SEQUENCE ANALYSIS OF ABO GENE IN KOREANS FOR ITS APPLICATION TO HUMAN IDENTIFICATION

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ABO blood system is widely applied to paternity test, forensic investigation as well as blood transfusion. Genotyping of ABO gene could be more informative and valuable than serological typing. This study intends to evaluate usefulness of ABO gene in forensic identification through population study. 100 unrelated Korean individuals were selected. DNA was extracted from sample and PCR and direct sequencing was performed to analyze the full sequence of exon 6 and exon 7 in the ABO gene.

The polymorphic nucleotide positions of ABO gene are 216 and 297 at exon 6 (2 position), and 467, 526, 579, 646, 657, 681, 703, 771, 796, 803, 829, and 930 at exon 7 (12 positions) in Koreans. Among, the Korean population, 18 ABO genotypes and 7 alleles were observed. O01 was in the most frequent (27.6%), followed by A102 (22.0%), B101 (22.0%), and O02 (21.0%) in that order. The observed heterozygosity and the expected heterozygosity of the ABO gene are 0.670 and 0.784 respectively. The polymorphism information content (PIC) is 0.744. The power of discrimination (PD) and the mean exclusion chance (MEC) are calculated to be 0.915 and 0.576.

Since the information of blood type can be obtained more easily than that of STR type, the determination of ABO genotype by sequencing may be useful in human identification such as finding an individual in relation to a criminal case, paternity test, and confirming possible relationships between family members.