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OPTICAL MAPPING FOR DE-NOVO GENOME ASSEMBLY AND PLANT COMPARATIVE GENOMICS

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Despite the enormous improvements accomplished in the sequencing methods over the last ten years, the next generation technologies are still characterized by consistent limits that hamper comprehensive genome analysis. Short reads sequencing approaches, such as Illumina, are indeed unable to properly resolve low complexity or highly homologous regions, to accurately detect large structural variants and to determine haplotype phasing.

Optical mapping analyses individual long (100s Kb) DNA molecules and produces fingerprints of DNA sequences in order to construct genome-wide maps. This allows a map to resolve complicated genome regions, including copy number variations and potentially homoeologous segments from polyploid genomes, more efficiently and unambiguously than other genomic technologies. Similarly, also low complexity part of the genome as repetitive regions, of which plants are particularly rich, can be accurately characterized.

We will present an array of our results demonstrating the ability of optical mapping to accurately reconstruct chromosomal pieces during *de-novo* genome assembly and to identify relatively large structural variants in genetic diversity studies. Such examples include the *de-novo* assembly of the *Solanum melongena* and scaffolding of the highly heterozygous *Vitis vinifera* cv. Cabernet-Sauvignon. Integration of optical maps to the draft assemblies allowed to consistently improve contig N50 from 640Kb to 2Mb and 1.4Mb to 5.7Mb, respectively, and to reduce the number of sequences up to 50 folds. In addition to improving the contiguity of draft reference genome assemblies, optical mapping served to identify assembly errors such as false joins, inversions, and translocations within the original assembly. In addition, using *Vitis vinifera* cv. Nebbiolo as test study, we will present the potential of optical mapping in identifying clone-specific structural variations, a 'hidden' diversity largely understudied so far but heavily contributing to phenotypic diversity. Direct comparison of maps unravelled the presence of several deletions/insertions present in a reciprocal manner in the two clones, but previously unrecognized using Illumina data.

Overall these results demonstrate that recent technical improvements in optical mapping, as well as reduction of error rate and costs, can potentially increase the use of this technology and thus facilitate our understanding of genome structural diversity and heritable agronomic traits.