Regional panel: IV Cape and (south)west coast

UPPSALA UNIVERSITET

Jakobsson Lab

Carina Schlebusch

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Jakobsson Lab, Department of Evolutionary Biology

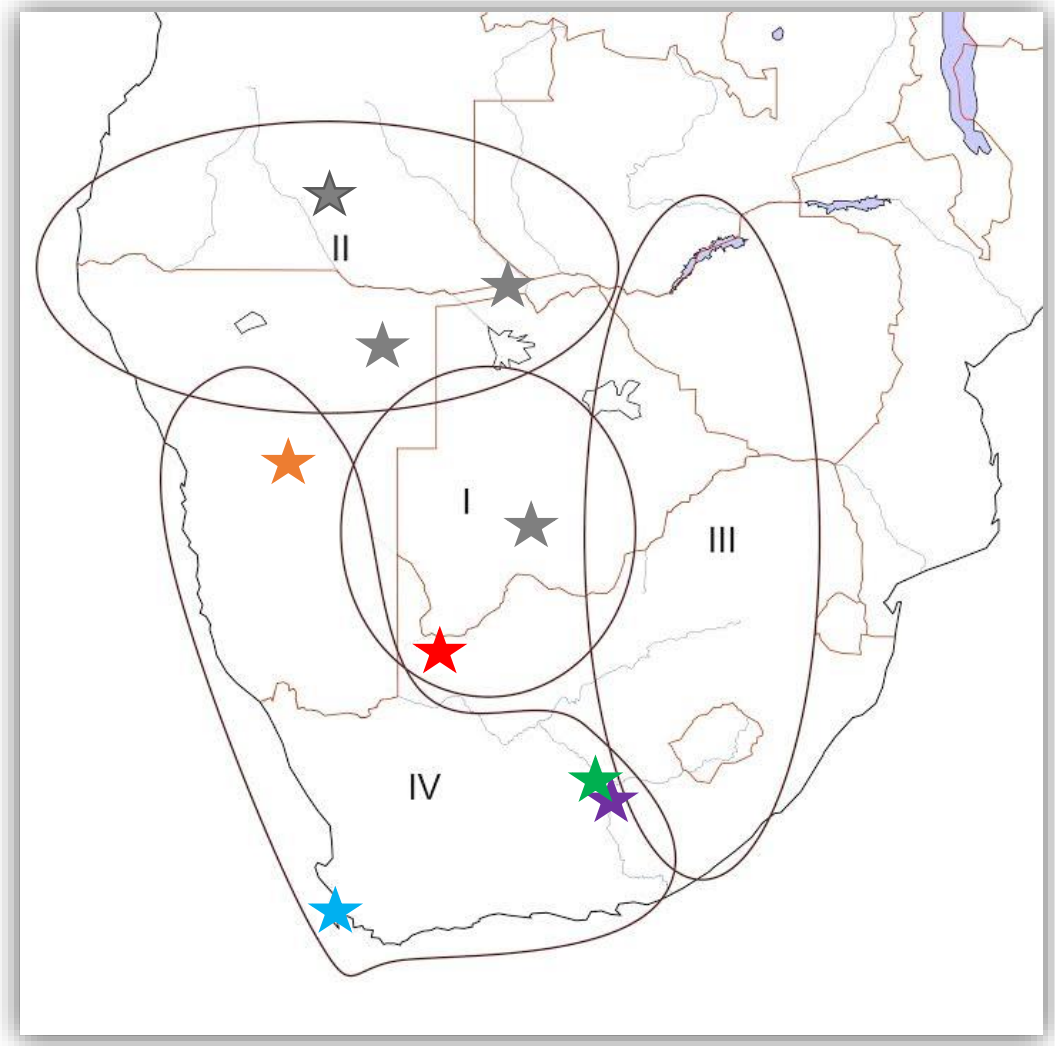
Evolutionary Biology Centre

Uppsala University, Sweden

UNIVERSITY OF THE WITWATERSRAND JOHANNESBURG

Speaking (of) Khoisan, Leipzig 14-16 May 2015

Regional panel: IV Cape and (south)west coast



Nama - Windhoek

≠Khomani - Askham

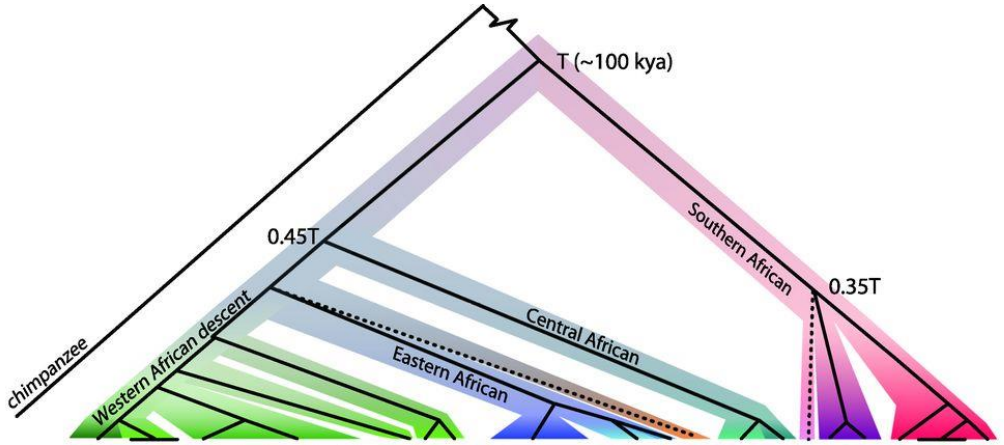
Karretjie People - Colesberg

“Coloured” - Colesberg

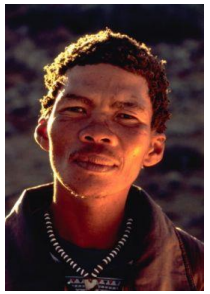
“Coloured” - Wellington


Great
Namaqua
Einiqua
Korana
Nama
Little
Namaqua
SAN
Guriqua
Cochoqua
Gonaqua
Peninsular
Khoikhoi
Attaqua
Inqua
Damaqua
Hessequa
KHOIKHOI
Gamtoos
Chainoqua
Gouriqua

Autosomal inferences





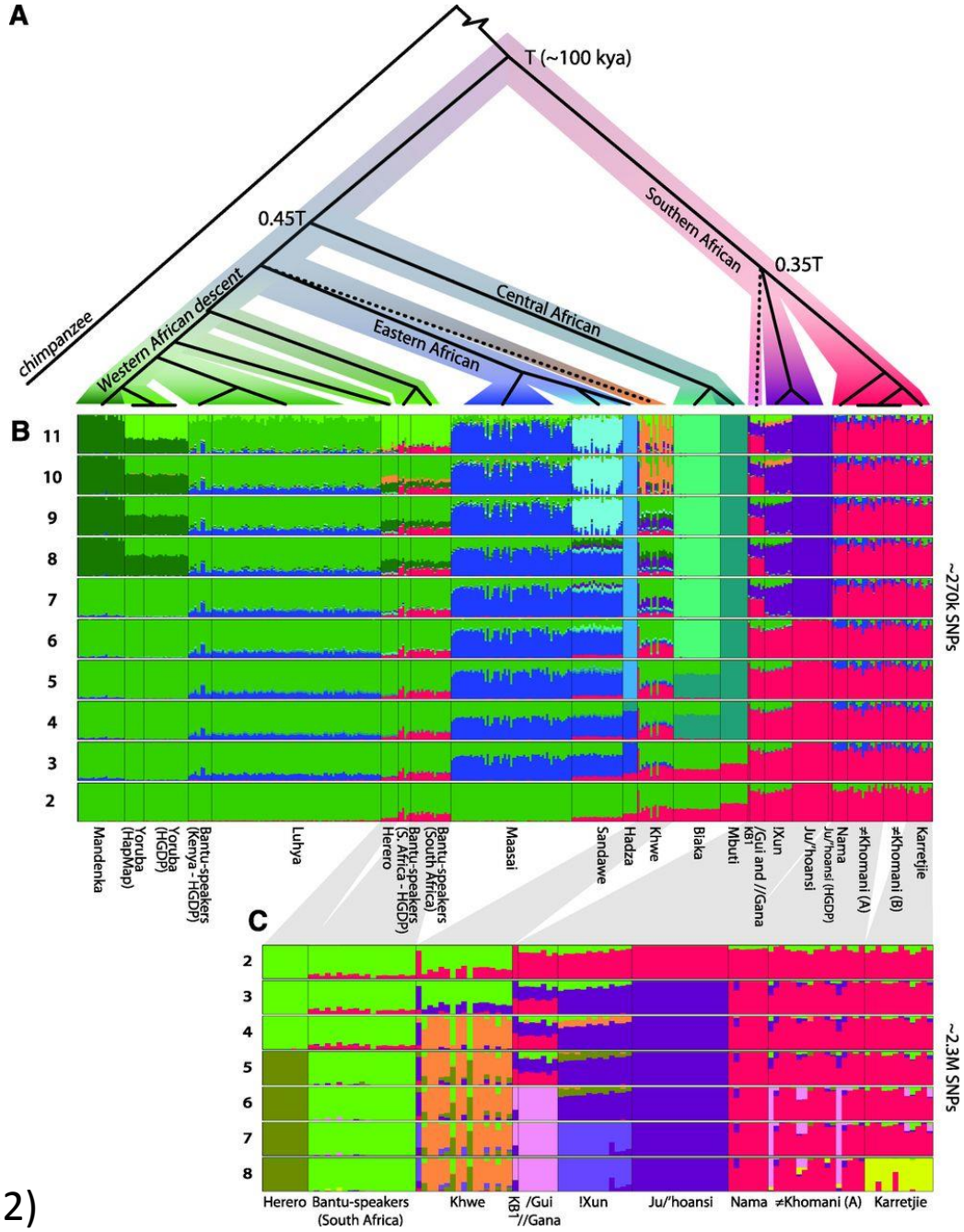



West Africans | East Africans | ”Pygmy” Central Afr | Khoe-San Southern Afr

Genomic Variation in Seven Khoe-San Groups Reveals Adaptation and Complex African History

Carina M. Schlebusch,[1]*† Pontus Skoglund,[1]† Per Sjödin,[1] Lucie M. Gattepaille,[1] Dena Hernandez,[2] Flora Jay,[3] Sen Li,[1] Michael De Jongh,[4] Andrew Singleton,[2] Michael G. B. Blum,[5] Himla Soodyall,[6] Mattias Jakobsson[1,7]*

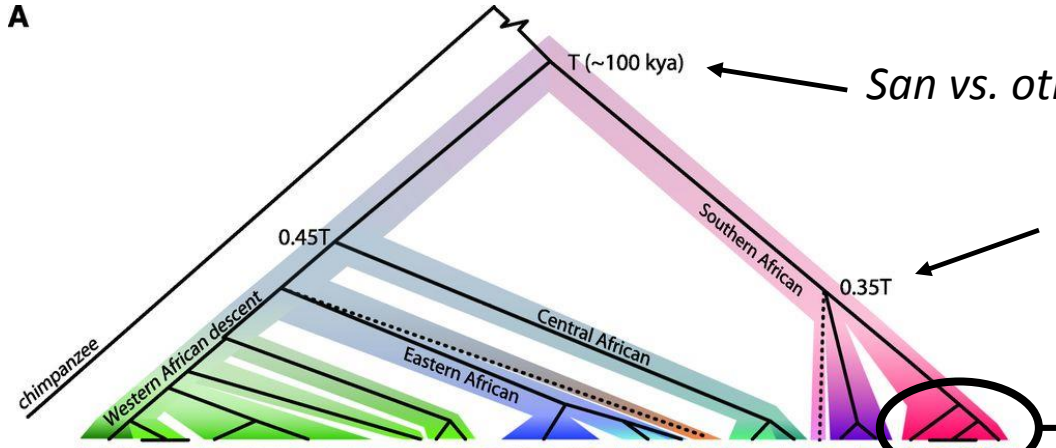
19 OCTOBER 2012 VOL 338 **SCIENCE** www.sciencemag.org



Africa
~260K SNPs

Southern Africa
~2,3M SNPs

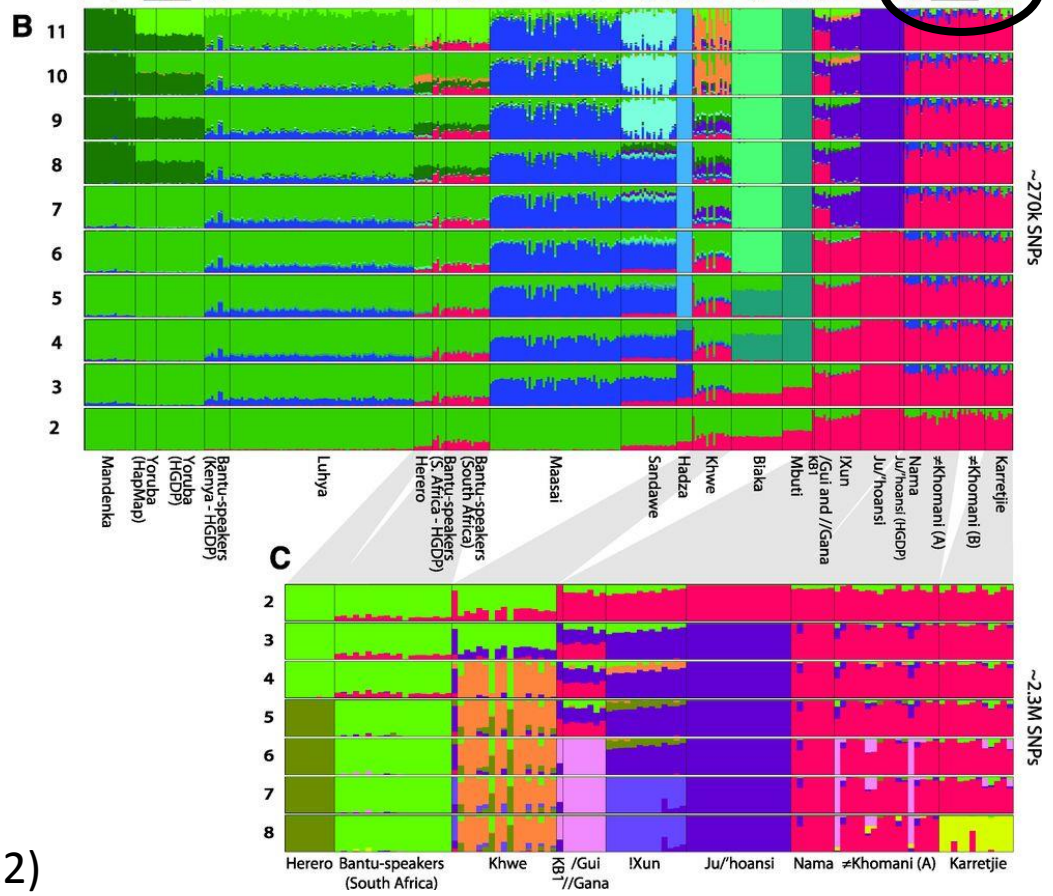
Schlebusch *et al.*
Science **338**, 374 (2012)



San vs. other split

"northern San" vs "southern San" "split"

Southern Khoe-San (Nama, Karretjie, Khomani, Coloured groups)



Africa
~260K SNPs

Southern Africa
~2,3M SNPs

Schlebusch *et al.*
Science **338**, 374 (2012)

CS
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11
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Mandenka
Yoruba (HapMap)
Yoruba (HGDP)
Bantu-speakers (Kenya - HGDP)
Luhya
Herero
Bantu-speakers (S. Africa - HGDP)
Bantu-speakers (South Africa)
Maasai
Sandawe
Hadza
Khwe
Biaka
Mbuti
/Gui and //Gana KB1
!Xun
Ju/'hoansi
Ju/'hoansi (HGDP)
Nama
≠Khomani (A)
≠Khomani (B)
Karretjie
C

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/Gui and //Gana KB1
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Ju/'hoansi
Nama
Ju/'hoansi (HGDP)
≠Khomani (A)
≠Khomani (B)
Karretjie
C
Nama

Current Biology

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Report

Lactase Persistence Alleles Reveal Partial East African Ancestry of Southern African Khoe Pastoralists

Gwenna Breton,[1,2,6] Carina M. Schlebusch,[1,6,*] Marlize Lombard,[3] Per Sjödin,[1] Himla Soodyall,[4] and Mattias Jakobsson[1,5,*]

Results and Discussion

We sequenced 360 bp of the lactase persistence (LP)-regula-
tory region, encompassing all known LP-regulatory variants
hern African popula-
ure 1, and the Sup-
e sample includes
which historically
San represent the
tors of current-day
tively as Khoe-San)
from the ancestors
subsequent admix-
mers arrived in the
housand years ago
some 1.3 thousand
ces of domesticated
about 2 thousand
to an introduction of

ease cite this article in press as: Macholdt et al., Tracing Pastoralist Migrations to Southern Africa with Lactase Persistence Alleles, Current Biology (2014), http://dx.doi.org/10.1016/j.cub.2014.03.027

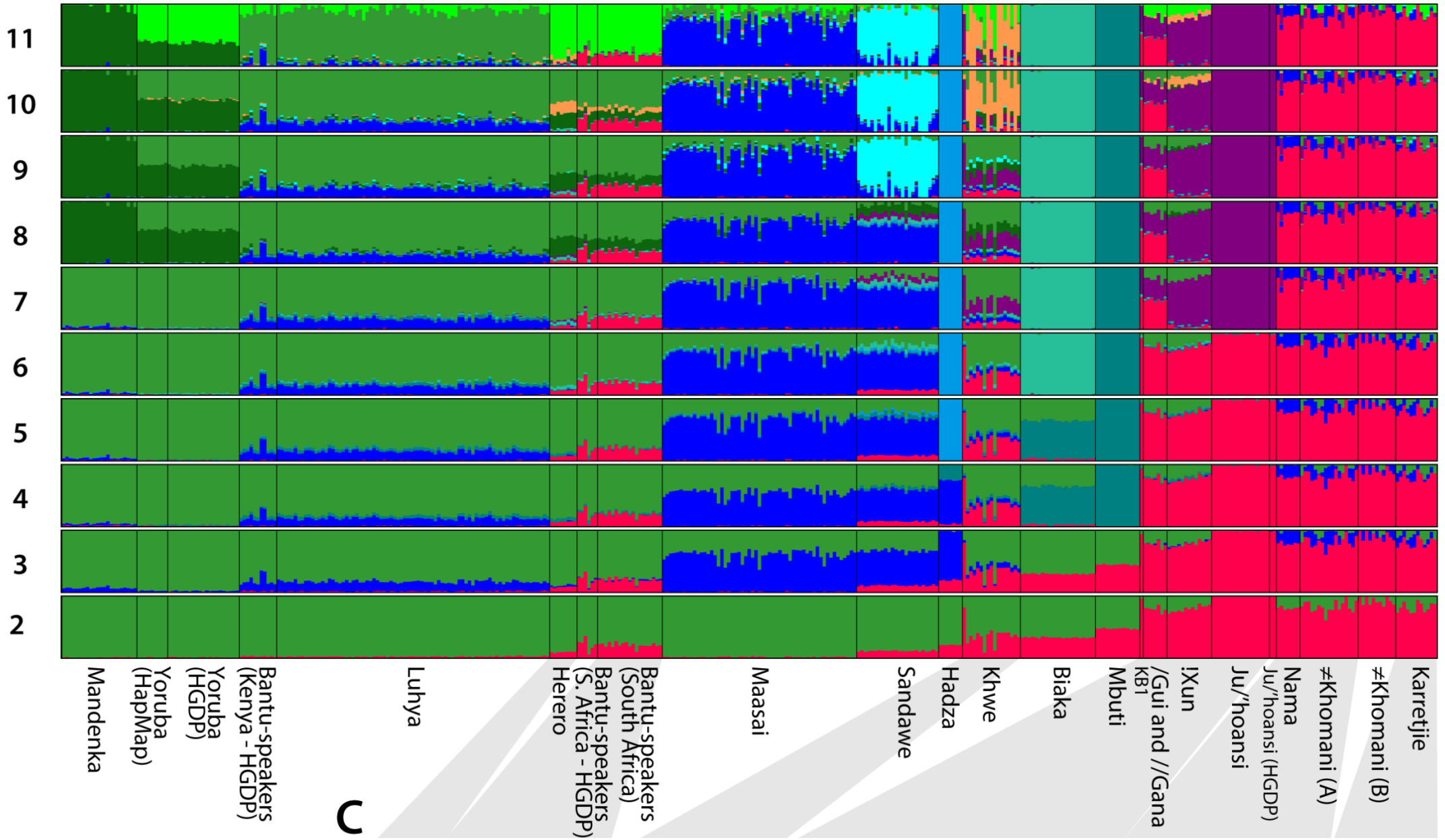
Current Biology 24, 1–5, April 14, 2014 ©2014 Elsevier Ltd All rights reserved http://dx.doi.org/10.1016/j.cub.2014.03.027

Report

Tracing Pastoralist Migrations to Southern Africa with Lactase Persistence Alleles

Enrico Macholdt,[1] Vera Lede,[1] Chiara Barbieri,[1,5] Sununguko W. Mpoloka,[2] Hua Chen,[3] Montgomery Slatkin,[3] Brigitte Pakendorf,[4,*] and Mark Stoneking[1,*]
[1]Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Deutscher Platz 6, 04103 Leipzig, ermany

click sounds, in fact Khoisan populations exhibit considerable diversity in languages, subsistence, and phenotype [13–15].
While it has been commonly assumed that Khoisan groups diverged early in the history of modern humans and have since remained relatively isolated, there is growing evidence of multiple migrations that have contributed to the current gene pool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Yoruba (HapMap)
Yoruba (HGDP)
Bantu-speakers (Kenya - HGDP)
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Ju/'hoansi (HGDP)
Nama
≠Khomani (A)
≠Khomani (B)
Karretjie
C
Karretjie People

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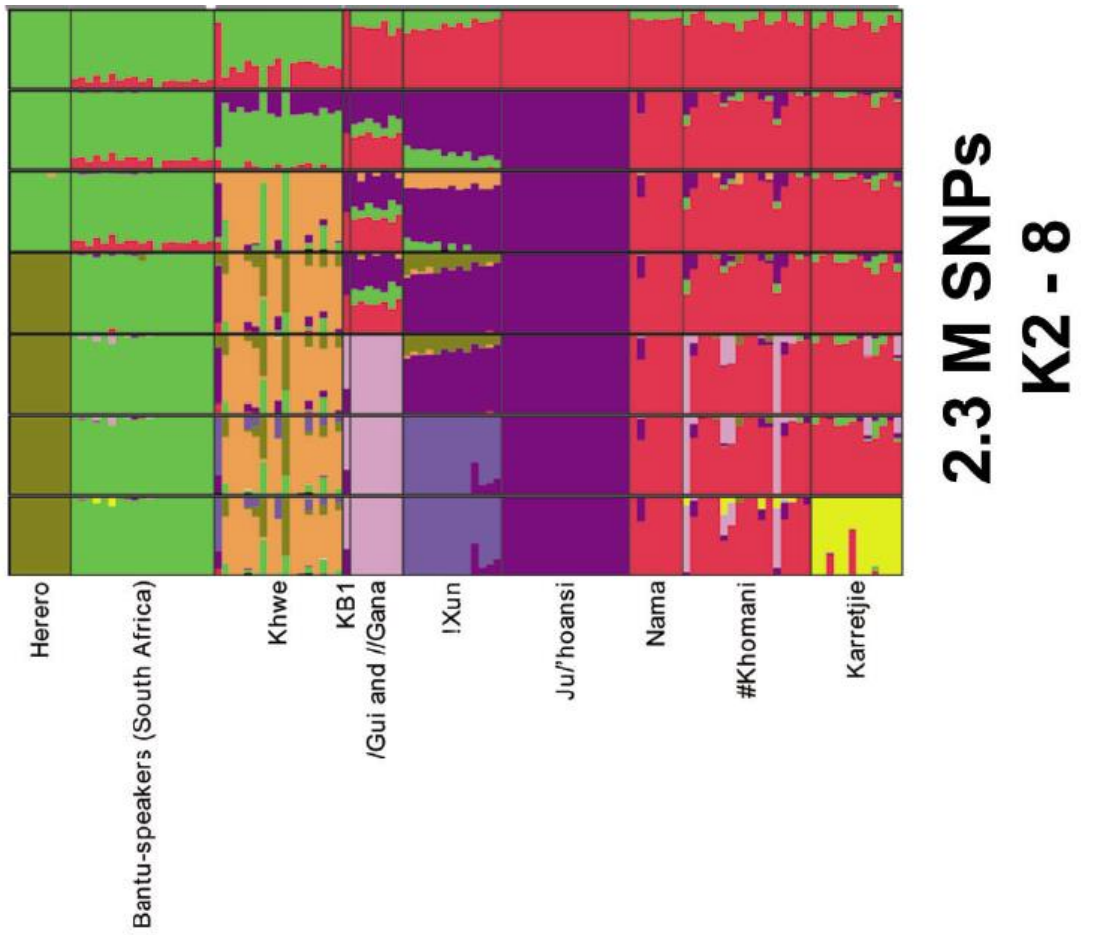
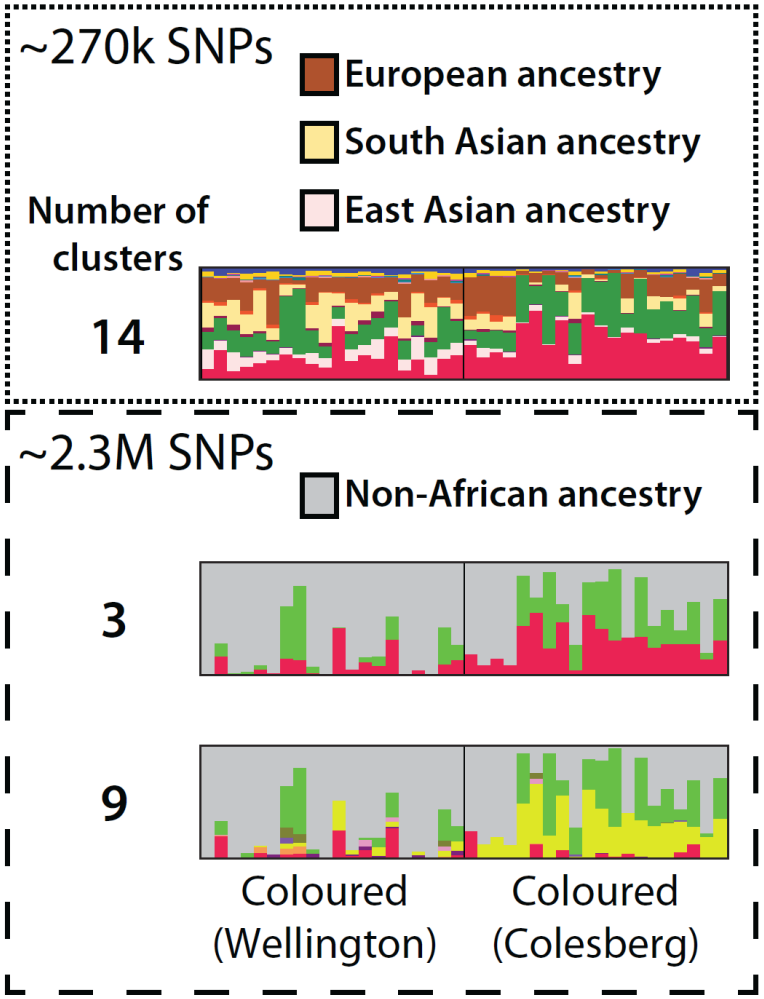
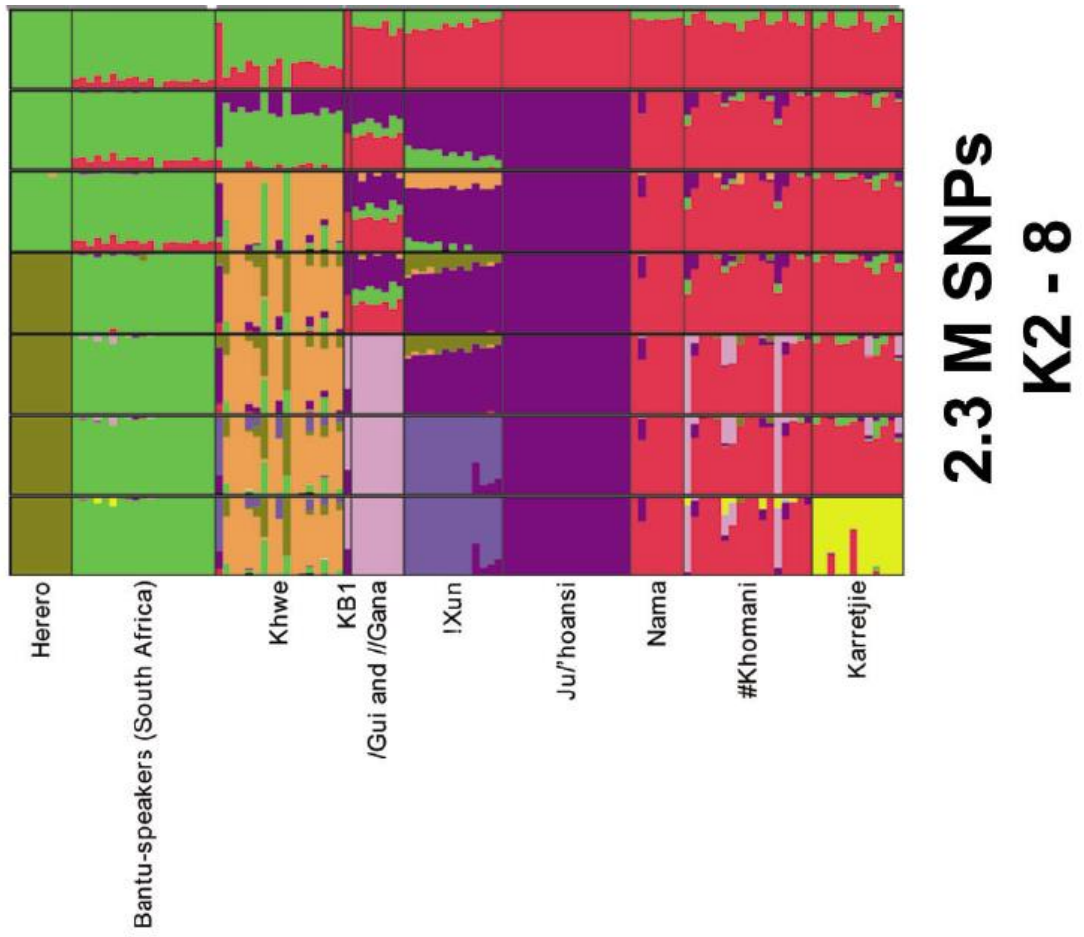
Herero
Luhya
Bantu-speakers
(Kenya - HGDP)
Yoruba
(HGDP)
Yoruba
(HapMap)
Mandenka
C
≠Khomani

High density dataset – Southern Africa

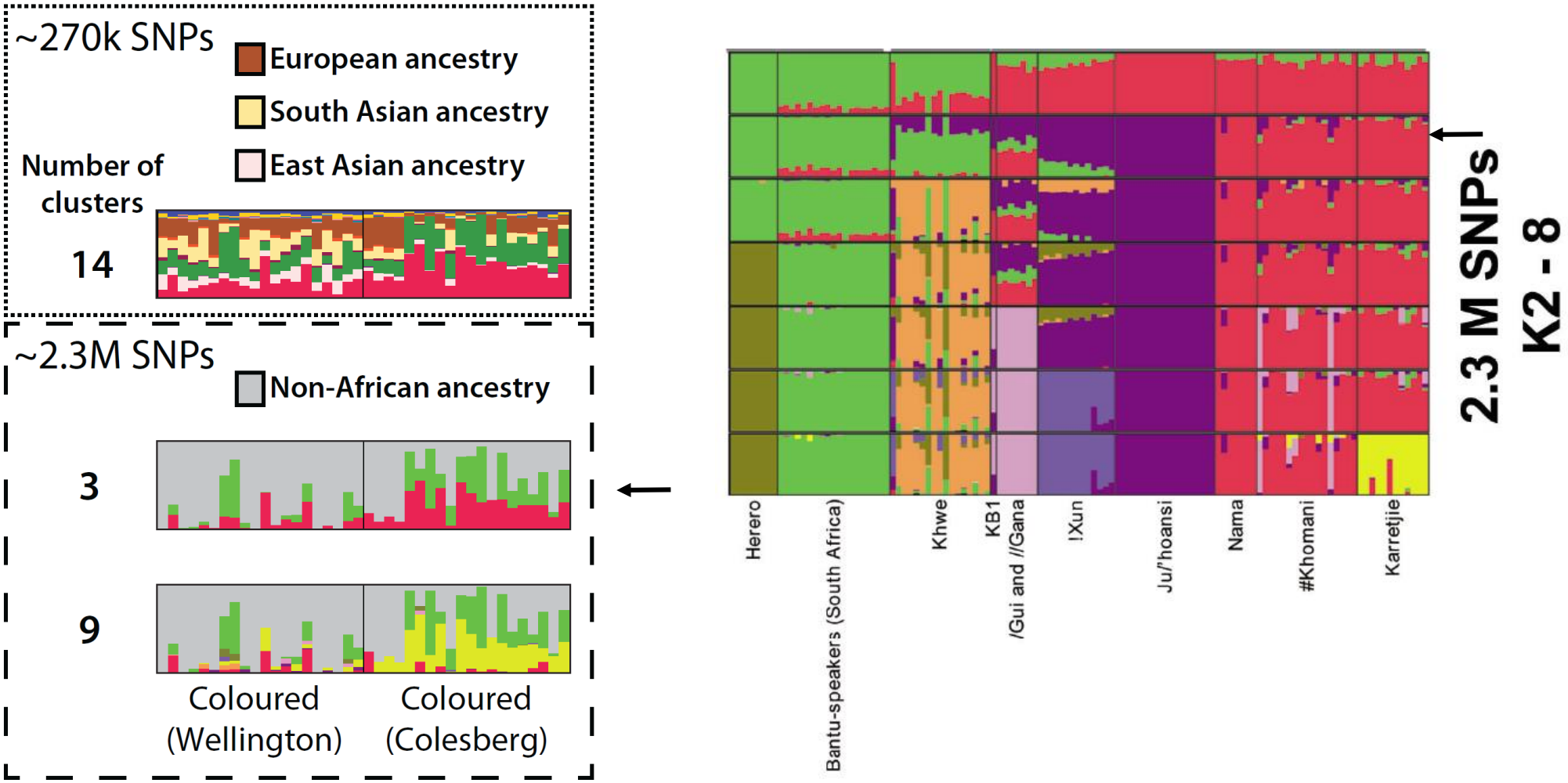


High density dataset – Southern Africa

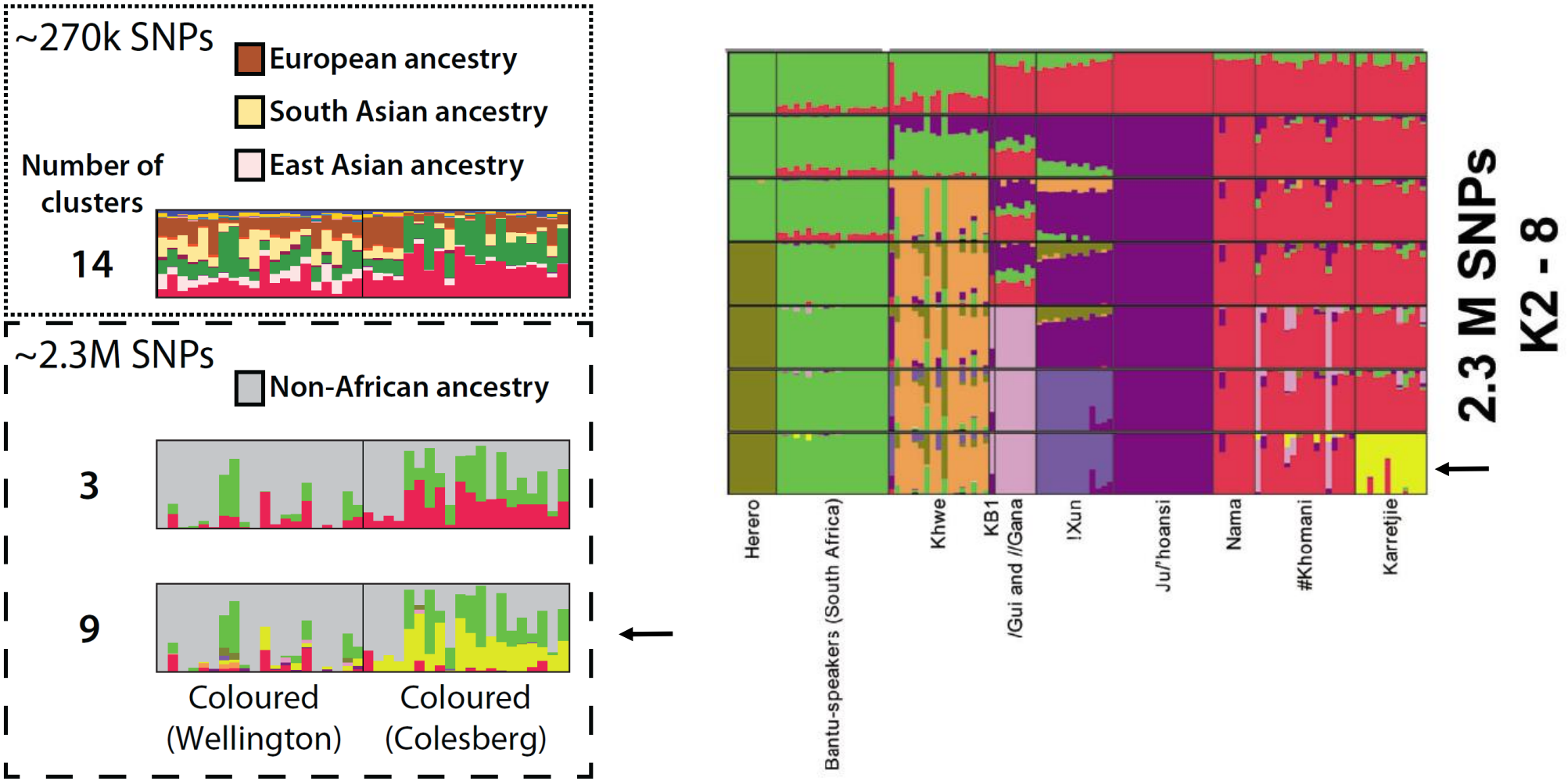




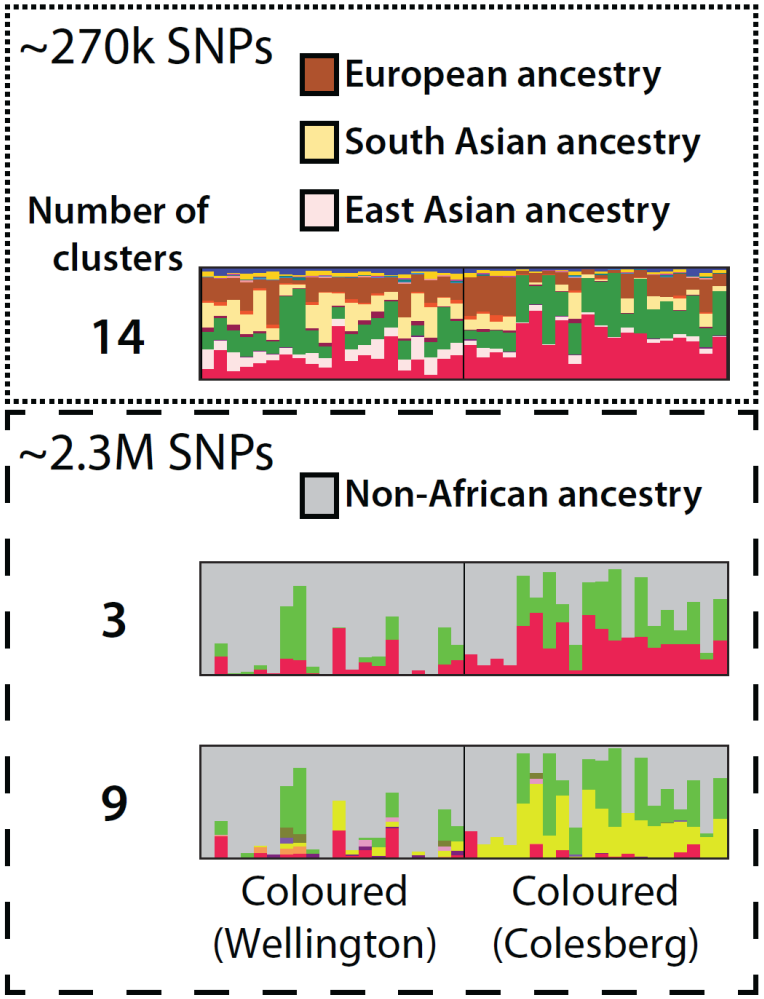
High density dataset – Southern Africa

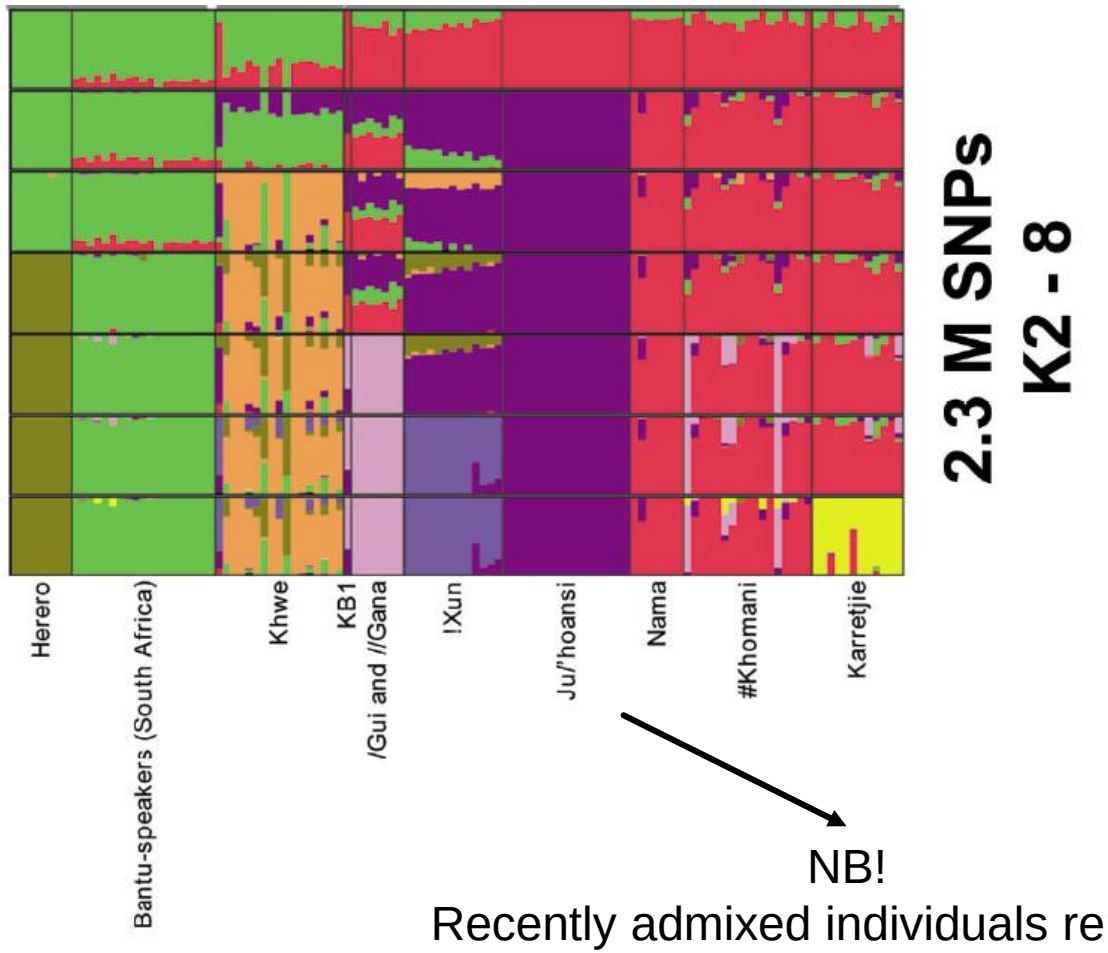


High density dataset – Southern Africa



High density dataset – Southern Africa

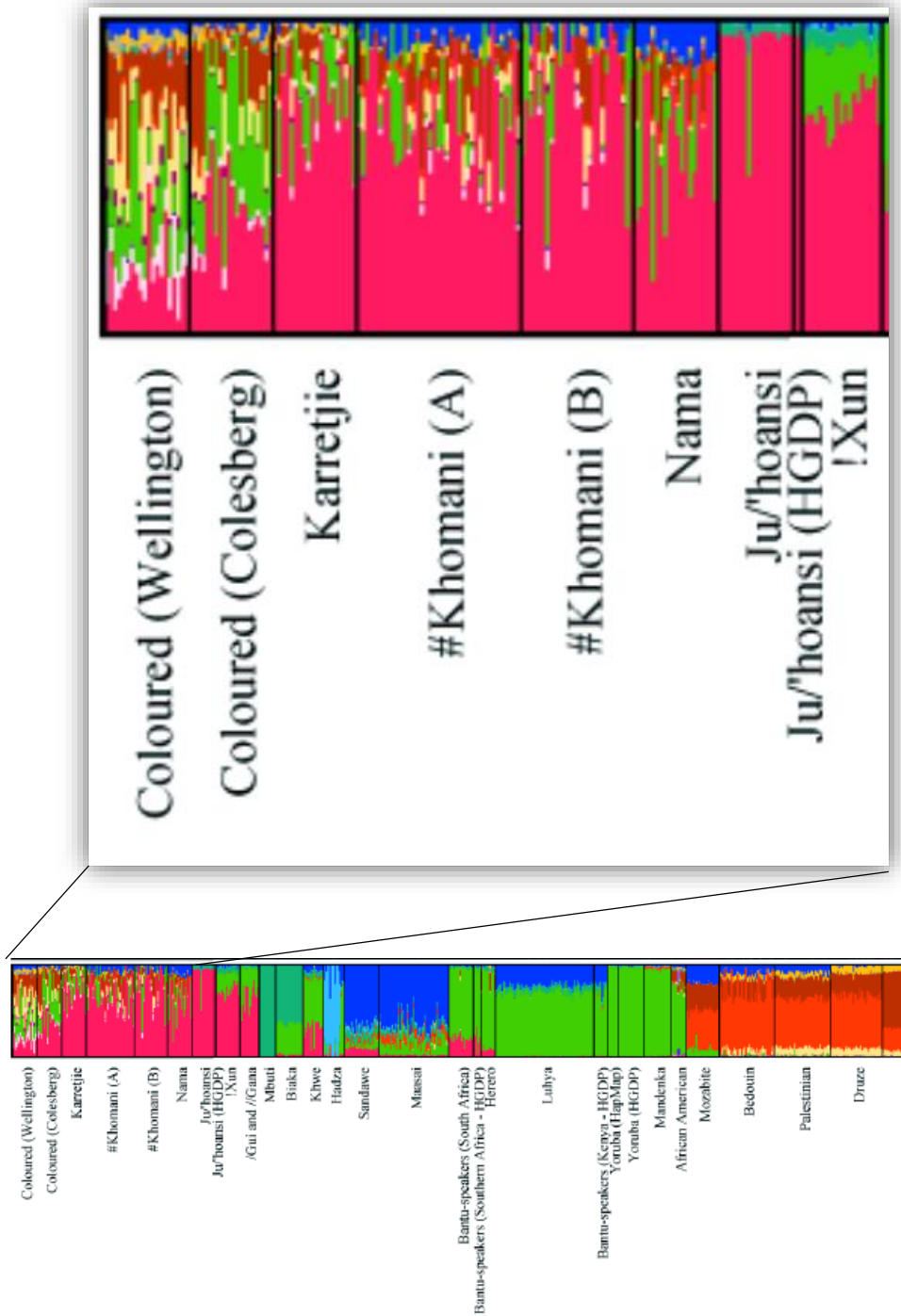




NB!

Recently admixed individuals removed.

Population genetic tool – not population composition today!

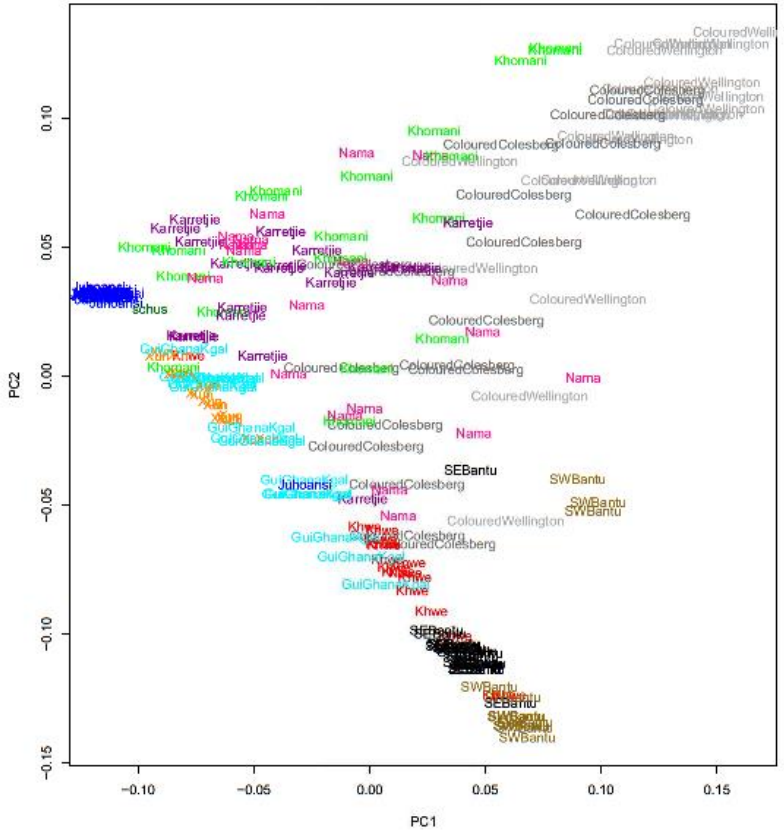


Non- admixed removed dataset Together with global comparative populations

K15

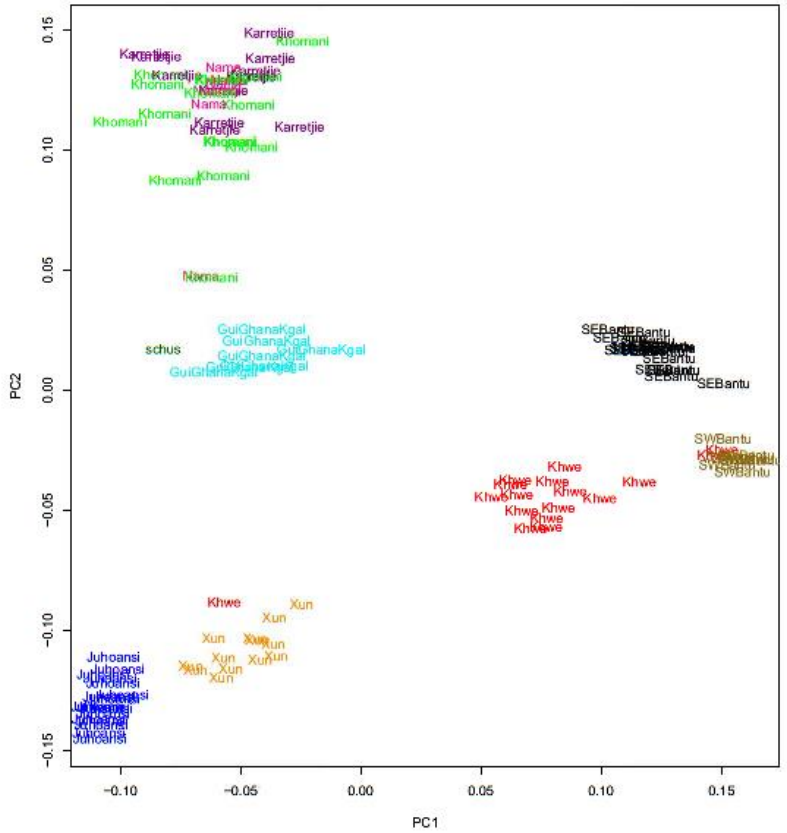
PCA

Full SA dataset



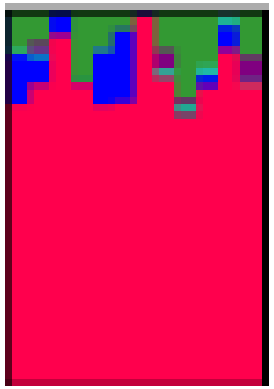
PCA

Admixed removed SA dataset

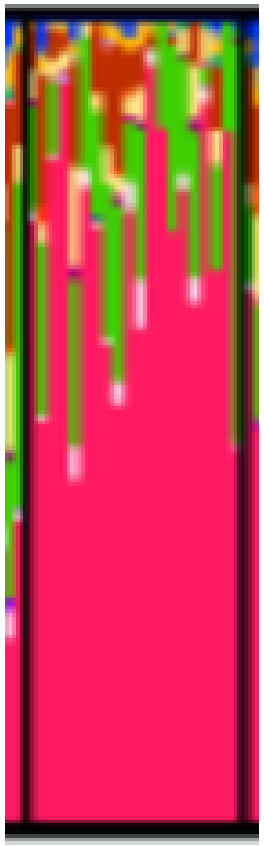


Karretjie People

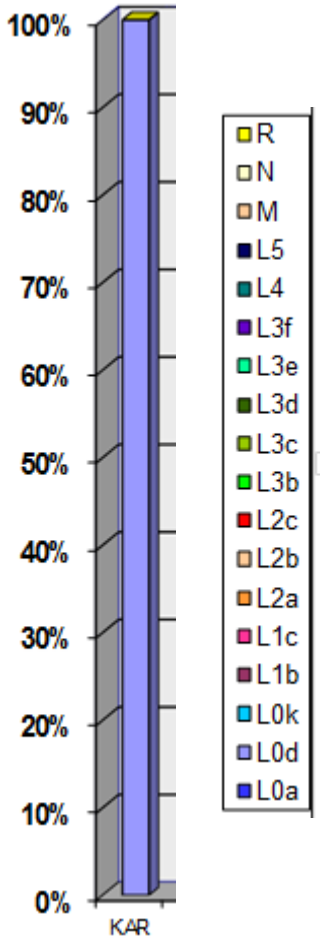
Autosomal
Recent Admixed
removed



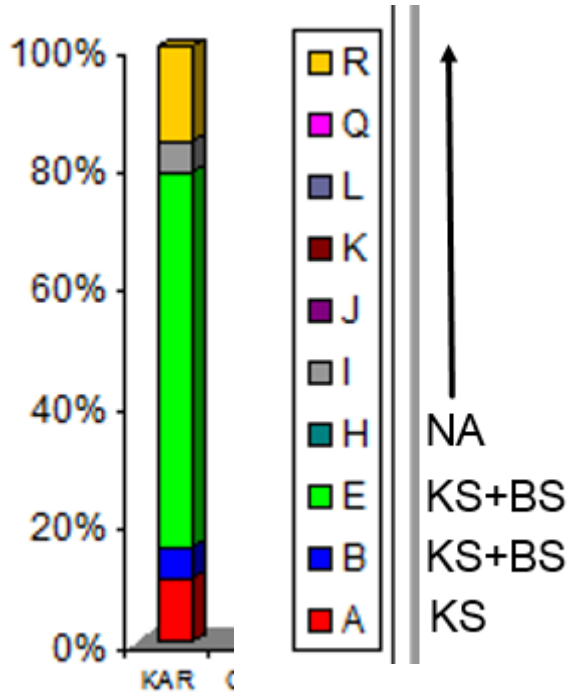
Autosomal
Unfiltered global



MtDNA

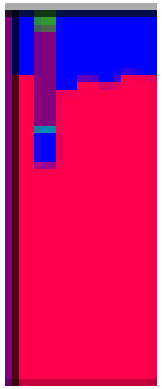


Y-chromosome

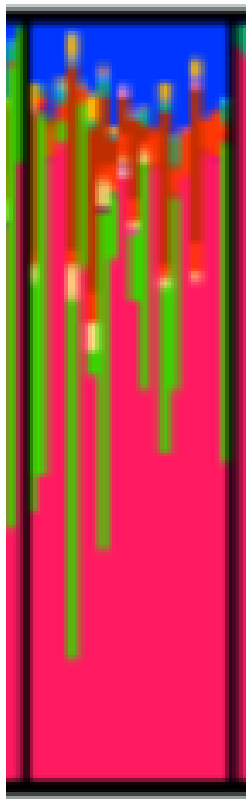


Nama

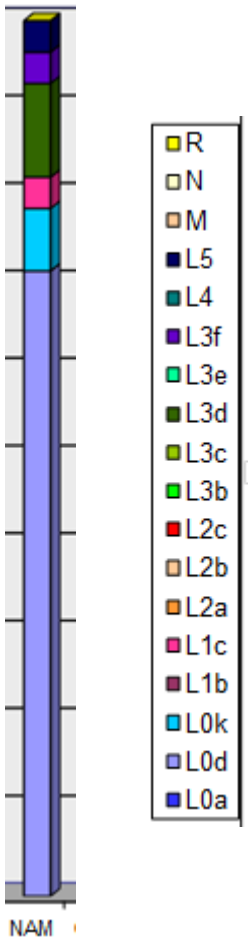
Autosomal
Recent Admixed
removed



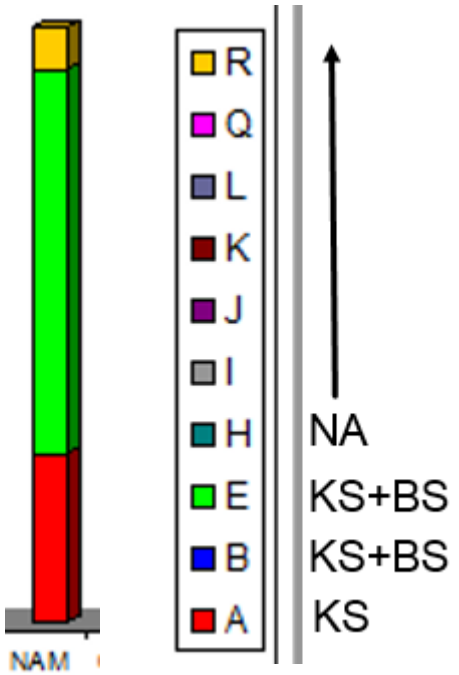
Autosomal
Unfiltered global



MtDNA

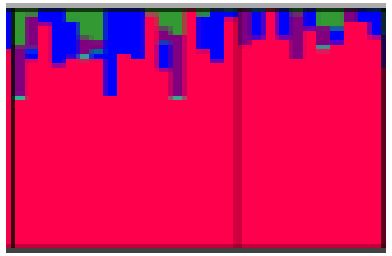


Y-chromosome

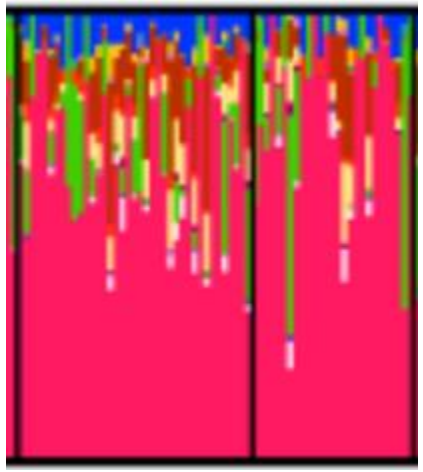


≠Khomani

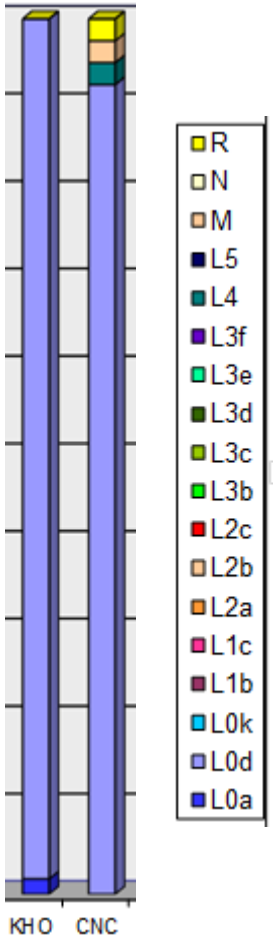
Autosomal
Recent Admixed
removed



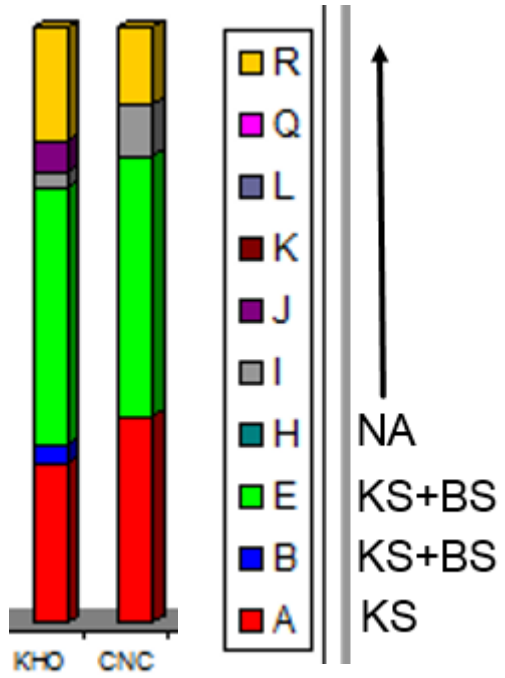
Autosomal
Unfiltered global



MtDNA

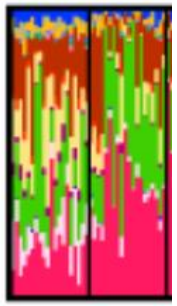


Y-chromosome

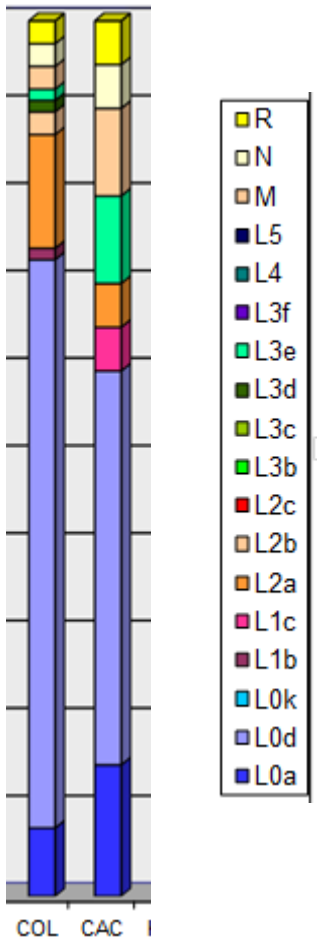


Coloured groups

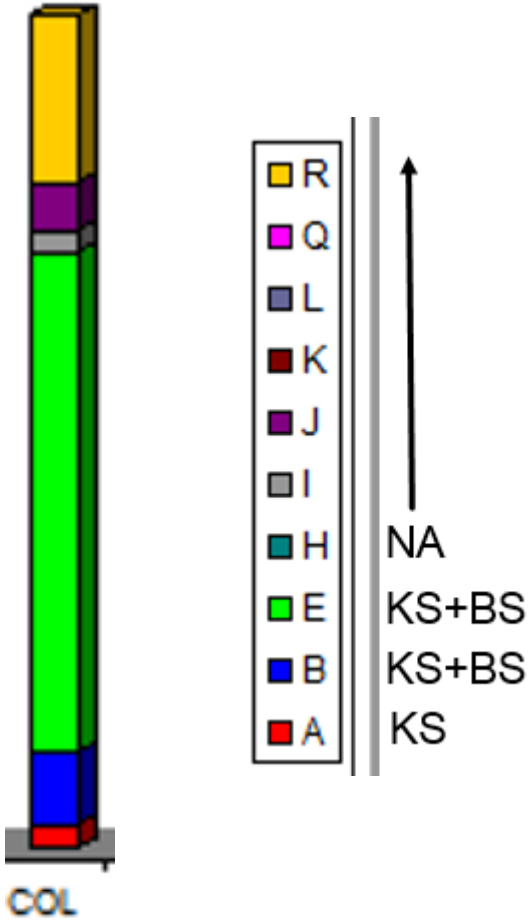
Autosomal
Unfiltered global



MtDNA



Y-chromosome



END - Thanks!

MtDNA and Y-chr details to follow

(If time allows of for information)

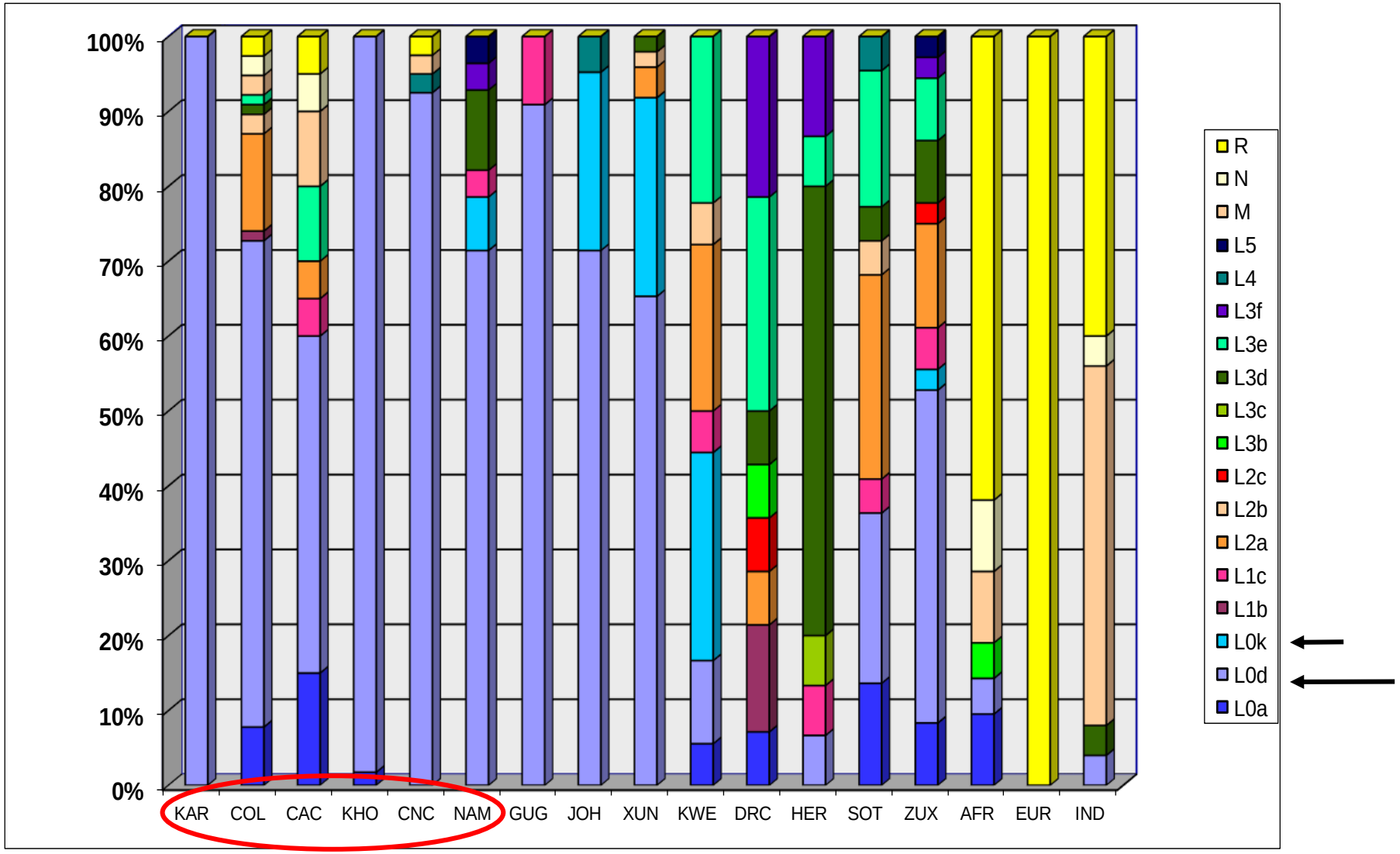
Mitochondrial DNA inferences

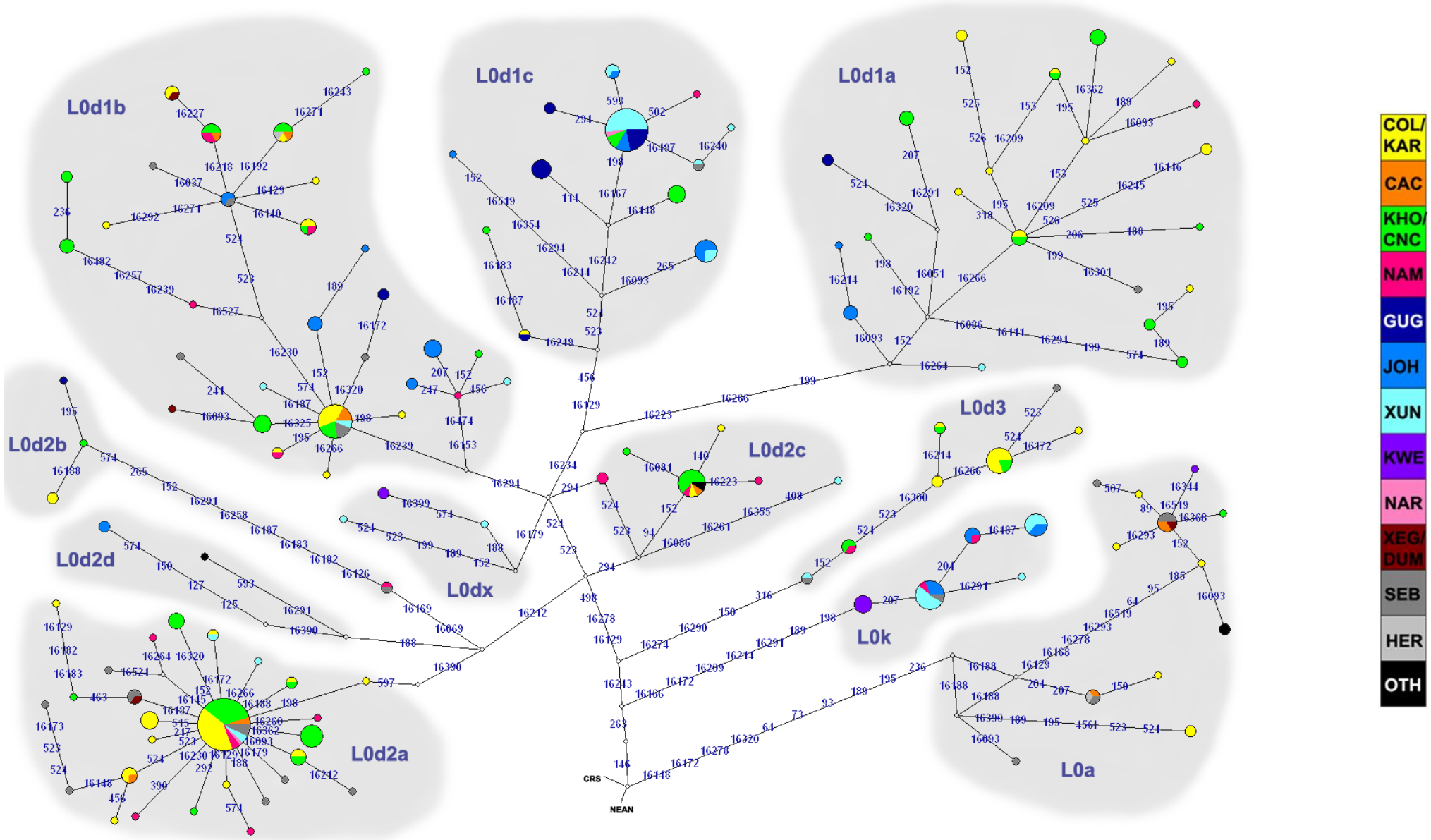
-Schlebusch, C. M. (2010). PhD Thesis: Genetic variation in Khoisan-speaking populations from southern Africa. Division of Human Genetics. Johannesburg, University of the Witwatersrand. **PhD thesis**.

-Schlebusch, C. M., M. Lombard, et al. (2013). "MtDNA control region variation affirms diversity and deep sub-structure in populations from Southern Africa." BMC Evol Biol **13**(1): 56.

-Schlebusch, C. M., M. de Jongh, et al. (2011). "Different contributions of ancient mitochondrial and Y-chromosomal lineages in 'Karretjie people' of the Great Karoo in South Africa." J Hum Genet **56**(9): 623-30.

Mitochondrial haplogroups

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L0d1c
L0d1a
L0d2b
L0d2c
L0d3
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L0dx
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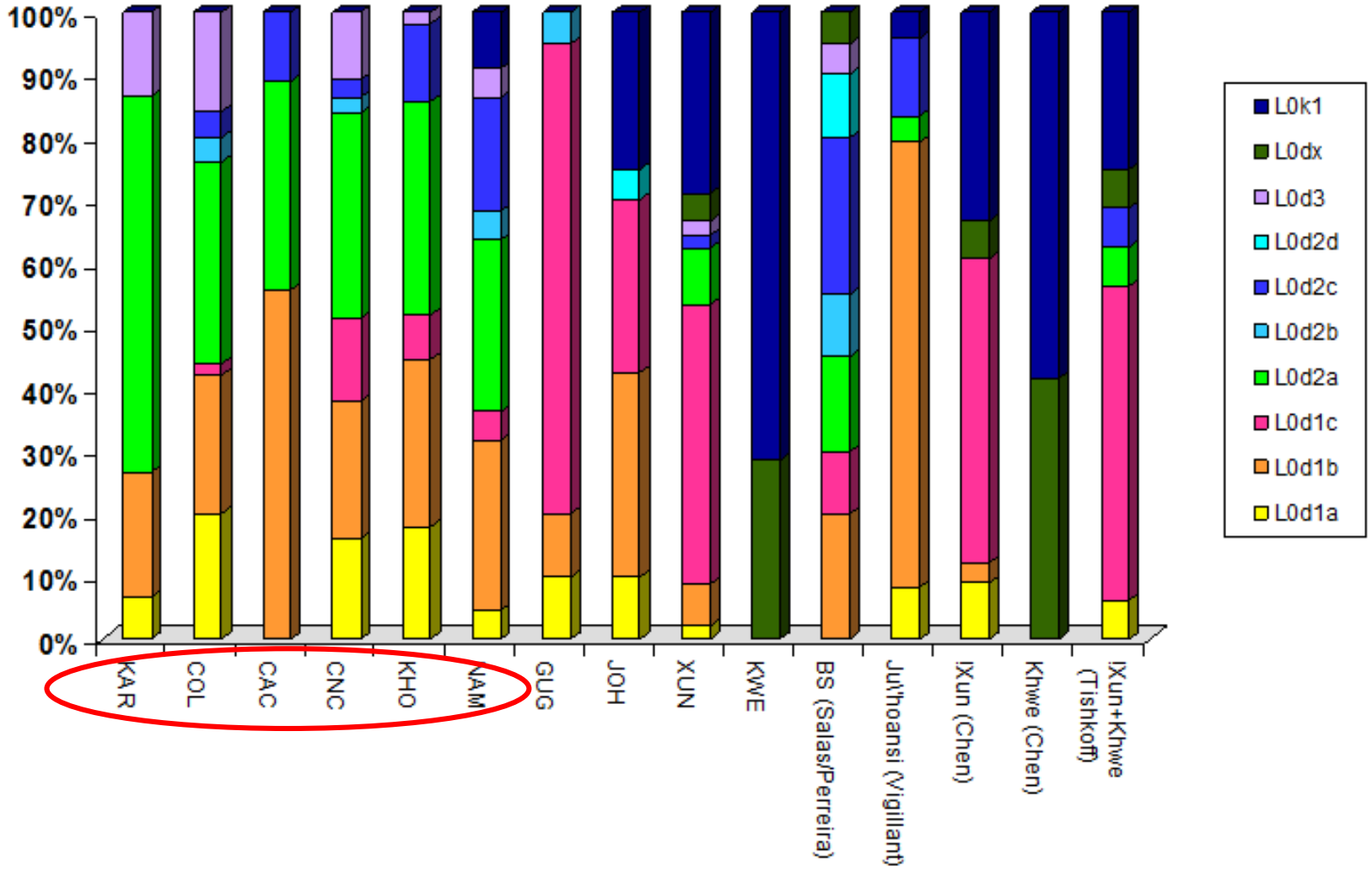
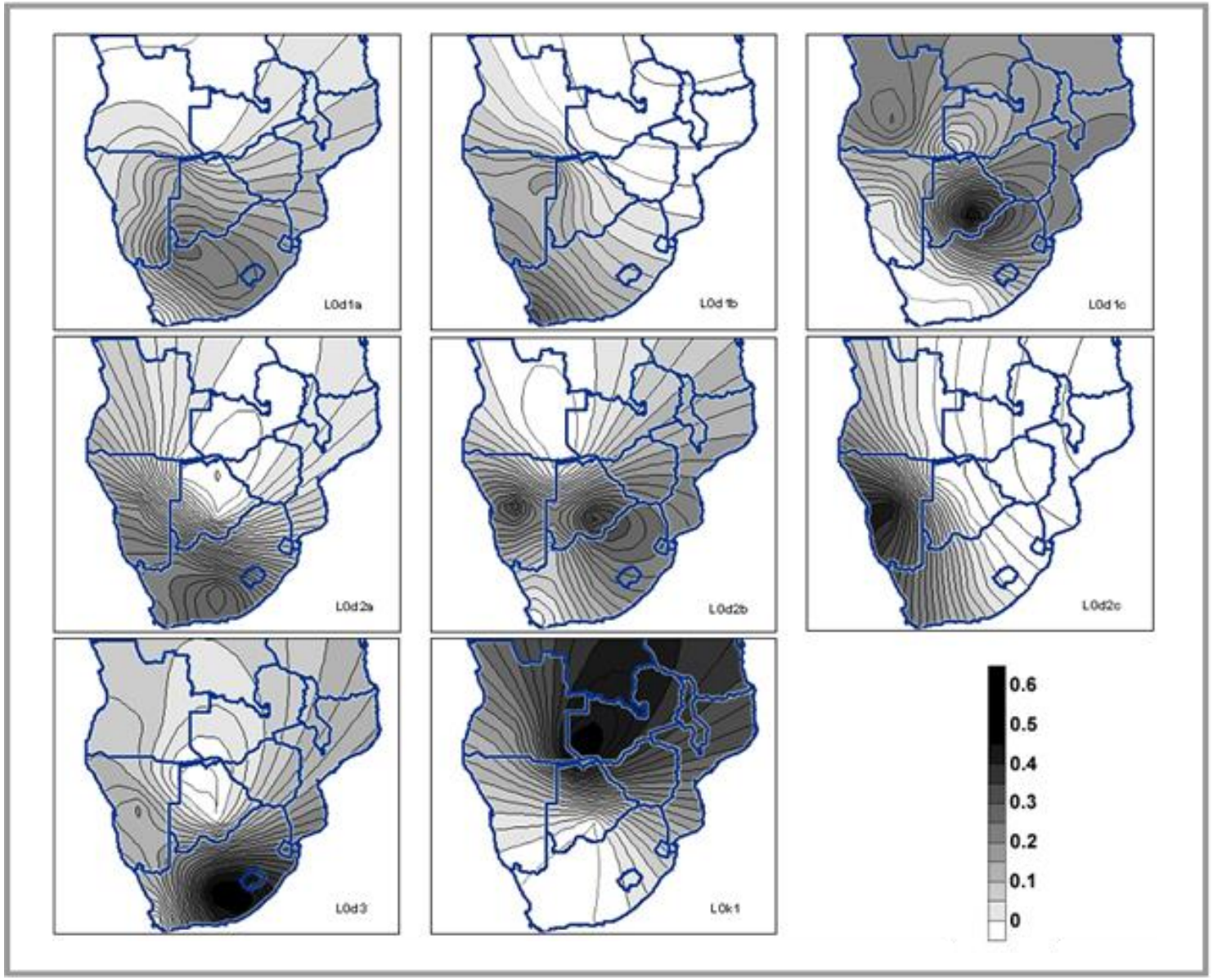
L0d2a
L0a
CRS
NEAN
COL/KAR
CAC
KHO/CNC
NAM
GUG
JOH
XUN
KWE
NAR
XEG/DUM
SEB
HER
OTH

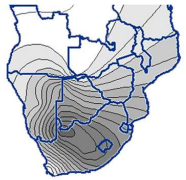
L0d/k sub-haplogroups

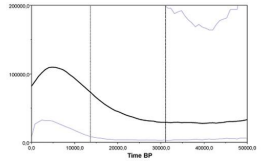


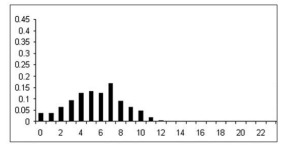
L0d/k sub-haplogroups frequency distributions



L0d1a







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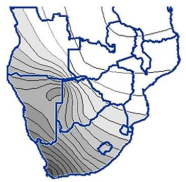

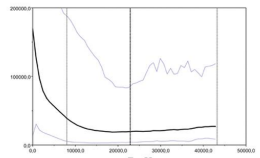

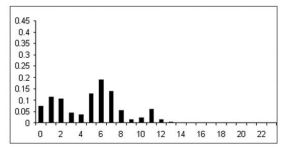

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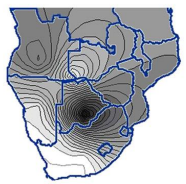

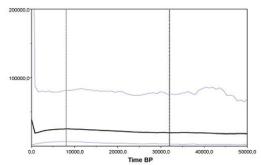

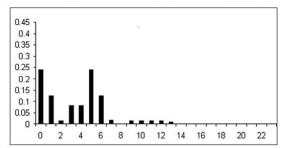

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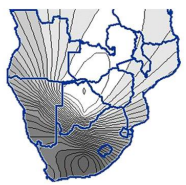

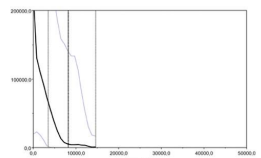

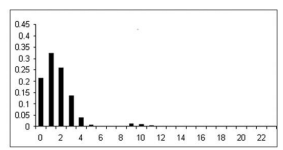

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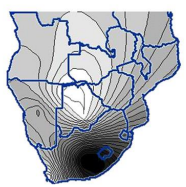

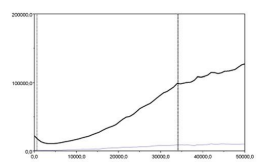

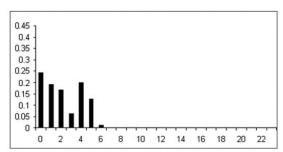

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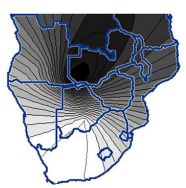

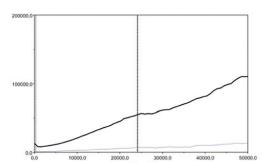

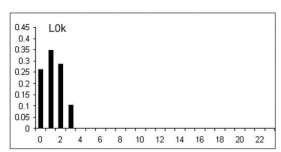

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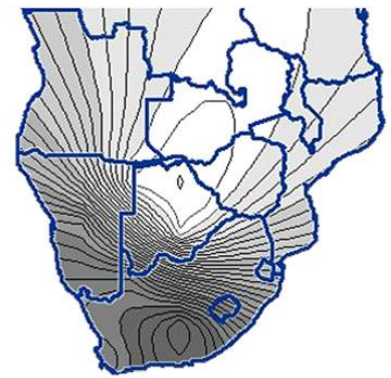
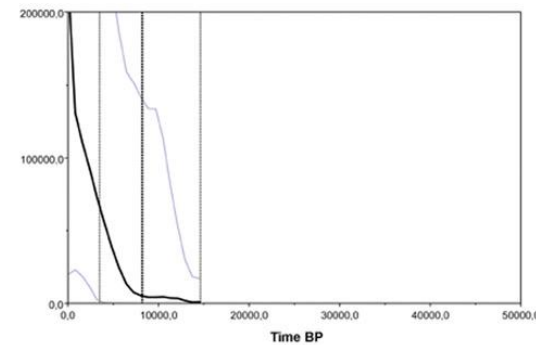




Y-chromosome inferences

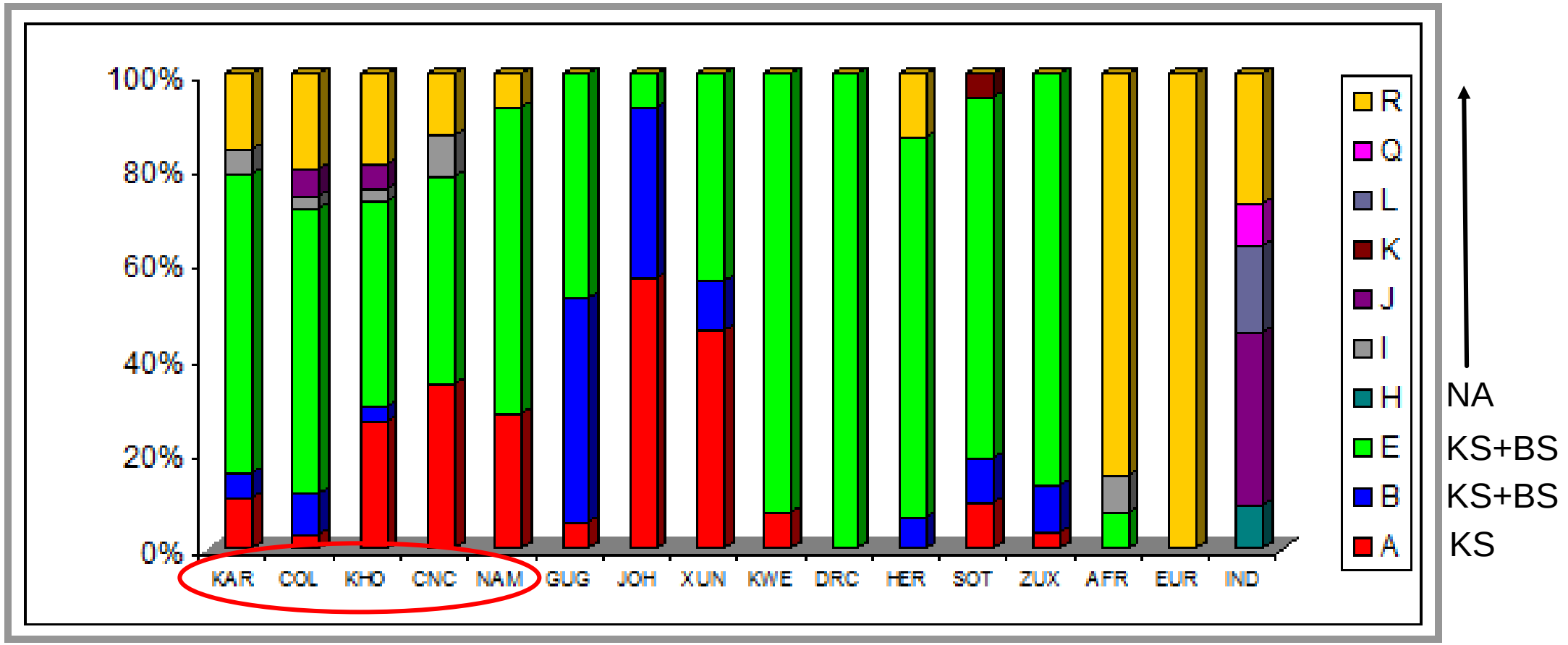
-Schlebusch, C. M. (2010). PhD Thesis: Genetic variation in Khoisan-speaking populations from southern Africa. Division of Human Genetics. Johannesburg, University of the Witwatersrand. **PhD thesis**.

-Schlebusch, C. M., M. de Jongh, et al. (2011). "Different contributions of ancient mitochondrial and Y-chromosomal lineages in 'Karretjie people' of the Great Karoo in South Africa." J Hum Genet **56**(9): 623-30.

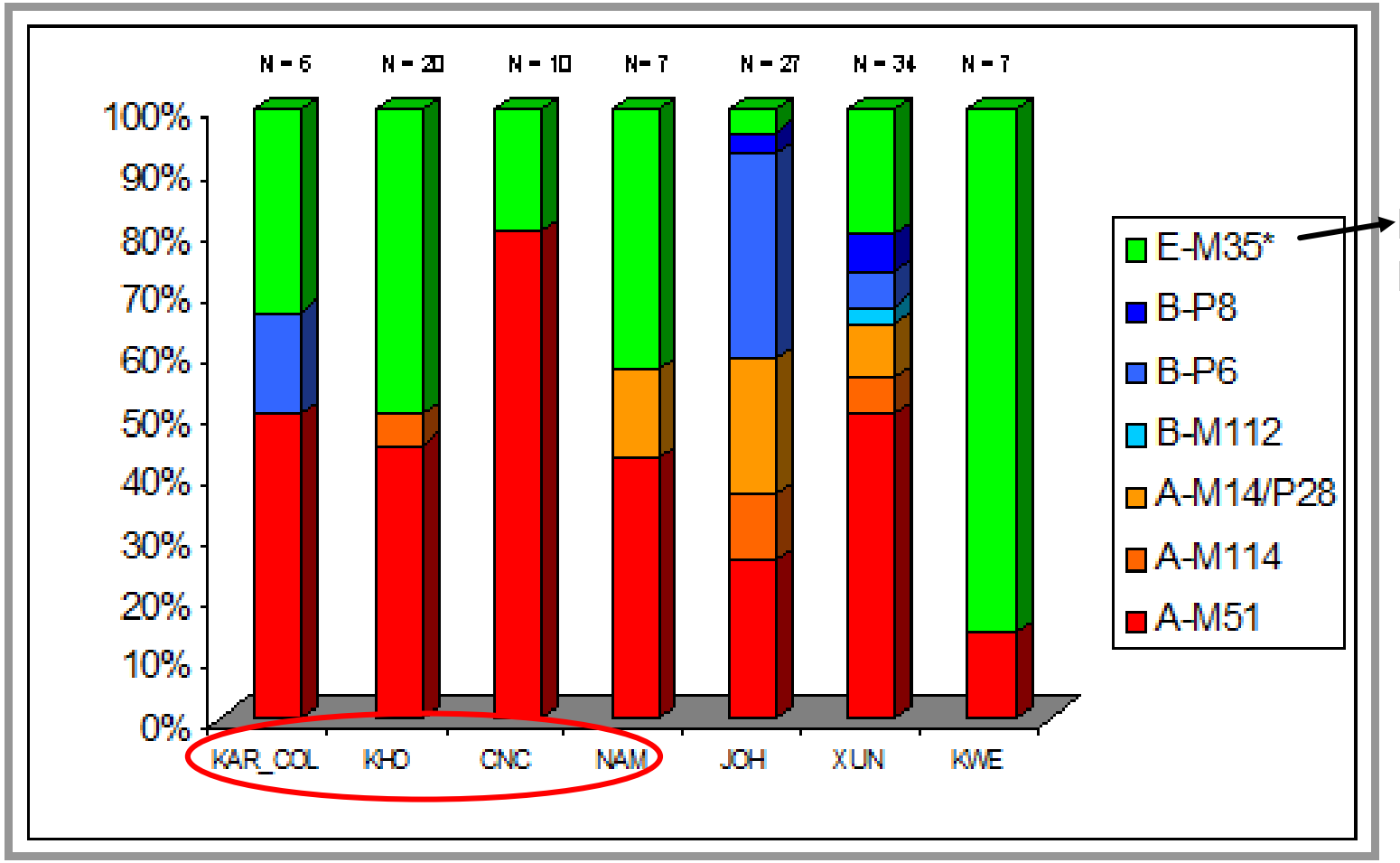
-Naidoo, T., C. M. Schlebusch, et al. (2010). "Development of a single base extension method to resolve Y chromosome haplogroups in sub-Saharan African populations." Investig Genet **1**(1): 6.

-Naidoo, T., (2014). Masters dissertation: Phylogeography of Y-chromosome haplogroups A & B in Africa. Division of Human Genetics. Johannesburg, University of the Witwatersrand. **Masters dissertation**.

Y-chromosome haplogroups



Y-chromosome haplogroups, only KS associated



East African?
Henn et al 2009

Y-chromosome haplogroup frequency distributions