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Mapdecode: inventory and benchmark of read mapping tools

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Paper 115 (abstract only)

Title:	Mapdecode : inventory and benchmark of read mapping tools
Track:	A: Sequencing and sequence analysis for genomics
Author	benchmark
keywords:	NGS mapping
Abstract:	<p>New sequencing technologies are able to produce enormous amounts of data, up to a billion reads per run. The first step of many analyses of these data, for instance the study of gene differential expression (RNA-seq), the study of gene regulation (ChIP-seq, Methyl-seq), the search for genomic variants (SNPs, chromosomal rearrangements) to name but a few, starts with mapping the reads on the reference genome. Over the last seven years, many mapping methods have been proposed to efficiently cope with the avalanche of data produced by the new generation sequencing (NGS) technologies.</p> <p>There exist exact algorithms for mapping reads on reference genomes, with or without indels but these algorithms are far too slow to be used with NGS data and large genomes. Therefore, most mapping methods implement heuristics that provide a trade-off between speed and accuracy. This trade-off often leads to the development of complex software with many ad-hoc options whose effects on the mapping results are usually difficult to predict beforehand.</p> <p>To help users choosing a particular mapping program that best suits their needs a number of benchmarks have been recently published that differs in the mapping tools they consider and in their methodology for carrying out the benchmarks (criteria for evaluating the mapping results, data used, etc.). In this work, we extend the work done in Schbath et al., 2012 by focusing, more specifically, on paired reads and extending the test cases (longer reads, consideration of indels, etc.)</p> <p>For the purpose of this study, we first compiled an inventory of 93 published mapping tools. However, only 25 of those tools seem actively maintained and have been updated since March 2013.</p> <p>From this list, we evaluated the performance of seven mapping tools on simulated datasets. We generated 3 different datasets from the human genome. The first one contains 10 millions 40 bp reads, the second one, contains 10 millions 100 bp reads. The last one contains 5 millions 2x100 bp paired reads. From these 3 datasets we then created further datasets with 1, 2 or 3 mismatches per reads. For the long and paired datasets, we also created a dataset with an insertion of three consecutive random nucleotides and a dataset with a deletion of three consecutive nucleotides. Finally, we generated a more realistic dataset by using a software that can simulate reads according to errors profiles observed in real datasets (we used bacterial data sequenced with an Illumina HiSeq2000). All the mapping tools were evaluated for correctness of mapping against these datasets.</p> <p>The inventory of the mapping tools and the benchmark results for the one tested are available on the Mapdecode website (http://mapdecode.genouest.org).</p> <p>This work is supported by the "France Génomique" project (ANR-10-INBS-0009)</p>

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Reviews

Review 1

Reviewer's confidence:

5: (expert)

Review:

The paper deals with the comparison of NGS mapping tools
It perfectly fit with ECCB program