ABO GENOTYPING BY SNAPSHOT MINISEQUENCING

C. Franchi[1], D. Podini[2], A. Berti[1], F. Fiorenrine[2], G. Largo[1]
[1]Reparto Carabinieri Investigazioni Scientifiche di Roma, Italy
[2]GENOMA Servizio Analisi del DNA, Italy

Since Yamamoto first sequenced, in 1990, the c-DNA of the I^A allele, many authors have developed new molecular methods to determine the ABO blood groups. One of the most popular methods is based on the PCR amplification of two regions containing allele specific nucleotide at positions 261 and 703 respectively, followed by a site specific restriction performed by two different enzymes. The interpretation of the allele specific patterns obtained by the enzyme digestion and gel separation provided a sensitive tool to genotype the ABO locus. The aim of our study is to develop a new method based on PCR and mini-sequencing by multiplex SnaPshot Technology. We considered three allele specific SNPs at position 251, 526 and 703 that are widely conserved and characterized among the population. The SNP at base 526 has been used to distinguish the rare 0^2 allele from the A allele. We successfully test the method with a control population.

The method can be extended to evaluate the presence of A and B rare subgroups by mini-sequencing other polymorphic position.

ABO mini-sequencing provides an important tool both in diagnostic and in forensic investigation.