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Multiallelic copy number variations of *MSH2-AT3G18530-AT3G18535* genes in *Arabidopsis thaliana*: In-depth analysis of genotypes, mechanisms leading to their formation and the functional implications

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Copy number variations (CNVs) are intraspecies duplications/deletions of large DNA segments (>1 kb). Because of the ability to alter the gene structure or copy number CNVs may influence gene expression and by the dosage effect, the interaction of gene products within the protein and metabolic networks. Such alterations may have no phenotypic effect, but often they account for adaptive or maladaptive traits and contribute to genome evolution or phenotypic variation. We adopted two experimental techniques, multiplex ligation-dependent probe amplification and droplet digital PCR to measure the copy number of specific sequences in Arabidopsis thaliana. With those approaches we performed an in-depth analysis of a complex multiallelic CNV encompassing 3 neighboring genes (*MSH2, AT3G18530* and *AT3G18535*). We evaluated the gene copy numbers in a large population of 189 A. thaliana ecotypes and analyzed the CNV breakpoints and the flanking regions. Based on the sequence data and eco-geographical distribution of *AT3G18530-AT3G18535* deletion and duplication genotypes we created a model of non-allelic homologous recombination (NAHR) mediated by low copy repeats flanking the two genes. We propose that the observed dupl-2 and del-2 genotypes originate from reciprocal products of interchromatidial/interchromosomal NAHR. Furthermore, we performed gene expression studies to evaluate the possible consequences of CNV on the evolution and functionality of *MSH2, AT3G18530* and *AT3G18535* genes. In light of those data, the perspectives of utilizing natural CNV phenomenon for functional gene analysis will be discussed.

Biography

Agnieszka Zmienko has completed her PhD in 2006. She works as a Research Scientist in the Institute of Bioorganic Chemistry, Polish Academy of Sciences and as an Assistant Professor in the Institute of Computing Science of Poznan University of Technology. She is a team Member of the ECBiG Regional Center which provides facilities, tools and databases for multi-level studies of biological systems. She has focused on plant genomics and transcriptomics and co-authored 15 papers in this field.

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