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Hereditary leiomyomatosis and renal cell carcinoma: A rare case report

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Abstract
Papillary Renal cell carcinoma is the second common histologic subtype of renal malignancy. Hereditary Leiomyomatosis and renal cell carcinoma are syndromes predisposing to papillary renal cell carcinoma type 2 along with leiomyoma uterus and skin. The prevalence of HLRCC is 1 in 200000. We present a case of HLRCC presenting with skin leiomyoma and renal cell carcinoma. This papillary renal cell carcinoma in HLRCC is associated with poor prognosis. Early and prompt excision of Renal mass and lymph nodes is the only treatment. Adjuvant treatment is not well established and not available for the common man in our part of the country. Post-surgical follow up and patient education is essential.

Keywords: hereditary leiomyomatosis and renal cell carcinoma, papillary RCC

Introduction

Papillary renal cell carcinoma is the second most histologic subtype (10-15%) after clear cell carcinoma [1]. It is of 2 types: type 1. Multicentre Common variant consists of basophilic cells with scanty cytoplasm, Type 2. Solitary, aggressive variety with eosinophilic cells. Hereditary Leiomyomatosis and renal cell carcinoma are syndromes predisposing to papillary renal cell carcinoma type 2 along with leiomyoma uterus and skin. The prevalence of HLRCC is 1 in 200000. Papillary RCC is avascular, as VHL mutations are lacking. Adjuvant therapies are lacking for papillary renal cell carcinoma; hence prognosis may be poor in type 2 [2].

We present a case of a young male presenting with papillary renal cell carcinoma type 2 with leiomyomas whole body.

Case summary

A 29 years old male presented with a history of abdominal pain, dull aching, intermittent. On examination, an abdominal lump was palpable. The patient has numerous nodules from 1mm to 3 cm on the back, both lower limbs. (Figure 1a-b) Eye examination was normal, Chest x-ray was normal. Blood investigation Hemoglobin 11.5 gram%, TLC 6700/mm3, urea 20, creatinine 0.8, SGOT/SGPT 23/34, serum alkaline phosphatase 113. Ultrasound abdomen showed a right renal mass. CECT abdomen and chest showed large heterogeneous mass mid pole with interaroto-caval and paracaval lymphadenopathy. Also, hypodense mass seen in the right adrenal. No IVC thrombus, metastases were seen. (Figure 2, 3)

Patient underwent biopsy of nodule from back and leg. Histopathology of nodules was Nodule leg Epidermis thinned out and hyperkeratosis, flattening of rete pegs, tumor composed of interlacing smooth muscle fibers and collagen fibers, these cells are mildly pleomorphic, with centrally located thin, long, blunt-Edged nuclei, bland chromatin, inconspicuous nuclei, eosinophilic cytoplasm.

CECT Abdomen Kidney Well circumscribed tumor predominately arranged in papillae having a fibrovascular core. The tumor cells lining these papillae show pseudo-stratification with vesicular chromatin, prominent nucleoli, and abundant eosinophilic cytoplasm. Occasional psammoma bodies are also seen. No necrosis or sarcomatoid change was seen. Surrounding renal parenchyma shows normal glomeruli and tubules. Overall features were suggestive of papillary renal cell carcinoma.

The patient underwent right open radical nephrectomy, under general anesthesia, supine position, on exploration large right renal mass 10 x 12 cm with hilar, paracaval, interaroto-caval lymphadenopathy. No peritoneal, liver metastases were seen.

The postoperative period was uneventful. Genetic counselled and testing was advised; however, patient refused for it. On follow up at 3, 6 months patient was asymptomatic, CECT scan at nine months was normal. The patient lost to follow up at one year and could not be contacted upon.

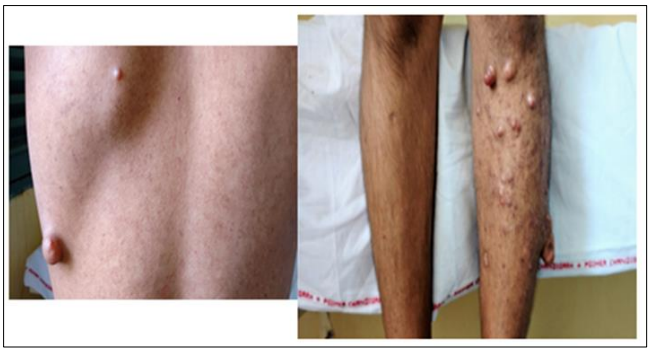

Fig 1: a-b Skin colored in nodules in back, bilateral legs of variable sizes less than 1mm to 3 cm

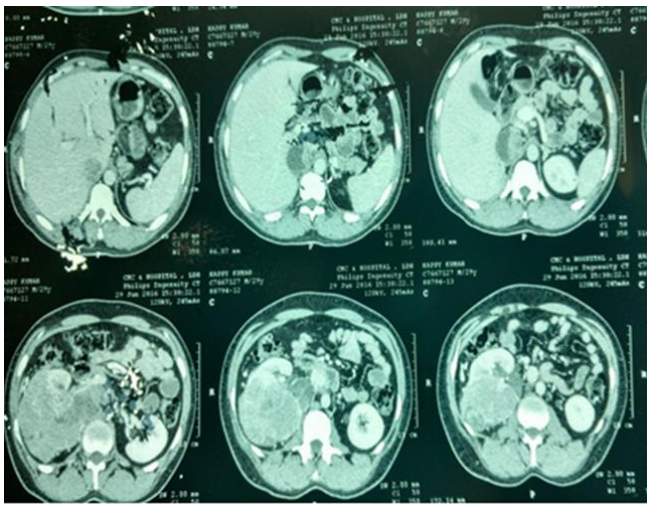

Fig 2: CECT abdomen showing rights renal mass along with lymphedenopathy, heterogeneous arising from posterior surface pushing normal kidney anteriorly

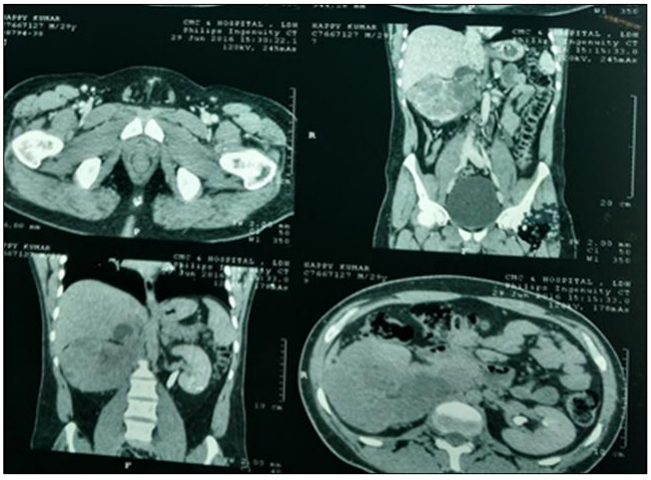

Fig 3: CECT abdomen axial and saggital showing rights renal mass along with lymphedenopathy, heterogeneous arising from posterior surface pushing normal kidney anteriorly

Discussion

Launomen *et al*. first described hereditary leiomyomatosis and renal cell carcinoma (HLRCC) syndrome in 2001. These patients presented at a mean age of 40 years with Leiomyomas (skin, uterine) and type II papillary renal cell carcinoma. Renal cell carcinoma associated with this syndrome was solitary and unilateral with an aggressive course [3].

HLRCC was associated with chromosome locus 1q42-44 with autosomal dominant inheritance. This locus was also associated with Fumarate hydratase, which is an enzyme in Kreb's cycle. Inactivation of Fumarate hydratase results in VHL independent upregulation of hypoxia-inducible factor and activation of proangiogenic growth factors [4]. Almost all patients develop leiomyomas (till 25-30 years), only a few (20%) will develop renal cell carcinoma. In females, uterine leiomyomas are almost always present. Rarely leiomyosarcomas are also reported in this syndrome [5].

Merino *et al*. reported in a series of 38 patients that these tumors are predominantly solitary and unilateral. HLRCC is a fulminant disease; hence an aggressive policy is adopted. Rapid diagnosis with early radical surgical treatment is advocated even for small tumors. The patient is followed for any recurrence. Surgical excision for recurrence is an only viable option [6].

In another study of 27 patients by Gardie *et al*., 74% of patients due to metastatic disease, the prognosis is reduced due to delayed diagnosis and aggressive renal cancer subtype.

In a study including 14 patients of advanced HLRCC, partial response was seen in 6(14%) patients. Further studies are undergoing for advanced papillary RCC [7].

Our young patient presented with advanced renal cell carcinoma with lymph node involvement. He was treated previously for skin leiomyoma; however, the renal mass was not suspected either evaluated as a syndrome. We were able to remove macroscopic disease, but adjuvant therapy was not available. The patient follows up was done with CECT abdomen and found to be normal. Patient was advised strict follow-up because of young age, advanced histopathology, and type 2 papillary RCC; patient was educated and counselled. However, the patient could not be traced after one year.

Conclusion

HLRCC is a rare diagnosis with an aggressive course of the disease. These patients should be treated promptly and advised regular follow up. Radical surgical is only viable option for these patients.

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