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**Start Codon Polymorphism in the Vitamin D Receptor is Associated with Colorectal Cancer**Mi Jung Woo<sup>P</sup>, Hee Jin Kim<sup>1</sup>, JungHyun Nam<sup>1</sup>, Kyung Sook Park<sup>C</sup>*Department of Biology, Sungshin Women's University, Seoul 136-742*

The expression of the nuclear vitamin D receptor (VDR) gene involved in regulating cell growth and proliferation may contribute to the development of the pathogenesis of colorectal cancer. The VDR has the start codon (FokI) polymorphism (ATG to ACG) in exon 2, the C allele at the second of two potential translation initiation sites, which is effective in transactivation of 1,25(OH)<sub>2</sub>D<sub>3</sub> signal, and a T1056C polymorphism (TaqI) in exon 9. This study evaluated the VDR germline mutation was responsible for colorectal cancer. Polymorphisms (FokI and TaqI) of the VDR gene were analyzed by the PCR-RFLP in 203 colorectal cancer and 222 controls. The \*C/\*C genotype frequency of the start codon polymorphism was significantly higher in colorectal cancer (\*C/\*C, 49.3%; \*C/\*T, 42.8%; \*T/\*T, 7.9%) than controls (\*C/\*C, 33.3%; \*C/\*T, 50.0%; \*T/\*T, 16.7%) (p=0.0009; OR=1.9; 95% CI=1.3-2.9) while the T1056C polymorphism was not different between colorectal cancer (\*T/\*T, 87.7%; \*T/\*C, 12.3%; \*C/\*C, 0.0%) and controls (\*T/\*T, 92.8%; \*T/\*C, 7.2%; \*C/\*C, 0.0%). The C allele (start codon), the short VDR transcript, interacts more efficiently binding to the basal transcription factor TF<sub>2</sub>B, and provides a mechanism for greater transactivation potency of the VDR gene. There is evidence that a high level of VDR expression is associated with prognosis of colorectal cancer. These results show that the VDR start codon polymorphism is associated with an increased risk for colorectal cancer.

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**Single Nucleotide Polymorphisms of the Matrix Metalloproteinase 1, 3, 7, 9 and 12 in Koreans**JungHyun Nam<sup>P</sup>, Hee Jin Kim<sup>1</sup>, Jin-Sun Choi<sup>1</sup>, Kyung Sook Park<sup>C</sup>*Department of Biology, Sungshin Women's University, Seoul 136-742*

The matrix metalloproteinases (MMPs) are a large family of proteolytic enzymes, which cleave extracellular matrix and non-matrix substrates. Increased MMPs expression has been reported in tumor invasion and metastasis. And MMP promoter variants have been known to be different from an effect of the MMPs transcriptional activities, such as MMP1-1607GG, MMP9-1562T, and MMP12-82A allele increased transcription or MMP3-11716A and MMP7-181A allele decreased transcription. Analysis of the MMP promoter MMP1, MMP3, MMP7, MMP9, and MMP12 single nucleotide polymorphisms were performed by PCR-RFLP using AluI, XmnI, EcoRI, SphI, and PvuII, respectively, in 219 unrelated Koreans. The genotype and allele frequency of five genes were compatible with the Hardy-Weinberg equilibrium. The allele frequencies of the MMP1-1607\*GG, MMP3-1171\*6A, MMP7-181\*A, MMP9-1562\*C, and MMP 12-82\*A were 0.724, 0.893, 0.932, 0.868, and 0.984, respectively. The allele frequencies of the MMP1 and MMP3 polymorphisms were different among ethnic distributions. The frequency of the MMP1-1607\*GG was different among Oriental (0.613-0.724), Caucasians (0.482-0.527), and Blacks (0.570). The allele frequency of the MMP3-1171\*6A was different between Orientals (0.803-0.893) and Caucasians (0.464-0.590). The haplotypes of the MMP7, MMP12, MMP1, and MMP3 on 11q21-23 were detected in 16 types. The common haplotypes were the MMP7A-181A, MMP12A-82A, MMP1GG-1607GG, MMP36A-11716A (35.1%), MMP7A-181A, MMP12A-82A, MMP1G-1607GG, MMP36A-11716A (27.8%), and MMP7A-181A, MMP12A-82A, MMP1G-1607GG, MMP35A-11716A (7.8%). These results suggest that single nucleotide polymorphisms of the MMP promoters may influence the expression of MMPs.

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**Genotypes of Mannose-binding Lectin in Behcet's Disease**Kyung Min<sup>P</sup>, KyungSook Park<sup>C</sup>*Department of Biology, Sungshin women's University, Seoul 136-742*

Mannose-binding lectin (MBL) is an acute phase protein of the innate immune response which participates in complement activation by binding to many pathogens surfaces. It has been well known that MBL exon and promoter polymorphisms affect MBL levels. We investigated the MBL polymorphism in 195 Behcet's disease (BD) and 200 healthy controls in Koreans. The genomic DNA analyzed the promoter region at G-550C(H/L variants), and G-221C(Y/X variants) and three missense mutations at codon 52 (C223T, Arg52Cys, A/D variants), 54 (G230A, Gly54Asp, A/B variants), and 57 (G239A, Gly57Glu, A/C variants) in exon 1 of the MBL gene by PCR-RFLP, SDM-PCR RFLP, and PCR-SSP. The difference was not found in exon 1 variants between BD and controls, the allele frequency of the MBL54\*Gly was 0.754 and 0.774 in BD and controls, respectively. No variants were found in MBL52\*Cys and in MBL57\*Glu either in BD and in controls, but only one MBL52\*Arg/\*Cys was found in controls. However -550G(H) allele was significantly higher in the BD patients than that in the controls (0.582 vs. 0.473, p=0.003, OR=2.3, 95% CI=1.32-4.09). G-221C(Y/X) was similar in BD patients and in controls. In the promoter region, high-MBL producing haplotype H.Y in the BD was significantly higher than that in the controls (58.0% vs. 47.3%, p=0.0026, OR=1.5 95% CI=1.16-2.04). We did not detect the H.X haplotype either in BD or in controls. High MBL concentration influence to complement activation consequently it may increase susceptibility to inflammatory disorders, including BD. These results suggest that the MBL -550G,-221G (H,Y) haplotype, high-MBL producing haplotype may contribute to BD susceptibility.

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**Characterization of P Element Insertion Mutant Lines Screened by otd Expression in Drosophila Central Nervous System**Sang-Hak Jeon<sup>C</sup>, Jae Yong Ahn<sup>P</sup>, Sang Hee Kim<sup>1</sup>, Sung Jin Kim<sup>2</sup>*<sup>P</sup>Department of Biological Sciences, Konkuk University, Seoul 143-701; <sup>C</sup>Department of Biology Education, Seoul National University, Seoul 151-748; <sup>1</sup>Department of Chemistry, Konkuk University, Seoul 143-701*

All complex nervous systems comprise two equally important cell types: neurons and glial cells. Glial cells play crucial roles in nervous system development, axonal guidance and fasciculation, and shaping of neuropilar structures. In order to find the genes involved in the gliogenesis, we screened the P enhancer trap lines which was primarily screened by the abnormal otd(orthodenticle) expression. 6 enhancer trap mutant lines out of 11 original lines affected the expression of gcm(glial cell missing), repo(reverse polarity), and pros(prospéro) that are involved in gliogenesis. As glial cells are known to regulate the differentiation process of axons, these lines were examined on the expression of FasII and BP102 that are markers on the central nervous system. These enhancer trap lines affected the expression of FasII and BP102. As gcm, a master gene in gliogenesis, is also involved in the hematopoiesis, the expression of Crq (croquemort) gene was investigated in these lines. croquemort is a marker of plasmatocyte. These lines also affected the expression of croquemort. Our results indicate that the screened lines are involved in gliogenesis, neuronal development process and hematopoiesis.