

Mutational signature analysis and its applications to the clinic

Abstract: Different mutational processes operative in cancer and other diseases leave distinct 'signatures' in the DNA. Mutational signature analysis is an attempt to deconvolve the mutational patterns from cancer sequencing data to better identify the factors that gave rise to cancer. Whereas previous work required a large amount of signal as found in exome and genome sequencing data, our new method SigMA enables accurate detection of mutational signatures even with >100-fold reduction in data size. This allows us to extend signature analysis to gene panels, the common platform used to profile tens of thousands of cancer patients each year. I will describe the methodology behind SigMA and how it can be used to identify patients with deficiency in the homologous recombination DNA repair pathway who should be considered for treatment with PARP inhibitors. This work was led by Dr. Doga Gulhan (PhD in heavy ion physics, MIT)

Bio: Dr. Park is Professor of Biomedical Informatics at Harvard Medical School and the director of its Bioinformatics and Integrative Genomics Ph.D. program. His group (<http://compbio.hms.harvard.edu>) specializes in computational and statistical analysis of high-throughput sequencing data in epigenetics, cancer genetics, and neuroscience. He was originally trained in applied math (B.A., Harvard; Ph.D., Caltech), but he stumbled upon molecular biology and genetics during his postdoctoral studies. He has multiple positions open in his group for students, postdoctoral fellows, and scientific programmers.