frequencies of *CYP2E1*C/*C*, *CYP2E1*C/*D* and *CYP2E1*D/*D* of *Dra*I RFLP in intron 6 were 3.2%, 25.8% and 71.0% respectively; the allele frequency of *CYP2E1*D* was 0.839;and the heterozygosities of *Pst*I/*Rsa*I and the *Dra*I SNPs of *CYP2E1* were both at 0.258. The test for Hardy-Weingberg equilibrium showed no significant deviation from our expectations. The allele frequencies in the 5′ flanking region and intron 6 of *CYP2E1* among Koreans were proven to be similar to those of other Oreintals, and the allele frequency of *CYP2E1*c2* in the 5′ flanking region in Caucasians is lower than that of Orientals including Koreans.

F803

Three Tetranucleotide Repeat Polymorphism at D16S539, D7S820 and D13S317 Loci in Korean Population

Byung-Sub Park* and Gwang-Sook Ahn
Dept. of Biology, College of Natural Science, Taejon University, Taejon 300-716

The markers of microsatellite are polymorphic and are mostly based on loci with tetranucleotide repeats, each locus commonly having between 5 and 15 alleles. Allele and genotype frequencies of D16S539, D7S820 and D13S317 were analyzed using PCR and denaturing polyacrylamide gel electrophoresis followed by silver staining for visualization. DNA extracts were obtained from about 80 unrelated Koreans. The frequency of allele 11 at D16S539 locus in Korean was the lowest among some racial populations. The frequency of allele 11 at D13S317 in Korean was similar to that of Asian populations. Comparing the results of present study with other racial populations, allele frequencies of three loci were different according to the loci. This study confirms that the irregularity in distribution of microsatellite alleles in different populations with the predominance of two or three alleles on these three investigated microsatellite loci.

F804

A Dinucleotide Repeat Polymorphism at the Calcium Sensing Receptor (CASR) Locus in Korean Population

Hye-Ja Kang* and Gwang-Sook Ahn
Dept. of Biology, Taejon University, Taejon 300-716

Calcium sensing receptor (CASR) in parathyroid gland regulates calcium homeostasis by sensing decreases in extracellular calcium levels and affecting an increase in secretion of parathyroid hormone and calcium reabsorption in the kidney. We analyzed a dinucleotide repeat polymorphism at CASR locus in normal individuals and osteoporosis patients by polymerase chain reaction and polyacrylamide gel electrophoresis. Eight alleles were detected and allele frequencies of A3 and A9 were 0.46 and 0.49 in the normal individuals. In osteoporosis patients, allele frequencies of A3 and A9 were 0.48 and 0.46, respectively. The observed heterozygosities were 0.552 in normal sample and 0.503 in osteoporosis one. Allele distribution and frequencies of normal group were similar to those of osteoporosis one in Koreans. Comparing this result with a Japanese population, some differences were found in the frequencies of main alleles, A3 (0.32) and A9 (0.57). It is concluded that there is significant difference in the allele frequency for normal groups between Korean and Japanese populations.

F805

Factor V Leiden (G1691A) and G20210A Prothrombin Variant by Multiplex PCR-RFLP

Eun-Hee Kim* and Kyung-Sook Park
Dept. of Biology, Sungshin Women's University, Seoul 136-742

A factor V mutation G1691A (FV Leiden) was found to destroy the vital cleavage site R506Q of the factor V, which is necessary for factor V deactivation, and thus causing thrombophilia. Mutation in prothrombin (factor II, G20210A) was discovered to be often associated with mutation in factor V (G1691A), thereby indicating an additional risk for thrombosis elevated to factor II levels. This experiment investigates the prevalence of these two mutations among two hundred nineteen unrelated Korean subjects using multiplex PCR-RFLP. Data revealed two heterozygous subjects for the factor V Leiden without prothrombin G20210A mutation. Both variations yield significant differences in relation to ethnic origins: the FV Leiden occurs at a frequency rate of 2.3% among Koreans, 11.0% among Swedish, 6.1% among Swiss people, and 19.0% among Jordanian people. Prothrombin (G20210A) mutation occurs at 0% frequency rate among Koreans, 1.2% among English, and 1.1% among Swiss people.

F806

Identification of the Mutation in the Patient with Dihydrolipoamide Dehydrogenase (E3) Deficiency

Moosik Kwon*
Department of Genetic Engineering, Sungkyukwan University, Suwon 440-746

The mammalian pyruvate dehydrogenase complex (PDC) plays a pivotal role in the decarboxylation of pyruvate derived from glucose and amino acids to acetyl-CoA, which is further utilized for energy production or for biosynthetic processes. This enzyme complex contains multiple copies of components including pyruvate dehydrogenase (E1), dihydrolipoamide acetyltransferase (E2), dihydrolipoamide dehydrogenase (E3), E1-kinase, phospho-E1 phosphatase, and E3-binding protein. Mammalian E3 is a common flavoprotein component of a-keto acid dehydrogenase complexes and catalyzes the reoxidation of dihydrolipoyl group; two cofactors, NAD and FAD, are utilized for the reoxidation. E3 deficiency leads to lactic acidosis, increased concentrations of branched-chain amino acids in the plasma and increased urinary excretion of a-keto acids. E3 deficiency also causes neurological degeneration due to the sensitivity of the central nervous system to defects in oxidative metabolism. In this study, E3 mutant cDNA from a patient showing PDC deficiency was amplified by RT-PCR and subcloned into pBlusscript SK- for further analysis. The clone was sequenced for the identification of mutation(s). Two substitutions were found. One was a single point mutation of the interface domain (CCT →CCC) which encodes Proline, but there was no change in amino acid sequence. The other was missense mutation due to substitution of thymine for guanine (GGT→ TGT), causing an Gly→Cys substitution at amino acid 229 of the mature protein. This substitution may interfere with the proper folding of E3 dimer by forming disulfide bridge with the other cystein(s) in NAD-binding domain. This mutation, therefore, causing diminished production of E3 protein and a possible structural change in the E3 dimer, probably leads to loss of activity.

F807

Construction of ESTs and Identification of a Stress-induced Gene in Chinese Cabbage

Moosik Kwon*
Department of Genetic Engineering, Sungkyunkwan University, Suwon 440-746