

## **RESEQUENCING OF A SECOND GRAPEVINE GENOME REVEALS FREQUENT SNP AND STRUCTURAL VARIATION**

CATTONARO F.\*, VENDRAMIN V.\*\*\*, SWAMINATHAN S.\*\*\*, POLICRITI A.\*\*\*\*, MORGANTE M.\*\*\*, THE FRENCH-ITALIAN PUBLIC CONSORTIUM FOR GRAPEVINE GENOME CHARACTERIZATION

\*) Istituto di Genomica Applicata, Via Linussio 51, 33100 Udine (Italy)

\*\*) Dipartimento di Scienze Agrarie e Ambientali, Università di Udine, Via delle Scienze 208, 33100 Udine (Italy)

\*\*\*) Dipartimento di Matematica e Informatica, Università degli Studi di Udine, Via delle Scienze 208, 33100 Udine (Italy)

*re-sequencing, grapevine, genome, nucleotide variation, structural variation*

The grapevine (*Vitis vinifera*) is economically the principal fruit plant in the world. Using a highly homozygous genotype, PN40024, derived from selfing of Pinot Noir a high quality draft sequence assembly was produced at 12X coverage with a whole genome shotgun approach. 90% of the 12X genome sequence assembly that totals approximately 485 Mbp is anchored in 192 pieces on the 19 chromosomes.

The re-sequencing of a second genotype, the Italian cultivar Tocai friulano, allowed the identification of single nucleotide polymorphisms and structural variations (SVs) between the two genotypes. This analysis was achieved by mapping over the reference sequence about 595.000 Tocai mate-pair Sanger reads corresponding to a sequence coverage of 0.9X and obtained from inserts of average size of 3.8 Kb. SNPs were identified between the two genotypes with a frequency of one every 118 bp and a total of more than 2 million SNPs in non repetitive regions. Structural variations were identified as significant size differences between the Tocai inserts and the corresponding regions of the reference sequence as identified by the pair-end reads. We observe several thousands structural variants between Tocai and the reference sequence that could frequently be attributed to the presence of transposable elements in the PN40024 sequence that are missing in Tocai or viceversa and could be attributed to recent insertions into either PN40024 or Tocai. LINE elements within introns appear to be especially polymorphic between the two genotypes. The computational analyses are experimentally validated by gel sizing of Tocai inserts and PCR analysis of transposable element insertion sites. The finding of SVs between two grapevine varieties confirms the dynamic nature of the plant genomes, revealing that transposable element activity is an important source for the generation of genetic diversity.