Identification and pattern analysis of SNPs involved in colorectal cancer

Jyoti Bhojwani
DAVV/Indore University, India

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 contributions of each unique SNP, ranging "from cause to effect" can be significantly assessed. The biological relevance of these SNP variations with respect to cancer prediction and predisposition, however, remains to be resolved, pending a better understanding of the impact of rational control design in SNP studies. Our results emerging from the analyses of significant SNP's reported here, demonstrates the utility of relevant bioinformatics tools and machine learning techniques in discriminating diseased populations based on realistic SNP data. In this study, we have primarily targeted critical members of Wnt signaling pathway, which play important developmental roles 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 techniques.

Biography

Jyoti Bhojwani is presently a Faculty of Genetics/Bioinformatics/ principal investigator of the M.Tech research programs (Bio-Informatics) at University of Indore, India. She obtained her BSc (Bachelor's degree) in Biological Sciences/Chemistry/Physics, MSc (Master's degree) in Life-Sciences and Doctoral degree (PhD) at School of Life-Sciences, University of Indore. She pursued her post-doctoral ventures at Max-Planck Institute for Biophysical Chemistry (FRG), University of California-Irvine and University of Pittsburgh (USA). Currently, her projects mainly focus on translational-research and extrapolation of basic developmental mechanisms from model-systems like fruitfly (Drosophila) to human. Apart from this, her thrust areas of research interest include; cancer Biology, stem-cell biology and homeotic-gene regulation. She is keen on studying in detail the genetic factors, which presumably aid in understanding of mechanism by which "cancer stem cells" function in transforming a tissue from normal to cancerous states. Her research has a motive to further facilitate the perception of stem cell potential/mechanistic in areas of regenerative medicine, translational research and anti-cancer therapy. 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. Owing to her immense interest in science journalism and writing potential, she is now on the editorial board of several international journals.

jbhojwani2005@gmail.com