Copy Number Implementation and Analysis of Ovarian Germ Cell Tumors

Ram, Rahul (School: Camas High School)

Ovarian germ cell tumors are tumors that arise from the egg cells of the ovaries. There are different types of ovarian germ cell tumors including mature and immature teratomas, metastatic tumors, and gliomatosis peritonei. Some of these tumors have a high potential of becoming malignant, and thus understanding the origination mechanism of these tumors is paramount. Currently, lab techniques are utilized to determine such mechanisms by finding zygosity and deducting which stage of meiosis failure caused the tumor. Though a tried and true methods, these techniques are expensive, inefficient, not widely available, and difficult to use. Thus to rectify this, bioinformatic computational methods were created in order to determine the origination mechanism and specific copy number events while being inexpensive, efficient, widely available, and easy to use. Biopsies from ovarian cancer patients were DNA sequenced in the form of reads. Then, duplicate reads were removed, and the remaining reads were mapped to a reference genome. Next, a bias correction was performed on the reads to prepare it for analysis. To determine the origination mechanism, the B-allele frequency was calculated for all reads. The resulting data was then used to determine mechanism of origin. To determine the copy number events, the reads were inputted into a Hidden Markov Model and machine learning algorithm. After the analysis was performed, the results were outputted in the form of copy number graphs. The results were verified with CNVKit. Finally, a task manager and pipeline were implemented to improve efficiency.

Awards Won: