

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



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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 bioinformatics: short reads obtained from DNA, the molecule encoding the genome of living organisms, are processed to provide insight into biological or medical questions. This thesis provides novel solutions to major topics within the analysis of NGS data, focusing on parallelization, scalability and reproducibility. The read mapping problem is to find the origin of the short reads within a given reference genome. We contribute the q-group index, a novel data structure for read mapping with particularly small memory footprint. The q-group index comes with massively parallel build and query algorithms targeted towards modern graphics processing units (GPUs). On top, the read mapping software PEANUT is presented, which outperforms state of the art read mappers in speed while maintaining their accuracy. The variant calling problem is to infer (i.e., call) genetic variants of individuals compared to 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 an algebraic approach and provide an intuitive solution for controlling the false discovery rate along with solving other challenges of variant calling like scaling with a growing set 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. These steps can be orchestrated by workflow management. We present the general purpose workflow system Snakemake, which provides an easy to read domain-specific language for defining and documenting workflows. Snakemake provides an execution environment that allows to scale a workflow to available resources, including parallelization across CPU cores or cluster nodes, restricting memory usage or the number of available coprocessors like GPUs. |...

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