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Hereditary leiomyomatosis and renal cell cancer: Diagnosis in patient after presentation of renal cell cancer in son

A 72 year-old Caucasian male with a past dermatologic history of malignant melanoma in situ at age 35 and multiple nonmelanoma skin cancers presents with several painful nodules, exacerbated by cold temperature, on the posterior left shoulder in the vicinity of previous excision sites. In his 40s and 50s, the patient reports several cutaneous leiomyomas were excised from his upper back. Some years later, genetic screening was triggered by the diagnosis of RCC in his then 31 year-old son. Site-specific analysis was positive for heterozygous p.N340K variant in the FH gene and a diagnosis of HLRCC. At present, physical exam was notable for two firm, translucent pink nodules on the left posterior shoulder. Histopathology of biopsy of posterior left shoulder nodules showed a well-differentiated spindle cell lesion within the dermis exhibiting classic smooth muscle consistent with a diagnosis of piloleiomyoma.

Hereditary leiomyomatosis and renal cell cancer (HLRCC) syndrome is a rare, autosomal dominant disorder in which affected individuals are predisposed to the development of cutaneous leiomyomas (CLM), uterine leiomyomas (ULM) in women, and early-onset of type 2 papillary renal cell carcinoma (RCC).

CLMs can occur sporadically and can be single or multiple. It has been reported that as much as 89% of patients with multiple CLMs have evidence of germline FH mutations. Surveillance of affected individuals is crucial in early detection of RCC. In contrast to other hereditary renal cancer syndromes, HLRCC-associated RCC requires excision at any size. Annual screening is recommended beginning at 10 years of age with contrast-enhanced MRI of kidneys.

Due to the association of HLRCC with aggressive RCC, it is crucial to properly evaluate patients presenting with cutaneous leiomyomas in order to ensure timely screening for possible development of RCC. Increased awareness of this syndrome is necessary in clinicians who may encounter these patients.