MOLECULAR ANALYSIS OF SMN AND NAIP GENES FOR SPINAL MUSCULAR ATROPHY PATIENTS IN MALAYSIA

RESEARCH CENTRE: Human Genome Centre, School of Medical Sciences, Universiti Sains Malaysia

CURRENT STATUS OF PROJECT: Ongoing

RESEARCHERS:

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TRACK RECORD:

1. Understanding the Role of Survival Motor Neuron 1 (SMN1) and Neuronal Apoptosis Inhibitory Protein (NAIP) Genes in the Pathogenesis of Spinal Muscular Atrophy (SMA) Patients in Malaysia (2003-2005).
2. Molecular Characterization of the SMN Gene; Towards Understanding The Transcriptional Control of The Promoter Region (2005).

Introduction:

Spinal Muscular Atrophy (SMA), a common neuromuscular disorder, is caused by degeneration of the anterior horn cells of the spinal cord. The major cause of this disease is deletion or mutation of the SMN1 gene. This autosomal recessive disease is classified into 3 clinical subtypes; Type 1, 2 and 3 based on onset age and disease severity. The SMN2 copy number and NAIP gene have been reported to be correlated to the severity of the disease.

Objectives:

This study focus on the deletion analysis of SMN1 and NAIP genes, and also the copy number of SMN1 and SMN2 genes in the family members' of the SMA patients.

Methodology:

Genomic DNA will be extracted from whole blood. The amplification of exons 7 and 8 of the SMN gene will be done using PCR-restriction enzyme digestion method (van der Steege et al, 1995) while NAIP exon 5 will be amplified according to Roy et al, 1995. After the confirmation of deletion of the SMN1 gene, the copy number of SMN1 and SMN2 genes will be quantified by relative quantification using real-time PCR.

Expected outcome:

We hope to get the baseline data for the deletion frequency of the SMN1 gene in Malaysian SMA patients. And also to correlate between the SMN2 copy numbers with the disease severity.