

Innumerable nevi with giant congenital melanocytic nevus clinically mimicking neurofibromatosis: A diagnostic challenge

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INTRODUCTION

Prior case studies have documented the combined phenotype of congenital melanocytic nevi with neurofibroma (NF)-like lesions.¹⁻⁴ This case is the first description of extensive nevi, occurring in the setting of a giant congenital melanocytic nevus (GCMN), clinically resembling neurofibromatosis.

CASE REPORT

A 31-year-old half Thai, half white woman presented to the dermatology clinic for evaluation of multiple disfiguring pigmented papules and nodules covering her entire body. Her dermatologic history was significant for a GCMN treated with curettage at age 2. The patient reported that she was born with many pigmented papules and nodules, which increased in number throughout her life. She was otherwise healthy with no cognitive deficits, seizures, evidence of neurocutaneous melanosis, or skeletal malformations. There was no family history of a similar skin disorder or neurofibromatosis.

Physical examination found a large plaque with residual skin-colored and pigmented nodules involving her left back (crossing midline), posterior neck, and left anterior chest. She had evidence of scar and residual pigmented patches and nodules consistent with regrowth after curettage of the GCMN, which covered an area with estimated greatest diameter of more than 60 cm. The patient also had greater than 1000 well-circumscribed, dome-shaped papules and exophytic, pedunculated nodules

Abbreviations used:

GCMN: giant congenital melanocytic nevus
NF: neurofibroma

diffusely throughout the body, which ranged from 0.5 to 2 cm in diameter and were skin-colored to dark brown. Clinically, many of these lesions resembled NFs (Fig 1). Examination was negative for café-au-lait spots, axillary or groin freckling, ocular abnormalities including Lisch nodules, skeletal abnormalities, and any other abnormalities. No ophthalmologists or neurologists were consulted at this time. Hematoxylin and eosin staining of a representative shave biopsy from the right thumb revealed a melanocytic nevus, with clear nesting, melanin production, and a well-circumscribed proliferation of chiefly epithelioid nevomelanocytes within the dermis (Fig 2). The cells showed maturation with descent and some neurotization in the deeper extent. Comparative genomic hybridization found no gains or losses in chromosomal material.

DISCUSSION

In this case, there was clinical concern of neurofibromatosis because of the sheer number of the lesions and the involvement of unusual anatomic sites, such as the palms and soles. The distinction between heavily neurotized melanocytic nevi and NFs, both benign cutaneous

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Fig 1. Presentation of melanocytic nevi on (A) plantar feet and (B) dorsal hands.

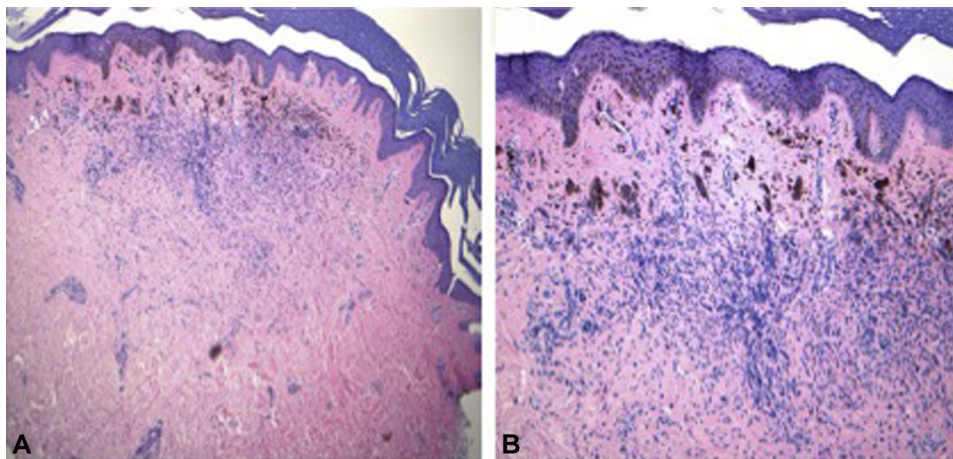


Fig 2. Melanocytic nevi biopsy from the right thumb. Panel B is a magnified view of Panel A. Note the well-circumscribed proliferation of mostly epithelioid nevomelanocytes. (Hematoxylin and eosin stain.)

neoplasms originating from neural crest–derived stem cells, can be difficult.^{5,6} Neurotization pertains only to dermal melanocytes and describes evolution of oval-shaped type A and B nevus cells with cytoplasmic melanin into type C cells that lack melanin and resemble neuroid structures of the dermis.^{5,6} In comparison, histologic examination of an NF finds a dermal proliferation of spindle-shaped cells with wavy nuclei in a background of loose reticulated collagen.⁵ In this case, the biopsy found some neurotization in the deeper extent, but the bulk of the samplings showed epithelioid cells with clear and copious melanin production. NFs were not a histologic concern.

Extensive nondysplastic melanocytic nevi and GCMN independently confer increased risk for melanoma.^{7,8} The relative risk of melanoma in patients with greater than 100 nondysplastic nevi is estimated at 9.8 ($P = .001$).⁷ Malignant

transformation of GCMN increases with greater size (diameter of 40 to 60 cm), numerous satellite nevi (>20), and truncal location (2.9% on trunk vs 0.3% on head or limb).^{9,10} GCMN removal with surgical excision to superficial fat, dermabrasion, laser therapy, curettage, and shave excision do not eliminate the risk of malignant transformation.¹⁰ The patient was counseled regarding the importance of close surveillance for melanoma. A plan was initiated to remove the disabling lesions from her palms and soles. However, the patient's family was relocated for work and she was lost to follow-up. When lesions clinically resemble NFs, pathologic examination is paramount. Prognostic implications of our patient's multiple nevi are not understood. Close monitoring of disease progression is critical and, unfortunately, difficult in patients with poor understanding of their diagnostic challenge.

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