



The faculty of Biotechnology and Food Engineering

Seminar

Dr. Yosef E. Maruvka

Massachusetts General Hospital Cancer Center and Department of Pathology

Broad Institute of Harvard and MIT

Harvard Medical School

The genomic landscape of microsatellite unstable tumors, detection and prevention

Abstract

Many tumor types contain a subtype with a deficiency in the DNA mismatch repair mechanism. These tumors have an increased rate of insertions and deletions (indels) in the repetitive loci of the genome called microsatellites, leading to what is called microsatellite instability (MSI). I developed a tool to read these difficult-to-read regions, called MSMuTect, that was able to find many indels missed by the previous tools, with medical import .

MSI tumors received attention recently, as the FDA approved the new immunotherapy PD-1/PD-L1 blockade treatment for all MSI tumors regardless of their tissue type. However, only about half of the MSI cases respond to immunotherapy. I demonstrated that it is the number of microsatellite indels that can predict the tumor response to immunotherapy .

Furthermore, using MSMuTect I was able to analyze the details of the mutations and to find new cancer genes in MSI tumors .

I was also able to leverage the unique features of MSI tumors in order to detect MSI tumors from cell-free DNA shed by tumors into the bloodstream .

By way of conclusion, I will describe an ongoing effort that uses the microsatellite indels that we found using MSMuTect to generate a preventive treatment for the hereditary version of MSI tumors called Lynch syndrome.

Sunday, 27.01.19, 13:00 – 14:00, Room 300

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