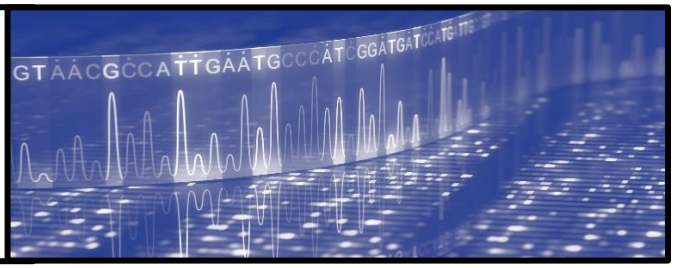
Yale Center for Biomedical Data Science



CBDS Seminar Series

"Evaluating Cancer Drivers with HotMAPS "

Rachel Karchin, PhD

The impact of somatic missense mutation on cancer etiology and progression is often difficult to interpret. One common approach for assessing the contribution of missense mutations in carcinogenesis is to identify genes mutated with statistically nonrandom frequencies. Even given the large number of sequenced cancer samples currently available, this approach remains underpowered to detect drivers, particularly in less studied cancer types. One approach to increase power is to focus on localized regions of increased missense mutation density or hotspot regions, rather than a whole gene or protein domain. Detecting missense mutation hotspot regions in three-dimensional (3D) protein structure may also be beneficial because linear sequence alone does not fully describe the biologically relevant organization of codons. Using a novel and statistically rigorous algorithm for detecting missense mutation hotspot regions in 3D protein structures, our group analyzed approximately 3 × 10(5) mutations from The Cancer Genome Atlas (TCGA) and identified 216 tumor-type-specific hotspot regions.

Rachel Karchin, Ph.D. is Professor in the Department of Biomedical Engineering at Johns Hopkins University. She received a Ph.D. in Computer Science from the University of California, Santa Cruz in 2003, spent three years as a postdoctoral fellow in the Department of Biopharmaceutical Sciences at University of California, San Francisco, and joined the Hopkins faculty in 2006. Her lab develops algorithms and tools to interpret and model genomic data, with a focus on cancer. Dr. Karchin has a joint appointment in the Department of Oncology and a secondary appointment in the Department of Computer Science. She is currently the William R. Brody Faculty Scholar at Johns Hopkins Whiting School of Engineering.

Wednesday, November 6, 2019
4 p.m. to 5 p.m. Seminar
The Anlyan Center TAC - N203
333 Cedar Street