



## Renal Cell Carcinoma in Hereditary Leiomyomatosis Syndrome: When the Web Makes the Difference

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### Clinical Image

A 68-year-old woman with a history of hysterectomy for leiomyoma at the age of 24 and cutaneous leiomyomas in the past few years discovered a renal multicystic mass without clear radiological signs of malignancy (Figure 1A and B). The patient, searching the web for possible links between the uterine leiomyoma and cutaneous leiomyomas, read about a potential syndrome and looked for genetic consultation. Family history showed cutaneous and uterine leiomyoma cases in her maternal lineage; an Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC) Syndrome was genetically confirmed through the Fumarate Hydratase mutational analysis on chromosome 1 [1].

Due to the genetic results, the CT scan was reviewed and, although the cystic mass did not show sign of malignancy, partial nephrectomy with intraoperative consultation (“frozen section”) was performed to confirm the nature of the lesion. The specimen was characterized by a 4.2 cm cystic tumour with mucoid appearance (Figure C). Histological examination showed a multicystic proliferation lined by oncocytic cells, with large nuclei, prominent orangiophilic nucleoli and clear perinucleolar halos (reminiscent of cytological modifications due to cytomegalovirus infection) (Figure D). Based on these cytological characteristics, hallmark of the HLRCC syndrome, diagnosis of syndrome-related renal cell carcinoma was made [5]. Due to the biologically aggressiveness of this subtype of tumor, total nephrectomy was performed. At 12 months the patient is alive and without evidences of disease.

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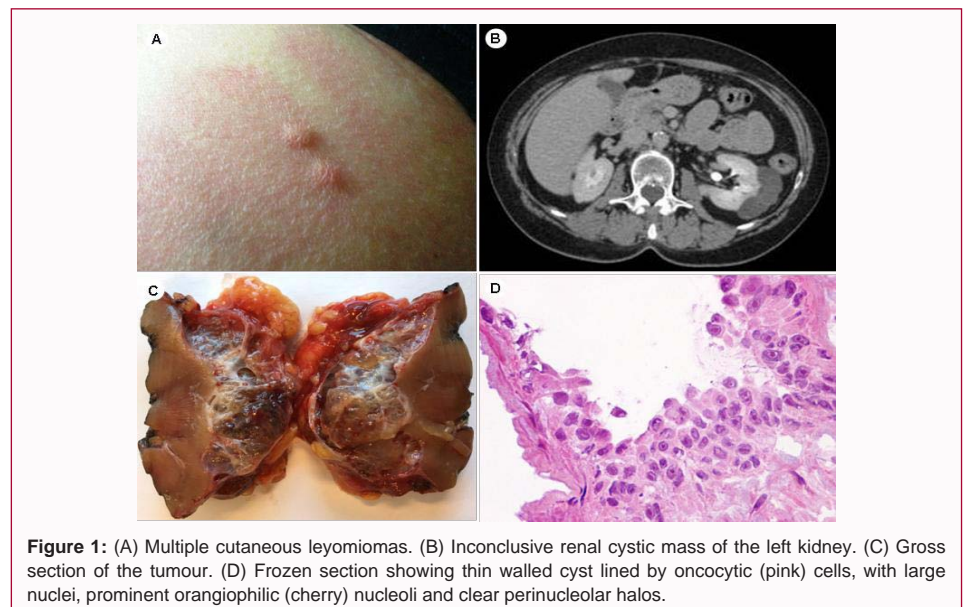
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**Figure 1:** (A) Multiple cutaneous leiomyomas. (B) Inconclusive renal cystic mass of the left kidney. (C) Gross section of the tumour. (D) Frozen section showing thin walled cyst lined by oncocytic (pink) cells, with large nuclei, prominent orangiophilic (cherry) nucleoli and clear perinucleolar halos.

but can be encountered in daily practice; an early diagnosis can be made only if this condition is suspected [1-5].

## References

1. Merino MJ, Torres-Cabala C, Pinto P, Linehan WM. The Morphologic Spectrum of Kidney Tumors in Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC) Syndrome. *Am J Surg Pathol.* 2007; 31(10):1578-1585.
2. Kiuru M, Launonen V, Hietala M, Aittomäki K, Vierimaa O, Salovaara R, et al. Familial cutaneous leiomyomatosis is a two-hit condition associated with renal cell cancer of characteristic histopathology. *Am J Pathol.* 2001; 159(3):825-829.
3. Launonen V, Vierimaa O, Kiuru M, Isola J, Roth S, Pukkala E, et al. Inherited susceptibility to uterine leiomyomas and renal cell cancer. *Proc Natl Acad Sci U S A.* 2001; 98(6):3387-3392.
4. Tomlinson IP, Alam NA, Rowan AJ, Barclay E, Jaeger EE, Kelsell D, et al. Germline mutations in FH predispose to dominantly inherited uterine fibroids, skin leiomyomata and papillary renal cell cancer. *Nat Genet.* 2002; 30(4):406-410.
5. Toro JR, Nickerson ML, Wei MH, Warren MB, Glenn GM, Turner ML, et al. Mutations in the fumarate hydratase gene cause hereditary leiomyomatosis and renal cell cancer in families in North America. *Am J Hum Genet.* 2003; 73(1):95-106.