Determining SNPs and demographic variables that impact level one fingerprint pattern

by Andrew Walton

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under the supervision of Dr Sebastien Moret, Dr Dennis McNevin and Dr Mark Barash

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Certificate of original authorship

I, Andrew Walton declare that this thesis, is submitted in fulfilment of the requirements for the award of Doctor of Philosophy, in the Faculty of Science at the University of Technology Sydney.

This thesis is wholly my own work unless otherwise referenced or acknowledged. In addition, I certify that all information sources and literature used are indicated in the thesis.

This document has not been submitted for qualifications at any other academic institution.

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Abstract

The current forensic use of fingerprints is for identification purposes and requires a reference sample for comparison to any unknown fingermark. Features of individual ridges can be used for identification however in combination they create overall level one fingerprint patterns (arches, loops and whorls). Past studies have indicated that some fingerprint characteristics such as level one pattern may occur at higher frequencies in some biogeographical ancestries (BGAs) and ridge density (the number of ridges within a defined area) may be used to determine the sex of an individual. The frequency at which these patterns and their subclassifications occur is largely unknown in the Australian population as there have been no modern studies utilising statistical analysis. Fingerprint experts would benefit from the publication of this information as it is the first step in building a statistical model that may add probabilities to their opinions on pattern rarity in a court setting. Previously, they may only rely upon their own observations from their experience and studies based on overseas populations.

This research aimed to represent the level one fingerprint pattern and ridge density frequencies of the diverse Australian population. This also provided the opportunity to assess the association of pattern and ridge density with BGA, sex, hands, fingers, and genetic markers. By assessing these associations, a new avenue of investigative potential could be unlocked from fingerprint evidence. For fingermarks that do not return a match it may be possible to predict which hand or finger the mark came from (provided it is not a full set), the ancestry, sex, or genotype of the depositor.

A total of 828 volunteers, 515 people from Sydney donated their fingerprints, DNA and self-declared BGA through a questionnaire and 313 people from Melbourne provided fingerprints with self-declared BGA information. The fingerprints in Sydney were collected via fingerprint scanner and those from Melbourne were provided as ink on card. The fingerprints were then classified using the National Crime Information Center (NCIC) classification system. Goodness of fit tests, multinomial logistic regression and general estimating equations were utilised for association of ancestry, sex, hands, and fingers with the pattern and ridge densities. Associations between fingerprint patterns and genetic markers were investigated for five genetic models.

The goodness of fit and multinomial logistic regression analyses revealed several patterns occurring at significantly higher and lower frequencies than expected for all independent variables. The general estimating equations also showed significant differences amongst ridge densities (radial, ulnar, and proximal positions) for all independent variables. A further investigation was made into the useability of ridge density in fingerprints of unknown finger or hand. Results showed that proximal and ulnar positions produced dissimilar results to right and left positions, indicating this characteristic would be limited in its usefulness in forensic casework.

In people of European and Middle Eastern biogeographical ancestry over 60 SNPs were significantly associated with fingerprint patterns and ridge densities and four genetic loci were amongst the hundreds of genetic markers that were not quite significant. The four loci included two distinct areas on chromosome six, an area on chromosome one and an area on chromosome 11. Several genetic markers were novel, and several replicated those found in previous studies. The hypothesis that non-coding regions and epigenetic regulation are causative of fingerprint development was tentatively supported. These results provide strong evidence that frequencies of level one fingerprint pattern and ridge density differ between ancestral populations and sex and occur with different frequencies amongst fingers and between hands. Genetic markers may be identified in the future with diverse and increased sample sizes and through DNA phenotyping the prediction of an individual's fingerprints may be possible, allowing the interrogation of a fingerprint database even if there are no physical fingerprints.

This research met the original aims to assess the association of pattern and ridge density with BGA, sex, hands, fingers, and genetic markers in the diverse Australian population. Many results were novel and created potential leads for future investigation. The use of this research by practitioners however would be premature as larger BGA groups are needed for both pattern to BGA and pattern to SNP association studies.

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Table of contents

Terms and abbreviations

BGA	Biogeographical Ancestry
SNP	Single Nucleotide Polymorphism
USA	United States of America
NCIC	National Crime Information Center
DNA	Deoxyribonucleic Acid
EVC	Externally Visible Characteristic
NCIDD	National Criminal Investigation DNA Database
ARC	Absolute Ridge Count
LOD	Logarithm Of Differentiation
QTL	Quantitative Trait Locus
ROES	Ridges Off the End Syndrome
PCA	Principal Component Analysis
FISWG	Facial Identification Scientific Working Group
FBI	United States Federal Bureau of Investigation
MLR	Multinomial Logistic Regression
GLM	Generalised Linear Model
GEE	General Estimating Equation
qPCR	Quantitative Polymerase Chain Reaction
HWE	Hardy-Weinberg Equilibrium
AIC	Akaike Information Criterion
CHR	Chromosome
BP	Base Pair
LL	Left Little finger
RL	Right Little finger
LR	Left Ring finger

RR	Right Ring finger
LM	Left Middle finger
RM	Right Middle finger
LI	Left Index finger
RI	Right Index finger
LT	Left Thumb finger
RT	Right Thumb finger
EMT	Epithelial Mesenchymal Transition

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Research communication

Publications

Walton AD, Moret S, Gunn P, Barash M. Comment on "Linkage analysis of a model quantitative trait in humans: Finger ridge count shows significant multivariate linkage to 5q14. 1" by Medland *et al.*,"Common Genetic Variants Influence Whorls in Fingerprint Patterns" by Ho *et al.* and "Hot on the Trail of Genes that Shape Our Fingerprints" by Walsh *et al.* Forensic Science International: Genetics. 2018.

Walton A, Moret S, Barash M, Gunn P. (2019). "The frequency of fingerprint patterns separated by ancestry and sex in a general population from Sydney, Australia." Australian Journal of Forensic Sciences: 1-6.

Presentations

ANZFSS 24th International Symposium – September 14th, 2018, Perth – "The frequency of fingerprint patterns separated by ancestry and sex in a general population from Sydney, Australia" – Walton A, Moret S, Barash M, Gunn P.

NSWPF Fingerprint Expert Conference – December 11th, 2019, Parramatta – "Determining the effect of ancestry and sex on fingerprint patterns" – Walton A, Moret S, Barash M, Gunn P.