Ruchchadol Sawangpanich 2007: Identification of Low Density Lipoprotein Receptor (*LDLR*) Gene Mutation in Thai Patients with Familial Hypercholesterolemia (FH). Master of Science (Genetic Engineering), Major Field: Genetic Engineering, Interdisciplinary Graduate Program. Thesis Advisor: Associated Professor Thanyachi Suru, M.D. 94 pages.

Hypercholesterolemia is one of the major risk factors for Coronary Artery Disease (CAD). Both hereditary and environmental factors lead to a wide variation in concentrations of low density lipoprotein cholesterol (LDL) in the general population. One form of primary in the *LDLR* gene causes an inherited primary hypercholesterolemia namely familial hypercholesterolemia (FH). Such mutation result in impaired clearance of plasma LDL and accumulation of LDL in bloodstream for a long time. At present, more than 700 mutations have been identified worldwide. In the present study, mutations at *LDLR* gene in Thai subjects with primary hypercholesterolemia were screened by PCR-SSCP. Different SSCP patterns in DNA samples from patients (n=19) were found as follows, fifteen in exon 10, six in exon 11 , eleven in exon 12 and another two in exon 15. Different SSCP patterns in exon 10, exon 11, exon 12 and exon 15 were characterized by automated DNA sequencing. Transition from A to G at nucleotide 24134, C to T at nucleotide 26669, C to T at nucleotide 27471 and A to G at nucleotide 33810 were found in exon 10, 11, 12 and 15 respectively. Single base change in each exon did not cause amino acid change and these changes have been reported previously.

Ruchchadol Sawangpanich. T. Suru 27 / May / 2007

Student's signature Thesis Advisor's signature