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THEERASAK PIMSAWAT : POLYMORPHISMS AT THE 5'-FLANKING REGION,
EXONS 1, 8, 13, AND 18 OF LOW DENSITY LIPOPROTEIN RECEPTOR GENE IN THAI
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The low density lipoprotein (LDL) receptor plays an important role in cholesterol homeostasis. A mutation in this gene causes the autosomal dominant disorder namely familial hypercholesterolemia (FH) and associated with coronary heart disease (CHD). In addition, a number of common LDL receptor gene polymorphisms have been identified and some of these polymorphisms were found to be associated with variation of cholesterol levels in several populations. However, this kind of data has never been reported in Thai subjects. In this study, the genotypes of some restriction sites: *HincII* (5'-flanking region), *AvaII* (exons 1,13), *StuI* (exon 8), and *NcoI* (exon 18) at the LDL receptor gene in 145 Thai subjects have been investigated by PCR-RFLP technique. The result shows that among the tested samples, there were no polymorphisms in *HincII* (5'-flanking region), *AvaII* (exon 1) and *StuI* (exon 8) sites. In contrast, there were polymorphisms at *AvaII* (exon 13), and *NcoI* (exon 18) sites. The genotype frequencies at the *AvaII* sites were 57.9%, 36.6%, and 5.5% for the (-/-), (+/-), and (+/+) genotypes, respectively. The genotype frequencies at the *NcoI* (exon 18) restriction sites were 17.9%, 50.4%, and 31.7% for the (-/-), (+/-), and (+/+) genotypes, respectively. Both of the *AvaII* and *NcoI* polymorphisms were not significantly associated with cholesterol levels ($P > 0.05$). However, the (+/+) *AvaII* genotypes tend to have higher total cholesterol and LDL-cholesterol and also tend to have lower HDL-cholesterol levels than (-/+ and (-/-) genotypes. Similarly, the similar trend was seen for *NcoI* genotypes except that the LDL-cholesterol levels were slightly lower in the (+/+) *NcoI* genotype when compared with the (-/-) genotype.