



**Development of data bases for
autosomal, Y-chromosomal and
mitochondrial DNA markers and
their application in forensic casework
and population genetics in
Sri Lankan populations**

A thesis submitted for the
Degree of Doctor of Philosophy

R. J. Illeperuma
Faculty of Science
University of Colombo
December 2009

ABSTRACT

The use of our hereditary material DNA as a biological tool has revolutionized forensic investigations and population genetic studies and has led to the emergence of molecular forensics and molecular anthropology. Analysis of polymorphisms in DNA enables the identification of individuals and the determination of relationships between groups of individuals with high degrees of sensitivity, accuracy and efficiency. The overall objectives of this study were to facilitate the use of autosomal, Y-chromosomal and mitochondrial DNA (mtDNA) markers in forensic case work and population genetics to identify individuals and to determine genetic affinities of Sri Lankan population groups.

A total of 648 unrelated individuals belonging to five Sri Lankan populations viz. Sinhalese, Sri Lankan Tamils, Indian Tamils, Moor and Veddahs (*Ādivāsis*) were sampled to study polymorphisms in 11 autosomal and 12 Y-chromosomal STR loci and the mitochondrial HVSI locus.

The allele (or haplotype) frequency distributions, the heterozygosities (or gene diversities) and polymorphisms of each loci tested in the above three DNA markers showed that Sri Lankan ethnic groups (Sinhalese, Sri Lankan Tamils, Indian Tamils and Sri Lankan moors) possess a relatively high genetic diversity, thus indicating the suitability of applying those markers in forensic human identification. The autosomal STR results of the present study have been successfully applied in more than 2600 paternity tests and 600 forensic cases of human identification and the statistics described in this thesis have been used to interpret DNA evidence submitted to Courts of Law in Sri Lanka.

Detailed phylogenetic analysis of the Sri Lankan populations using these three marker sets revealed that the indigenous *Ādivāsi* population of Sri Lanka, who are considered to be one of the "relic" population groups of early modern humans, forms a distinct cluster separate from the other Sri Lankans. This suggests that they have experienced genetic drift, a population bottle neck, or combination thereof. Their cryptic population structure and unique allele signatures as revealed in the present study would make significant implications in forensic investigations where *Ādivāsi* individuals are involved. The *Ādivāsis* possess haplotypes that separate them from the rest of the Sri Lankans with multiple mutational steps suggesting that they might be the surviving descendents of the earliest inhabitants of Sri Lanka.

The suitability of using autosomal, Y-chromosomal and mtDNA markers for discriminating between Sri Lankan individuals in DNA based forensic case work was established. As a consequence of the development of databases for Y-STRs and mtHVSI haplotype frequencies as well as the evaluation of applying and interpreting results in casework as discussed herein, Y-chromosome and mitochondrial based human identification can now be used as valuable additions to the current autosomal test panel to improve the exclusion probabilities in forensic case work in Sri Lanka. In summary, Sri Lankan ethnic groups possess considerable genetic homogeneity and are genetically similar to each other. The observation that genetic differentiation among ethnic populations grouped by linguistic affiliation was relatively higher than among populations delimited by ethnicity suggests language has been a stronger barrier to gene flow than ethnic identity in Sri Lanka.