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Molecular karyotypes: SVA tools for junction detection and copy number variation of genome-wide chromosomal rearrangements by mate-pair sequencing (MPseq)

We have developed MPseq all long-insert next-generation sequencing approach for the detection of genomic structural variants. SVA tools is a set of algorithms to detect both chromosomal rearrangements and large (>10kb) copy number variants (CNVs) in genome-wide MPseq data. SVA tools can also predict disrupted genes and gene fusions and characterize the genomic architecture of complex rearrangements. SVA tools with MPseq provides comprehensive and accurate whole-genome junction detection with improved breakpoint resolution, compared to karyotype FISH and CMA combined. Copy number variation (CNV) is a common form of structural variation detected in aberrant human genomes, such as those observed in cancer. Cytogenetic techniques like chromosomal microarray (CMA) are widely used in analyzing these structural variations. An algorithm will also be presented, capable of performing copy number analysis from mate-pair sequencing (MPseq) data. The algorithm uses a step-wise procedure involving normalization, segmentation and classification of the sequencing data. The segmentation technique is novel in that it is the first technique to combine both read depth and discordant mate-pair reads to increase the sensitivity and resolution of CNV calls. This allows for the classification step to accurately calculate copy number.

Speaker Biography

George Vasmatzis is an Associate Professor in the Department of Molecular Medicine and a Member of the Mayo Clinic Cancer Center, as well as the Co-Director of the Biomarker Discovery Program, within the Center for Individualized Medicine. He is also the Founder of a software company called WholeGenome. His research program consists of bioinformatics specialists, molecular biologists, epidemiologists and pathologists. This team has demonstrated success in discovery and translation of several biomarkers as well as developing evidence-based models that should help clinicians stratify (cancer) patients in order to provide each individual with the appropriate care. With the recent advances in Next Generation Sequencing (NGS) technologies, his laboratory has been engaging in massive sequencing to scan the genome of cancer cells for abnormalities that can be used for clinical purposes such as diagnosis and stratification of patients for optimal treatment. He has published papers in Journal of Clinical Oncology, Cancer Research and BLOOD, further demonstrate our discovery, validation and translation capabilities. Recently, Mayo Clinic has launched a whole genome mate-pair sequencing test that was primarily developed by his program.

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