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

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Speaking (of) Khoisan, Leipzig 14-16 May 2015
Genomic Variation in Seven Khoe-San Groups Reveals Adaptation and Complex African History

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19 OCTOBER 2012 VOL 338 SCIENCE www.sciencemag.org
High density dataset – Southern Africa
- 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 Southern 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)
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Chromosome 6 – Immunity – Southern San groups
Chromosome 10 – MYPN – Muscle growth and contraction – Ju’/hoansi
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.
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.
• two types of muscle fibers,
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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
Stronger signal of recent selection for lactase persistence in Maasai than in Europeans

Carina M Schlebusch*1,3, Per Sjödin1,3, Pontus Skoglund1,3 and Mattias Jakobsson1,2
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,2 Per Sjödin,1 Himla Soodyall,4 and Mattias Jakobsson1,5,*

Tracing Pastoralist Migrations to Southern Africa with Lactase Persistence Alleles

Enrico Macholdt,1 Vera Lede,1 Chiara Barbieri,1,5 Sununguku W. Mpoloka,2 Hua Chen,3 Montgomery Slatkin,3 Brigitte Pakendorf,4,6 and Mark Stoneking1,7

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Results and Discussion

We sequenced 360 bp of the lactase persistence (LP)-regulatory region, encompassing all known LP-regulatory variants in the Southern African population (Figure 1, and the Supporting Information) from 35 Khoe-San samples including representatives of current-day Khoe-San (referred to hereafter as San), which historically represented the majority of the indigenous population of southern Africa. The sample includes 16 San individuals (9 % of the 1,968 modern San represent the ancestral genetic background of current-day San) who are direct descendents of San who were present in Africa before the arrival of Europeans. Additional 20 Khoe-San individuals arrived in the southern African region about 2,000 years ago and some 1,300 years ago. It is likely that these were the result of domestications of domesticated livestock, which were introduced to the region about 2 thousand years ago. We identified about 300 of the domesticated livestock, which were introduced to the region about 2 thousand years ago. We identified about 300 of the domesticated livestock.
East African LP-mutation (red) in high frequency in the Nama (Khoe) – lower in San groups