qBIO Summer School abstract

Comparison of tools for genetic variant detection from exome sequencing data

Justyna Mika, Wojciech Łabaj, Kinga Leszczorz, Katarzyna Sieradzka, Andrzej Polański

Sequencing of matched tumour and normal sample pairs is nowadays a standard procedure used for detecting somatic mutations and copy number variations. There is a lot of algorithms design for both mentioned issues.

The aim of the following study was to implement and compare different tools for genetic variant detection from exome sequencing data. The study focused on somatic alterations and copy number variations detection. The data consisted of 5 sample pairs of thyroid carcinoma and normal control downloaded from The Cancer Genome Atlas database. Following tools were used for somatic variant detection: Mutect1, Mutect2, VarScan2 and SomaticSniper. Common mutations were searched and similarity of results was checked. Interestingly only few mutations were common for all of the tools. The biggest similarity of detected mutations was observed between VarScan2 and Mutect1.

In order to detect copy number variation heuristic based VarScan2 and probability based ExomeCNV algorithms were ran and compared. Both tools require sample coverage input, which was calculated regarding the newest reference genome GRCh38. Common variations, ie. deletions and amplifications, were searched between the tools. As before, most of the CNV regions were unique for each tool with only a little part being common. More CNV regions were detected by ExomeCNV when compared to VarScan2.

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