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FINAL REPORT: LIVING HISTORY PROJECT
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LIVING HISTORY PROJECT: FINAL REPORT

SAMPLING

Sampling was conducted in Cape Town, University of Kwa-Zulu Natal (Pietermaritzburg) and Johannesburg by the AGEI in conjunction with the Human Genomic Diversity and Disease Research Unit (National Health Laboratory Service & Wits). Altogether, 483 individuals were included in the study (see Table 1). [Note: The samples collected by Prof Soodyall (129 individuals from Ermelo, Marabastad & Johannesburg) was not included as part of this study]. A list of participants, their contact details and AGEI codes assigned to the individuals were given to the AGEI on request.

TABLE 1: Sample size by sex and self identified ethnicity.

	Female	Male	Total
Black	70	98	168
White	79	74	153
Coloured	44	53	97
– Cape Malay	(6)	(4)	(10)
Asian	10	11	21
Unknown/Other	17	17	34
	226	**257**	**483**

INDIVIDUAL RESULTS AND REPORTS

All subjects were given their individuals results in sealed envelopes and summaries of the results for the sampling conducted in Cape Town and UKZN were given to Prof Wilmot James at the AGEI in February and March 2008, respectively (see inclusions). Those individuals who could not attend the scheduled events to receive their results from Prof Soodyall and her team were given their reports in sealed envelopes from the AGEI office in Cape Town (Jacqui Le Roux) and by Jewel Koopman (Alan Paton Centre & Struggle Archives, University of KwaZulu-Natal). The 13 individuals sampled in Johannesburg were given their results in April; given the small sample size a summary report was not generated for this sample but is included in the final summary report.

DATABASE

A de-identified database with the complete mtDNA and Y chromosome DNA results using new LHP codes for each individual sampled was given to Prof Wilmot James in May 2008.

MTDNA DATA

TABLE 2: Distribution of mtDNA haplogroups in sample showing the most likely geographic region of origin of haplogroups (use colour scheme in key).

Key:
Southern Africa
sub-Saharan Africa
Northern Africa
European
Eurasian
Asian

mtDNA HG	Black	White	Coloured	Cape Malay	Asian	Unknown/Other	*Total*
L0d	12	6	17		2	5	*42*
L0d1	9		8			2	*19*
L0d2	5	2	5			4	*16*
L0d3	1	2					*3*
L0a	4		7				*11*
L0a1a	9						*9*
L0a2	2						*2*
L0f	2		1				*3*
L1b	1						*1*
L1b1	2		1				*3*
L1c*	1						*1*
L1c1	3		1				*4*
L1c1a			1				*1*
L1c1b	1						*1*
L1c2	7						*7*
L1c2b						1	*1*
L1c3	3		1			1	*5*
L2*		1					*1*
L2a	1		3				*4*
L2a1	3	1			1		*5*
L2a1a	2					1	*3*
L2a1b	16	1	7	1		1	*26*
L2b	2						*2*
L2c1	1						*1*
L2c2	3						*3*
L3*	2						*2*
L3b	2						*2*
L3b1	8						*8*
L3d1	6						*6*
L3d1	1						*1*
L3d2	2						*2*
L3d3	3						*3*
L3e1	2		1				*3*
L3e1a	3		2				*5*
L3e1b	6		2				*8*
L3e2			1				*1*
L3e2b	11		2		1		*14*
L3e3	5	1					*6*
L3f			1				*1*
L3f1	2						*2*
L3f2	1						*1*
L4b2	4						*4*
L5a	2						*2*
L5b	1						*1*

TABLE 2: (CONTINUED)

I		5					*5*
V		4	1			1	*6*
F1a			2				*2*
H	2	50	3	2		1	*58*
HV1		2					*2*
J		5	1			1	*7*
J1		3				1	*4*
J1a		2				1	*3*
J1b1		1					*1*
K		20	1	2		1	*24*
N	1	2				1	*4*
N1		1					*1*
N1a	1	2					*3*
N1b						1	*1*
R		1		3		1	*5*
T		3				1	*4*
T1		3					*3*
T2		11	1				*12*
T3	1						*1*
T4					1		*1*
U	1	4	3	1		1	*10*
U1a		1					*1*
U2					1		*1*
U2e		1					*1*
U4		2		1			*3*
U5		3					*3*
U5a		2					*2*
U5a1a		3				1	*4*
W		1	2				*3*
X		1	1	1			*3*
B	1		2			1	*4*
B4			2				*2*
D					1		*1*
M	8	1	4	7	4	1	*25*
M2			1				*1*
M2a			2			1	*3*
M3	1		2			1	*4*
M5			2	1		2	*5*
M6a			1				*1*
M7b2					1		*1*
M9a			1				*1*
U2b			1				*1*
U2c		5	3				*8*
U7		1			1		*2*
	167	*154*	*97*	*19*	*13*	*33*	*483*

Y CHROMOSOME DATA

TABLE 3: Distribution of Y chromosome DNA haplogroups in different ethnic groups. The most likely geographic region of origin of the Y chromosomes is shown in colour (use in conjunction with the key)

Key:
Southern Africa
sub-Saharan Africa
Northern Africa and Meditarreanian
European
Eurasian
Asian

Y Haplogroup	Black	White	Coloured	Cape Malay	Asian	Unknown/Other	*Total*
A3b1	1		1			1	*3*
A3b2*	2						*2*
B2a1	3						*3*
E*	1						*1*
E2b	8						*8*
E3a*	39	2	7				*48*
E3a1	1		1				*2*
E3a7	34		1				*35*
E3b1*	1		2				*3*
E3b1a*	2	2				1	*5*
I*	1	10	7			2	*20*
F*		1	2			2	*5*
G*		3	1				*4*
J*	2	2	1		1		*6*
J2*		2	3		1		*6*
K2		2					*2*
KM		1					*1*
R*	3	32	15	2	2	7	*61*
R1a*		5					*5*
R1a1*		7	3	2	3	2	*17*
R2		4				1	*5*
C*			1		2		*3*
H1			3		1	1	*5*
L*					2		*2*
O*			3				*3*
O3*		1	1				*2*
	98	*74*	*52*	*4*	*12*	*17*	*257*

CONTRASTING PATTERNS OF MTDNA AND Y CHROMOSOME DNA VARIATION

The overall distribution of mtDNA (**Fig.1**) and Y chromosome DNA **(Fig. 2**) haplogroups derived from southern African Khoisan (SA), other regions in sub-Saharan Africa (SSA), North Africa (NA), European (EU), Eurasian (EA) – ie., haplogroups found both in Europe and Asia, and Asia (AS) are given for each group below.

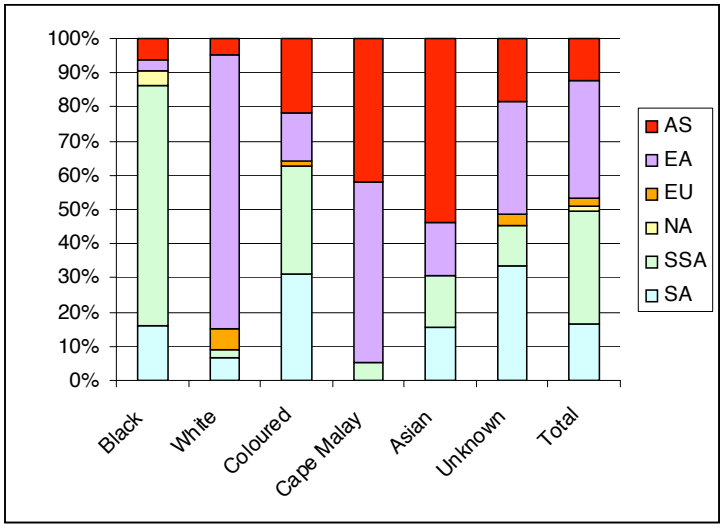

Fig. 1. MtDNA diversity in ethnic groups sampled.

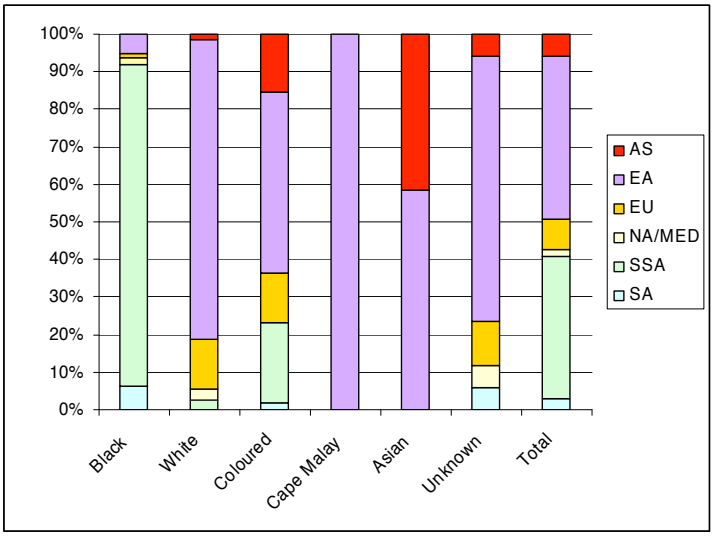

Fig. 2. Y chromosome diversity in ethnic groups sampled.

CONCLUSION

In the total sample of 483 individuals, 36% self-identified as Black (1), 32% as White (2), 21% as Coloured (including those who identified as Cape Malay)(3), 4% as Asian (4) and 7% did not disclose their ethnic group affiliation (5) (refer to Fig 3a). All individuals were tested for mtDNA variation: 51% of mtDNA haplogroups originated in Africa (17% Khoisan, 33% sub-Saharan African and 1% North African); 2% were traced to European origins, 35% Eurasian origin and 12% were derived from Asia (refer to Fig 3c).

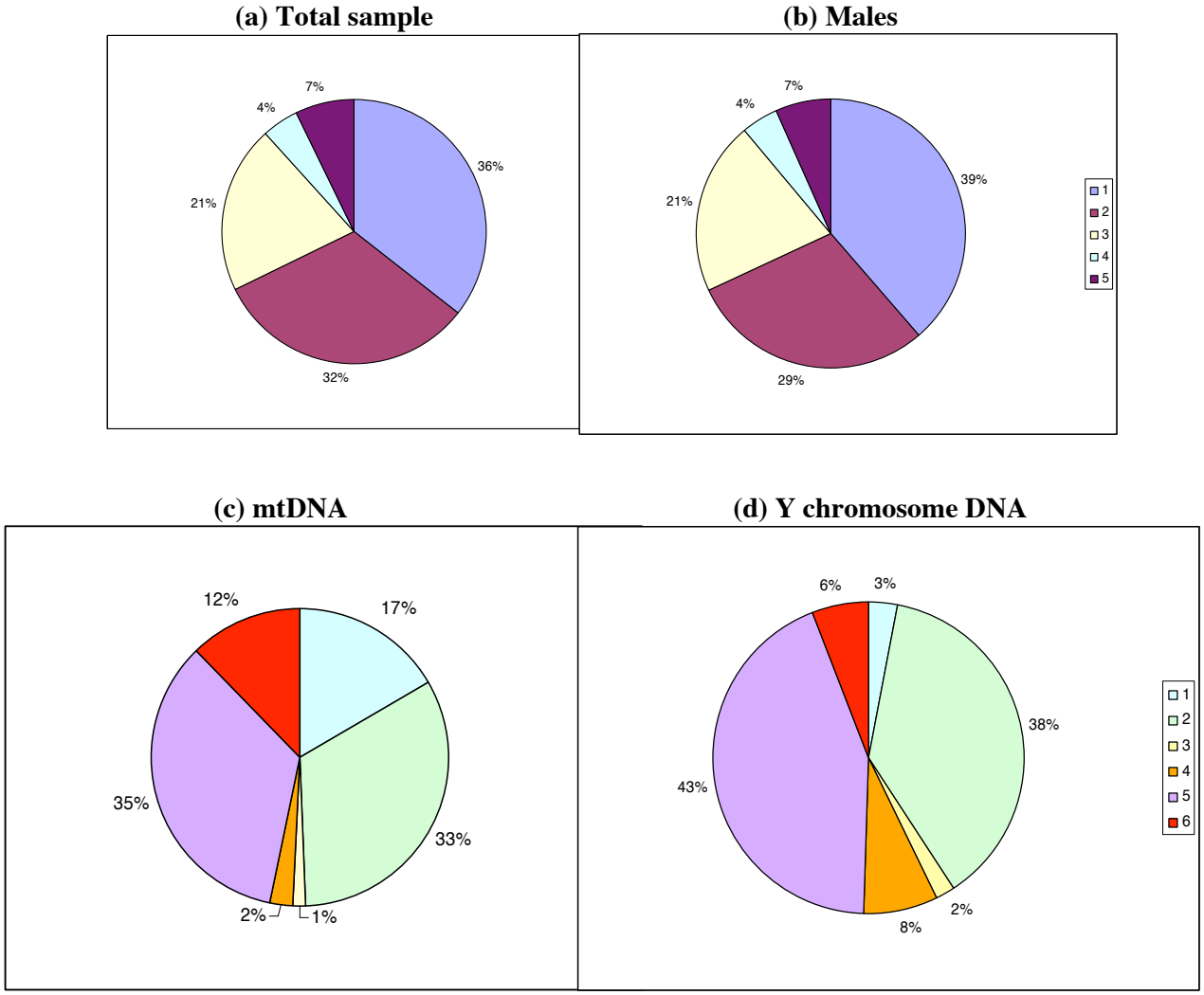

Fig. 3. Ethnic breakdown of total sample – both males and females (a) and only males (b) and the apportionment of mtDNA (c) and Y chromosome (d) haplogroups in sample traced to geographic regions of origin (same colour scheme used earlier).

Among the males tested, 39% self identified as Black, 29% as White, 21% Coloured (including Cape Malay), 4% as Asian and 7% did not disclose their ethnic affiliation (refer Fig. 3b). Using the Y chromosome haplogroups we find that 43% trace to African origins; 8% to European, 43% to Eurasian and 6% to Asian sources. These

results indicate that males and females have contributed differently to the gene pool of populations in southern Africa.

These results clearly demonstrate that a person's genetic ancestry may differ from their concept of identity. One of the major messages of this study is to drive home the point that the genetic markers used for genetic ancestry tests **cannot** reveal a person's identity or explain why they may look the way they do. The study also reaffirms the rich genetic diversity that exists in the southern part of Africa which is the result of different migrations from other parts of Africa to the region as well as contributions from sea-borne immigrants since mid 1600s.

ACKNOWLEDGEMENTS

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