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JIRAWAT AKKANIT : MOLECULAR ANALYSIS OF RB1 GENE IN RETINOBLASTOMA IN THAI PATIENTS. THESIS ADVISOR : PIMPICHA PATAMASIRIWAT , Ph.D., PA-THAI YENCHITSOMANUS , Ph.D. 82 p. ISBN 974-589-114-2

Genetic changes of retinoblastoma (RB1) gene were analyzed in 9 patients with retinoblastoma. Mutations within intron 1 and intron 17 of RB1 gene were screened by polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) technique, and mutations within intron 20 were screened by polymerase chain reaction based microsatellite analysis technique. Southern blot hybridization was used for analysing gross structural changes within RB1 gene in tumors.

Mutations were detected in 8 of 9 tumors (89%). Five of nine tumors (55.5%) showed deletions within intron 1 and in one out of five tumors the deletions occurred within both alleles. Four tumors showed deletions within intron 17 (44.4%), and the deletions of both alleles were found in two of these four tumors. Five of nine tumors (55.5%) exhibited deletions within intron 20 and all of the deletions were found in only one allele compare to normal DNA from patients' white blood cells. Loss of heterozygosity (LOH) was shown in tumors with deletion in one allele. Fifty-six DNA samples from white blood cells of patients and patients' family members were also analysed using both methods. Abnormality was not observed in these samples. No gross structural change was found in any tumors when analysed by Southern blot hybridization under probe condition and restriction enzyme was used.