Parallelization, Scalability, and Reproducibility in Next-Generation Sequencing Analysis

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Reviews

This is the best book i have read until now. It can be filled with knowledge and wisdom Once you begin to read the book, it is extremely difficult to leave it before concluding.

(Nadia Konopelski)

PARALLELIZATION, SCALABILITY, AND REPRODUCIBILITY IN NEXT-GENERATION SEQUENCING ANALYSIS

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Book Condition: New. Publisher/Verlag: epubli | This PhD thesis provides novel solutions to major topics within the analysis of next-generation sequencing data, focusing on parallelization, scalability and reproducibility. | The analysis of next-generation sequencing (NGS) data is a major topic in bioinfor-matics: short reads obtained from DNA, the molecule encoding the genome of livingorganisms, are processed to provide insight into biological or medical questions. Thisthesis provides novel solutions to major topics within the analysis of NGS data, focusingon parallelization, scalability and reproducibility.The read mapping problem is to find the origin of the short reads within a given referencegenome. We contribute the q-group index, a novel data structure for read mapping withparticularly small memory footprint. The q-group index comes with massively parallelbuild and query algorithms targeted towards modern graphics processing units (GPUs).On top, the read mapping software PEANUT is presented, which outperforms state ofthe art read mappers in speed while maintaining their accuracy.The variant calling problem is to infer (i.e., call) genetic variants of individuals comparedto a reference genome using mapped reads. It is usually solved in a Bayesian way.In this work, we show how to integrate filtering of variants into the calling with analgebraic approach and provide an intuitive solution for controlling the false discoveryrate along with solving other challenges of variant calling like scaling with a growingset of biological samples.Depending on the research question, the analysis of NGS data entails many other steps,typically involving diverse tools, data transformations and aggregation of results. Thesesteps can be orchestrated by workflow management. We present the general purposeworkflow system Snakemake, which provides an easy to read domain-specific languagefor defining and documenting workflows. Snakemake provides an execution environmentthat allows to scale a workflow to available resources, including parallelization acrossCPU cores or cluster nodes, restricting memory usage or the number of availablecoprocessors like GPUs. |...

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