IDENTIFICATION OF SINGLE NUCLEOTIDE POLYMORPHISM (SNP) OF THE RB1 GENE IN MALAY CHILDREN WITH RETINOBLASTOMA

Research center: Human Genome Center, School of Medical Sciences, Universiti Sains Malaysia, Kelantan, Malaysia.

Current status of project: Ongoing

Researcher:
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Track record:
Retrospective review: Clinical features and survival of children with retinoblastoma in Hospital Universiti Sains Malaysia (2004).

Introduction:
Retinoblastoma is the commons primary intraocular tumor that occurs in early childhood. Mutation of a tumor suppressor gene, RB1 gene is reported the cause of this disease. A diallelic polymorphism was previously detected as a novel variant (an A -> G change) at nucleotide 153,104 in intron 18 of the Rb1 gene and effect a Tsp 5091 restriction site exclusively among Asians.

Objectives:

1. To identify the presence of A -> G single nucleotide polymorphism (SNP) in exon 19 of Rb1 gene in Malay children with retinoblastoma.
2. To determine the allele frequency of the mutated allele G in exon 19 of RB1 gene in Malay children with retinoblastoma.

Methodology:

We planned to obtain 5cc of the blood from the 57 selected patients and do the DNA extraction. The amplification of the Rb1 gene will be performed using the method described by Kadam Pai P et al. The PCR product will be digested with Tsp5091 enzyme for rapid genotyping of the Rb1 SNP. Direct sequencing of randomly selected samples will be performing by RFLP.

Expected outcome:

1. Identification of the A->G SNP in exon 19 of the Rb1 gene in Malay children with Retinoblastoma.
2. Determination of frequencies of the mutated allele G in exon 19 of Rb1 gene in Malay children with Retinoblastoma.