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Next Generation Sequencing: where big data and high-performance computing meet

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The advent and evolution of next generation sequencing (NGS) has considerably impacted genomic research, including precision medicine. High-throughput technology currently allows for the generation of billions of short DNA or RNA sequence reads within a matter of hours. This becomes extremely important in the case of genetic disorders where rapid and inexpensive access to a patient's individual genomic sequence is imperative and enables target variant identification. NGS technologies results in the generation of large data sets which require extensive bioinformatic and computational resources. Computational life sciences therefore relies on the implementation of well-structured data analysis pipelines as well as high-performance computing (HPC) for large-scale applications. Here, we report the sequencing of the first six whole human genomes in South Africa and the processing of the data in collaboration with the Centre for High Performance Computing (CHPC). Efficient parallel and distributed implementations of common time-consuming NGS algorithms on modern computational infrastructures are imperative. The latter becomes pivotal as NGS will continue to transcend from research labs to clinical applications in the near future.

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