(7) Kalahari Basin prehistory before the advent of food production – Genetics – Selection and Khoisan “split”

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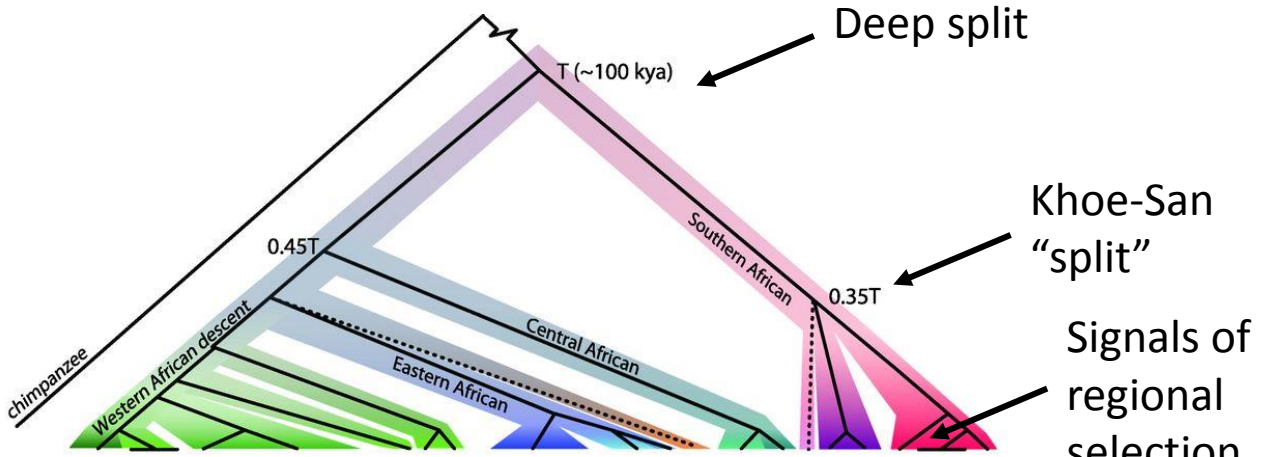
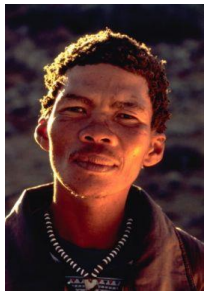
UNIVERSITY OF THE WITWATERSRAND JOHANNESBURG

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





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*]

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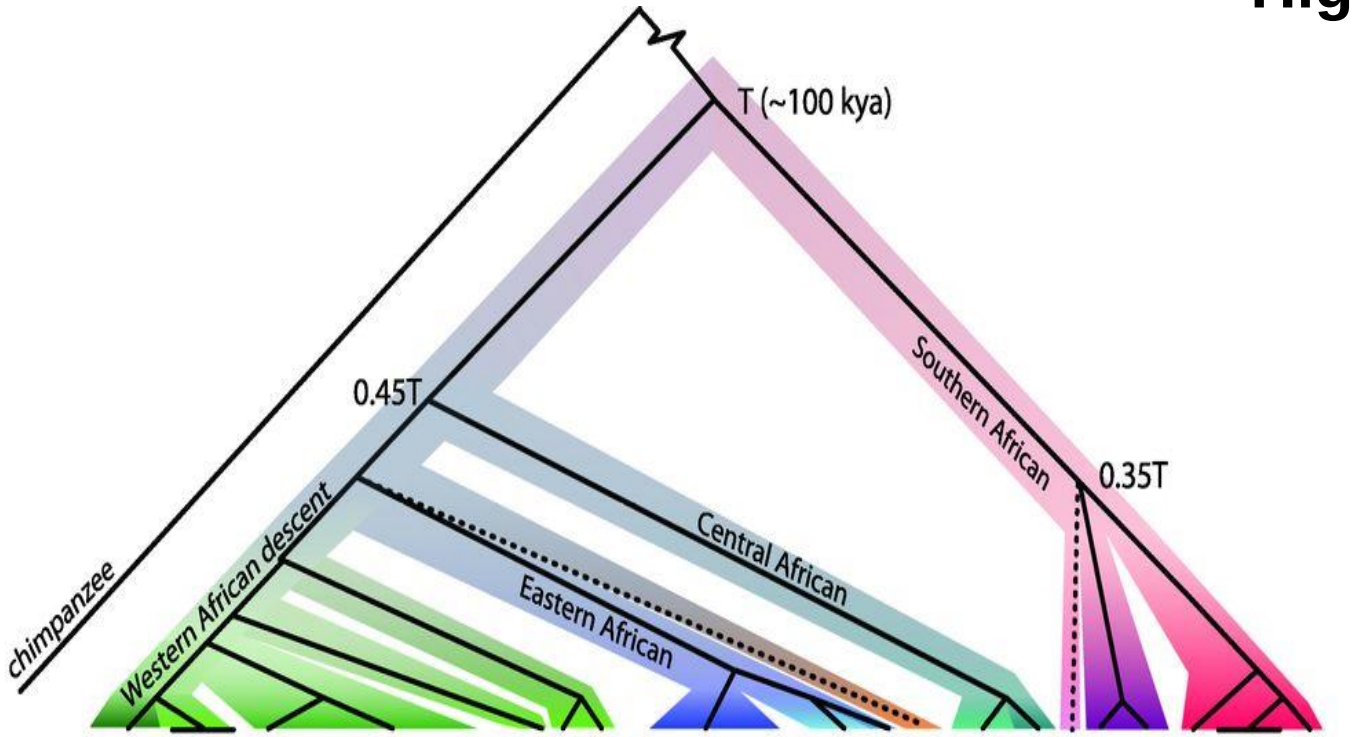
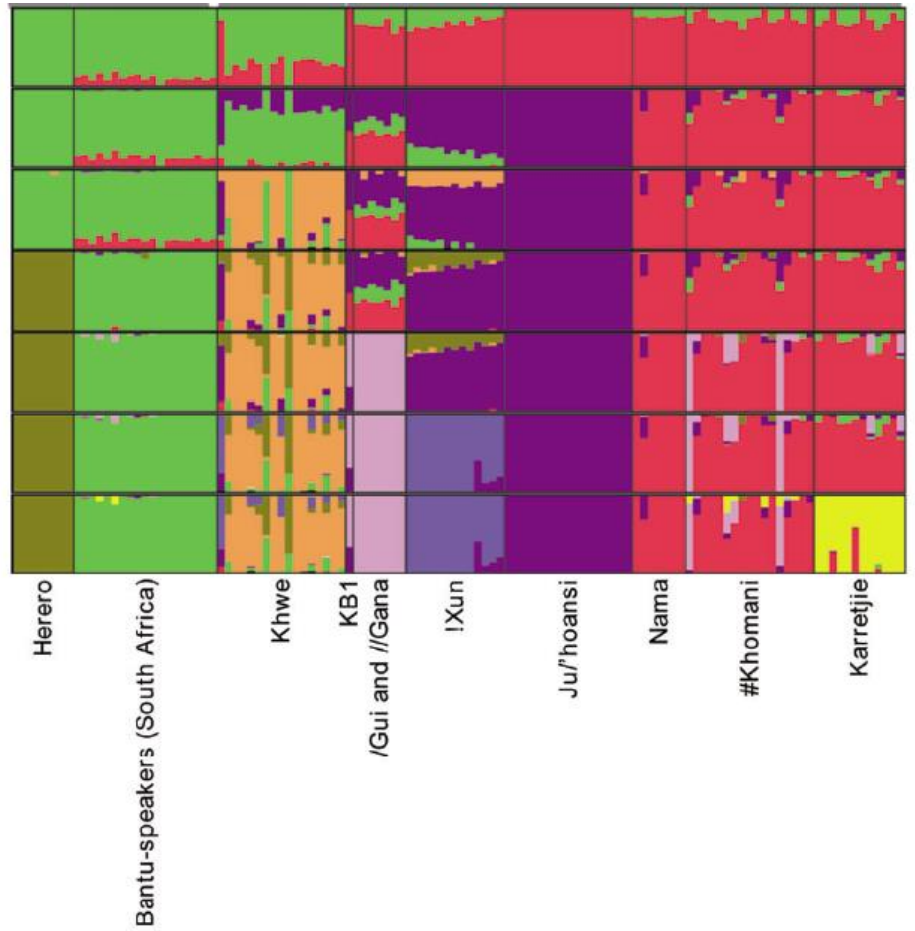
High density dataset – Southern Africa

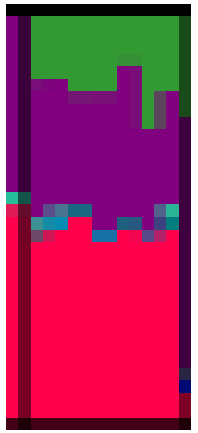


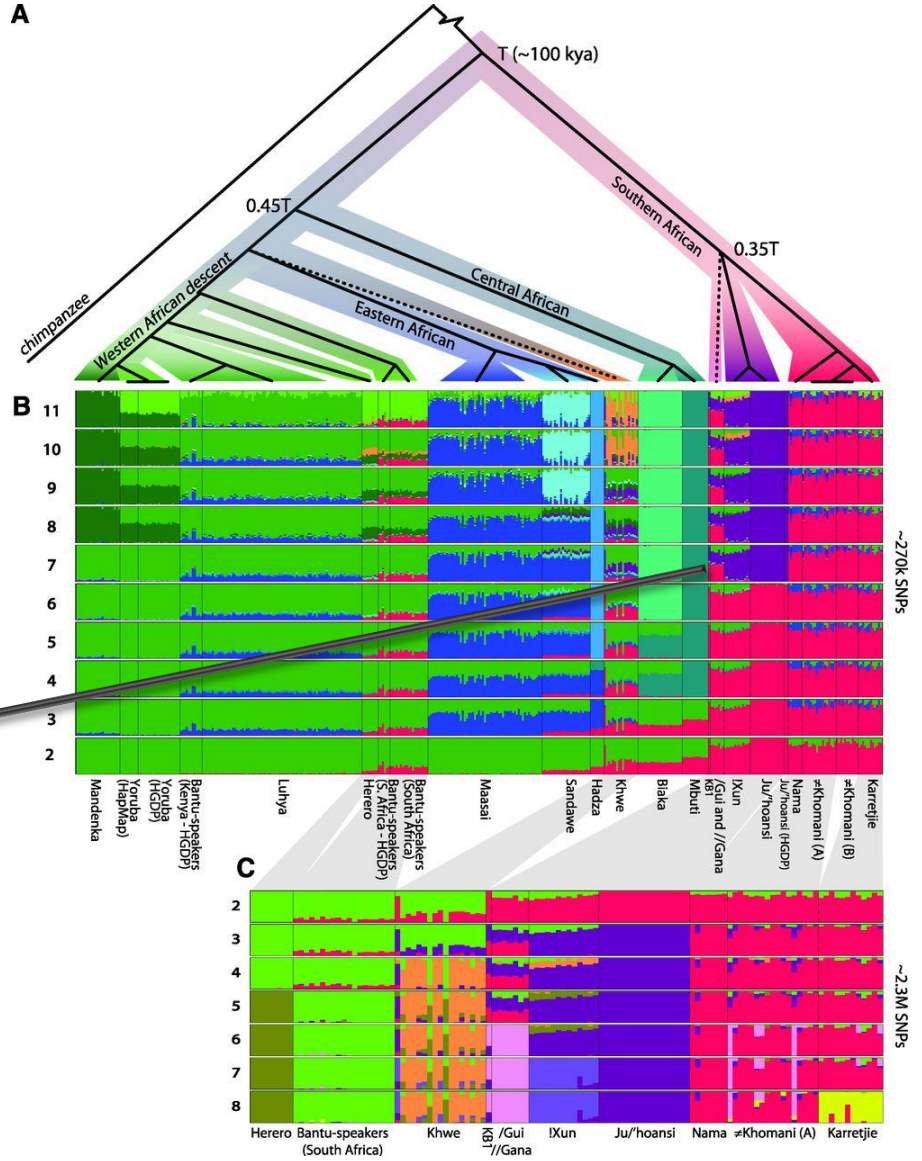


2.3 M SNPs

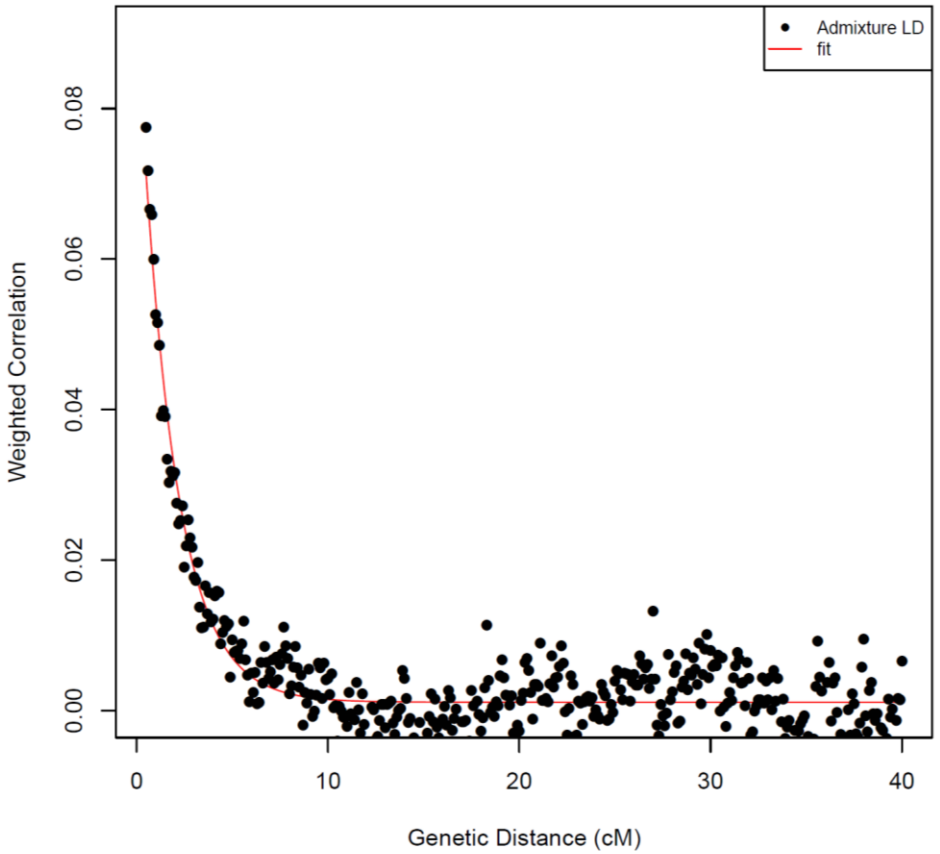
K2 - 8

- Central San Mid-Way or “split” between northern (purple) and southern (red) group
- Questions – Split times – Admixture – Isolation by distance

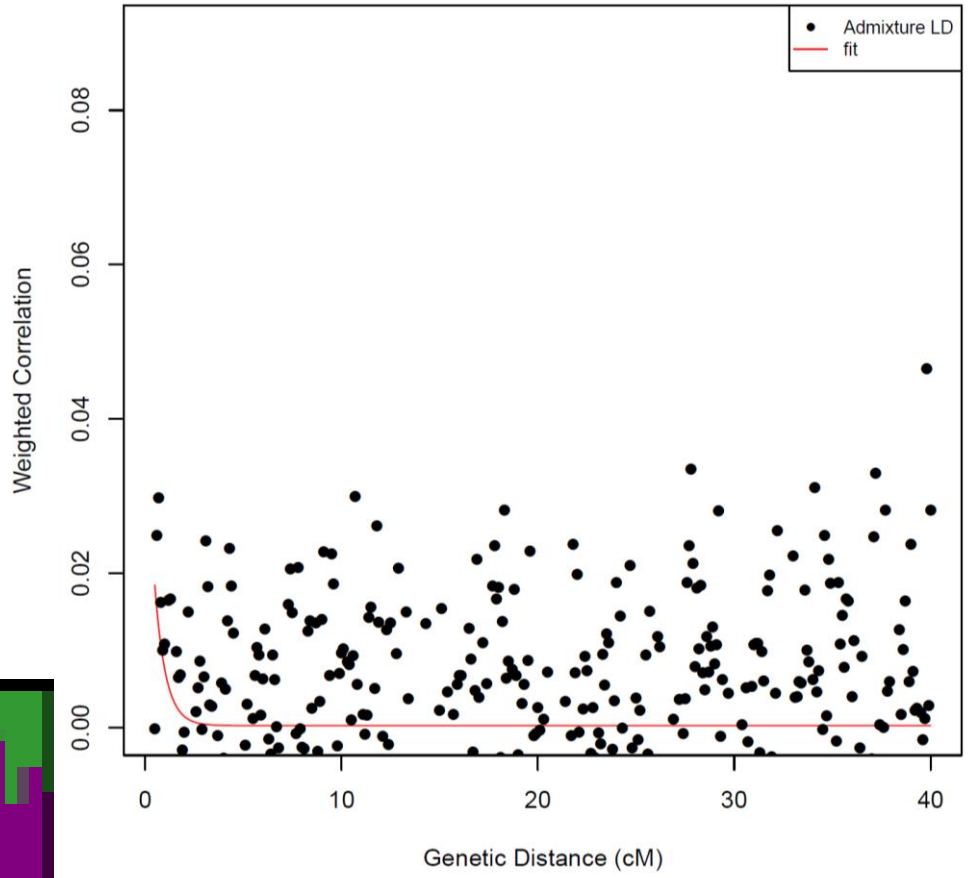




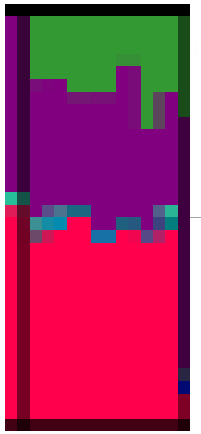
Bantu-sp with central San admixture



Northern with Southenrn San admixture

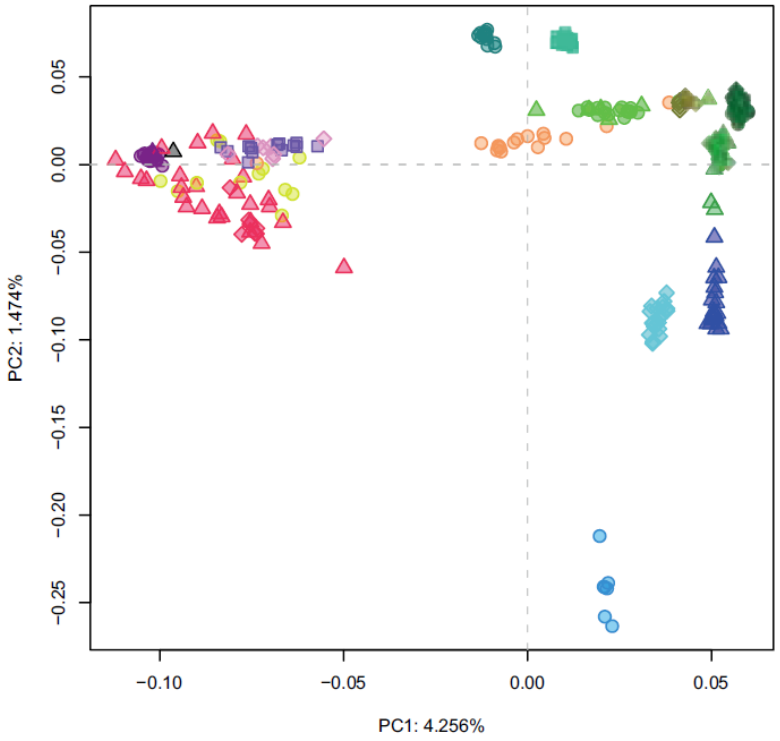


14 generations

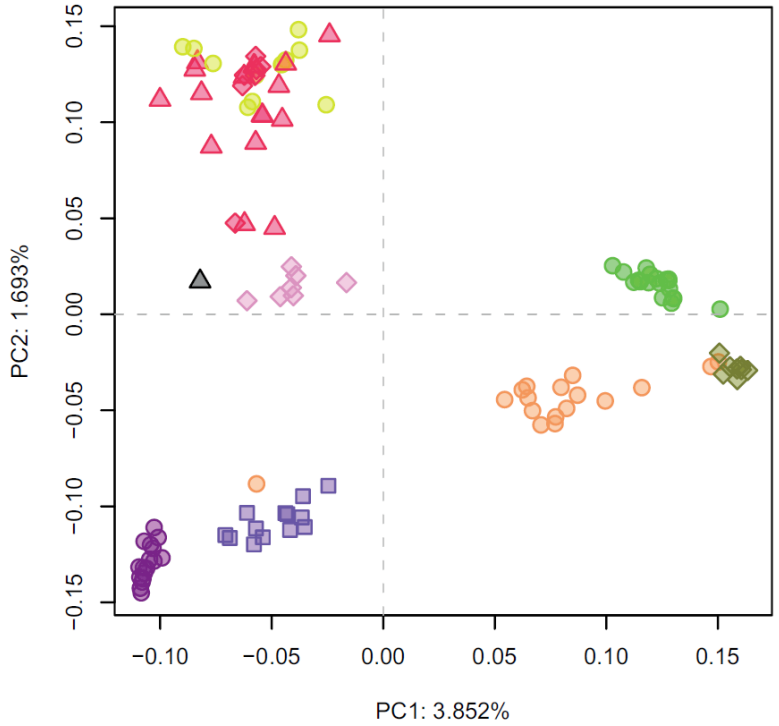


Principle component analysis

African dataset

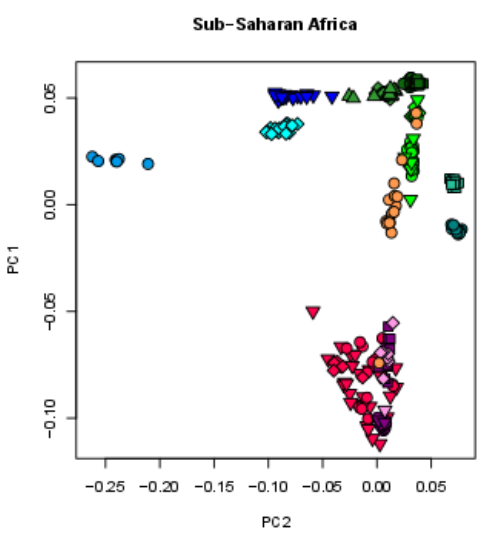


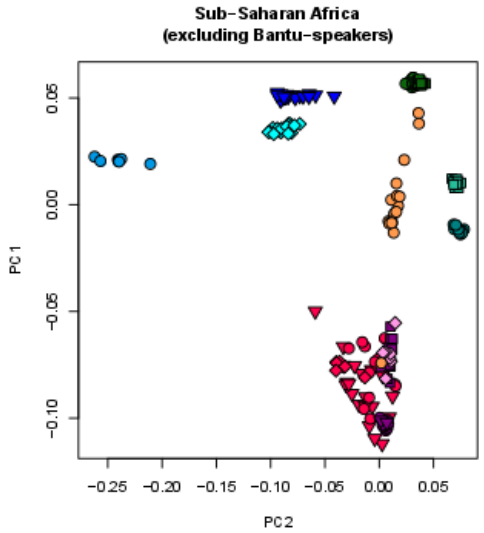
Southern African dataset

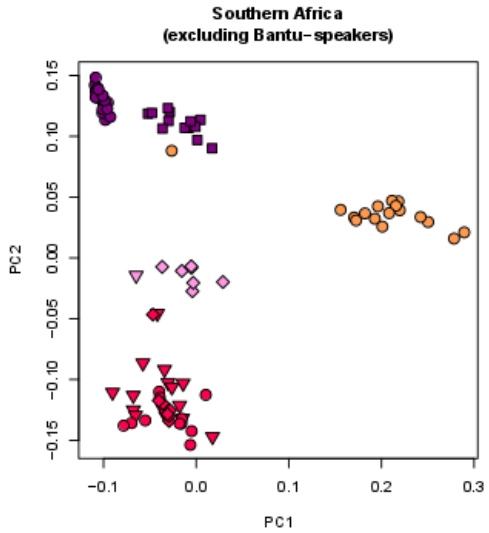


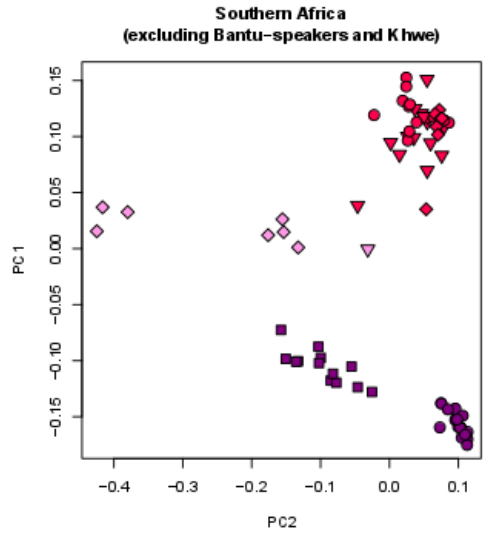
- Karretjie
- Nama
- Khomani (A)
- Khomani (B)
- /Gui and //Gana
- Ju/hoansi
- Ju/hoansi (HGDP)
- !Xun
- Khwe
- KB1
- Mbuti
- Biaka
- Hadza
- Sandawe
- Maasai
- Luhya
- Bantu−speakers (Kenya − HGDP)
- Bantu−speakers (Southern Africa − HGDP)
- Bantu−speakers (South Africa)
- Herero
- Yoruba (HapMap)
- Yoruba (HGDP)
- Mandenka

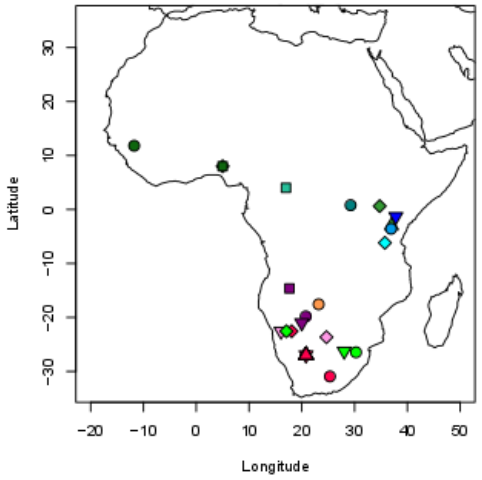
Principle component analysis - Procrustus

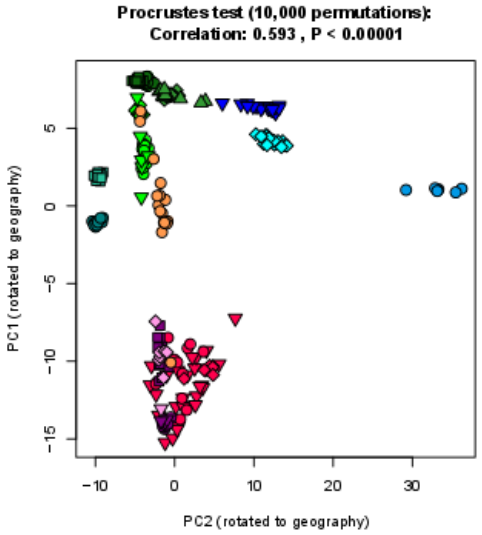


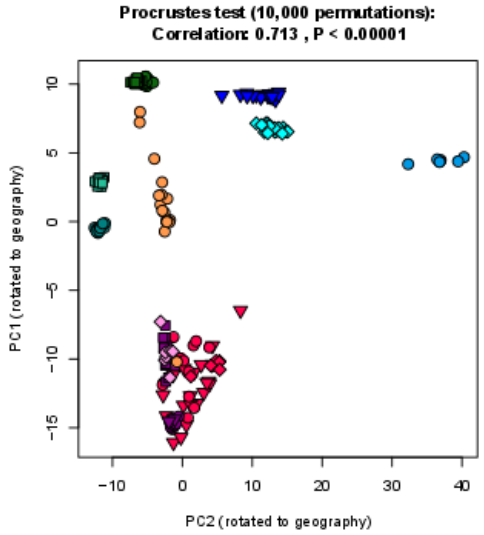


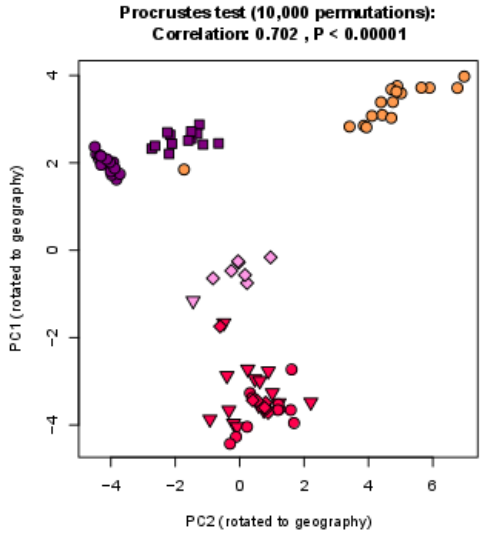


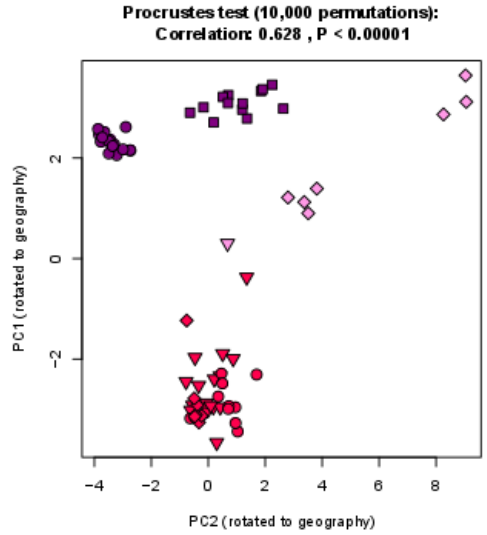






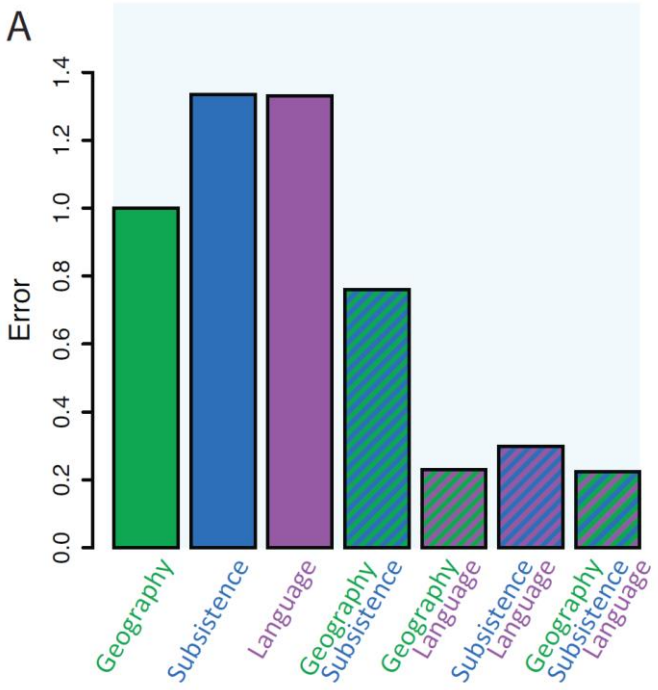




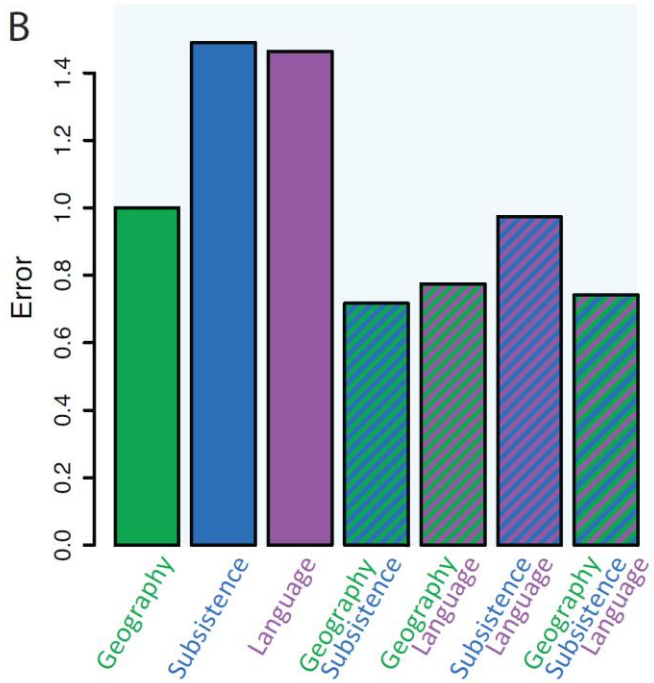


Predictive error relative to geography

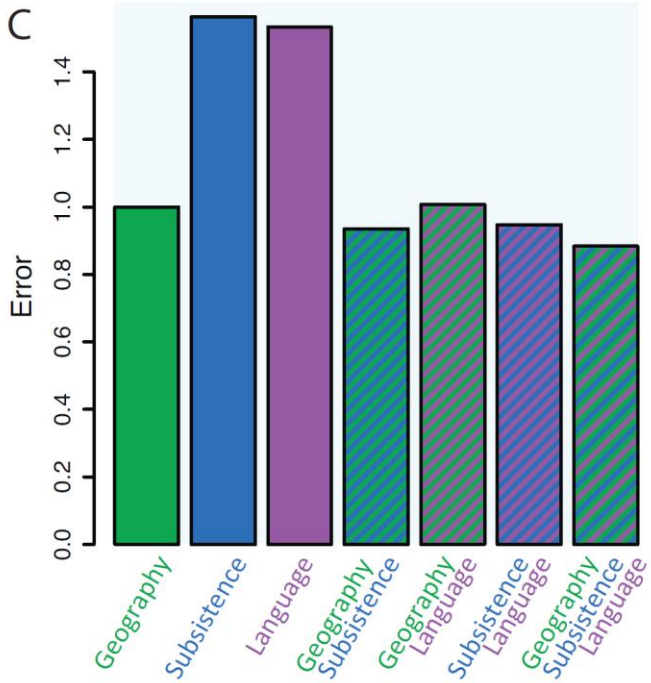
African dataset



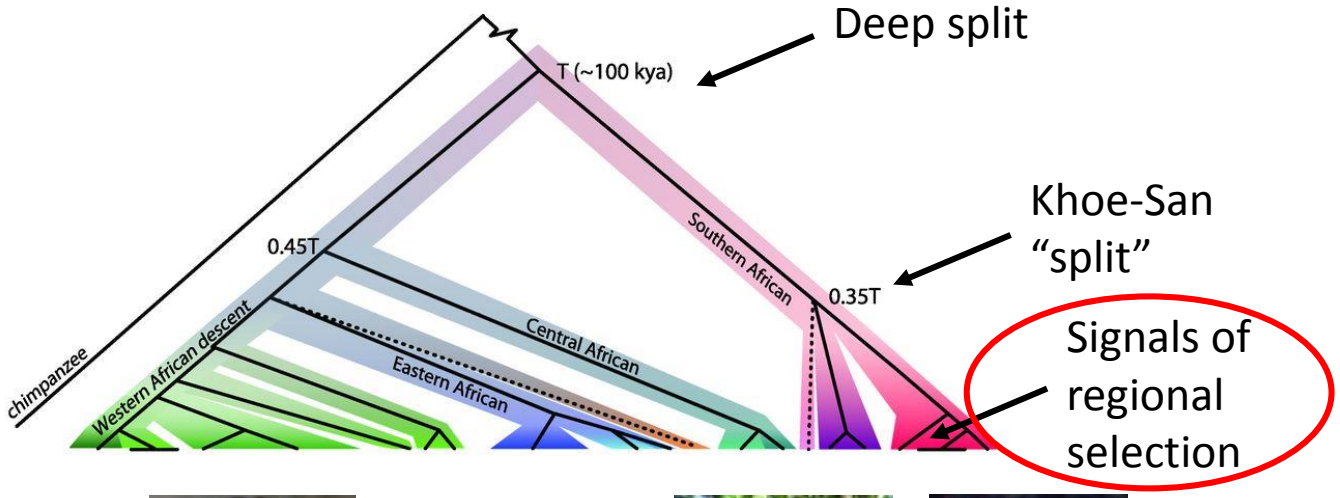
Southern African dataset
Incl. Bantu-speakers



Southern African dataset
Excl. Bantu-speakers



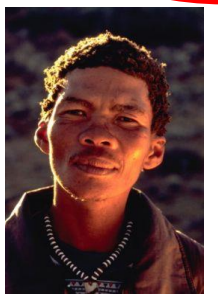
(values <1 show improved predictive capacity as compared to that of geography)



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]*

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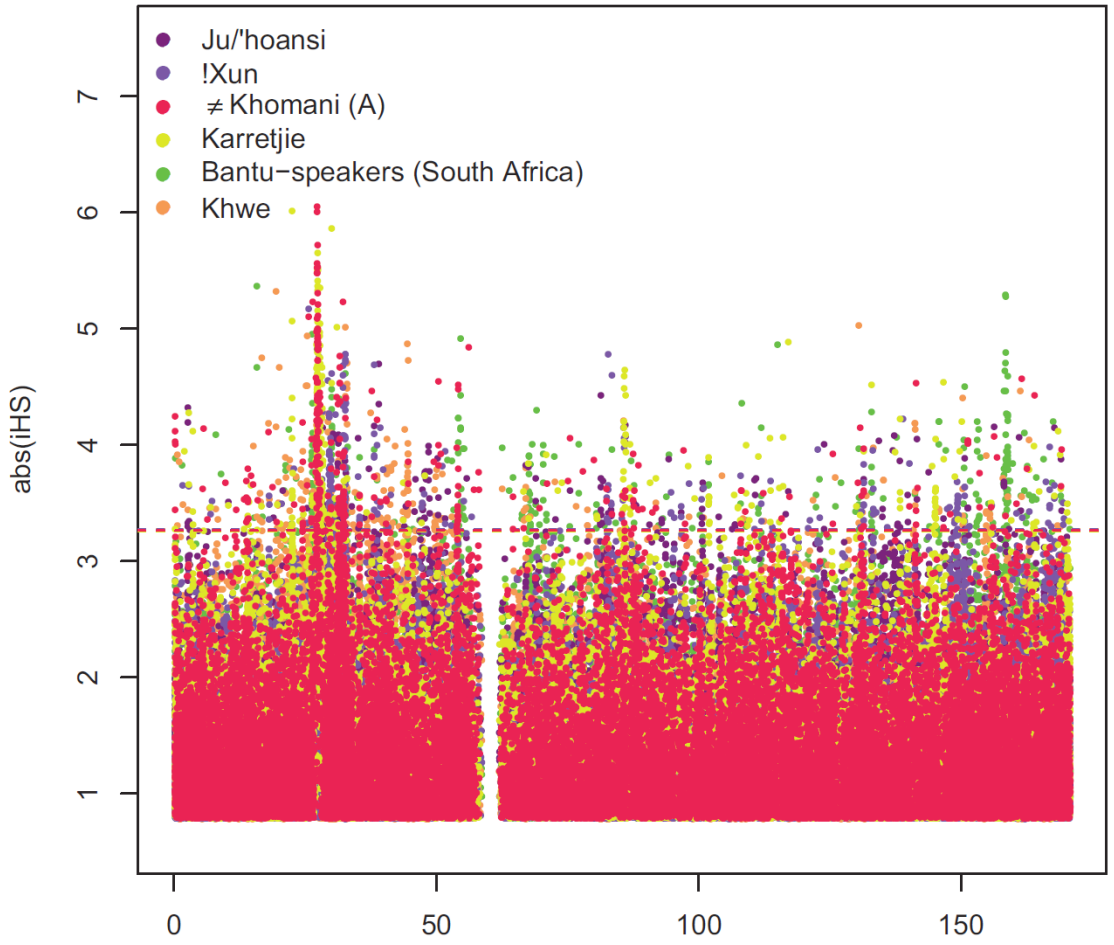
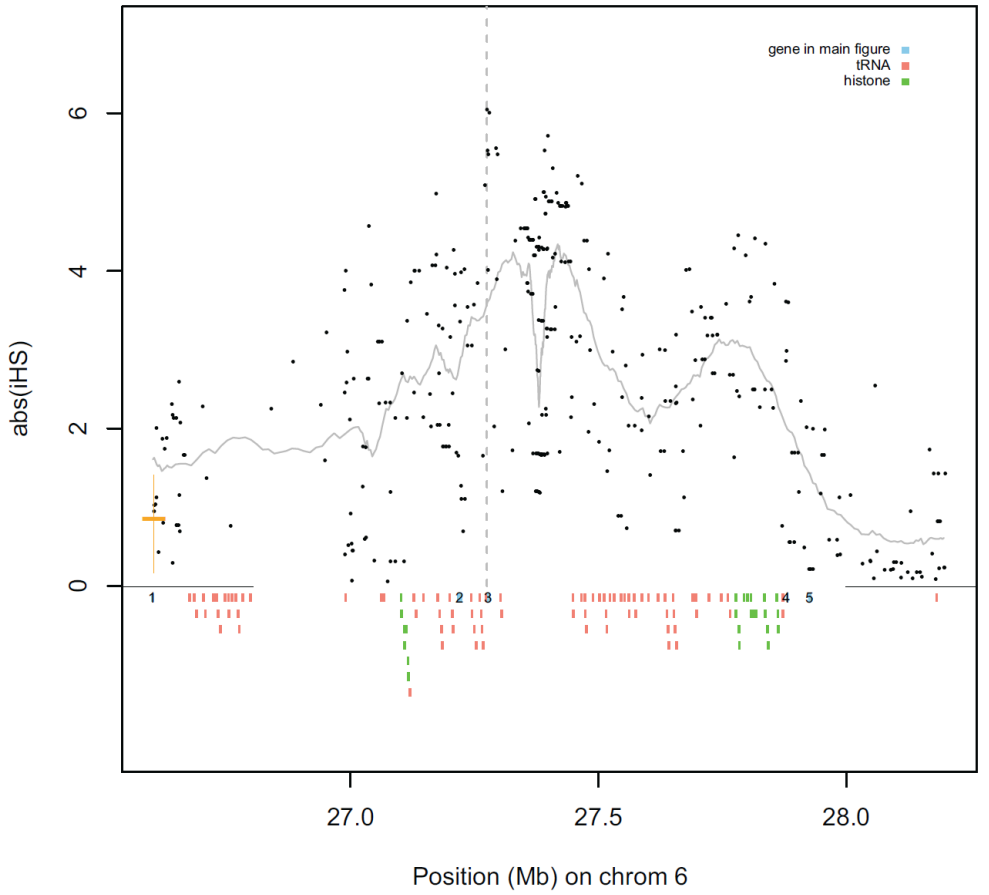
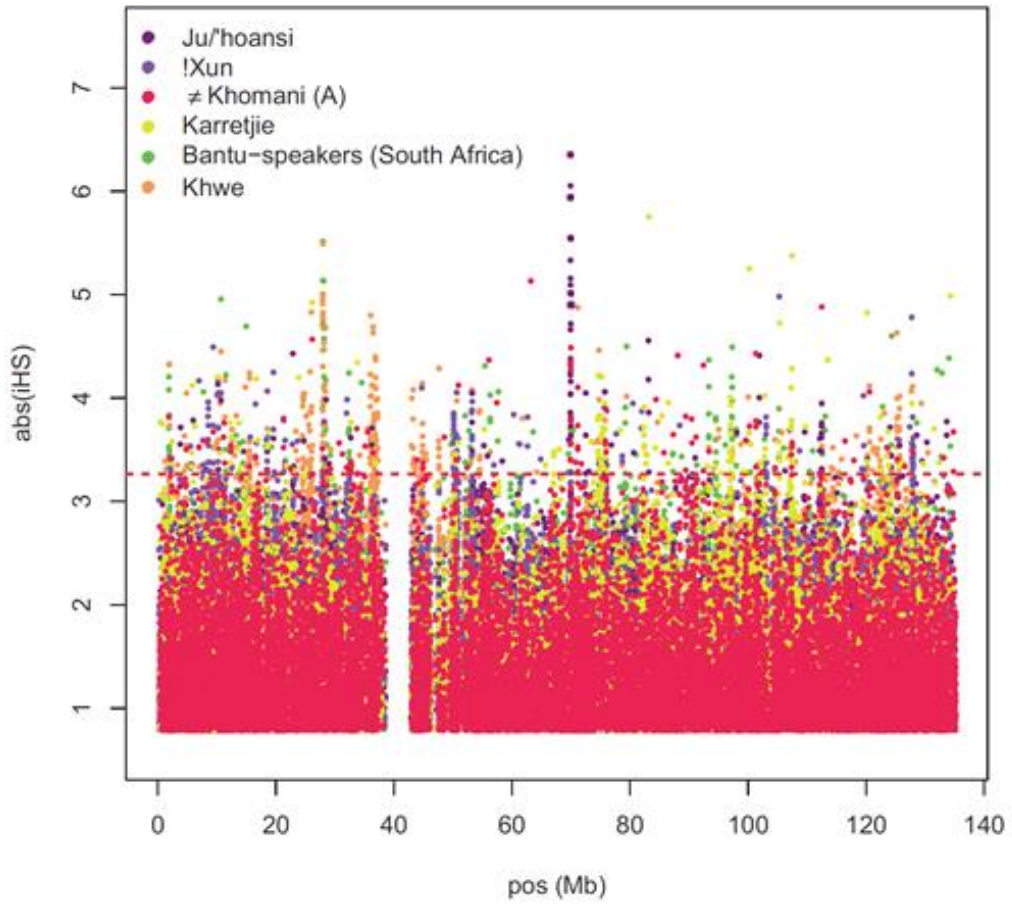
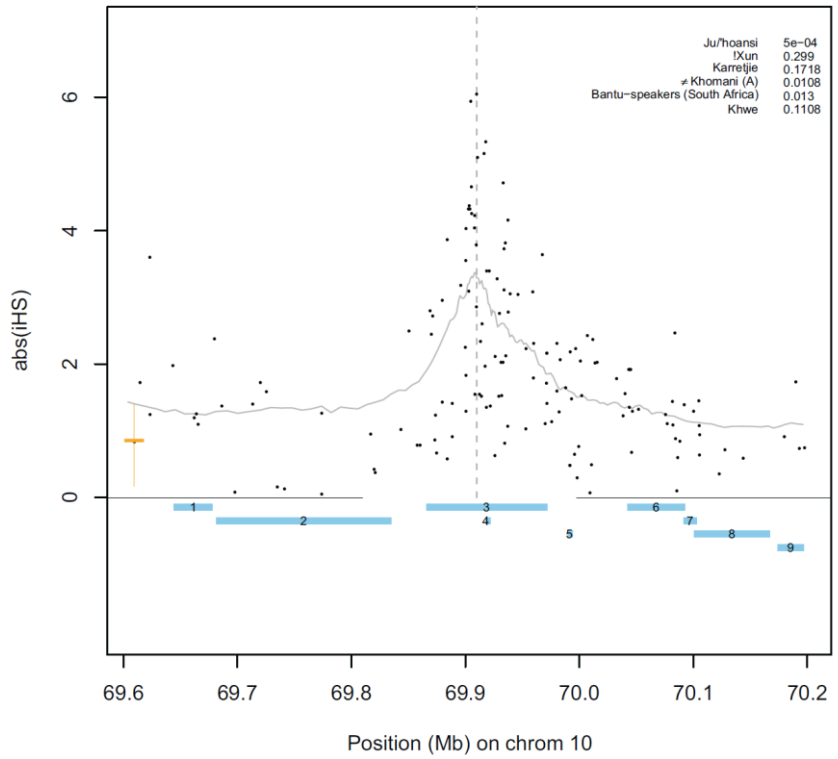
Chromosome 6 – Immunity – Southern San groups





Chromosome 10 – MYPN – Muscle growth and contaction – Ju’/hoansi





ACTN3

rs1815739 T/T kids dropped from sports programs everywhere – no chance for Olympic glory

November 30, 2008 by dendrite

I was irked to see, in today's New York Times, a picture of a young child having his cheek swabbed so that his parents could ascertain his status at the rs1815739 C/T variant . T-alleles at this site give rise to a premature stop codon

Image by sean dreilinger via Flickr

ACTN3

- two types of muscle fibers,
- slow twitch and fast twitch
- Slow twitch fibers more efficient in using oxygen to generate energy, fast twitch fibers are less efficient
- Fast twitch fibers fire more rapidly and generate more force.

ACTN3

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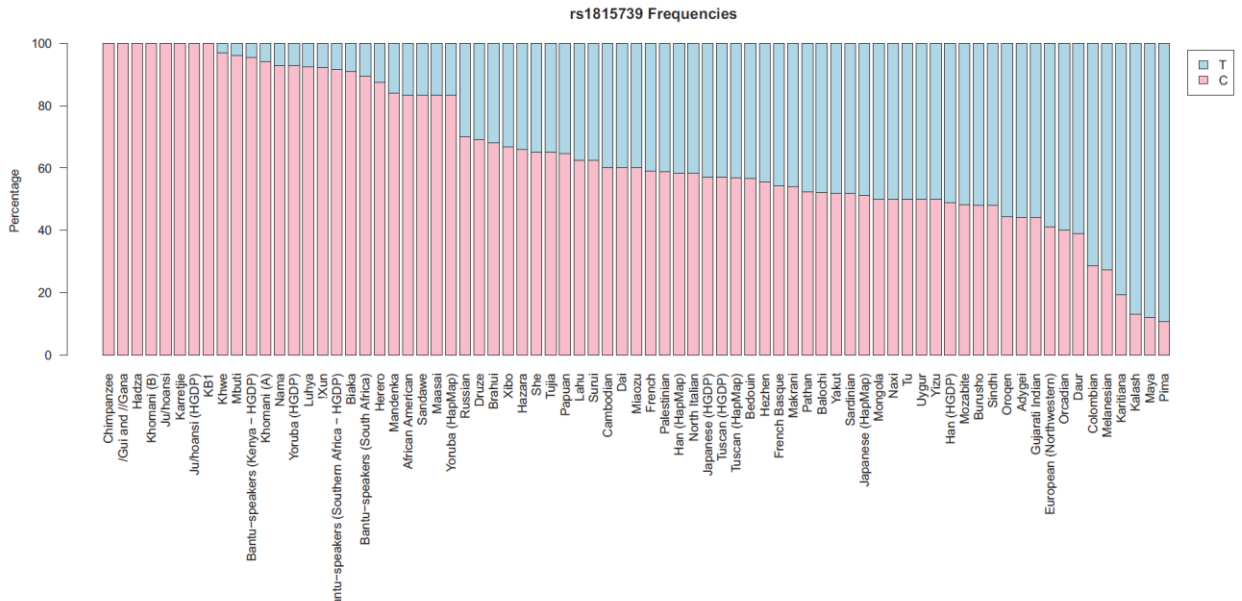
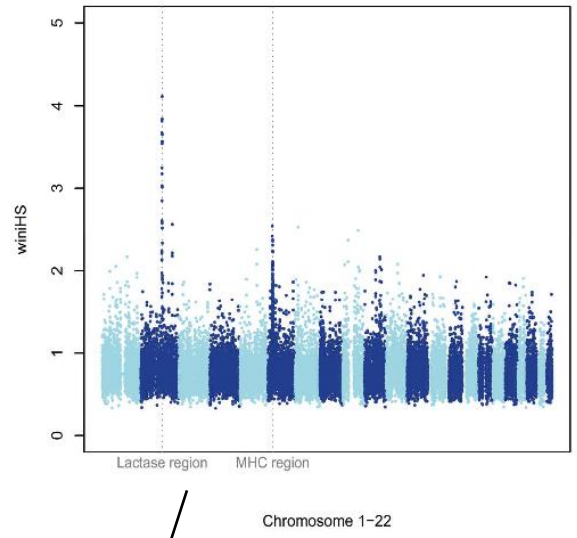


Acknowledgements

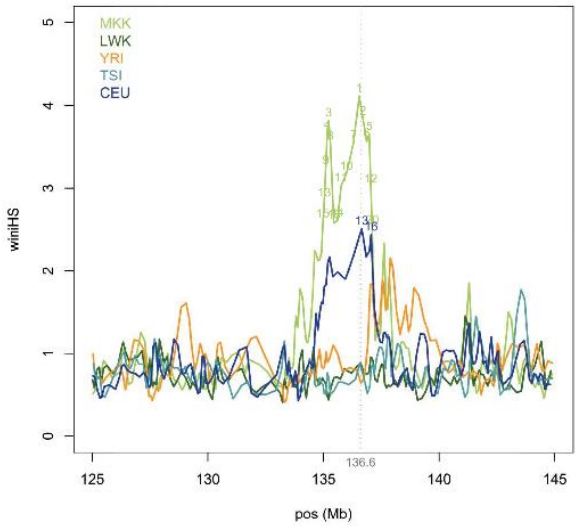
- San + Khoe + Coloured individuals
- South African San Council (SASC)
- Working Group of Indigenous Minorities in Southern Africa (WIMSA)
- Prof. Himla Soodyall and the HGDDRU lab – NHLS and University of the Witwatersrand, Johannesburg, South Africa
- Jakobsson group, Dept Evolutionary Biology, Uppsala University, Sweden
- Funding: Swedish Research Council, Wenner Gren Foundation

Lactase persistence - Maasai

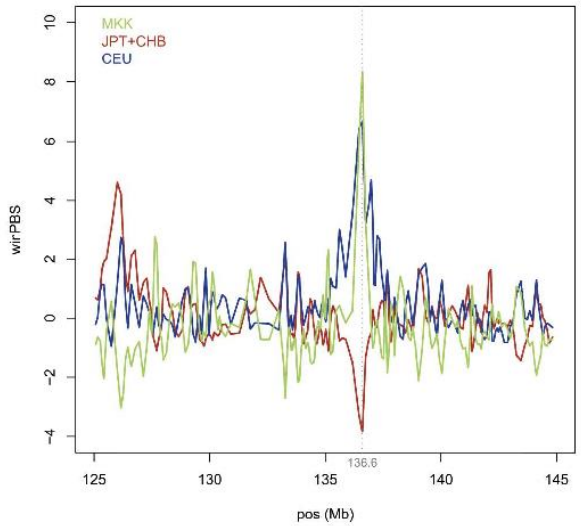
Genome-wide iHS



Local zoom iHS



Local zoom PBS



Lactase

Lactase

Lactase

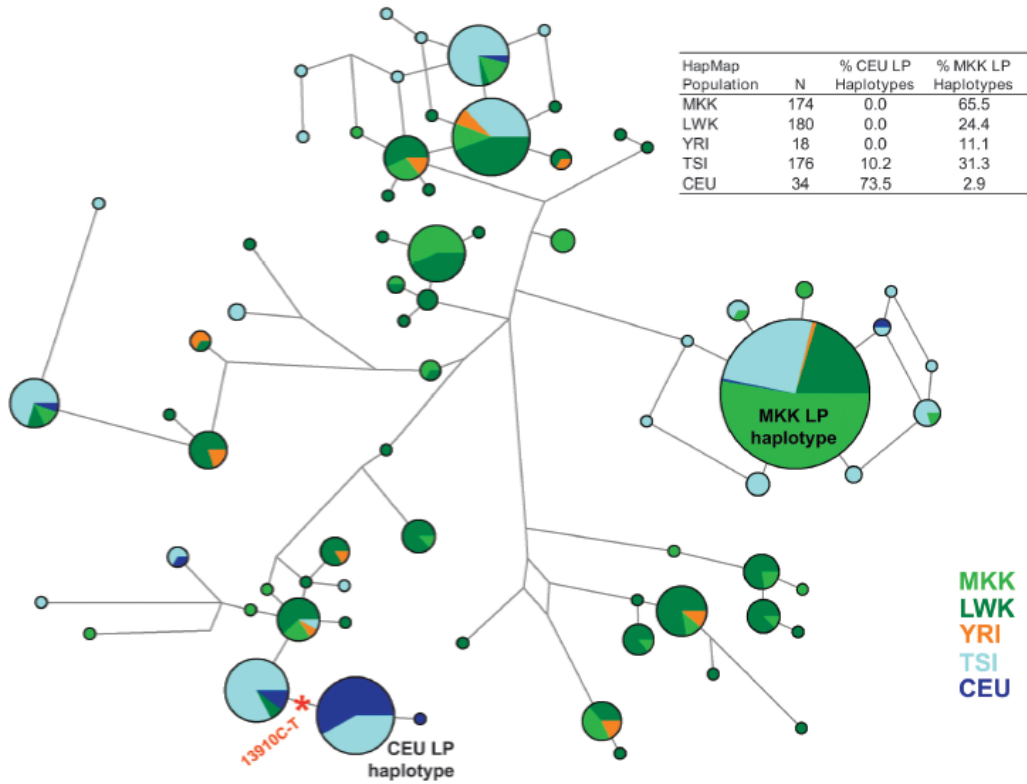
European Journal of Human Genetics (2012), 1–4

www.nature.com/ejhg

ARTICLE

Stronger signal of recent selection for lactase persistence in Maasai than in Europeans

Carina M Schlebusch*[,1,3], Per Sjödin[1,3], Pontus Skoglund[1,3] and Mattias Jakobsson[1,2]



HapMap Population	N	% CEU LP Haplotypes	% MKK LP Haplotypes	% Other Haplotypes
MKK	174	0.0	65.5	34.5
LWK	180	0.0	24.4	75.6
YRI	18	0.0	11.1	88.9
TSI	176	10.2	31.3	58.5
CEU	34	73.5	2.9	23.5

Current Biology

Please cite this article in press as: Breton et al., Lactase Persistence Alleles Reveal Partial East African Ancestry of Southern African Khoe Pastoralists, Current Biology (2014), http://dx.doi.org/10.1016/j.cub.2014.02.041

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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

Please 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

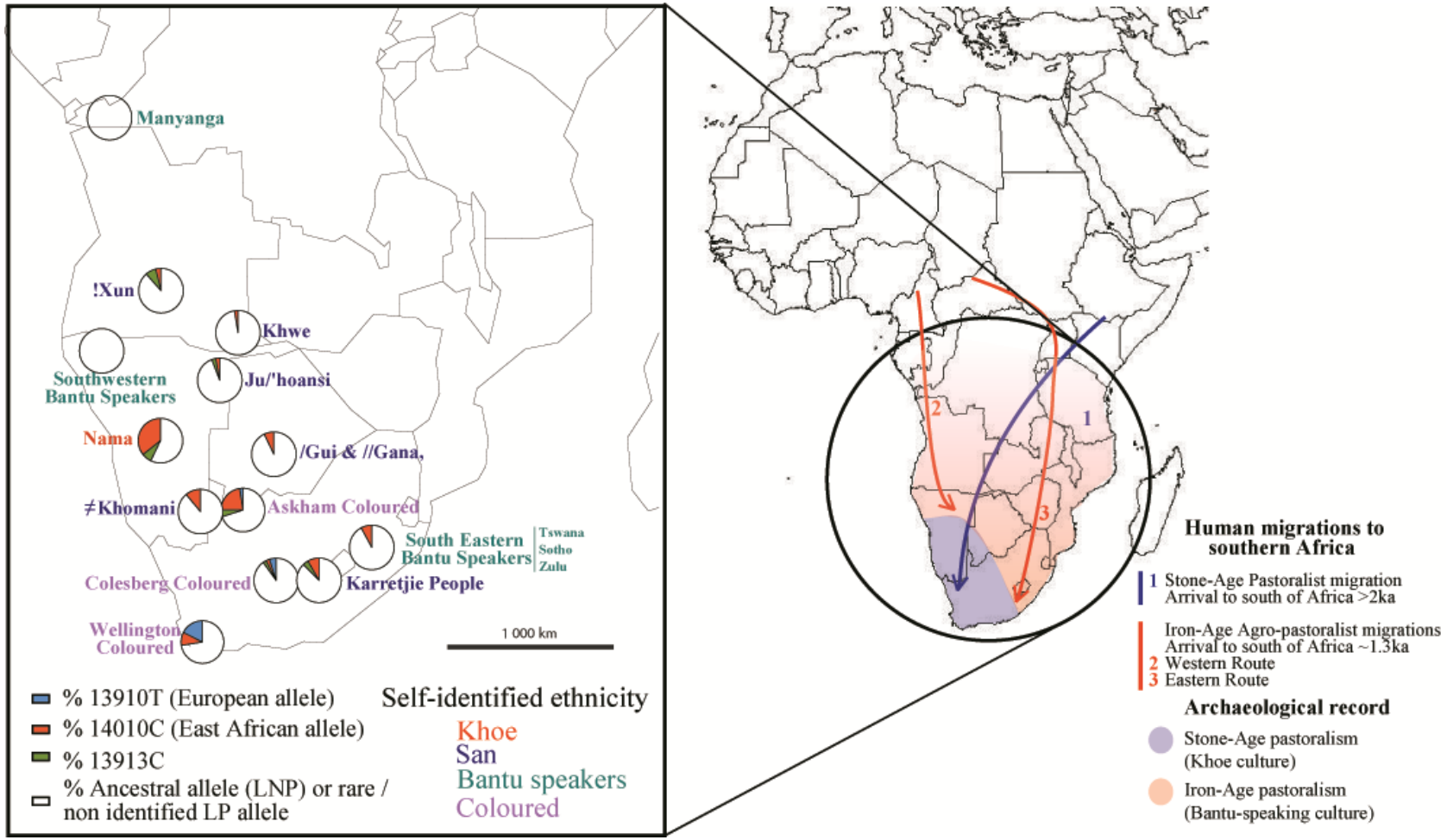
Report

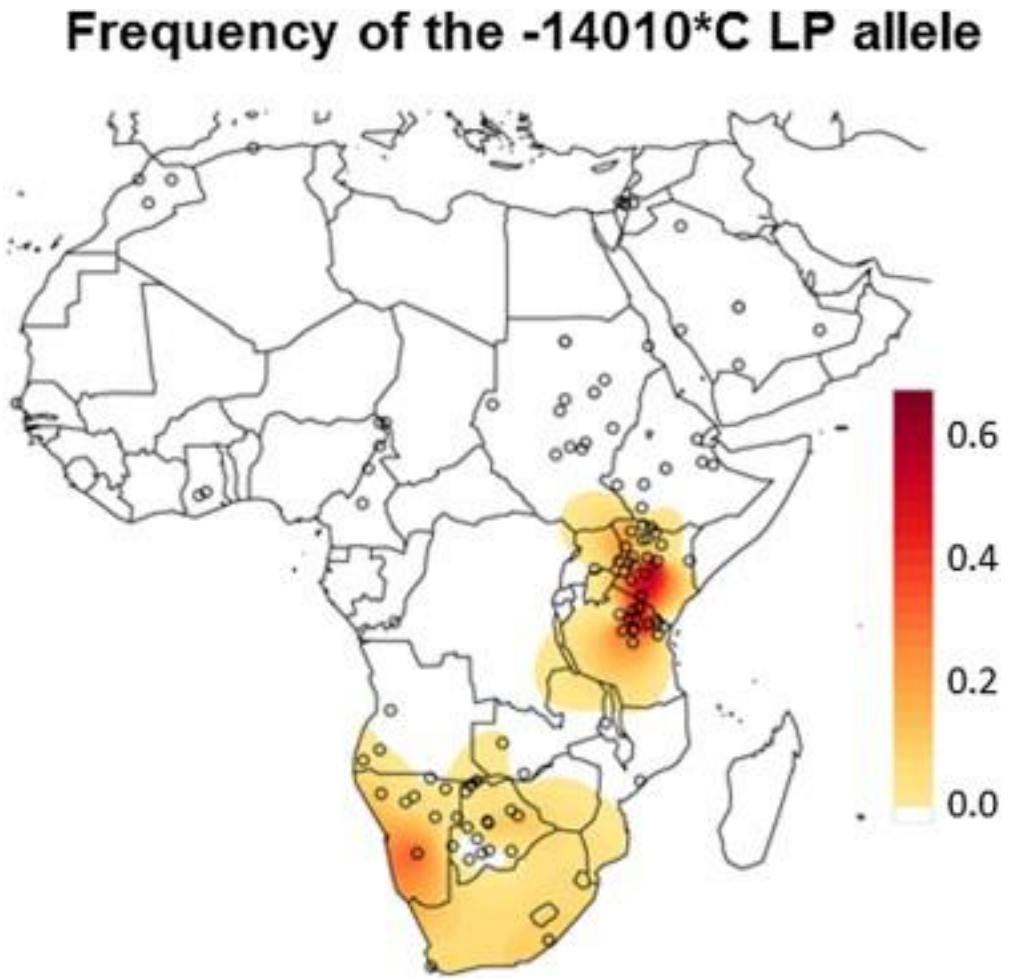
Tracing Pastoralist Migrations to Southern Africa with Lactase Persistence Alleles

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, Germany

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

East African LP-mutation (red) in high frequency in the Nama (Khoe) – lower in San groups





Macholdt et al 2014,
Current Biology 24:875–879