CytoMeth – a tool to perform DNA methylation analysis of bisulfite-converted Next Generation Sequencing data

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DNA methylation [5-methylcytosine (5mC)], currently one of the best-investigated epigenetic modifications, participates in epigenetic regulation of gene expression, genes sustained inactivation e.g. X chromosome, genomic imprinting, cell development and differentiation. 5mC is also associated with aging, disease-related processes, and higher mutation rate being the hot spot of 5mC to thymine transition. There are several methods to determine the methylation level of DNA samples in a single base-pair resolution. Up to now, bisulfite conversion followed by sequencing is considered the "golden standard" in DNA methylation studies. Our aim was to develop bioinformatic tool enabling fast and transparent rough bisulfite-sequenced data processing. First of all, we compiled a set of open source software listed in Roche's pipeline to automate multistep analysis of each sample: from rough FASTQ data to set of beta values and corresponding set of conclusive figures presenting DNA methylation variability across samples and various genomic regions. Our CytoMeth GitHub project performs simple automatic installation that downloads and installs all needed open source tools and reference files on the user's machine. CytoMeth is a set of R/python/shell scripts but can be easily started from command line by non-programmers users. The automatic batch processing of input data is also transparent and based on default set of parameters recommended by Roche. However some more sophisticated users can configure the most crucial processing steps and tune up some parameters. Additionally, we provide the example study of over 20 glioma samples that have been evaluated in the context of their quality and DNA methylation level variability by CytoMeth. Supported by the NCN grant 2015/16/W/NZ2/00314.

(Author(s) will also present a poster)
