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Hassan Ashktorab

Howard University, USA

Targeted cancer gene sequencing identifies potential causative novel candidate mutations in colon carcinogenesis

Colorectal cancer is the second cause of death in the world and genomic alteration plays an important role in this disease. Much of the underlying genetic cancer driver mutations in sporadic colorectal cancer (CRC) have not been characterized by race. Here, we report the identification of distinct novel variants from CRC patients in mismatch repair (MMR) genes *MHS3* and *MSH6* and *APC*. We developed a panel of 20 frequently altered colon cancer genes for targeted sequencing in 138 colon tissues using next generation sequencing to examine 98.8% of the targeted exons and splice junctions at a depth of sequencing that allowed for high confidence variant calling. After alignment and variant calling, we annotated the variants with information from the 1000 Genomes Project, COSMIC, Polyphen2 and PFAM domain and transcription factor motifs. Excluding synonymous SNVs, 212 deleterious variants in adenoma, 760 in advanced adenoma and 2624 variants in tumors were detected. Novel variants (1591 and 1363) were found in MMR genes (*MSH6* and *MSH3*) and *APC* gene, respectively. These findings further highlight the relevance of *APC* gene in CRC onset but also the potential underestimation of the MSI-H in sporadic CRC as many of the novel mutations in MMR genes detected here were of a deleterious nature with a therapeutic interest.

Biography

Hassan Ashktorab has completed his PhD from Utah University and Postdoctoral studies from Indian University and University of Florida, School of Medicine. He is the Director of Microarray lab and a Member of Gastro-interstitial Research group. He has published more than 100 papers in reputed journals and has been serving as an Editorial Board Member of many journals including *DDS*, *GUT*, *PlosOne* and others.

hashktorab@Howard.edu

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