Original Article

A Sporadic Association Of Naevus Spilus And Neurofibromatosis (Type 5) - A Case Report

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Abstract

Nevus spilus, also known as speckled lentiginous nevus or nevus on nevus, are congenital nevus with uniformly coloured hyper-pigmented patchy background, superimposed with multiple dark pigmented spots. Two different types of nevus spilus have been reported in the literature, namely nevus spilus maculosa and nevus spilus papulosa. Our patient along with naevus spilus had segmental café-au-lait macules and neurofibroma affecting more than one dermatome which according to Riccardi's classification, comes under NF-type5. There are reports of the association of nevus spilus with neurofibromatosis 1 in literature . But there are no reports of association of nevus spilus and segmental NF in the same patient.

Case report

A 31-year-old male patient came to the dermatology department for an opinion before his orthopaedic surgery for trauma over his left foot. The patient reported a large pigmented lesion over the right side of the trunk and right arm present since birth. The lesions were progressively increasing in size, with more number of superimposed multiple dark spots. No other focal or systemic complaints. No similar lesions in any family members.

On examination, a large hyper-pigmented patch was seen over the right side of the abdomen, chest, back and right arm. Multiple hyperpigmented macules and a few hyperpigmented papules were seen over the patch. Around 20-22 hyperpigmented nodules involving the back and arm were seen. Multiple café-au-lait macules were also seen over the left side of the back. Bilateral palmar frecklings were also present. An ophthalmological examination was done to look for lisch nodules and were not detected. The skeletal survey and MRI brain were normal. An excisional biopsy was done from one of the nodules over the right arm. Differential diagnoses of a nodular variant of nevus spilus and neurofibroma were considered. Histopathology examination showed normal epidermis, presence of bland spindle-shaped cells arranged as diffuse sheets in the dermis. The stroma appeared oedematous. Cells showed wavy to plump nuclei. Thickened collagen bundles were seen. Immunohistochemistry with S100 was positive which suggests the cell of origin was melanocytes or Schwann cells. To further differentiate, we did HMB-45 and it was negative. Thus narrowing down the cell of origin as the Schwann cell. The patient was diagnosed to have giant Naevus spilus with a co-existing segmental neurofibromatosis (NF type V) based on the clinical and histopathological findings. As the lesion was present since birth and as he was not concerned about the lesion at present no specific treatment was given. Counselling played an important part in our case as patients with giant nevus spilus (macular variant) as well as segmental NF are at an increased risk of malignant melanoma and malignant peripheral nerve sheath tumour. Hence we educated our patient about regular skin examination for

early detection of any malignancy. The patient was also informed that they do not have neurofibromatosis type-1 and their risk of disease-associated complications is low.

Discussion

Nevus spilus, also known as speckled lentiginous nevus or nevus on nevus or zosteriform lentiginous nevus, are congenital nevus with uniformly coloured hyper-pigmented patchy background, superimposed with multiple dark pigmented spots. Recently, activating mutations in HRAS were detected in all eight cases of nevus spilus that were examined [1]. Two variants of nevus spilus are known [2]. Nevus spilus maculosa i.e. macules over a hyperpigmented patch giving a polka dot appearance. Nevus spilus papulosa or Papular variant i.e. papules or nodules are scattered unevenly over a hyperpigmented background. In addition, hyperpigmented macules may be present. Literature review showed association of nevus spilus with speckled lentiginous nevus syndrome (ipsilateral dysesthesia, muscular weakness or hyperhidrosis) phakomatosis pigmentovascularis, phacomatosis pigmentokeratotica, neurofibromatosis 1 ,Vitiligo Vulgaris, melanoma [3][4][5]. In addition to nevus spilus, our patient had segmental café-au-lait macules and neurofibroma affecting more than one dermatome which according to Riccardi's classification, comes under NF-type5 [6]. The most common manifestation of SN is neurofibroma, and less frequently, café au lait spots. Lesions are generally unilateral (approximately 6% of cases are bilateral). There are reports of the association of nevus spilus with neurofibromatosis 1 in literature [7]. But there are no reports of association of nevus spilus and segmental NF in the same patient.

Conclusion

Nevus spilus is an entity that requires proper diagnosis and investigations to rule out other associated syndromes and malignancy. This case is reported for the following specific findings - Giant nature of nevus spilus, association with segmental NF at the same site. Segmental NF in more than one dermatome.

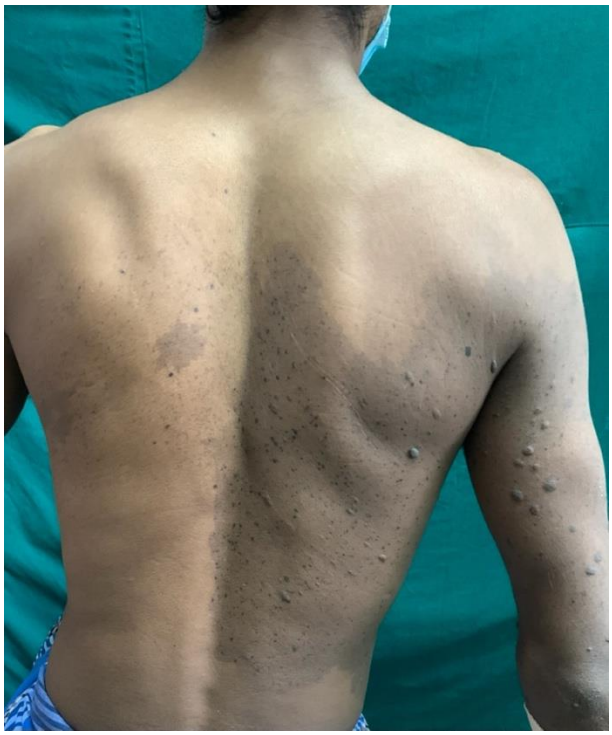

Figure 1b Clinical photograph showing a giant naevus spilus, café au lait and multiple neurofibromas over the right side of the back and posterior aspect of the arm.

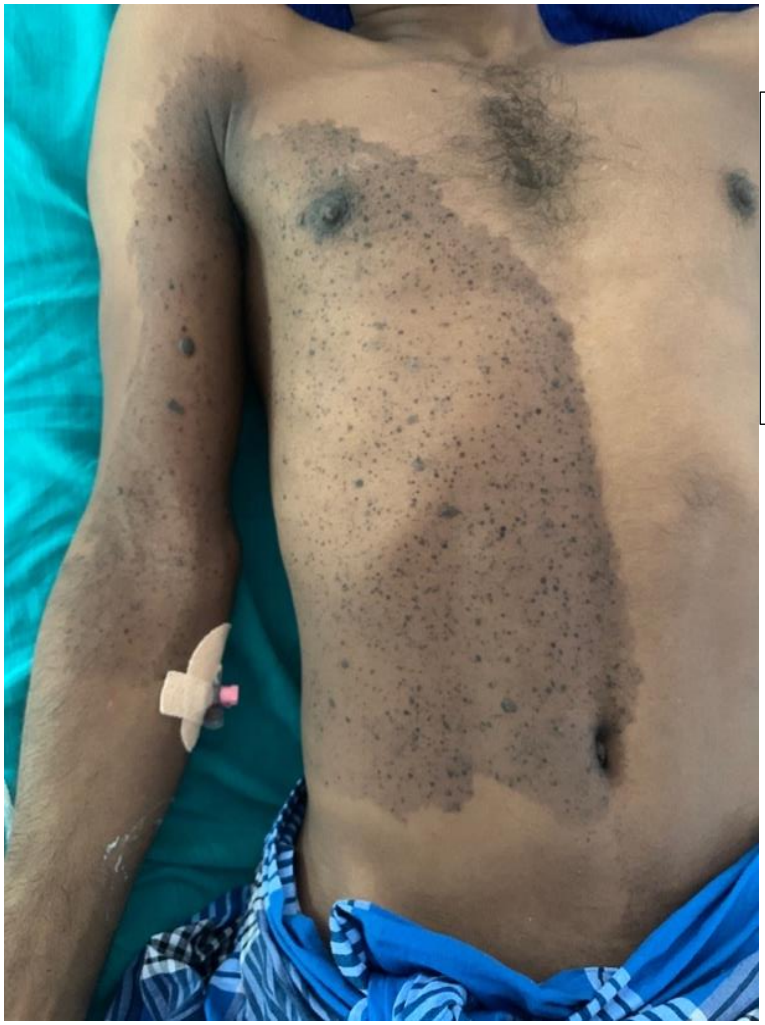

Figure 1b- clinical photograph showing giant hyperpigmented patch with superimposed hyperpigmented macules, papules and nodules, representing giant

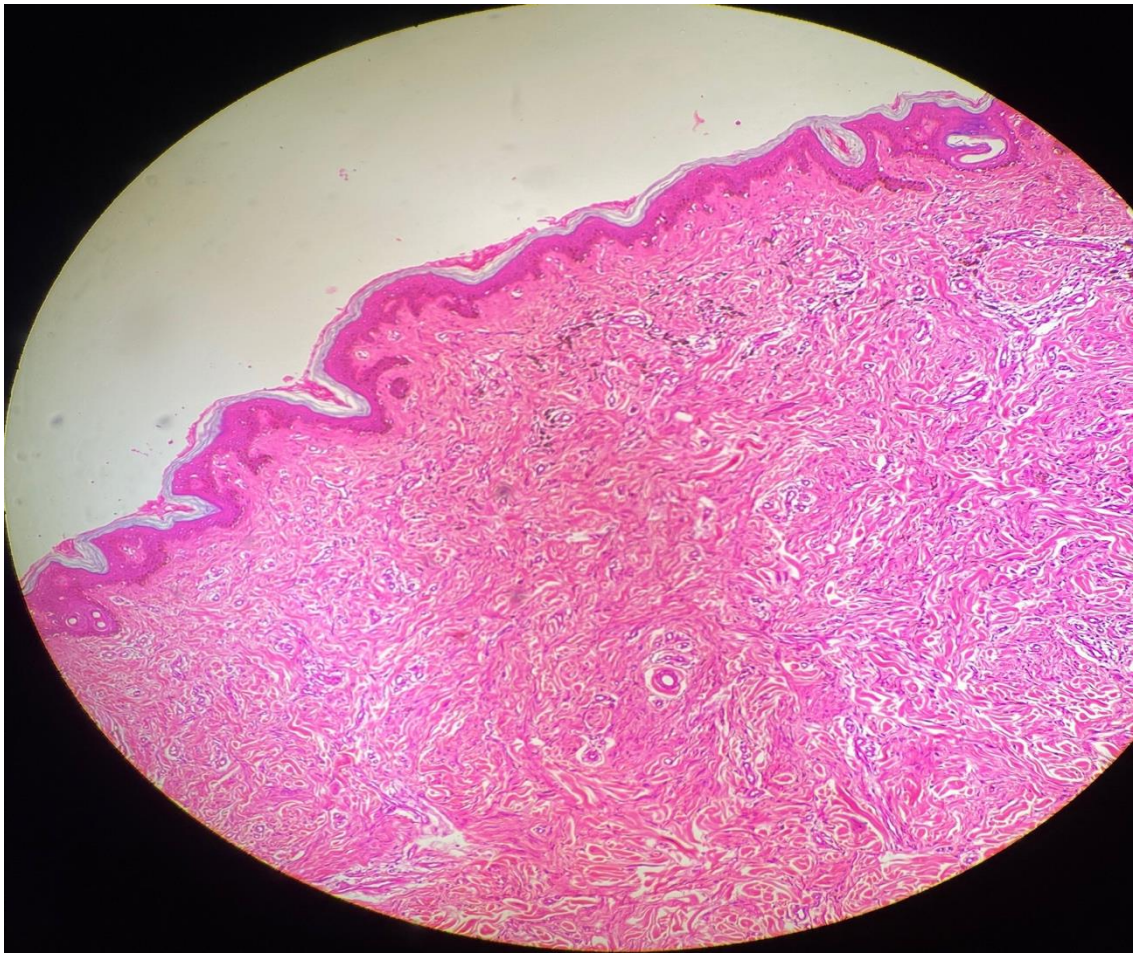

Figure 2 HPE: H&E 400X- The dermis composed of bland spindle shaped cells arranged as diffuse sheets. The cells show wavy to plump nuclei

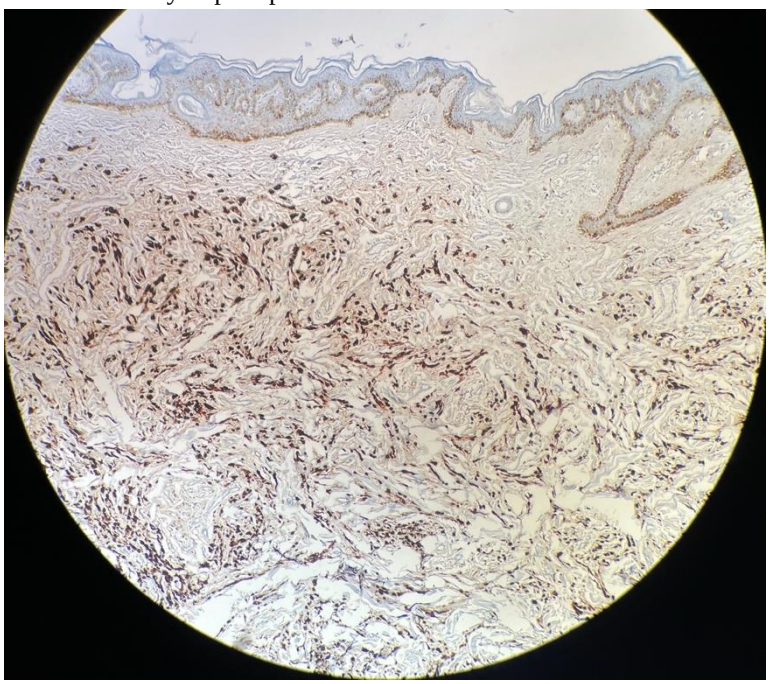

Figure 3A IHC- S100 stain. 40X magnification. S100 positivity which suggests the origin as melanocytes or Schwann cells

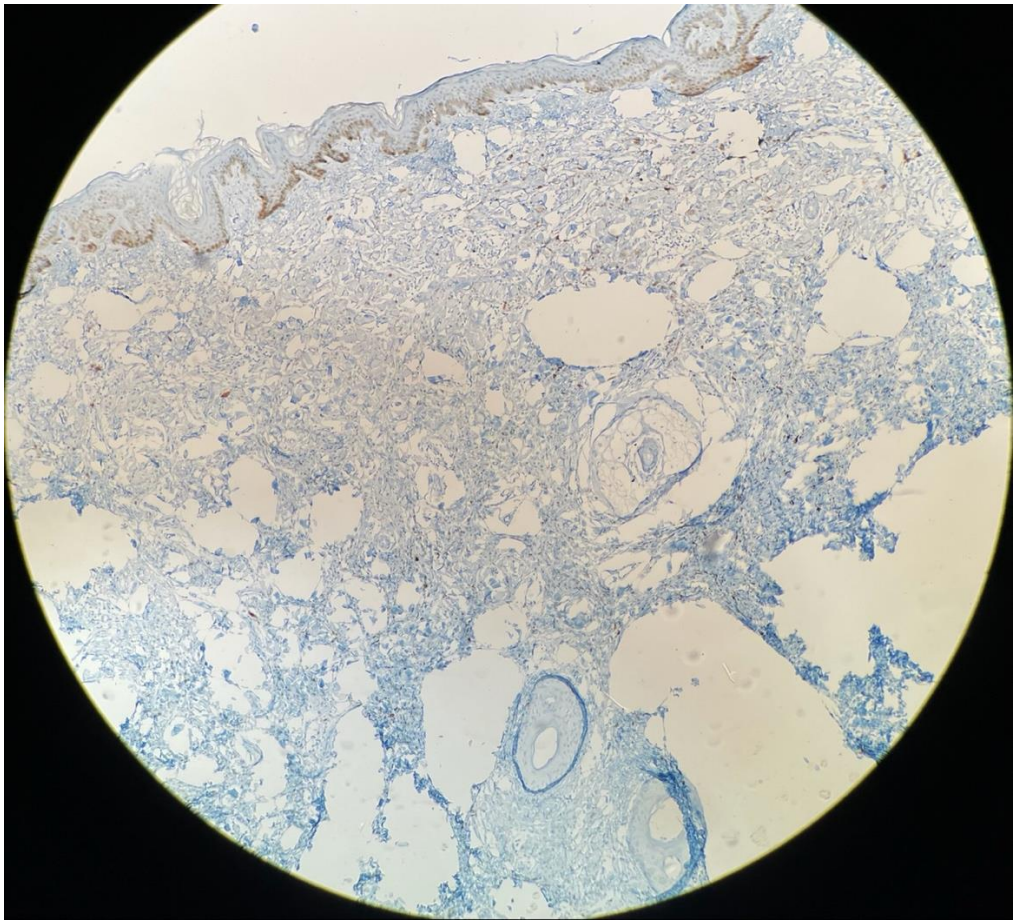

Figure 3B IHC – HMB 45 stain. 40X magnification. HMB-45 was negative suggesting the cell of origin as schwann cells

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