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A Pipeline for Detecting Copy Number Variation from Whole Exome Sequencing Using Comprehensive Tools

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Abstract : Copy number variations (CNVs) have played an important role in many kinds of human diseases, such as Autism, Schizophrenia and a number of cancers. Many diseases are found in genome coding regions and whole exome sequencing (WES) is a cost-effective and powerful technology in detecting variants that are enriched in exons and have potential applications in clinical setting. Although several algorithms have been developed to detect CNVs using WES and compared with other algorithms for finding the most suitable methods using their own samples, there were not consistent datasets across most of algorithms to evaluate the ability of CNV detection. On the other hand, most of algorithms is using command line interface that may greatly limit the analysis capability of many laboratories. We create a series of simulated WES datasets from UCSC hg19 chromosome 22, and then evaluate the CNV detective ability of 19 algorithms from OMICtools database using our simulated WES datasets. We compute the sensitivity, specificity and accuracy in each algorithm for validation of the exome-derived CNVs. After comparison of 19 algorithms from OMICtools database, we construct a platform to install all of the algorithms in a virtual machine like VirtualBox which can be established conveniently in local computers, and then create a simple script that can be easily to use for detecting CNVs using algorithms selected by users. We also build a table to elaborate on many kinds of events, such as input requirement, CNV detective ability, for all of the algorithms that can provide users a specification to choose optimum algorithms.

Keywords : whole exome sequencing, copy number variations, omictools, pipeline
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