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How can clinician be precise in era of precision medicine? Case of colon cancer

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/varaints 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 MSH2, 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, Catalogue of Somatic Mutations in Cancer (COSMIC), Polymorphism Phenotyping v2 (Polyphen2) and PFAM domain and transcription factor motifs. Excluding synonymous SNVs, 212 deleterious variants in adenoma, 760 in advanced adenoma, and 2624 variants in tumours 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 so called "uncertain significance" in MMR genes detected here were of a deleterious nature with a therapeutic interest.

Biography

Ashktorab has completed his PhD from Utah University and Postdoctoral studies from Indina University and University of Florida, School of Medicine. He is the director of Microarray lab, a member of Gastrointestical Research group. He has published more than 100 papers in reputed journals and has been serving as an editorial board member of many Journal including DDS, GUT, PlosOne and others.

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