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Clinical Image
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Renal Cell Carcinoma in Hereditary Leyomiomatosis Syndrome: When the Web Makes the Difference

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Clinical Image

A 68-year-old woman with a history of hysterectomy for leyomioma at the age of 24 and cutaneus leyomiomas 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 leyomioma and cutaneus leyomiomas, read about a potential syndrome and looked for genetic consultation. Family history showed cutaneous and uterine leyomioma 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.

Despite the progresses in patient care, a careful overview of clinical history is often lacking, especially when dealing with benign disease in outpatient or in day-hospital settings; a virtuous use of the internet by the patient allowed an early diagnosis of an aggressive neoplasm. Moreover the clinical hypothesis, based on history, physical examination and genetics, even with a benign radiological and macroscopic appearance, was only confirmed intraoperatively and a more aggressive surgery was undertaken. Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC) Syndrome is rare

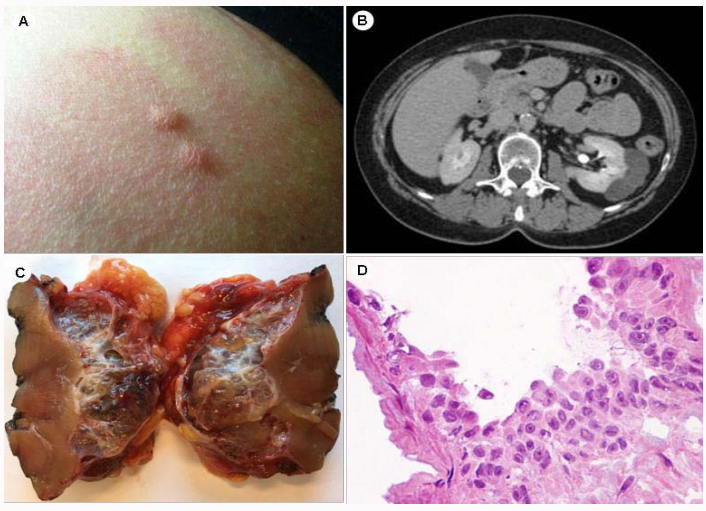

Figure 1: (A) Multiple cutaneous leyomiomas. (B) Inconclusive renal cystic mass of the left kidney. (C) Gross section of the tumour. (D) Frozen section showing thin walled cyst lined by oncocytic (pink) cells, with large nuclei, prominent orangiophilic (cherry) nucleoli and clear perinucleolar halos.

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

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