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Methods to determine copy number variation in Brassica species

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Many genes in diploid species Brassica rapa and B. oleracea are present in two or more copies as a result of ancestral polyploidy events, and therefore in four or more copies in recent allopolyploid species B. napus. Some gene families tend to show rapid reversion to only a single working gene copy after polyploidy events, while other gene families show retention and even expansion of gene copy number after polyploidization. Additionally, structural rearrangements such as duplications and deletions of particular chromosome segments resulting from non-homologous recombination events are highly abundant in Brassica species. In B. napus, “balanced” translocations (where a segment of an A-genome chromosome is replaced by a segment of a C-genome chromosome, or vice versa) are now known to be extremely prevalent, and significant presence-absence variation for single gene copies has been detected in the diploid species. These copy number variants can have severe consequences on the underlying genes, and putatively hence on plant phenotypes. However, not a lot of supportive data has been provided demonstrating the influence of gene copy number variation on traits, either in Brassica or in other crops. This is partly due to lack of established methods and widespread understanding of how to call and integrate copy number data into genetic analyses. Here we show some easy-to-implement bioinformatic pipelines for copy number variation calling from next-generation sequencing data as well as from SNP array genotyping data. Moreover, we present examples for the use of such results in studying the phenotypic effects of copy number variants in Brassica. In future, generation and investigation of additional data linking traits to copy number variants in polyploids may shed light on the impact of copy number variants on crop phenotypes.