Segmental Neurofibromatosis: Isolated Eruption of Neurofibromas on the Scapula in a Middle-Aged Man

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Introduction

Neurofibromatosis is an autosomal dominant inherited disorder that results from a mutation of the *neurofibromin* protein, a negative regulator of the RAS protein, an all-important protein for cell-cycle regulation. Composed of five clinically distinct subtypes, neurofibromatosis has often been associated with either neurofibromatosis type 1 or 2, two of the more common tumor-prone inherited diseases. Our case presents a patient with segmental neurofibromatosis, also known as neurofibromatosis type V, a rare clinical variant of the neurofibromatosis spectrum with a reported prevalence of approximately 0.0014%-0.002%, often presenting as an amalgam of features both clinically and histologically.

Case Presentation

HPI: A 63-year-old Caucasian male was referred to the clinic for the evaluation and management of multiple skin tags that first presented in childhood. Certain lesions were easily irritated by clothing and the patient wanted them removed.

PMHx: Though the patient initially denied any relevant personal or family history of dermatologic disease, specific questioning revealed his siblings had similar findings of varying degrees.

Physical Exam: Physical examination was relevant for a grouped collection of pedunculated skin-colored papules overlying the patient's left scapula (Figure 1). Lesions were soft and non-tender to palpation. Two shave biopsies were performed to confirm the diagnosis. A referral to a geneticist was offered but deferred by the patient, as he has no plans to conceive. Evaluation by ophthalmology was also recommended but declined, citing no active visual complaints.

Diagnosis: Two shave biopsies were performed to confirm the diagnosis

Discussion

Segmental neurofibromatosis (SNF), has a prevalence of approximately 0.0014%-0.002% and may present with characteristics of type 1, type 2, or mixed neurofibromatosis.[1]

SNF is commonly seen in middle-aged women, and rarely has symptoms or associated family history. Genetic mosaicism via postzygotic mutations in the *NF1* gene on chromosome 17 is responsible for disease.[1]

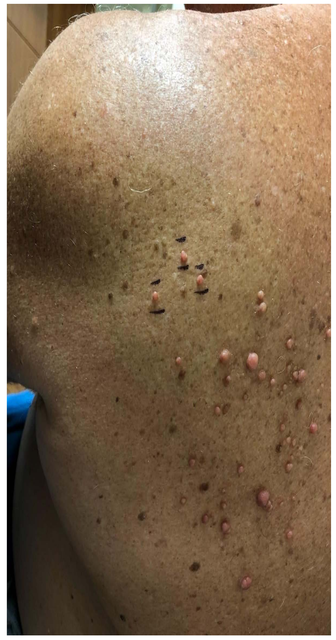
Histopathology reveals well-circumscribed, non-encapsulated aggregations of spindle cells with wavy, "s-shaped" nuclei arranged in a whorled-pattern. Immunohistochemistry identifies S100+ Schwann cells, CD34+ fibroblasts, and EMA+ perineural cells.[2]

Treatment options for SNF are limited. Surgical excision is the gold-standard for cutaneous manifestations. Clinical trials with a variety of pharmacologic agents are ongoing, with promising reports emerging for mTOR and kinase inhibitors.[3] As 3-13% of patients with SNF develop peripheral nerve sheath tumors, routine observation of these patients is recommended.[4]

Referral to an ophthalmologist is paramount, as NF1 has a high association with ocular hamartomas. NF1 is among the most common tumor-prone autosomal dominant diseases, as such, genetic counseling should be offered and should extend to instances of egg or sperm donor pregnancies where gonosomal mosaicism in donor cells has been reported to result in offspring affected by NF1.[5]

Figures

Figure 1: grouped, skin-colored papules on the left back.



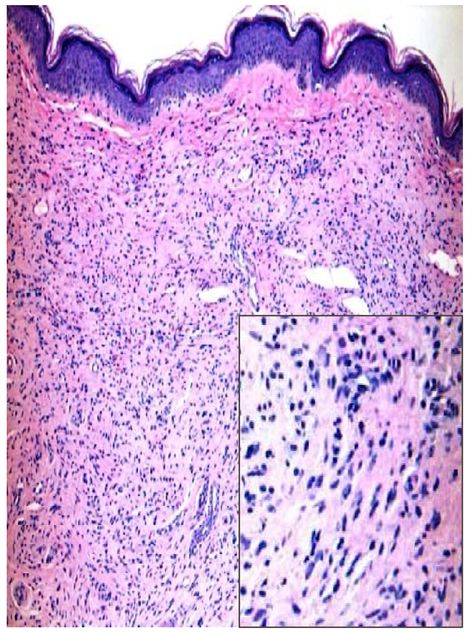


Figure 2: characteristic well-circumscribed aggregate of spindled neural cells in a whorled pattern (inset)

Conclusion

Segmental neurofibromatosis is an uncommon clinical variant which may have predominant features of type 1 or type 2 disease. Once the diagnosis has been confirmed, referral for ophthalmologic and genetic assessment is recommended. Routine dermatologic evaluation for evolution of cutaneous symptoms is warranted, and enlarging or symptomatic lesions should prompt imaging and surgical referral. While no treatment of choice exists, multiple systemic kinase inhibitors have shown variable success and may be considered.

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