

April 6<sup>th</sup>, 2018

## Diagnosis of low burden tumors using circulating cell-free DNA

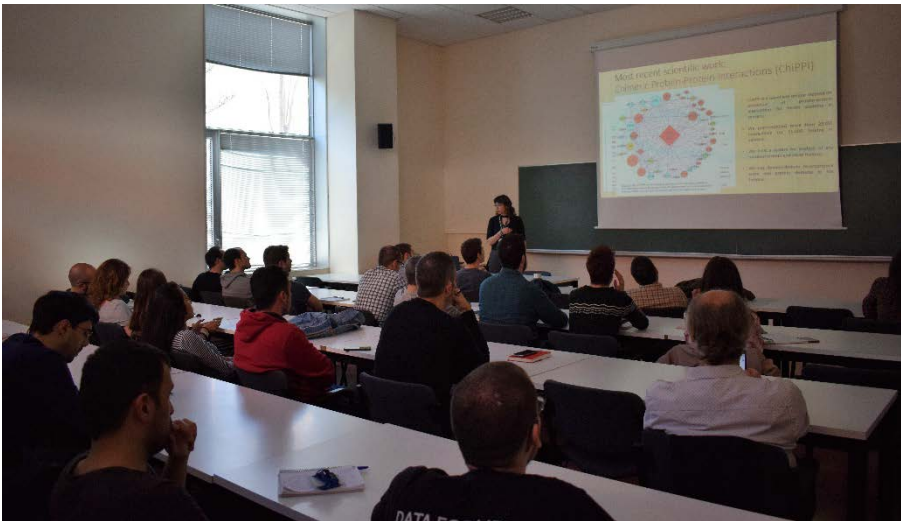
*This seminar is part of the "Bioinfo4Women" series.*

**Milana Frenkel-Morgenstern**

The Azrieli Faculty of Medicine, Bar-Ilan University, Safed, Israel

### Abstract

Gliomas are the most frequent brain tumors worldwide. Gliomas make up about 30% of all brain and central nervous system tumors, and 80% of all malignant brain tumors. Diagnosis of the glioma tumor type and its grade is a most essential step in order to suggest a right treatment for the glioma patients.



We present a comprehensive study of the different types of the tumors with a low burden in plasma matched with the cfDNA extracted from a clinical cohort of patients' plasma in order to find unique tumor mutations as biomarkers. We successfully detected the glioma specific mutations for the highly frequently mutated genes such as IDH2, PDGFRA, NOTCH1, PIK3R1 and 30 other genes. We identified the particular mutations of the cfDNA isolated from the plasma of the glioma patients, followed by the DNA-sequencing

and our predictive bioinformatics analysis. We have collected the matched tumor and cfDNA mutations to uncover the tumor grade as well as its heterogeneity using our unique measurement of the mutations coverage by the DNA-seq reads. Moreover, we used our previously published methods to uncover unique fusions in the glioma patients and its alterations in the protein-protein interactions networks to understand the tumor prognosis. For the best of our knowledge, our study is the most advanced study in the field of the liquid biopsy for the brain cancer tumors, and it will provide a quick and safe non-invasive diagnostic method for the glioma patients, as it uncovers the tumour subtypes using unique biomarkers. This will provide the best personalized treatment for the highly complicate disease and will eventually bypass the existing "wait-and- see" method for prognosis.

### Short bio



Milana Frenkel-Morgenstern has completed her Ph.D at the age of 32 years from Weizmann Institute of Science and postdoctoral studies from Spanish National Cancer Research Centre (CNIO). She has published more than 20 papers in reputed journals and serving as an editorial board member of repute. She is a founder of the Art in Science competition at the ISMB conference since 2008, a chair of the ISCB affiliated Israeli Bioinformatics group, and a head of the Cancer Genomics and BioComputing group in the Azrieli faculty of Medicine, Bar-Ilan University.