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Usefulness of dinucleotide polymorphism markers in genetic analysis of Duchenne muscular dystrophy in Korean

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Dinucleotide polymorphisms are short tandem repeat sequences that can be used as probes for haplotype analysis in Duchenne's muscular dystrophy (DMD). There are approximately a total of 50,000 to 100,000 such loci in the human genome, and they are highly informative due to the variability of allele lengths at these loci. Primers can be designed to amplify across such repeats located in the dystrophin gene to provide diagnostic information when RFLP analysis is uninformative. We report the usefulness of six such loci for analysis of DMD families in Korean. Four (CA)n repeats is located in introns 44, 45, 49, 50 of the dystrophin gene while 5'DYS-II marker is located upstream to the transcriptional start site for the brain dystrophin promoter and 3'CA marker is identified in the 3'UTR of the gene. As for 5'DYS-II and four (CA)n repeats, the numbers of alleles in our cases were less than in Caucasians, and the heterozygosities were lower than in Caucasians. However, the 3'CA polymorphisms showed almost the same frequencies and heterozygosities as in Caucasians. In addtion, all of female cases showed a heterozygous pattern for at least one locus, with the combination of the six markers. Therefore, they were useful for linkage analysis, identification of deletion mutations, confirmation of paternity and mapping of gene recombination.

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Identification of Differrentially Expressed Genes in Human Normal Liver Cell Line between Under Normoxic and Hypoxic Condition by DD-PCR

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Hypoxia occurs in various pathological conditions and this state in cells or tissues often leads to neovascularization. Especially, solid tumor growth, e.g., hepatocellular carinoma, is much related to angiogenesis and hypoxia is thought to be a main signal in solid tomor angiogenesis.

To know the initial state of liver tumor, we investigated the contribution of hypoxic condition in normal liver using human normal liver cell line, Chang liver. To identify differentially expressed genes under normoxic (21% O_2 tension) or hypoxic (1% O_2 tension) condition, we used differential display of polymerase chain reaction (DD-PCR) technique using total RNA extracted from both of the condition. Five different polymerase chain reaction (PCR) primers were used to compare the gene expression patterns of cells under each condition.

Our results revealed several up-regulated and down-regulated partial cDNA fragment. We isolated 10 different kinds of cDNA fragments which showed marked differences in two conditions for additional analysis. The DNA sequence homology matching analysed using GenBank and SWISS-Prot data base revealed six clones showed high similarities to the already known sequences. Among 6 clones, 2 clones shows sequence similarities with *P. falciparum* topoisomerase I gene whereas other clones to human 18S ribosomal protein (HKE3) mRNA, human mRNA for protein involved in DNA double-strand break repair and H+/peptide cotransporter mRNA, respectively. Northern blot analysis using each clone as a probe, or RT-PCR confirmed that the differentially detected PCR products reflected the differential expression of each mRNA in each condition. These differentially detected genes might be related to liver tumorigenesis at early stage and will be a useful genes for analysing contribution of hypoxia to liver cancer.