CHARACTERISATION OF VARIANT ALLELES AT THE HUMD21S11 LOCUS

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CERTIFICATE OF AUTHORSHIP/ORIGINALITY

I certify that the work in this thesis has not previously been submitted for a degree nor has it been submitted as part of requirements for a degree except as fully acknowledged within the text.

I also certify that the thesis has been written by me. Any help that I have received in my research work and the preparation of the thesis itself has been acknowledged. In addition, I certify that all information sources and literature used are indicated in the thesis.

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ABBREVIATIONS

Acronym	Definition
DNA	Deoxyribonucleic Acid
dNTP	Dinucleotidetriphosphate
KYA	Thousand Years Ago
$MgCl_2$	Magnesium Chloride
ηg	Nano gram
PCR	Polymerase Chain Reaction
RPM	Revolutions Per Minute
SSM	Slipped Strand Mispairing
SUPAMAC	Sydney University Prince Alfred
UCO	Unequal Crossing Over
µL	Micro Litre

Abstract

Significant genetic substructure within a population can affect the evidential weight of a DNA profile to the detriment of a defendant. To prevent this from occurring, forensic examiners continually look to understand more about the degree and structure of genetic variation within a population. A part of this involves the characterisation of the microsatellite loci applied in forensic testing.

The HUMD21S11 microsatellite is commonly used in forensic examinations in conjunction with a suite of other microsatellite loci in order to identify individuals who may have been present at a crime scene, or who cannot be identified through more traditional non-genetic means (such as visual, dental or medical records).

This research confirmed the existence of an ancestral relationship between the indigenous populations of Australia and Papua New Guinea as well as the presence of regional differentiation within the Australian Aboriginal population. The sequence variation present at the HUMD21S11 microsatellite locus makes it a suitable candidate to further understand and describe the regional differentiation within the Australian Aboriginal population.

This study also confirmed that microsatellites are able to retain their variability after structural change (Möller et al. 1994, Brinkmann et al. 1996, Griffiths et al. 1998, Walsh et al. 2003), and that a single mutation event can involve single repeat units or multiple repeat units.

The structural complexity of microsatellites like HUMD21S11 could be used to further develop mutation models as well as investigate the proposal that the mutation rate of microsatellite loci is be dependent on the DNA sequence present.

The sequence variation at the HUMD21S11 microsatellite is prevalent enough in the Australian Aboriginal population, to warrant a more complete investigation of the genetic variation at this locus. However, in order to better understand the genetic diversity present in the Australian Aboriginal populations, examination of the

population based on traditional tribal boundaries (rather than judicial boundaries) is recommended.

Additional population studies at the sequence level will increase our level of understanding about the genetic relationships of the Australian Aboriginal population on a local and global level. The sequence data obtained will also assist in the understanding of the mutation process and aid in the development of statistical models.