1130-0108/2011/103/9/500-501
Revista Española de Enfermedades Digestivas

Rev Esp Enferm Dig (Madrid)
Vol. 103, N.° 9, pp. 500-501, 2011

Letters to the Editor

Association between Lynch syndrome and renal carcinoma

Key words: Lynch syndrome. Colorectal cancer. Clear cell renal carcinoma. Von Hippel Lindau syndrome.

Dear Editor,

We read with interest the study published by Ferrer et al. about a patient with Lynch syndrome (LS) who presented a colorectal carcinoma (CRC) and a synchronous clear cell renal carcinoma (CCRC) (1). From our point of view, it would be interesting to emphasize some aspects of this patient, such as the type of mutation carried by the patient, and its cancer familial tree. In addition, we would like to point out that the most common cancer of the urinary system associated with LS is the urinary tract (urothelial) carcinoma (renal pelvis, ureter) (2), and that there are no references on the association of this syndrome with CCRC. In the same way, the identification of microsatellite instability, or more directly, the absence of protein expression of the DNA repair system (so the importance of knowing the mutated gene), not only in the CRC, but also in the CCRC, would help to classify both tumors as caused by the LS (3). If the result of this study is positive for renal carcinoma, it would be very interesting, since it would be the first reported case of the association of LS with CCRC.

The need to know the family tree of this patient is based on the type of renal neoplasm. In sporadic cases, CCRC is associated with abnormalities in the VHL gene. But the presence of family history of CCRC, with the appearance at early ages, as is the case of the commented patient, it would require to exclude a germline mutation of the VHL gene, which defines the Von Hippel Lindau syndrome (4,5). Identification of this mutation not only would give a special importance to this case (two germline mutations in two different genes, and consequently two syndromes of hereditary cancer), but it would also determine the need to carry out the same identification in other family members of the latter syndrome. In addition, the Von Hippel Lindau syndrome is also associated with other disorders, such as retinal hemangioblastomas, pheochromocytomas, pancreatic lesions, endolymphatic sac tumors or benign epididymis cystadenomas (males) or more rarely, of the broad ligament (females) (6). If the case of a positive identification of a Von Hippel-Lindau syndrome, it would be mandatory monitoring, not only the Lynch syndrome, but also the potential manifestations of the latter syndrome.

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