F811

Applicability of Short Tandem Repeat Genotyping by Fluorescent Detection for the Individual Identification

선 문 숙*, 강 순 자#, 최 상 규
국립과학수사연구소 유전자분석실, #이화여자대학교 사범대학 과학교육학과

The polymorphisms of genetic markers based on the variable number of repeat units are displaying considerable polymorphisms and providing the complementary information. In the present study, twenty-one short tandem repeat (STR) markers based on (dC-dA)n(dG-dT)n dinucleotide repeat sequences were analyzed for their polymorphisms in the Korean population using fluorescently labeled primers and automated sequencer with Genescanner software. The potential usefulness of these polymorphisms for the determination of individual identification was calculated from the allele frequencies of twenty-one STR markers. The most common genotype frequency combined of twenty-one markers was $6.09X10^{-11}$ and the least common genotype frequency combined of twenty-one markers was $7.88X10^{-51}$. And the probability of a random match (PM) combined of the twenty-one markers was very low with $7.48X10^{-24}$. The discrimination power was high showing above 0.8 with the exception of D20S115 (0.737) and was the highest showing 0.954 in D12S85 and D20S117. The STR genotyping by the fluorescence-based system significantly facilitates large-scale databasing for the individual identification. Therefore we can obtain the high discrimination power for the individual identification within a short time frame.

F812

Detection of Factor IX Gene Mutations in Korean Hemophilia B Patients by Polymerase Chain Reaction-Single Strand Conformation Polymorphism(PCR-SSCP)

김봉윤*, 조율희[1], 서동상
성균관대학교 생명자원과학대학 유전공학과, [1]한양대학교 의과대학 유전학교실

Hemophila B is an X-linked bleeding diease, resulting from sequence alterations of the coagulant factor IX gene. Hemophilia B is caused by a variety of mutations , which can be found in the whole coding regions . We screened mutations of factor IX gene in 8 Korean hemophilia B patients. Amplifications of eight exons, promoter region, and their intron boundaries, were performed with polymerase chain reactions (PCRs) and the PCR products were analyzed by single strand conformation polymoyphism (SSCP) with PhastSystem™ employing silver staining protocol. PCR-SSCP is a powerful tecnique that can be used to detect base subsitutions. Now this tecnique was used expendingly for molecular biology and medical part. Here, we have detected six mutations of an altered migration pattern of single strand DNA. Each patients have different mutations in exon B, G and H. And now, we are identifying six different hemophilia B mutations by direct sequencing.