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Dermatology

A CASE OF PIGMENTED NEUROFIBROMA WITH HYPERTRICHOSIS

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ABSTRACT This study explores the uncommon overlap of neurofibromas and hypertrichosis. Neurofibromas, benign tumors from nerve tissue, occasionally exhibit melanin-rich pigmented cells, a rarity in clinical cases. Hypertrichosis, marked by excessive hair growth in non-androgen-dependent regions, is another central theme. The coexistence of these conditions is rarely seen in clinical practice. We detail an unusual case of a 22-year-old male with hyperpigmented macules, nodules, and hypertrichosis on his trunk. While signs hinted at neurofibromatosis type 1 (NF1), the lack of standard criteria complicated diagnosis. Key observations included Lisch nodules, absence of typical neurofibromas, and no family history of similar skin conditions. MRI scans negated malignancies. The patient's examination revealed extensive terminal hair growth and subcutaneous anomalies. The discussion sheds light on hypertrichosis, its potential origins, and its link with neurofibromas, especially around the periorbital area. The study emphasizes the importance of thorough clinical evaluations for unconventional tumor presentations and delves into the mechanisms behind hypertrichosis. Conclusively, the research points to emerging treatments for hypertrichosis, with lasers and chemical agents leading the way.

KEYWORDS : Neurofibromatosis, Neurofibroma with Hypertrichosis

INTRODUCTION

Neurofibromas are benign tumors that arise from the nerve tissue and often present with characteristic clinical features. Less than 1% of all occurrences of neurofibromas have melanin-rich pigmented cells.[1] The cause of this malignancy is poorly understood. Hypertrichosis is a medical disorder marked by excessive hair growth on non-androgen-dependent areas of the body.[2] The association of pigmented neurofibromas and hypertrichosis is not frequently encountered in clinical practice.

This article describes a peculiar case of a 22-year-old male who exhibited an unusual presentation of hyperpigmented macules and nodules with hypertrichosis primarily on his trunk. While the clinical manifestations raised suspicions of neurofibromatosis type 1 (NF1), the absence of its typical criteria posed a diagnostic challenge.

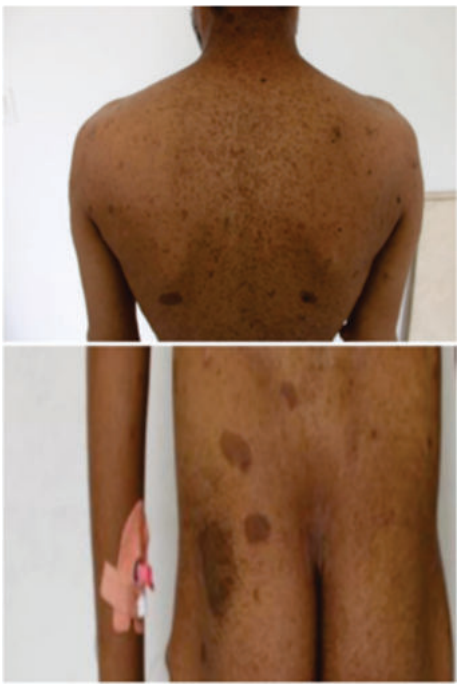

Fig. 1 Café-au-lait Macules With Overlying Tuft Of Hair Seen Over The Back And Buttocks

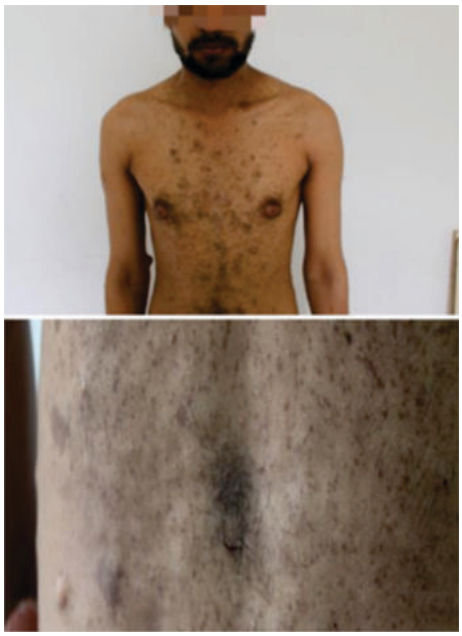

Fig. 2 Multiple Pigmented Neurofibroma With Tuft Of Hair Seen On The Trunk

CASE STUDY

A 22-year-old male displayed abnormal hyperpigmented macules and nodules on many areas of his upper body since 8 years of age (Fig. 1). On examination, the nodules were soft and rubbery in consistency, with no tenderness and were covered with numerous terminal hairs (Fig. 2). Lisch nodules were found on slit lamp examination, the patient's growth and development were normal, and there was no family history of similar skin lesions. No other clinical features fulfilling the NF1 criteria were observed, including typical neurofibromas, axillary or inguinal freckling. To better understand the extent of soft tissue involvement, a magnetic resonance imaging was performed; however, there were no indications of the tumour or other malignancies extending into the surrounding tissue. During examination, the patient's body was covered with numerous ill-defined regions of extra terminal hair growth that ranged in size from 0.2 cm x 0.2 cm to 5 cm x 6 cm. On closer inspection, beneath the area of additional hair, was a palpable, soft, non-tender fullness of the

subcutaneous tissue. In conclusion, this unique case highlights the importance of thorough clinical examination and the potential for unusual presentations of benign tumors. While further investigation is necessary, the absence of malignancy suggests a favorable outcome for this patient.

DISCUSSION

Hypertrichosis refers to the excessive hair growth on areas not typically affected by androgens of the body. This condition is typically classified based on its onset, which can either be congenital or acquired, as well as its distribution, which can be either localised/circumscribed or generalised. In prepubertal children, hypertrichosis may occur alone or in combination with other congenital abnormalities.[3] Restricted hypertrichosis can be caused by a plexiform or diffuse neurofibroma, which is not often described in patients. Although NF-1 is a congenital illness, hypertrichosis was not apparent in our patient until the age of 18. A clinician can diagnose localized hypertrichosis by examining a patient who has complained of it. A soft subcutaneous mass, with or without subcutaneous hyperpigmentation, was palpable in our patient. The most common location for diffuse and plexiform neurofibromas associated with localised hypertrichosis is the periorbital area.[1,4] Clinicians were able to diagnose neurofibromatosis in one case because hypertrichosis was the initial presenting symptom.[1] Areas of friction, irritation, or trauma are frequently present in patients with hypertrichosis, suggesting that the same underlying mechanism may be at play. Increased regional blood flow has been proposed by some specialists as a potential contributing element.[5] Congenital localised hypertrichosis in hamartomas or underlying neurological conditions are two more potential processes that could result in hypertrichosis. Complex interactions between the epithelial and mesenchymal components of a hair follicle during morphogenesis dictate the fate of the cell, and these signals are repeated throughout each hair follicle cycle. The signals involved in the hair follicle cycle have been the subject of several recent research, which may help us understand the mechanisms behind hypertrichosis. The anagen, or developing hair follicle, has been shown to be influenced by growth factors like bone morphogenic protein, platelet-derived growth factor, and bone epidermal growth factor.[6] Regional variables, such as surface injury or inflammation, may have an impact on these growth mediators and lengthen the anagen phase. The size of the hair follicle is also affected by differences in the volume and cell count of the follicular papilla, the Mesenchymal component of the hair.[7] This process may cause hypertrichosis because the follicular papilla may be impacted by congenital anomalies, regional circumstances, or paracrine causes. Recent investigations have shown that the mesenchyme and connective tissue sheath that surround the hair follicle are extraordinarily flexible, causing important alterations in the hair follicle.[7] Localised hypertrichosis may result from abnormal signaling for follicular papilla expansion, prolonged anagen phase, or both. Although there are some limitations, the range of treatments for hypertrichosis is expanding. Currently , the most common methods include lasers and chemical hair-removal agents.[7,8]

CONCLUSION

The intricate relationship between pigmented neurofibromas and hypertrichosis remains an area of investigation in dermatology. This case study of a 22-year-old male, presenting with hyperpigmented macules, nodules, and hypertrichosis, underscores the diagnostic challenges posed by atypical clinical manifestations. While the patient's presentation bore semblance to neurofibromatosis type 1 (NF1), the absence of its hallmark criteria necessitated a more nuanced approach to diagnosis. The study highlights the importance of a comprehensive clinical examination, especially when confronted with unconventional tumor presentations.

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