F101 Single nucleotide polymorphisms and variable number of tandem repeat polymorphism of CYP2E1 in Koreans

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Cytochrome P450 2E1 (CYP2E1) is a toxicologically important enzyme with a high extent of interindividual variability in activity. Genetic polymorphism represents a major determinant for the interindividual variation of the CYP2E1 activities. We analyzed the six polymorphic sites to elucidate *CYP2E1* combined allele types, which are at -1293 G>C (*PstI*) and -1053 C>T (*RsaI*) of 5'-flanking region, 1132 G>A7632 (*HhaI*) of exon 2, 7732 T>A (*DraI*) of intron 6 and single strand-conformational polymorphism at position 10023 G>A (V389I) of exon 8 and one VNTR marker between -2178 and -1945 nucleotide position in 323 Koreans. The *-1293*G* (*PstI*), -1053*C (*RsaI*) and *6 (VNTR) alleles in 5'-flanking region were common alleles, and the allele frequencies were 77.2%, 77.2% and 79.3%, respectively. *-1293*G* and *-1053*C* polymorphisms in 5'-flanking region were shown in complete linkage. The *1132*G* in exon 2, *10023*1* in exon 8, and *7632*T* in intron 6 were 99.7%, 83.4% and 75.5%, respectively. No deviation from the expectation according to the Hardy-Weinberg equilibrium was found in six sites. In combination of six sites of *CYP2E1* polymorphisms, 28 genotypes were observed and *6*6,*G*G,*C*C,*G*G,*T*T, *1*1 (VNTR/-1293/-1053/1132/7632/10023, 30.3%) was found to constitute the majority.

F102 Functional roles and acceptor molecules of tyrosylprotein sulfotransferase-A in Caenorhabditis elegans.

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Tyrosine O-sulfation is a kind of post-translation modification of proteins in all eukaryotic organisms. Many proteins have been shown to contain tyrosine sulfate. Caenorhabditis elegans tyrosylprotein sulfotransferase (TPST-A) catalyzes the transfer of the sulfuryl group from 3\'-phosphoadenosine 5\'-phosphosulfate (PAPS) to tyrosine residue(s) within highly acidic motifs of polypeptides. Biochemical evidence indicates that the enzyme is a membrane-associated protein with a lumenally oriented active site localized in the trans-Golgi network. To gain insights regarding the biological function of TPST-A, we used the reverse genetic tool, RNA interference (RNAi), to \"knock out\" enzyme in C. elegans. RNAi was carried out in C. elegans. In a typical experiment >70% of RNAi animals were affected by various phenotypes. Specific phenotypes similar to pull in worm\'s belt in random site on body (>10% of animals) and some worms are stationary and finally dead in L2, L3, and L4 stages. Phenotypes also cause an arrest of growth usually at the molt from the third to the fourth larval stage (>40% of animals). Adults are dumpy worms with retained many eggs and larvae and finally dead (>20% of animals). These phenotypes indicate that TPST-A may catalyze tyrosine O-sulfation of a variety of protein substrates involved in diverse physiologic functions.

F103 Assessment of Substrate specificity of Hepatitis G Virus NS3 protease and turnip mosaic virus Nla protease by a Genetic Method

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The HGV NS3 protease is responsible for the cleavage of the HGV polyprotein at four

different locations for its replication. The TuMV NIa protease is also responsible for the cleavage of the TuMV polyprotein at seven different locations. To determine the substrate specificity of the NS3 and NIa proteases, amino acid sequences cleaved by the NS3 and NIa proteases were obtained from randomized sequence libraries by using a screening method referred to as GASP. Based on statistical analyses of the obtained cleavable sequences, consensus substrate sequences were deduced: Gln-Glu-Thr-Leu-Val▽Ser for HGV NS3 protease and Yaa-Val-Arg-His-Gln▽Ser for TuMV Nia protease, with Yaa being one of aliphatic amino acids. The relevance of this peptide as a cleavable substrate was further supported by molecular modeling of the HGV NS3 protease.

F104 Molecular Characterization of Blue Pigment Binding Protein-1 from Pieris rapae.

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We cloned and molecularly characterized a blue pigment binding protein-1 (BP-1) cDNA from cabbage white butterfly. The cDNA has a length of 474 bp coding for a 158-residue protein with a predicted molecular mass of 18,051 Da. The calculated isoelectric point is 8.2. Multiple alignment analysis of amino acid sequence revealed that BP-1 is most similar to bilin-binding protein (BBP) from *Pieris brassicae* (93%) followed by BBP of *Galleria mellonella* (48.7%). The BP-1 transcript was detected by Northern blot analysis in fat body and midgut.

F105 Mutations of Methyl-CpG binding protein 2 gene in Rett syndrome patients

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Rett syndrome (RTT [OMIM#312750]) is an X-linked dominant neurodevelopmental disorder affecting 1/10,000 - 15,000 females. RTT results from mutations in *MeCP2*, methyl-CpG binding protein 2, in 80% of patients. *MeCP2* gene spans 76kb in Xq28 and encodes two functional domains, a methyl-binding domain (MBD) and transcriptional repression domain (TRD). Authors have already reported the results of mutation screening of *MeCP2* in 20 Korean patients with RTT. In this presentation, we will report the newly identified 10 mutations including five missense mutations (D97Y, L100V, R133C, T158M, R306C), four nonsense mutations (R168X, R255X, R270X, R294X), and one frameshift mutation (a 41-bp deletion at 1157~1197). Two of these (D97Y and L100V) were novel mutations. We also established 21 lymphoblastoid cell lines from RTT patients. These cell lines will be used as valuable materials to ascertain whether *MeCP2* is related to the X chromosome inactivation, if any, what is its function.

F106 Comparative analysis of deleted region at 7q11.23 in WS and SVAS patients

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Williams syndrome (WS) is a complex developmental disorder with multisystem manifestations including supravalvular aortic