TITLE:
A STUDY OF E1A BINDING DOMAIN MUTATIONS OF RB1 GENE AMONG RETINOBLASTOMA PATIENTS IN MALAYSIA

RESEARCH CENTRE: Human Genome Center, School of Medical Science, Universiti Sains Malaysia

CURRENT STATUS OF PROJECT : Ongoing

RESEARCHERS:
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TRACK RECORD:
Clinical presentation and survival rate of children with retinoblastoma in Hospital Universiti Sains Malaysia. Oral presentation 2nd USM ophthalmology

Introduction:
Retinoblastoma is an intraocular tumor in children under four years old. Development of this tumor requires both alleles of RB1 gene to be functionless. RB1 gene is a tumor suppressor gene. It encoded nuclear phosphoprotein (Rb protein) which responsible to inhibit uncontrolled cell division. The Rb protein contains several functional domains: the N-terminal region, the small pocket (consisting of domain A and B), the C-terminal region, and the large pocket (consisting the small pocket plus part of C-terminal). The large pocket binds several viral oncoproteins and cellular regulatory proteins and is critical for controlling cell growth. Although there are no single 'hot spot' in the RB1 gene, many mutations were reported to affect the function of large pocket domain.
Early detection of this disease among patients with retinoblastoma and their siblings can help to improve the survival rate and preserve vision.

Objectives:
1. To study the mutations of E1A binding domains in RB1 gene among retinoblastoma patients in Malaysia
2. To determine the mutation characteristic by using DNA sequencing strategy.
3. To correlate the mutation characteristics with laterality of the disease

Methodology:
DNA extraction will be amplified in separate PCR assay in for each exons except for exons 15-16 which amplified in same reaction. PCR product will be purified and undergo for screening mutations using dHPLC and SSCP.

Expected outcome:
Identification of the mutations in target region using dHPLC and SSCP analysis.