



Segmental distribution of nodules on trunk

The lesions on this patient's trunk had been growing in size—and number—over the past 42 years. The pattern of these lesions proved helpful in making the diagnosis.

A 70-YEAR-OLD CAUCASIAN MAN presented with a longstanding history of numerous nontender, fleshy, skin-colored papules on his trunk, ranging from 3 to 8 mm in size (FIGURE). They were noted incidentally during an examination of unrelated nonhealing lesions on the patient's left cheek. He said the lesions on his trunk first appeared when he was 28 years old and had continued to grow in size and number. The patient said his son had

at least one similar lesion on his upper back, but otherwise there was no family history of these lesions.

A biopsy was performed on one of the nodules.

- WHAT IS YOUR DIAGNOSIS?
- HOW WOULD YOU TREAT THIS PATIENT?

FIGURE

Papules on trunk



The patient had numerous nontender, fleshy, skin-colored papules on his trunk.

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TABLE

Differential diagnosis for dermatomally distributed nodules⁴

Neoplasms	
Benign	Syringocystadenoma papilliferum, trichoepithelioma, cutaneous schwannoma
Malignant	Basal cell carcinoma, cutaneous metastases, lymphoma, plasmacytoma, squamous cell carcinoma
Other mucocutaneous conditions	Granuloma annulare, neurofibromatosis type 1, pseudolymphoma, rheumatoid nodules, sarcoidosis, xanthomas

Adapted with permission from: Hager CM, Cohen PR, Tschen JA. Segmental neurofibromatosis: case reports and review. *J Am Acad Dermatol.* 1997;37:864-869.

Diagnosis:
Segmental neurofibromatosis

Dermatopathologic evaluation of the tissue sample indicated that the lesion was a neurofibroma, and clinical correlation fine-tuned the diagnosis to segmental neurofibromatosis (NF). The diagnosis of segmental NF is clinical with biopsy to confirm the lesions are neurofibromas. Segmental NF is a mosaic form of neurofibromatosis type 1 (NF1) that results from a postzygotic mutation of the NF1 gene. While NF1 is a relatively common neurocutaneous disorder that occurs with a frequency of one in 3000,¹ segmental NF is more rare, with an estimated prevalence of one in 40,000.²

NF1 often follows an autosomal dominant inheritance pattern, although up to 50% of patients with NF1 arise de novo from spontaneous mutations.³ NF1 is characterized by multiple café-au-lait macules, axillary freckling, neurofibromas, and Lisch nodules (pigmented iris hamartomas).

Systemic findings that are associated with NF1 include malignant peripheral nerve sheath tumors, optic gliomas, and vasculopathy.³ While patients with segmental NF may exhibit some of these same findings, the distribution of neurofibromas is often limited to one dermatome. Additionally, patients with segmental NF typically do not exhibit extracutaneous lesions, systemic involvement, or a family history of NF.

Rule out these dermatomal lesions

This case highlights a unique pattern of neoplasm development along a dermatome, an area of skin where innervation derives from

a single spinal nerve. Symptoms that follow a dermatome often point to a pathology involving the related nerve root.

■ **This differs from Blaschko lines**, which form a specific surface pattern that is believed to reflect the migration of embryonic skin cells. Blaschko lines do not follow any known vascular, nervous, or lymphatic structures of the skin. Interestingly, when patients with segmental NF have associated pigmentary lesions, such as café-au-lait macules, these lesions may border Blaschko lines.

■ **Herpes zoster**, also known as shingles, is the most common infectious process that presents in a dermatomal pattern. Herpes zoster is caused by reactivation of the varicella-zoster virus, which lies within the dorsal root ganglion of a spinal nerve. This condition commonly results in a dermatomal distribution of vesicles/bullae on an erythematous base.

■ **Neoplasms**—including common cutaneous malignancies, such as basal cell carcinoma, as well as rare benign cutaneous conditions, such as cutaneous schwannoma, may have a distribution similar to that of segmental NF. A biopsy can help distinguish the diagnosis. See the **TABLE**⁴ for a complete differential diagnosis for dermatomally distributed nodules.

Classifying neurofibromatosis

It's important to classify the type of NF in order to get a better handle on the patient's prognosis and to facilitate genetic counseling. In particular, the much more common NF1 comes with an increased risk of systemic findings such as malignant peripheral nerve sheath tumors, optic gliomas, other gliomas, and leukemia. Few patients with segmental NF, on the other hand,

➤ The distribution of neurofibromas with segmental NF is usually limited to one dermatome.

will have these systemic findings.⁴ Segmental NF treatment typically focuses on symptomatic management or cosmetic concerns.

■ **Our patient** did not have any of the systemic complications that occasionally occur with segmental NF as discussed above, so no medical treatment was required. We informed him that the cutaneous and subcutaneous neurofibromas do not require removal unless there is pain, bleeding, disfigurement, or signs of malignant transformation. Our patient was not interested in removal of the nodules for cosmetic reasons, so we recommended follow-up as needed. **JFP**

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