



## 2021 Annual Spring Virtual Meeting | Abstract Submission

### How a Routine Skin Check Led to a Diagnosis of Reed syndrome

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A 69-year-old Caucasian male presented to dermatology for an annual skin cancer screening. On exam, there were multiple pink to skin-colored smooth papules and plaques on the arms and superior back, some of which were tender (Fig.1&2). The patient had multiple similar lesions excised decades ago, which were reportedly smooth muscle tumors. Because the current lesions were symptomatic, they underwent excision, and pathology revealed piloleiomyomas. Given the significant number of smooth muscle tumors, genetic counseling was recommended. A heterozygous pathogenic variant in fumarate hydratase was identified, which confirmed the diagnosis of hereditary leiomyomatosis and renal cell cancer (HLRCC), also known as Reed syndrome. The patient is undergoing imaging workup and his siblings have initiated genetic counseling. Reed syndrome is a rare autosomal-dominant syndrome, which manifests as cutaneous leiomyomas (CLM), uterine leiomyomas, and renal cell carcinomas (RCC).<sup>1</sup> CLMs are the most specific and sensitive clinical indicators of HLRCC, and piloleiomyomas, as seen in our patient's case, are the most common type of CLM associated with Reed syndrome.<sup>1</sup> Almost all patients develop cutaneous leiomyomas by the age of 40 and the mean age of renal tumor development is 44.<sup>1,2</sup> Symptomatic piloleiomyomas may be treated with surgical excision, carbon dioxide laser, cryotherapy or electrodesiccation.<sup>1</sup> Given the predisposition to RCC, annual renal MRI is recommended.<sup>1</sup> Women with uterine leiomyomas should undergo annual gynecologic examinations.<sup>1</sup> Siblings of affected individuals should be notified. Dermatologists should be aware of the manifestations of this rare syndrome to identify and diagnose patients who are unknowingly affected by Reed syndrome.

### References:

1. Patel VM, Handler MZ, Schwartz RA, Lambert WC. Hereditary leiomyomatosis and renal cell cancer syndrome: An update and review. *J Am Acad Dermatol.* 2017;77(1):149-158. doi:10.1016/j.jaad.2017.01.023
2. Stewart L, Glenn G, Toro JR. Cutaneous leiomyomas: a clinical marker of risk for hereditary leiomyomatosis and renal cell cancer. *Dermatol Nurs.* 2006;18(4):335-342.

**Figure 1:** Skin-colored plaques on the right arm



**Figure 2:** Pink papule on the right superior back

