Upper urinary tract tumors associated to Lynch syndrome: A case report

Dear Editor

Lynch syndrome (LS) or hereditary nonpolyposis colorectal cancer (HNPCC) is a dominant autosomal multiorgan cancer syndrome caused by mutations in the genes responsible for DNA mismatch repair (MLH1, MSH2, MSH6, or PMS2), which is characterized by the occurrence of colorectal (63%) and endometrial (9%) cancer, although other tumors such as urothelial carcinoma of the upper urinary tract can be associated, which is in third place (5%).

In this respect, we present the case of a 47-year-old woman diagnosed with left renal pelvis tumor after episode of renal colic.

This is a patient diagnosed with LS after performing DNA sequencing, followed-up by the digestive service and controlled with regular colonoscopies, with paternal family history of colon cancer: great-grandmother (40), grandfather (38), grandfather’s brother (60), and father (42). She attends the emergency department due to colic-type picture in left renal fossa, without analytical impact. After expulsion of stones spontaneously, imaging tests are performed for the anatomo-functional study of the urinary system, with the following findings:

- Intravenous urography: filling defect of rounded morphology and 1.5 cm in diameter in the left renal pelvis.
- Abdominal ultrasound with contrast: 1.6 cm focal thickening in left renal pelvis, which protrudes into the lumen of the urinary tract (Fig. 1A).
- Abdominopelvic contrast CT: lesion of approximately 1.5 cm in left renal pelvis, with contrast uptake, suggesting tract tumor (Fig. 1B).

In response to the imaging tests and after performing the preoperative study, left nephroureterectomy is performed through retroperitoneoscopy with 3 trocars (1 × Hasson, 1 × 10 mm and 1 × 5 mm), conducting specimen removal, with prior bagging, by means of Gibson incision. The pathology report of the surgical specimen was a transitional cell carcinoma of the renal pelvis with inverted growth, pT2 stage, grade 2, with free surgical margins. The patient progressed favorably being discharged at 72 h of the surgery. Currently the patient is asymptomatic and periodically checked-up by our service, without signs of recurrence objectified.

Fig. 1 Tumor of the left renal pelvis (arrows). (A) Abdominal contrast ultrasound. (B) Abdominal pelvic contrast CT.
Due to ignorance, many urothelial tumors of the upper urinary tract are misclassified as sporadic when they really are a manifestation of HNPCC. An estimated 21.3% of patients diagnosed with upper urinary tract tumor can associate a HNPCC, especially when they are under 60 and have a family or personal history.

In addition, controversy exists as to the association with bladder tumors. Some studies report that patients diagnosed with HNPCC with MSH2 mutations not only have an increased risk of upper urinary tract tumors but also of bladder tumors, which is important to conduct a proper study. While others point out that patients with HNPCC are more likely to develop high but not bladder urothelial tumors.

Therefore and in conclusion, we can say that it is important that the urologist is aware of the association of urothelial tumors with hereditary nonpolyposis colorectal cancer to perform a proper evaluation of the patient and thus identify those cases associated with heritable genetic damage, allowing for better clinical management of both patient and family.

References


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