Abstract

Massive parallel sequencing (MPS) data analysis tasks are often computationally demanding and their execution time would take too long using standard computing machines. Thus there is a need for parallelization of this tasks and ability to execute them on a sufficiently powerful computing machines. In the first chapter we describe a newly created platform for resequencing analysis of MPS data - MOLDIMED and novel annotation tool, which is ready to deploy on HPC infrastructure.

The second chapter describes MPS approaches in Diamond-Blackfan anaemia (DBA), which is predominantly underlined by mutations in genes encoding ribosomal proteins (RP); however, its etiology remains unexplained in approximately 25% of patients. We performed panel sequencing of all ribosomal genes in DBA patient without previously known molecular pathology. A novel heterozygous *RPS7* mutation coding RPS7 p.V134F was found in one female patient and subsequently confirmed in two asymptomatic family members, in whom mild anemia were detected on further examination. Subsequently, we performed whole transcriptome analysis in all family members and patient with RPS7 mutation in comparison with healthy control group and with DBA patients with known mutation in *RPS19.* We observed dysregulation mainly in signal pathways of translation, cell cycle, rRNA processing and in genes of immune system.

The third chapter demonstrate MPS deployment in whole transcriptome analysis of novel transgenic barley lines overexpressing cytokinin dehydrogenase 1 gene from Arabidopsis. These lines showed drought-tolerant phenotype mainly due to alteration of root architecture and stronger lignification of root tissue. In optimal conditions of revitalized leaves, comparative transcriptomic analysis displayed up-regulation of genes encoding enzymes and proteins involved in photosynthesis, and especially those encoded by the chloroplast genome.