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Multiple Mutations in a Family with Charcot-Marie-Tooth (CMT) Disease

Mi Sun Lee[P], Byung Ok Choi[1], Il Nam Sunwoo[2], Ki Wha Chung[C]

[PC]Department of Biological Science and RRC for New Materials Recycling, Kongju 314-701; [1]Department of Neurology, Ewha Womens University, Seoul 110-783; [2]Department of Neurology, Yonsei University, Seoul 120-752

Charcot-Marie-Tooth (CMT) disease is the most common form of hereditary motor and sensory neuropathy. It is largely classified into demyelinating form (CMT1) and axonal form (CMT2). CMT1 is most frequently caused by 1.4 Mb tandem duplication of 17p11.2-p12 containing PMP22 gene. Mutations of PMP22, MPZ, EGR2 and Cx32 genes also cause a variety of distinct CMT phenotypes. From the mutation screen of the CMT-causable genes in 48 CMT families, we found a particularcase of a family having several different mutations. The family has a severe CMT with bilateral facial palsies. The detected mutations were Val136Ala and Ser198Ser in Cx32, Arg359Trp and Arg362Arg in EGR2 and Gly200Gly in MPZ. Of them, Val136Ala and Ser198Ser in Cx32 were identified to novel mutations. Particularly, Val136Ala was determined as *de novo*mutation found only from one family member. Arg359Trp in EGR2 was previously reported from foreign CMT patients. Arg362Arg in EGR2 and Gly200Gly in MPZ were regarded to polymorphisms.

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Allelic Distribution and Linkage Analysis of X-linked Microsatellites in Korean Population

Sang Hee Shin[P], Ki Wha Chung[C]

Department of Biological Science and RRC for New Materials Recycling, Kongju 314-701

Microsatellites are widely applied to forensic and linkage analysis. We have investigated allele frequencies and linkage disequilibriums of 20 X-linked microsatellite markers, DXS6807, DXS8378, DXS9895, DXS9902, DXS1214, DXS6810, DXS7132, DXS981, DXS6800, DXS9898, DXS6789, DXS101, DXS6797, GATA172D05, GATA165B12, HPRTB, GATA31E08, DXS1227, DXS8377 and DXS7423 in Korean population. The genotype distributions of most markers were not significantly deviated from the Hardy-Weinberg equilibrium, except for DXS6789. The all examined male samples showed completely different haplotypes. The test for pairwise linkage disequilibrium between two neighboring markers showed no significant disequilibrium. Four cases of mutation were identified at GATA172D05, GATA31E08, DXS7132 and HPRTB from the analysis of 21 family including 53 father-child-mother-child trios. Details of X chromosomal microsatellites in Koreans would be useful in paternity tests, forensic purposes and whole X-chromosomal mapping studies.

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A Genetic Screen for Suppressors of Calcineurin B Mutant (*cnb-1*)

Hyun-Ok Song[P], Jungsoo Lee[1], Joohong Ahnn[C]

Department of Life Science, K-JIST, Gwangju 500-71

Calcineurin is a Ca2+/calmodulin-dependent serine/threonine protein phosphatase that has been implicated in various signaling pathways. *C. elegans* calcineurin binds calcium and functions as a heterodimeric protein phosphataseestablishing its biochemical conservation in the nematode. Calcineurin is expressed in hypodermal seam cells, body-wall muscle, vulva muscle, neuronal cells, and in sperm and the spermatheca.The null mutant of calcineurin B [*cnb-1(jh103)*] showed pleiotropic defects including small body size, defective movement and delayed egg-laying. To identify genes involved in controlling body size in *C. elegans*, we used the small phenotype of *cnb-1(jh103)* as the criterion for a genetic suppressor screening. We have isolated several candidates which restore the body length nearly up to wild type level. We are currently in the process to map these suppressor mutants.

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Characterization of the Repair Gene Encoding RAD51B from *Arabidopsis thaliana*

Min-Yong Jang[P], Jeong-Seob Kim[1], Sang-Gu Kim[C]

School of Biological Sciences, Seoul National University, Seoul 151-742

Eukaryotic cells possess several mechanisms for repairing double-strand breaks(DSBs) in DNA. There are at least two pathways for DSBs repair : homologous recombination(HR)-mediated recombination repair and non-homologous end joining(NHEJ). In eukaryotes, the RAD52 epistasis group proteins serve the main role for meiotic recombination and/or homologous recombinational repair. RAD51, one of the RAD52 epistasis group proteins, is a homologue of the bacterial RecA recombinase. Five human RAD51 paralogs have been identified(XRCC2, XRCC3, RAD51B, RAD51C, and RAD51D), and each interacts with one or more of the others. But in plants repair the repair genes are not well known. Sequencing of an EST clone(Genbank accession no BE530642) was revealed it contains an open reading frame for *AtRAD51B*which is 1017 bp and encodes approximately 38 kDa protein. By southern blot analysis it was shown that *AtRAD51B* exists as one copy in the genome of Arabidopsis. As a result of RT-PCR, *AtRAD51B* is highly expressed in flower. Expression of *AtRAD51B*in a yeast rad52 mutant did complement the sensitivity to methyl methanesulphonate(MMS). These results suggest that the gene product of *AtRAD51B*is involved in the homologous recombination repair. The exact function of *AtRAD51B* is being investigated using transgenic *Arabidopsis thaliana*.