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MRC/NHLS/WITS HUMAN GENOMIC DIVERSITY AND DISEASE RESEARCH UNIT (HGDDRU)

THE LIVING HISTORY PROJECT: REPORT ON GENETIC ANCESTRY TESTING CONDUCTED IN PIETERMARITZBURG (OCTOBER, 2007)

by
Professor Himla Soodyall

In October 2007, 98 people responded to the invitation extended by the African Genome Education Institute (AGEI) in partnership with Ancestry24.com to volunteer a cheek swab sample for genetic ancestry testing. Genetic ancestry testing involves the use of specific regions of the human genome, which by virtue of their unique mode of inheritance, have been used widely as tools to shed light on population affinities and human evolution. Mitochondrial DNA (mtDNA), which is inherited exclusively through our matriline (mother, maternal grandmother, maternal great-grandmother, and so on), provides us with the opportunity to trace our maternal ancestry several thousands of years back into the past. Both sons and daughters inherit their mother's mtDNA, but only the daughters (not sons) will pass their mtDNA to their offspring. All surviving mtDNA lineages ultimately trace back to a common ancestor who lived in Africa approximately 150,000 years ago.

Over time, as humans migrated out of Africa to populate the rest of the world, they took with them a subset of the divergent mtDNA lineages found in Africa which eventually mutated or changed to produce the various lineages found in living people outside of Africa. At the same time, those ancestors who remained in Africa continued to evolve newer lineages. As people migrated and settled throughout the continent, their contact with other groups contributed to gene exchange between groups thereby contributing to shaping the gene pool found in living people.

In the past 20 years or so, researchers have studied several populations throughout the world and have mapped the global patterns of mtDNA variation. There is reasonable concordance between one's geographic region of origin and the mtDNA lineages found in that region. For example, the lineages found in indigenous Africans belong predominantly in haplogroup (or branch) L of the mtDNA tree whereas haplogroup M is found most commonly among Asians from India.

In addition to mtDNA, Y chromosome DNA offers an equivalent tool to trace paternal heritage (father, father's father, great grandfather, etc) in males. Y chromosome patterns of variation are also highly structured by geographic region. Also, specific Y chromosome changes or mutations have been identified that contribute to the branching pattern of the Y chromosome tree.

In the present study, the Human Genome Diversity and Disease Research Unit at the National Health Laboratory Service and University of the Witwatersrand under the Directorship of Professor Himla Soodyall was invited to conduct the laboratory testing and analysis on the samples collected for this project. The project was conceived with the goal of giving members of the public an opportunity to learn about their ancestry and to engage with scientists on this journey of discovery. Given the rich diversity and history of the people of the South Africa, mtDNA and Y chromosome DNA was used to assess how females and males, respectively, have contributed to shaping the gene pool among its inhabitants.

Sample

The sample consisted of 98 individuals from 15 different countries (11 African countries, including South Africa; New Zealand, USA, Cuba and Romania). All participants had completed and signed an informed consent form which included among others questions related to the individuals self identified ethnicity, home language and gender. Some of these results are given in Fig. 1.

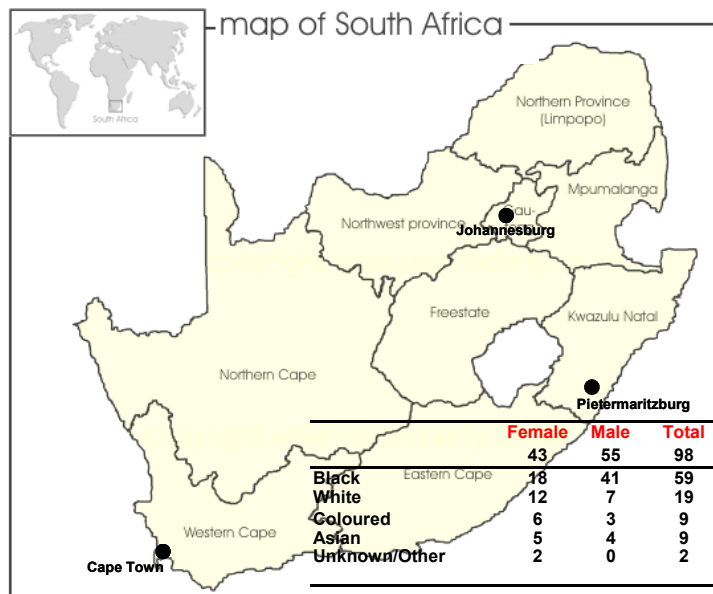


Fig.1. Demographic details of participants

Only two individuals did not to complete the section on identity and we have grouped them under the heading for ethnic group “Unknown/Other” in this analysis.

Mitochondrial DNA (mtDNA) results

The Southern African mtDNA haplogroup (branch) L0 and its sub-haplogroups (sub-branches) altogether made up 8% of mtDNA lineages in the total sample (see Fig. 2). Haplogroup L0 is commonly found in Khoe and San people and their maternal descendants, and can therefore be used as a marker to estimate Khoesan (termed used in reference to the combined group) maternal gene admixture in the other groups (see Fig. 3).

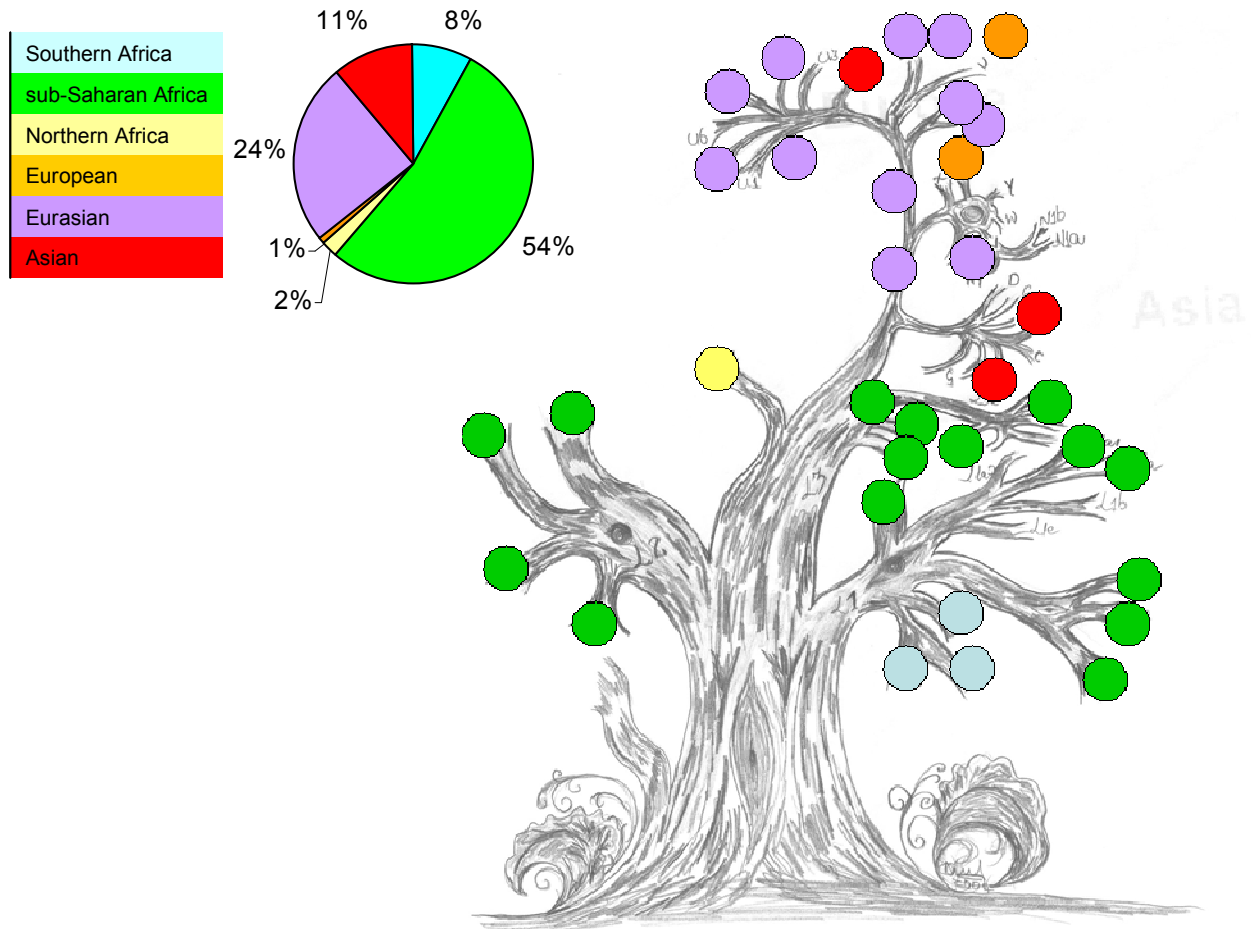


Fig. 2 Schematic diagram showing the branching pattern of mtDNA haplogroups on a tree and their geographic regions of origin (colour circles on tree correspond to geographic region). The pie chart summarizes the apportionment of the mtDNA variation derived from the different geographic regions in the total sample (98 individuals).

Of the 59 individuals who self-identified as Black, 19 came from other African countries (Nigeria, Ghana, Zimbabwe, Eritrea, Kenya, Zambia, Mozambique, Malawi, Namibia and Rwanda). Haplogroup L0d is commonly found in San and Khoe people and is almost exclusively found in Southern Africa, with a few

exceptions in Tanzania, Chad and Sudan. In the present study it was only found in the South African Black population who are Bantu-speakers and was not found in individuals from the other African countries (Fig. 3). Also, about 1 out of 9 Black people have mtDNA lineages derived from Eurasian (3.5%) and Asian (5.1%) origins. The newly defined haplogroup L4 that make up the North African mtDNA lineages was only found in the Black sample in an individual from Eritrea.

Geographic region of origin of mtDNA haplogroups	Black 59	White 19	Coloured 9	Asian 9	Unknown/Other 2	Total 98
Southern Africa	0.136					0.082
sub-Saharan Africa	0.746		0.667		1.000	0.531
Northern Africa	0.034					0.020
European		0.053				0.010
Eurasian	0.034	0.895		0.556		0.245
Asian	0.051	0.053	0.333	0.444		0.112

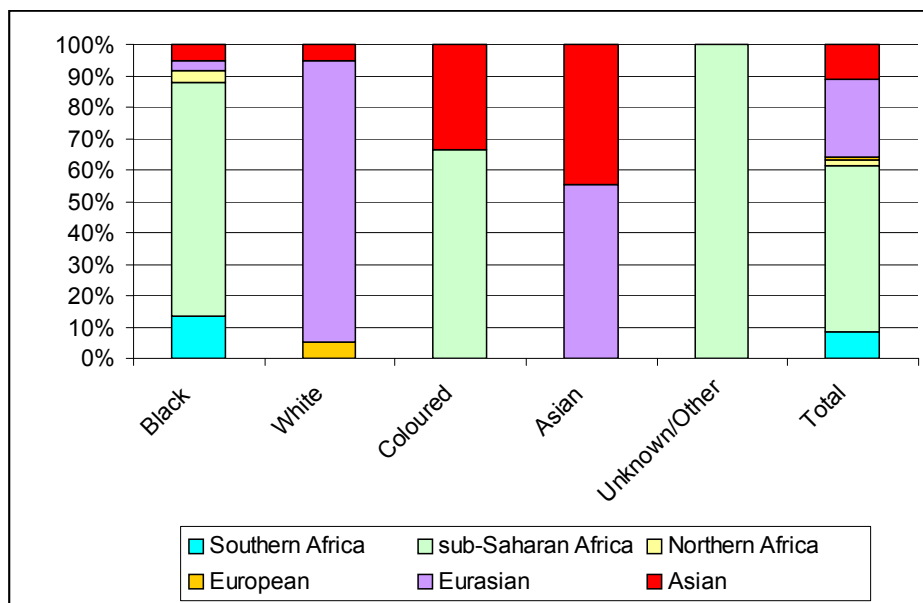


Fig.3 Geographic origins and distribution of mtDNA haplogroups

People who self-identified as White, Coloured or Asian were represented in the total sample as small groups with sample sizes of 19, 9, and 9, respectively, and were therefore not analysed in any detail at the population level.

For the most part there is good concordance between the self identified ethnic origins indicated by participants and the origins of their mtDNA haplogroups.

Y chromosome DNA results

Y chromosome haplogroup data was obtained for 55 males in the sample. The number of males within each group is given in Fig.4, and like for the mtDNA above, the sample sizes for the White, Coloured and Asian groups are very small to discuss in a meaningful way. Given that 75% of the males who participated

were Black, it was not surprising that the proportion of Y chromosomes that trace back to sub-Saharan Africa is found at the highest frequency (see Fig. 4). A small percentage of Eurasian and European derived Y chromosomes are found in the Black sample (~5%).

We have not described the haplotype diversity as deduced from the short tandem repeat analysis in this report. This part of the analysis would be done in the final report after combining the results from the different sampling locations.

Geographic region of origin of Y chromosome DNA haplogroups	Black 41	White 7	Coloured 3	Asian 4	Total 55
Southern Africa	0.049				0.036
sub-Saharan Africa	0.878		0.333		0.673
Northern Africa and Meditarreanian	0.024	0.143			0.036
European	0.024	0.143	0.667		0.073
Eurasian	0.024	0.714		0.750	0.164
Asian				0.250	0.018

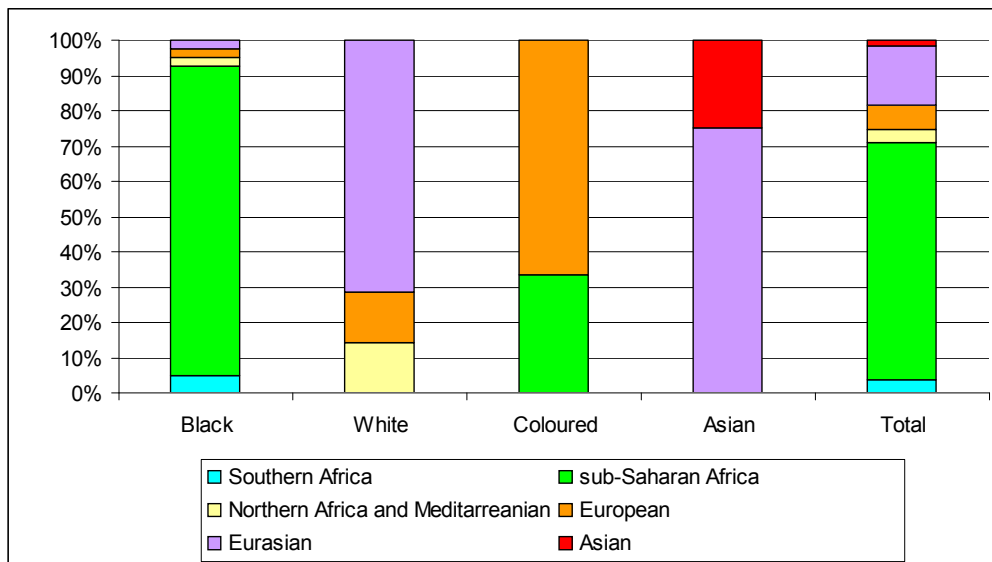


Fig. 4. Summary of Y chromosome DNA haplogroup variation in males sampled at Pietermaritzburg. Those Y chromosome haplogroups found both in Europe and Asia are referred to as Eurasian.

Acknowledgements

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