# PATTERN OF ALU INSERTION POLYMORPHISM IN MALAYSIAN MAJOR ETHNIC GROUPS



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#### 5.2 Enhanced Executive Summary

An identical by descent (IBD) Alu insertion polymorphisms provides a useful genetic marker for the study of human population genetic. Four human specific Alu loci (HS3.23, HS4.14, HS2.25 and HS4.69) were analyzed to determine the pattern of Alu insertion polymorphism in Malaysian major ethnics namely Malay, Chinese and Indian. Buccal samples were collected from 75 unrelated individuals using FTA paper and later undergo PCR amplification. The amplified product were detected and separated by 2% gel electrophoresis. All loci studied were polymorphic except for HS2.25. The mean of Alu insertion frequencies were 0.2077 in Malay and 0.2765 in Chinese. Interestingly, the insertion of all loci were completely absent from Indian ethnic group. All ethnic groups show trend toward lower heterozygosities. Across all subpopulations, the loci produced value of  $F_{\rm ST}$  ranging from -0.7719 to 0.1022. Analysis of F-statistic had suggested that moderate genetic differentiation existed between Malay, Chinese and Indian ethnic population in Malaysia.

#### 5.3 Introduction

Genetic loci which exhibit high degree of polymorphism give the forensic scientist a powerful tool for characterizing biological evidence. The generation of genetic profile of a person through DNA analysis of biological evidence has been a revolutionary development in solving of criminal cases. DNA profiles from the crime scene are compared with the database from the general population to estimate whether there are probability that unrelated individual would have the identical DNA profile. The genetic markers including variable numbers of tandem repeats (VNTRs), short tandem repeats (STRs), mitochondrial DNA (mtDNA) and Y-chromosome (Jobling & Gill, 2004).

Recently, there has been an increasing interest among forensic investigators in *Alu* insertion polymorphisms. Since the young *Alu* subfamilies did not become active until after the human lineage diverged from the last common ancestor with the non human apes, insertion events of these *Alus* are restricted to humans and the ancestral alleles is the absence of an insertion (Watkins *et al.*, 2001). At each polymorphic locus, the chromosome containing the *Alu* insert is the derived allele, and different individuals with an *Alu* insert at a given locus share a chromosomal region that is identical by descent (Batzer & Deininger, 2002). Thus, permit tracing of population ancestral heritages. Information about the likely ethnicity of the sources of the sample is the information that investigators may use when persuing leads base on the genetic analysis of crime scene evidence (Pillay *et al.*, 2007).

Malaysia population consists of three major ethnics namely Malay, Chinese and Indian. Traditionally, these three primary ethnics group are distinguished based on their language spoken, religion and physical appearance such as skin color and facial features. To date, there are only a few published reports on the study of Malaysian population based on DNA markers. Previously we have reported on six Alu loci to study Malaysian major ethnic groups and it suggested that genetic differentiation existed between ethnics of Malay, Chinese and Indian at some locus although the value is considered as low (Wan Nurhayati et al., 2009). The present study furnishes new information on another four Alu loci to determine the genetic differentiation among Malaysian major ethnic groups.