Gastroenterology and Hepatology From Bed to Bench

LETTER

XPD gene polymorphism and colorectal cancer risk

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TO THE EDITOR

We examined Rezaei et al's paper (1) and we are interested in the subject. The study was about the functions of repair genes, their mechanisms in cancer development and associated polymorphisms which increase susceptibility to CRC. As we know Single nucleotide polymorphisms (SNPs) are the most common genetic sequence variation in human genome. SNPs are associated with population diversity, disease susceptibility and individual response to medical therapy (2, 3). In the other hand, the development of CRC is associated with environmental factors, genetic susceptibilities, and their interaction (4). Polymorphisms in DNA repair genes contribute to the variations in individual genetic susceptibility to many types of cancers, such as lung cancer, breast cancer and gastric cancer (5–9).

The study population of this paper was heterogeneous in terms of colorectal cancer. The authors mentioned that of 88 colorectal cancer patients enrolled in this study, 32 patients (45%) had a positive family history for colorectal cancer in their first-degree relatives. They didn't determine the type of familial CRC in these patients. We didn't understand whether patients with history of CRC in the first degree relatives were matched the criteria for HNPCC or FAP or not. In familial subgroup, mutations screening in related genes is more important than evaluating SNPs and this is the hallmark of the molecular screening for familial and hereditary diseases. The authors could at least evaluate the SNPs in the disease-causing mutations occurring in genes which totally account for 15%-20% of CRC familial population, including Mismatch Repair (MMR) genes, Tumor suppressor gene APC or the base excision repair gene MUTYH (10).

In present study, the authors evaluated the SNP in two groups of sporadic and familial patients with CRC and as we know, some polymorphisms might be more common in familial or hereditary forms of disease than the sporadic forms, and this would affect the result. Moreover, SNP distribution among sporadic or familial cases wasn't present. In the other hand, SNP studies need larger sample size to get more accurate results. Therefore even if, all 88 patients were sporadic or familial cases, a reliable conclusion would not be feasible. So, a case- control study with adequate sample size and well- defined groups is recommended to compare the frequency of the specific allele with patients and healthy subjects.

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