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Identification and pattern analysis of SNPs involved in colorectal cancer

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Colorectal cancer (CRC) is the second leading cause of cancer related deaths globally posing a lifetime risk of 80-100% in every individual. Genetics and relevant mechanisms underlying some key signaling pathways like *Wnt, TGF, p53, K-ras* etc. play a detrimental role in governing the predisposition for CRC. A high percentage of colorectal tumors (adenomas and carcinomas) show activating mutations in *beta-catenin* or *axin*, whereas, loss of certain tumor suppressor genes (TSGs), like APC cause the initiation of random polyps in the colon. All of these molecules incidentally are critical components of an evolutionarily conserved *Wnt* signaling pathway, which is instrumental at various time points in the development of this disease. Differences in SNP profiles amongst sample groups in the genomic landscape can be recognized through a smart and efficient use of machine learning techniques. The statistics and pattern analyses of these SNP profiles, interestingly provides us with a concrete and logical platform upon which, relative contribution/s of each unique SNP, ranging "from cause to effect" can be significantly assessed. The biological relevance of such SNP variations with respect to cancer prediction and predisposition, however, remains unexplored, pending a better understanding of the impact of control design in SNP studies. Our results emerging from critical analyses of significant SNP's, demonstrates the utility of relevant bioinformatics tools in discriminating diseased populations based on realistic SNP data. In this study, we have primarily targeted members of *Wnt* signaling pathway, which play important developmental role/s during different stages of colorectal cancer, depicting a classical "multigene-multistep nature" of cancer. We have identified and related common genetic variants for the "early-acting" and "late-acting" members of this pathway, that are most prevalent in patients with CRC disease. Complex relationships and correlations hidden in large data sets have been dug and analyzed here, by deploying various data-mining tools.

Biography

Jyoti Bhojwani, is presently a Faculty of Genetics/Bioinformatics, Principal Investigator of the M.Tech Research Programs (Bio-Informatics) and is engaged in teaching the curriculum in M.Sc Life-Sciences/Bio-Technology at University of Indore, India. She obtained her B.Sc. degree in Biological Sciences/Chemistry/Physics, M Sc degree in Life-Sciences, and Doctoral degree (Ph.D.) at School of Life-Sciences, University of Indore. Thereafter, Dr. Jyoti Bhojwani pursued her post-doctoral ventures at Max-Planck Institute for Biophysical Chemistry (FRG), University Of California-Irvine and Univeristy of Pittsburgh (USA). Currently, Dr. Jyoti Bhojwani's projects mainly focus on translational-research and extrapolation of basic developmental mechanisms from model-systems like fruitfly (*Drosophila*) to human. Apart from this, her major thrust areas of research interest have been Cancer Biology, Stem-Cell Biology and Homeotic-Gene Regulation. She is really keen on studying in details the genetic factors, which presumably aid in understanding of mechanism by which "cancer stem cells" function in transforming a tissue from normal to neoplastic or hyperplastic states. Especially, the members of genetic pathways, which bear special relevance and convergence to both tumorigenesis and stem cell renewal/differentiation/lineage progression, are of prime interest, in her research-regimen. Her research has a motive to further facilitate the perception of stem cell potential/mechanistic in areas of regenerative medicine and anti-cancer therapy. Currently, being involved in Clinical informatics, her students are also training a Cancer model and a Stem cell model, deploying Systems Biology approach and other Gene Networking Bioinformatics tools. This novel area of research will hopefully lead to further understanding the tipping of balance from a stem cell/normal cell to a transformed cancer cell. Besides being involved with research and teaching, she is also deeply inclined towards Science Journalism. Owing to her publications and useful insights in scientific field, she is now on the editorial-board of an international journal (IJUDH). Her Specialties Include: Research/Teaching/Mentoring/Science-Journalism

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