A Case of a Solitary Segmental Neurofibroma in a 31-year-old Woman

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Background

The presence of cutaneous neurofibromas are the hallmark finding in neurofibromatosis type 1 (NF1). However, cutaneous neurofibromas are common in individuals without the disorder.

The etiology of neurofibromas is largely unknown, but the presence of a cutaneous neurofibroma alone is not associated with any complications or malignancies. Though many are asymptomatic, cutaneous neurofibromas may cause pain or discomfort and may lead to cosmetic concerns. A patient may be recommended to have surgical excision if experiencing any discomfort or other concerns.

In contrast, NF1 is an autosomal dominant genetic disorder associated with a number or complications and malignancies such as optic pathway gliomas, pseudoarthrosis, hypertension, and tumors within the central nervous system and/or gastrointestinal system.

Distinguishing between a solitary neurofibroma and NF1 is important to providing appropriate surveillance and care.

Objective

To better differentiate the diagnosis of solitary segmental neurofibroma vs NF1, which is associated with internal malignancies and would require additional screening.

Methods and Case Presentation

A 31-year-old woman without significant past medical history presented to the dermatology clinic for evaluation of multiple skin-colored and pink papules that coalesce into a plaque on her right lower back.

The patient states she first noticed this plaque five years prior, following a motor vehicle accident. She noted the plaque as asymptomatic, however was slightly enlarging over the past five years. The patient finally sought dermatologic care as she was under the impression the plaque on her back were keloids and she was looking for further evaluation and management. She otherwise had no personal history of melanoma or keratinocyte carcinomas. She did report a family history positive for basal cell carcinoma in her father. Review of systems was unremarkable.

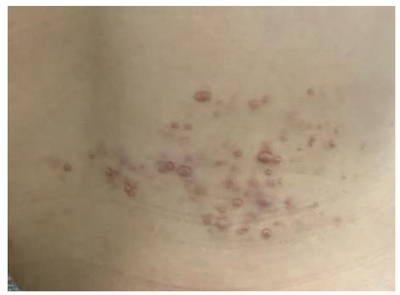

Figure 1. Right superior medial midback lesion comprised of multiple skin-colored and pink papules which coalesce into a plaque.

Physical examination demonstrated multiple small 4 mm skin colored to pink papules coalescing into a plaque on the right lower back. The papules were soft, rubbery, and demonstrated a buttonhole sign on palpation. No other pertinent cutaneous or ocular findings noted.

Results

A tangential shave biopsy was performed. Hematoxylin and eosin (H&E) stain revealed a well-circumscribed dermal nodule composed of delicate wavy fibrils of neural origin. Surrounding this dermal proliferation of benign neural components, there were elongated fibroblasts and a mucinous stroma. The findings depicted on H&E were consistent with a benign neurofibroma.

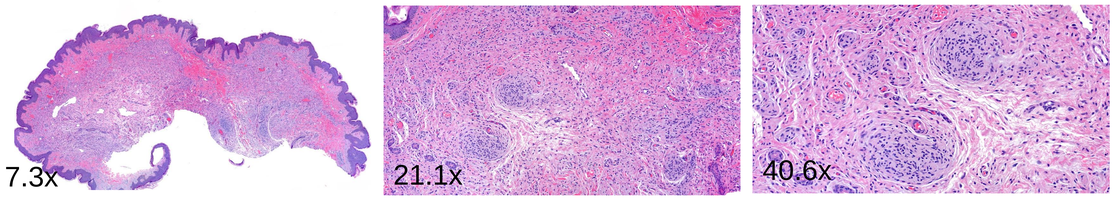

Figure 2. H&E staining of biopsy tissue.

Conclusions

When combining the clinical features of several neurofibromas coalescing into a plaque in a dermatomal distribution, the dermatologist was able to render a diagnosis of solitary segmental neurofibroma. Given this unique presentation, it was critical to ensure that this was not additionally a presentation of segmental neurofibromatosis type 1 (NF1). After reviewing both major and minor criteria of NF1 with the patient, there were no additional findings to confirm a diagnosis of NF1. It is important to be able to differentiate a solitary segmental neurofibroma from NF1, as there are important internal organ malignancies and additional medical screening that must accompany a diagnosis of the latter.

Diagnostic Criteria for Neurofibromatosis Type 1
CATEGORY 1
• ≥6 café-au-lait macules over 5 mm in greatest diameter in pre-pubertal individuals, or over 15 mm in greatest diameter in post-pubertal individuals
• Axillary or inguinal freckling
• ≥2 neurofibromas or 1 plexiform neurofibroma
• Optic pathway glioma
• ≥2 iris Lisch nodules or ≥2 choroidal abnormalities
• A distinctive osseous lesion such as sphenoid dysplasia, anterolateral bowing of the tibia, or pseudarthrosis of a long bone
• Presence of a heterozygous pathogenic NF1 variant in apparently normal tissue
CATEGORY 2
• A child of a parent who meets the diagnostic criteria specified in category 1

Figure 3. A diagnosis of NF1 requires at least 2 features from category 1, or a single feature from category 2.

Disclosures

The authors have no financial relationships to disclosure.

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