F814 Mutation of Thr16Pro in Human Lysosomal Acid Lipase(LAL) Gene

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Lysosomal acid lipase (LAL) is a key enzyme involved in intracellular hydrolysis of cholesteryl esters and triglycerides. Here we report a novel restriction fragment length polymorphism (RFLP) of Thr16Pro ($A^{+46}\rightarrow C$) in human LAL gene exon 2. The mutation was genotyped by the PCR and subsequent digestion using *Hae*III. We examined the genotype distributions in subjects with Korean normal (n=198) and hypercholesterolaemia groups (n=81). The observed *AA*, *AC* and *CC* genotype numbers in control group were 95, 88 and 15, respectively (frequency of *A*=0.702, *C*=0.298). The genotype numbers in disease group were 28, 41 and 12, respectively (*A*=0.599, *C*=0.401). Each group showed some frequency differences, but it was no significantly different. However, significant differences were found in genotype distributions between control and disease groups (χ^2=6.28, df=2, 0.025<P<0.05).

F815 Elavated Promoter Activity of Mouse IL-3 Gene by the Upstream Granulocyte-Macrophage Colony-Stimulating Factor Enhancer

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Granulocyte-macrophage-colony-stimulating factor (GM-CSF) and IL-3 are cytokines that have roles promoting the proliferation, differentiation, and function of numerous classes of hemopoietic cells and whose genes are closely linked in mice. To investigate the function of cyclosporine A-inhibitable enhancer located 2 kb upstream of the mouse GM-CSF gene on the transcriptional regulation of IL-3 gene, we isolated the enhancer locus from genomic DNA of mouse EL4 T cell line by PCR and inserted it into the previously cloned IL-3-CAT reporter plasmid (pCIL3-1003), and named as pEIL3-1003 (having enhancer). In transiently transfected mouse IC-2 mast cells, the promoter pCIL3-1003 was induced 15-fold or 2-fold in the presence of A23187 alone or A23187 plus PMA, however, in case of pEIL-1003, these inductions were enhanced as 40-fold or 55-fold respectively. This induction was in each case reduced to near basal levels in the presence of cyclosporin A. These results clearly show that this cyclosporin A sensitive enhancer region acts as an enhancer of the mouse IL-3 promoter.