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**THREADS FROM THE PAST:  
A GENETIC STUDY OF  
AFRICAN ETHNIC GROUPS  
AND HUMAN ORIGINS**

A THESIS PRESENTED IN PARTIAL FULFILMENT OF THE REQUIREMENTS  
FOR THE DEGREE OF  
DOCTOR OF PHILOSOPHY IN BIOLOGICAL SCIENCE  
*BIOLOGICAL ANTHROPOLOGY*

AT MASSEY UNIVERSITY

ELIZABETH EVELYN WATSON

1996

*I am the vessel of fate  
I sail between the reefs of the warp  
that stand for life  
from the right bank I pass to the left  
unreeling my gut  
to add to the fabric  
then back from the left bank I pass to the right  
unreeling my gut  
life is perpetual coming and going  
a permanent gift of the self*

-a Fulbe weaver's invocation (Hampâté Bâ, 1990)

## 1. ABSTRACT

Mitochondrial DNA (mtDNA) provides an efficient tool for investigating human pre-history and evolution. In this thesis Hypervariable I sequences from 241 individuals representing nine diverse African ethnic groups are presented, and analysed together with published sequences. Each ethnic group studied represents a different combination of geography (East and West Africa), linguistic phylum (Niger-Kordofanian, Nilo-Saharan, and Afroasiatic), and subsistence mode (agricultural and pastoral). The African mtDNA dataset is expanded considerably by the addition of these samples. Questions concerning human evolution and human pre-history are readdressed.

These samples demonstrate a higher diversity in Africa than previously reported. An ethnic group such as the Turkana has a higher mitochondrial diversity than the rest of the world combined. There is some evidence of regionalism, in that some of the clusters are specific to West or East Africa; however, there is also sharing of lineages across Africa suggesting substantial migration and admixture both in the past and more recent times. In Africa, the !Kung being the exception, the mtDNA from one ethnic group does not show a common unique ancestor, rather the ethnic groups are composites of several mtDNA types, with distinct origins and population histories. Combined analysis with Median networks and pairwise distance methods, demonstrate that within Africa approximately 60,000 years ago, there was a phenomenal expansion of one cluster, which was associated with the people who subsequently moved out of Africa. Almost all non-African populations are derived from this cluster. The Out of Africa hypothesis is supported, but there may have been more than one expansion of modern humans out of Africa. Further sampling in Asia and Australia is crucial to conclude the Multiregional-Out of Africa debate.

## ABBREVIATIONS

°C	degrees Celsius
μl	microlitre
A.L.F.	Automatic Laser Frequencer (Pharmacia)
AD	<i>anno Domini</i> years after the birth of Christ
APS	ammonium persulphate
BC	years before Christ
bp	base pairs
BP	years before present
<i>c.</i>	<i>circa</i>
ddNTP	de-deoxyribonucleotide triphosphate
DNA	deoxyribonucleic acid
dNA	deoxyribonucleotide acid
DTT	dithiothreitol
EDTA	ethylenediamine tetraacetic acid
g	grams
HVR-I	Hyper-variable Region I
HVR-II	Hyper-variable Region II
kb	kilobase
kya	thousand years before present
M	Molar
Mya	million years before present
mg	milligram
ml	millilitre

mtDNA	mitochondrial DNA
NaCl	Sodium Chloride
np	nucleotide position with reference to Anderson <i>et al.</i> , (1981)
°C	degrees Celsius
PCR	polymerase chain reaction
RFLP	restriction fragment length polymorphism
RNA	ribonucleotide acid
SDS	sodium dodecyl sulphate
TEMED	N,N,N,N, tetramethylethylenediamine
tRNA	transfer ribonucleotide acid

## NOTES

The term “race” is something which can not be visualised genetically; in humans there is usually more diversity within a population than between populations. Terms which have racial overtones have been replaced by the geographic origin of the peoples i.e. Asian, European, African, American.

- “African” replaces the term Negroid.
- “European/Middle Eastern” replaces the term Caucasoid and refers to people living in the geographical area of Europe and the Middle East, and who are not known to be recent immigrants.
- “Asian” replaces the term Mongoloid and refers to people living in Asia.
- “San” replaces the term Bushman and refers to hunter-gatherers who speak a Khoisan language. In this study the San population are known as the !Kung.
- “Dama” replaces the term Hottentot and refers to pastoralists who speak a Khoisan language.

I have consciously avoided describing the physical attributes of the people as most groups exhibit high variation, and physical attributes generally do not show genetic affinity. Stature and skin colour are, for instance, more an adaptation to the environment than determining any genetic relationship.

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## CONTRIBUTION

The thesis is my own work. For some of the chapters, however, I have collaborated with other people. The following list summarises my contribution to the following chapters.

### *SAMPLE COLLECTION AND ETHICS*

**target:** no journal targeted  
**purpose:** to introduce the fieldwork and design of project  
**contribution:** my own work

### *SEQUENCE DESCRIPTION*

**target:** no journal targeted  
**purpose:** to give a general overview of the sequences  
**contribution:** my own work

### *THE TURKANA PEOPLE FROM KENYA HAVE THE WORLD'S DIVERSITY IN THEIR MTDNA GENEPOOL*

**target:** Nature  
**purpose:** to emphasis diversity within Africa  
**contribution:** my own work with supervisorial advice from David Penny  
**authors** Watson, E.E. and Penny, D.

### *HUMAN MITOCHONDRIAL FOOTPRINTS OF EXPANSIONS WITHIN AFRICA*

**target:** to be decided  
**purpose:** to trace mitochondrial relationships within Africa and date expansion events  
**contribution:** written by myself with assistance from Hans-Jürgen Bandelt, Peter Forster and Martin Richards  
**authors** Watson, E.E., Forster, P., Richards, M. and Bandelt, H.-J.

***TREES FROM LANGUAGES AND GENES ARE VERY SIMILAR***

**target:** Systematic Biology 42(3):321-323

**purpose:** to compare trees generated globally from genetics and language.

**contribution:** my original idea and tree construction

**authors** Penny, D., Watson, E., and Steel, M.A.