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PHYSICAL INTERACTION BETWEEN CHROMATIN REGIONS AND STRUCTURAL VARIATION OCCURRENCE IN *VITIS VINIFERA*

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Sequence variation among individuals of the same species is partly due to large insertions or deletions, known as structural variants (SV), that can derive either from the movement of transposable elements or from defective repair of double strand breaks through Non Homologous End Joining (NHEJ) or unequal Homologous Recombination (HR). Copy Number Variants (CNV) are one of the most common types of SV, in which different individuals possess the same DNA sequence in a different number of copies. CNV can affect the structure and the regulation of genes, giving rise to either disadvantageous or favorable traits (such as resistance to stress, pathogens or chemicals).

SV formation involves changes in chromosome structure, creating a junction between two formerly separated DNA sequences. Thus, the physical contact between the involved regions is a key point in the investigation of CNV occurrence.

We use grapevine (*Vitis vinifera*), an economically significant crop, to investigate the role of chromatin three-dimensional organization in SV formation. We used Hi-C data from Pinot noir and Rkatsiteli, two grapevine varieties, to compare interaction frequencies across the borders of regions in which a CNV was present in one variety but absent in the other one (we called such regions as CNV and CNV-prone, respectively). We focused exclusively on regions where one of the varieties showed a deletion in comparison to the reference sequence.

We find that both CNV and CNV-prone regions have significantly higher interaction levels across their borders than do background regions. Moreover, there was no significant difference in interaction levels across the borders of CNV and CNV-prone regions . A k-mers analysis of both the CNV and background borders confirmed that these results were not due to mapping bias. We show that the presence of CNV can be visually confirmed in the Hi-C interaction matrix heat map, with increased signal seen across the CNV borders with respect to the surrounding regions. This signal increase is also observed in the unaffected variety.

While these results were to be expected for CNV regions because the deletion brings in physical proximity regions that in the reference sequence are distant apart from one another, they were not expected for those chromosomes where the CNV is not present, pointing to a physical interaction as a prerequisite for the occurrence of the deletion.

Our results provide evidence that the three-dimensional organization of a genome can have a dramatic effect not only on the functioning of the genome but also on its structure and variation.