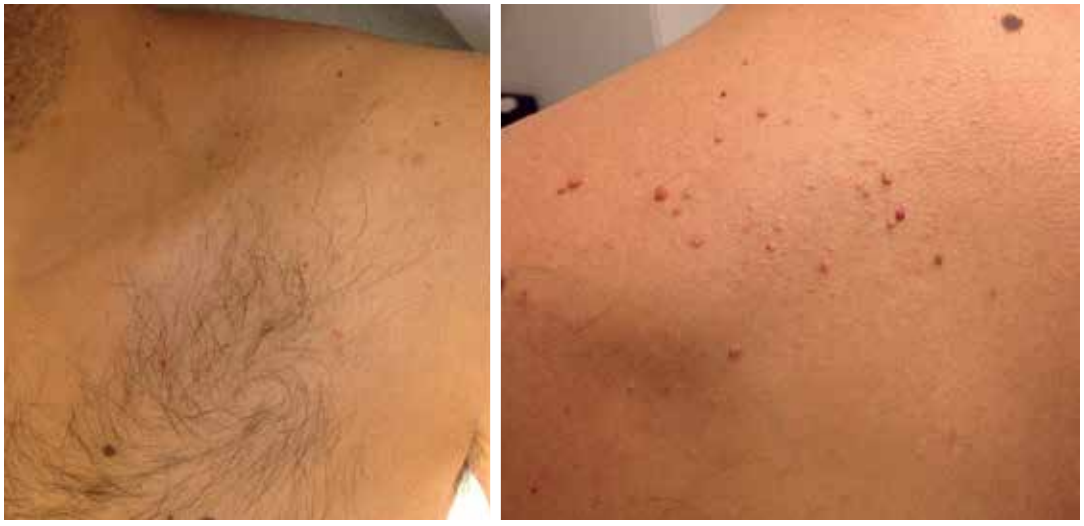

Figure 1: Lesions appearing in dermatomal fashion on the anterior chest wall and back

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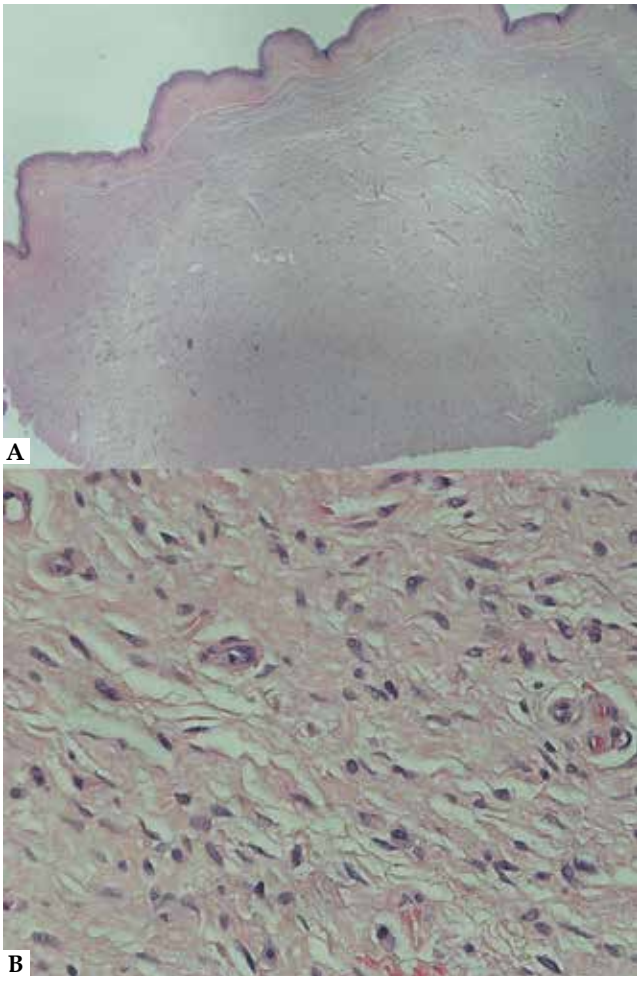

Figure 2: A. Proliferation of dermal, non-encapsulated spindle cells (Hematoxylin-eosin; X2). B. Spindle cells with wavy nuclei, interspersed in a stroma of fibrillary collagen. No nuclear pleomorphism or mitoses were seen (Hematoxylin-eosin; X40)

DISCUSSION

Neurofibromatosis (NF) is extremely variable in its presentation. Segmental neurofibromatosis (SNF) is a rare disorder (prevalence between 0.0014 and 0.002%) characterized by café-au-lait spots and/or neurofibromas in a single, unilateral segment of the body. It does not entail crossing of the median line, systemic involvement, or family history.[1,2] Furthermore, it may go unrecognized if few lesions are observed.

First described in 1956, SNF was initially termed sectorial NF. In 1982, Riccardi included SNF in his NF classification to represent NF type 5.[3,4] It is thought to arise from a postzygotic NF type 1 gene mutation located on the proximal long arm of chromosome 17, and may occur in both somatic and gonadal cell lines. Patients whose manifestations are limited to neurofibromas are believed to carry the mutation in Schwann cells (explaining its dermatomal distribution). If only pigmentary manifestations are present, the mutation occurs in the fibroblasts (explaining the blaschcoid distribution). Gonadal mosaicism is thought to be responsible for localized disease in patients whose children have generalized NF type 1.[5] Clinically, it presents solely with pigmentary changes or neurofibromas, with both pigmentary changes and neurofibromas, also with both pigmentary changes and neurofibromas, and with isolated plexiform neurofibromas. Most commonly, patients only present with mainly cervical or thoracic neurofibromas.[3] In rarer cases, the face may be the only affected site.[6] Systemic complications typically associated with NF type 1 are rare (under 10% of cases) and include: learning difficulties, plexiform neurofibromas, optic pathway gliomas and pseudarthrosis.[1] It has been proposed that the risk of malignancy in SNF may be comparable to NF type1 and therefore, age-appropriate malignancy screening is indicated. Individuals with SNF have a low risk of transmitting the disease to their offspring, although genetic counseling and evaluation of offspring for NF skin lesions and cognitive impairment are advisable.[5] SNF may be underreported since it is almost asymptomatic and patients only seek medical attention for cosmetic concerns. Physicians must be vigilant and able to recognize and manage the condition.❑

Abstract: Neurofibromatosis is extremely variable in its presentation. Segmental neurofibromatosis (SNF), which corresponds to NF-type 5 in the Riccardi classification, is a rare disorder. It may go unrecognized if few lesions are observed. We present a case of segmental neurofibromatosis in a 28 year-old patient who presented with multiple papules and nodules distributed in dermatomal fashion on the trunk. The histopathological examination of the lesions revealed a non-encapsulated, well-circumscribed spindle cell neoplasm, which was consistent with neurofibromas.

Keywords: Neurofibroma; Neurofibromatosis; segmentar neurofibromatosis

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