

# PD Dr. Rajiv Kumar

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## Current Research

Major research interests are cancer genetics. The current research projects involve study of somatic molecular events in malignant melanoma. Malignant melanoma that mainly afflicts Caucasian population is the major cause of death due to skin cancers. The aim of our research is to understand the somatic genetic events involved in the initiation and progression of melanoma. The impact of somatic alterations on global genomic expression is also investigated using microarrays. In addition, our research is also involved in investigation of germline genetic variants involved in the susceptibility and disease outcome of malignant melanoma. These goals are being pursued using both candidate gene as well as whole genome approach using SNP microarrays. Our studies have identified and characterized the correlation between major somatic genetic events in malignant melanoma. While the oncogenic mutations in the B-RAF and N-RAS genes constitute the initiating somatic events that are followed by loss of a major check point gene mainly CDKN2A or in some cases p53 or PTEN. The major susceptibility genes identified, besides the MC1R include ASIP and TYR. Some of the genetic variants in the DNA repair gene XRCC1 that influence the disease outcome have also been identified and such variants can be potentially used as prognostic markers.

Other major cancers being studied currently include basal cell carcinoma of skin, childhood acute lymphoblastic leukemia, esophageal and gastric cancers.

## Contact:

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## Future Projects and Goals

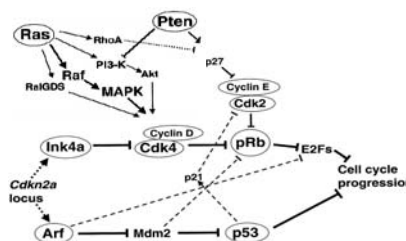
The aim of our research is to identify and characterize the sequential genetic events that occur from initiation through progression to metastasized melanoma in order to identify targets of intervention. Further, we aim to characterize the variants that cause the population susceptibility to the disease and influence disease outcome. The markers can be eventually used as markers for identification of potentially high-risk groups and for predicting prognosis that could influence in treatment decisions

## Selected Publications

Gudbjartsson DF et al. ASIP and TYR pigmentation variants associate with cutaneous melanoma and basal cell carcinoma. *Nat Genet.* 2008; 40:886-891.

Akslen LA et al. Mutation analysis of the EGFR–NRAS–BRAF pathway in African melanomas and other subgroups of cutaneous melanoma. *Melanoma Research* 2008;18:29-35.

Scherer D et al. MC1R variants associated susceptibility to basal cell carcinoma of skin: Interaction with host factors and XRCC3 polymorphism. *Int J Cancer* 2008;122:1787-1793.



Genetic pathways involved in melanoma