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Cross tumor type chromosomal instability subtype analysis

We conducted a study on the tumor subtypes chromosomal instability (CIN) and genomic stable (GS), which are not well-studied in endometrial, colorectal, and gastric cancers. To distinguish between CIN and GS tumors, we developed a new computational tool called CINDetect that uses various genomic sequencing methods, including targeted panel sequencing. With a database of 4618 tumor samples that includes multiple types of omics data, such as whole exome sequencing, panel sequencing, RNA sequencing, and single-cell RNA sequencing, we searched for differences between CIN and GS samples by tumor type and combined them.

We found that CIN and GS samples in endometrial cancer differed in numerous ways, while in colorectal cancer, the main difference was a high frequency of TP53 and its effects on gene expression and copy number changes. Gastric cancer fell between the two. However, CIN samples in all tumor types had many deletions and amplifications that offer opportunities for targeted therapy, including genes such as ERBB2 and VEGFA, which were amplified in all three tumor types.

Our single-cell RNA sequencing data showed that the CIN micro-environment changes to support faster angiogenesis, further supporting VEGFA as a potential therapeutic target. We developed an AI algorithm that uses H&E histological images to classify samples as CIN vs GS, with an AUC score of 0.8.

In conclusion, our study sheds light on the unique characteristics of CIN and GS tumors and their potential therapeutic targets.

Biography:

Dr. Yoesef Maruvka earned a dual major in Physics and Math from Bar-Ilan University, followed by a Masters and PhD in physics focused on applying statistical mechanics to population genetics and evolutionary biology. He then completed a post-doctoral fellowship at the Broad Institute of MIT and Harvard where he developed methods to detect mutations in repetitive regions of the genome. In 2020, he founded the Cancer Genomics Evolution lab at the Technion, leading a team focused on understanding early tumorigenic steps and developing methods for analyzing genomic data.

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