

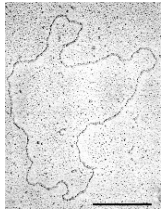
Links with Eastern Africa: Genetic evidence

**Mark Stoneking
on behalf of the KBA geneticists**

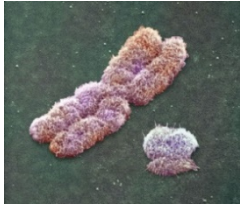
Kalahari Basin Area

Endangered Language & Population History Research

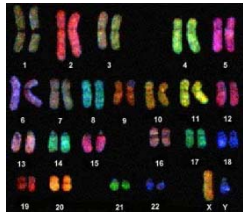
Outline



mtDNA: **maternal** history



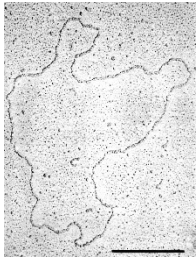
Y chromosome: **paternal** history



Genome-wide data:
the other 99.5% of our ancestry



Lactase persistence



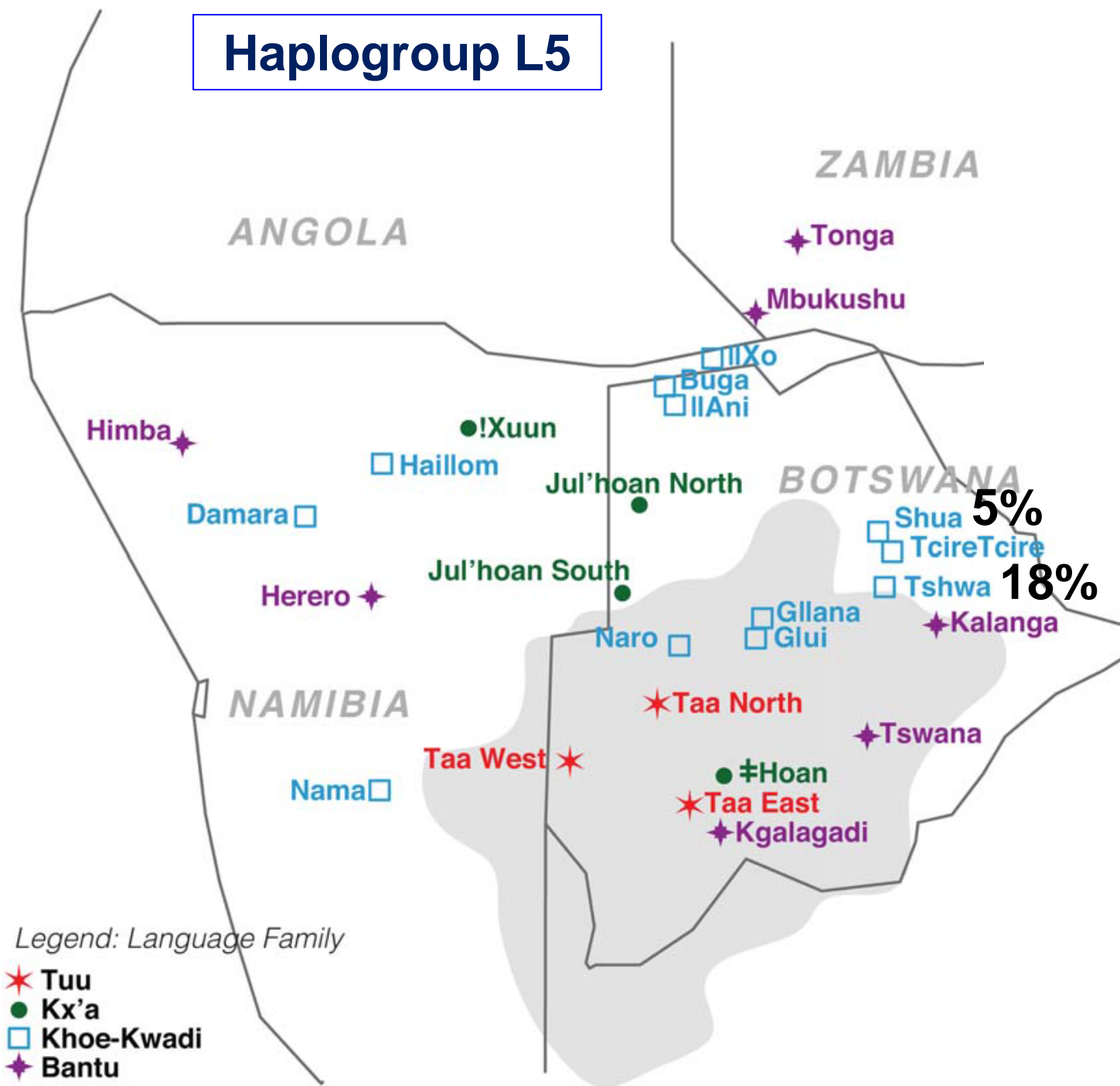
MtDNA

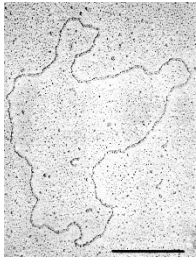
Unraveling the Complex Maternal History of Southern African Khoisan Populations

Chiara Barbieri,^{1*} Tom Güldemann,^{2,3} Christfried Naumann,^{2,3} Linda Gerlach,¹ Falko Berthold,¹ Hiroshi Nakagawa,⁴ Sununguko W. Mpoloka,⁵ Mark Stoneking,⁶ and Brigitte Pakendorf^{1*}

- **potential signals of eastern African mtDNAs hampered by paucity of data from eastern Africa**
- **best candidate: haplogroup L5**
 - **found in Tanzania (including Sandawe), Kenya, Ethiopia, Egypt, Sudan**
 - **(but also found in Central African Pygmies)**

Haplogroup L5

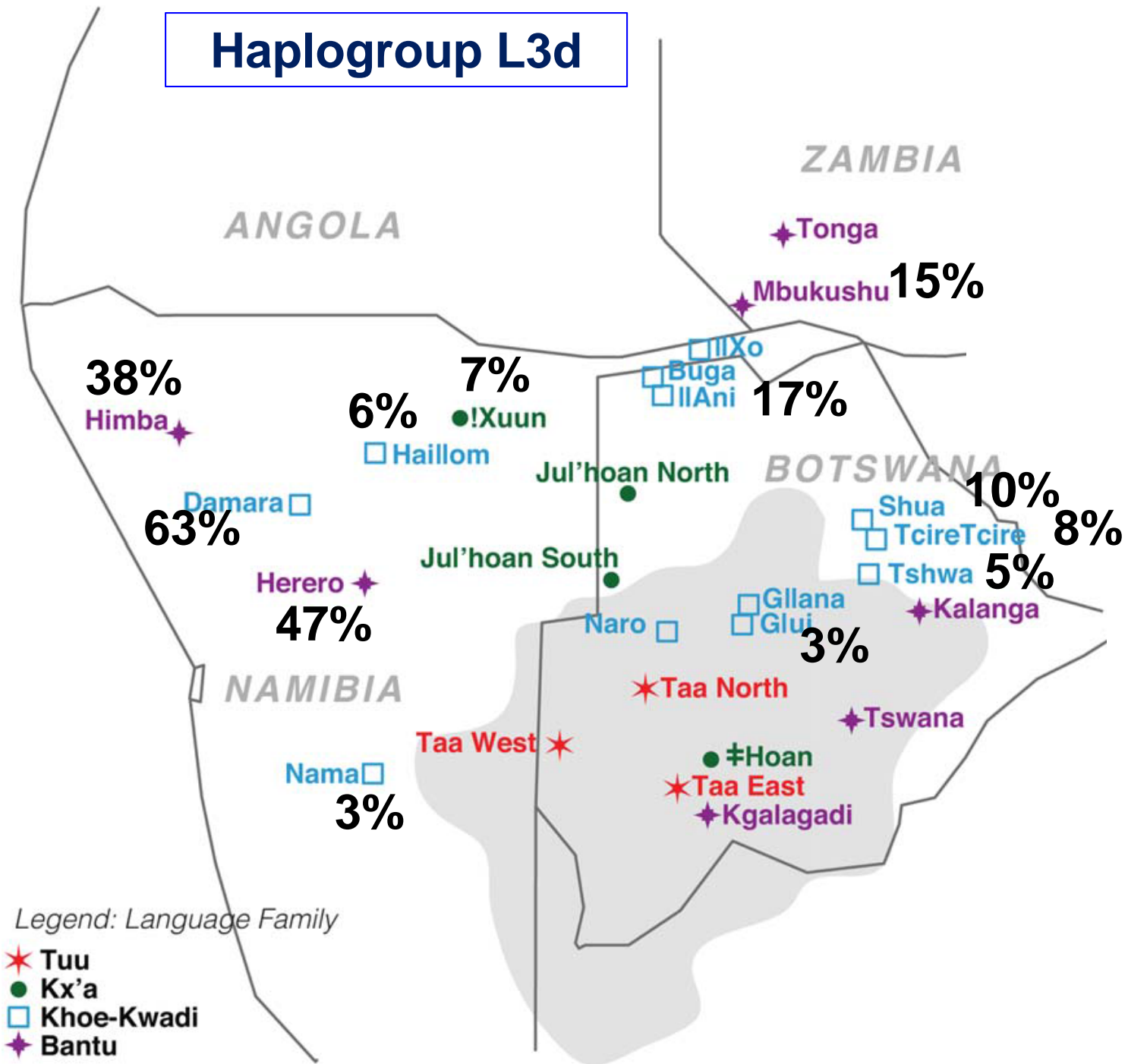




MtDNA

- another candidate: haplogroup **L3d**
 - mostly West African, but also occurs in Ethiopia

Haplogroup L3d

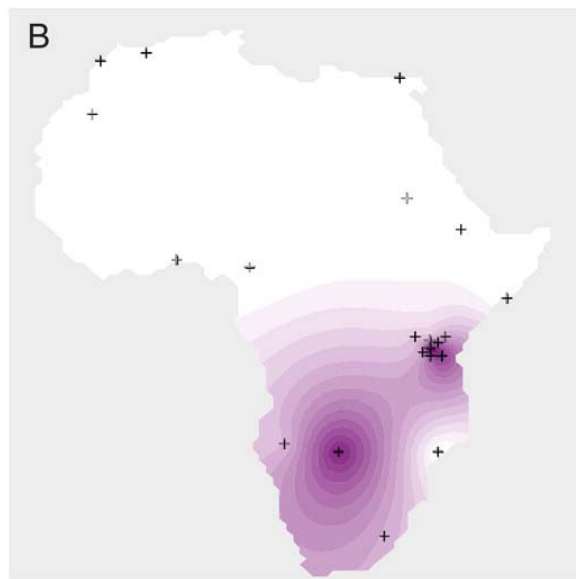




Y-chromosome

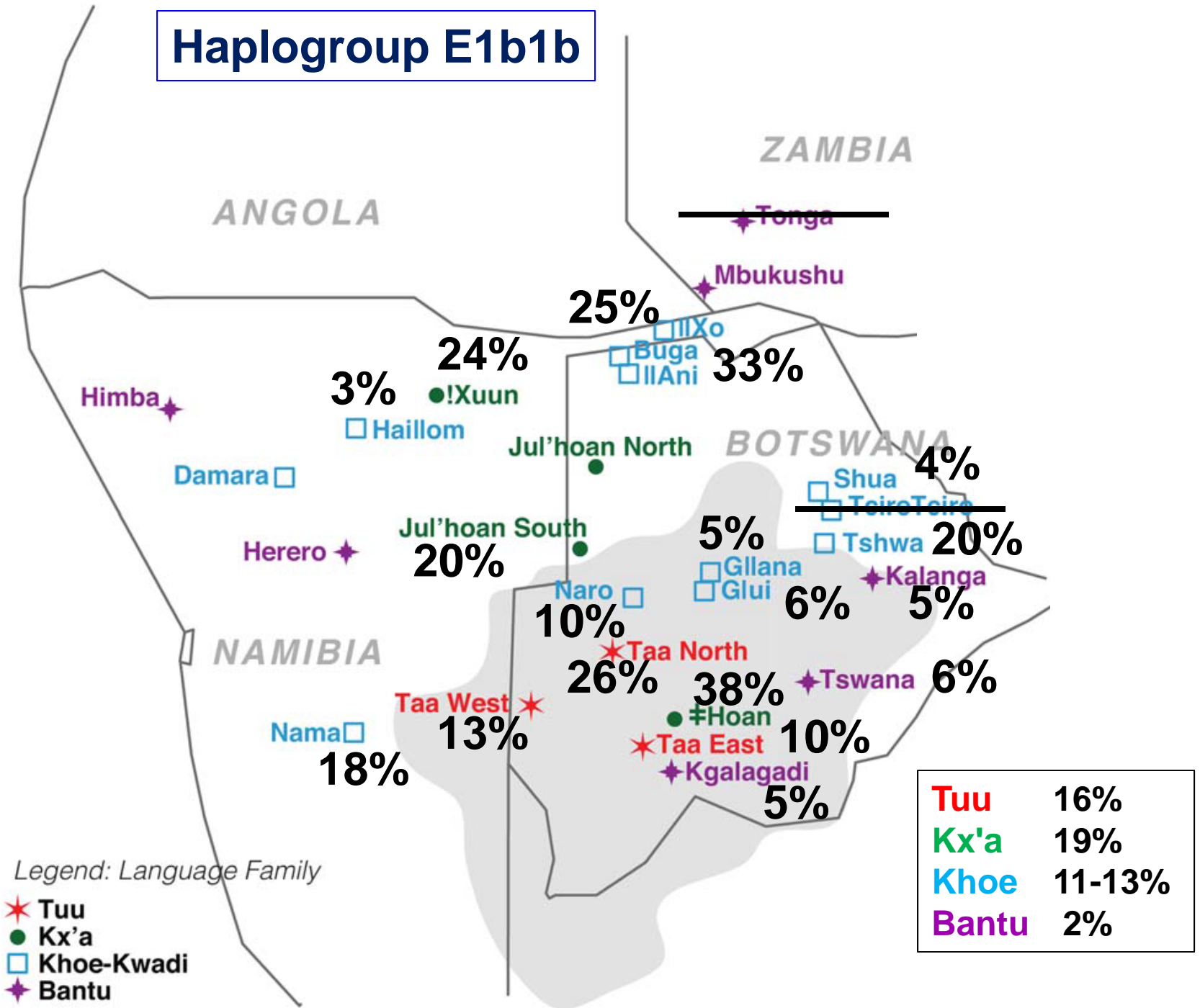
Y-chromosomal evidence of a pastoralist migration through Tanzania to southern Africa

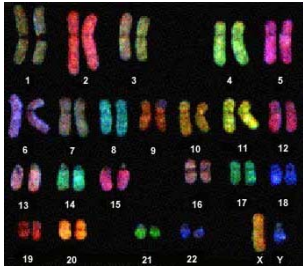
Brenna M. Henn^{†‡}, Christopher Gignoux^{†§}, Alice A. Lin[¶], Peter J. Oefner[¶], Peidong Shen^{††}, Rosaria Scozzari^{††}, Fulvio Cruciani^{††}, Sarah A. Tishkoff^{§§}, Joanna L. Mountain^{§¶}, and Peter A. Underhill[¶]



Khwe: 31%
!Kung: 11%
HGDP Bantu: 13%

Haplogroup E1b1b





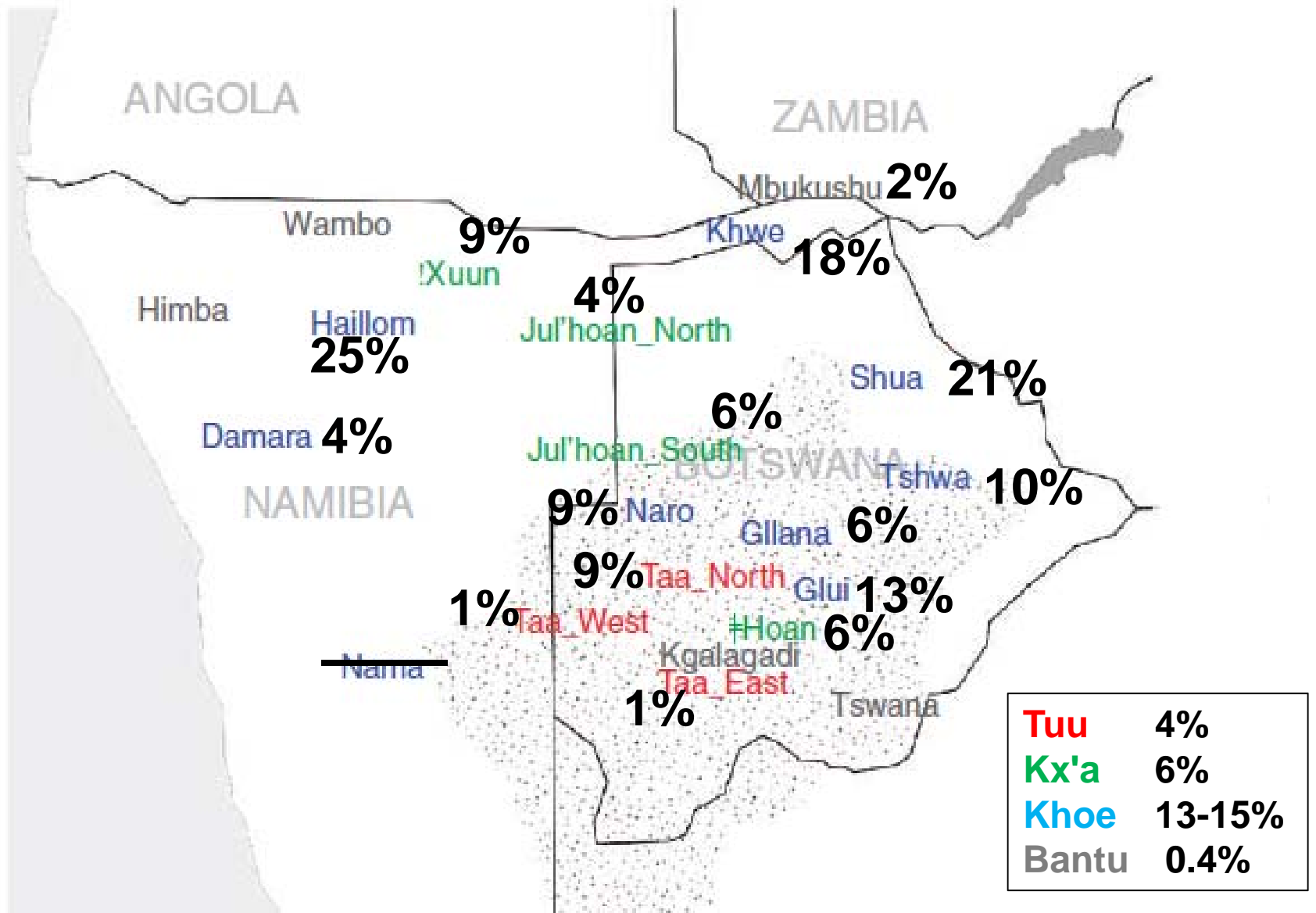
Genome-wide data

Ancient west Eurasian ancestry in southern and eastern Africa

Joseph K. Pickrell^{a,1,2}, Nick Patterson^b, Po-Ru Loh^c, Mark Lipson^c, Bonnie Berger^{b,c}, Mark Stoneking^d, Brigitte Pakendorf^{e,1}, and David Reich^{a,b,f,1}

- showed that southern African populations harbor Eurasian ancestry that is relatively old (~900-1800 years) and related to a signal of Eurasian ancestry in eastern Africa that is even older (~2700-3300 years)
- used this Eurasian ancestry to estimate east African ancestry in southern African populations

"East African" ancestry





Lactase persistence

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,*}

Brief Communication: New Insights into the History of the C-14010 Lactase Persistence Variant in Eastern and Southern Africa

Enrico Macholdt,¹ Montgomery Slatkin,² Brigitte Pakendorf,^{3*} and Mark Stoneking^{1*}

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,*}

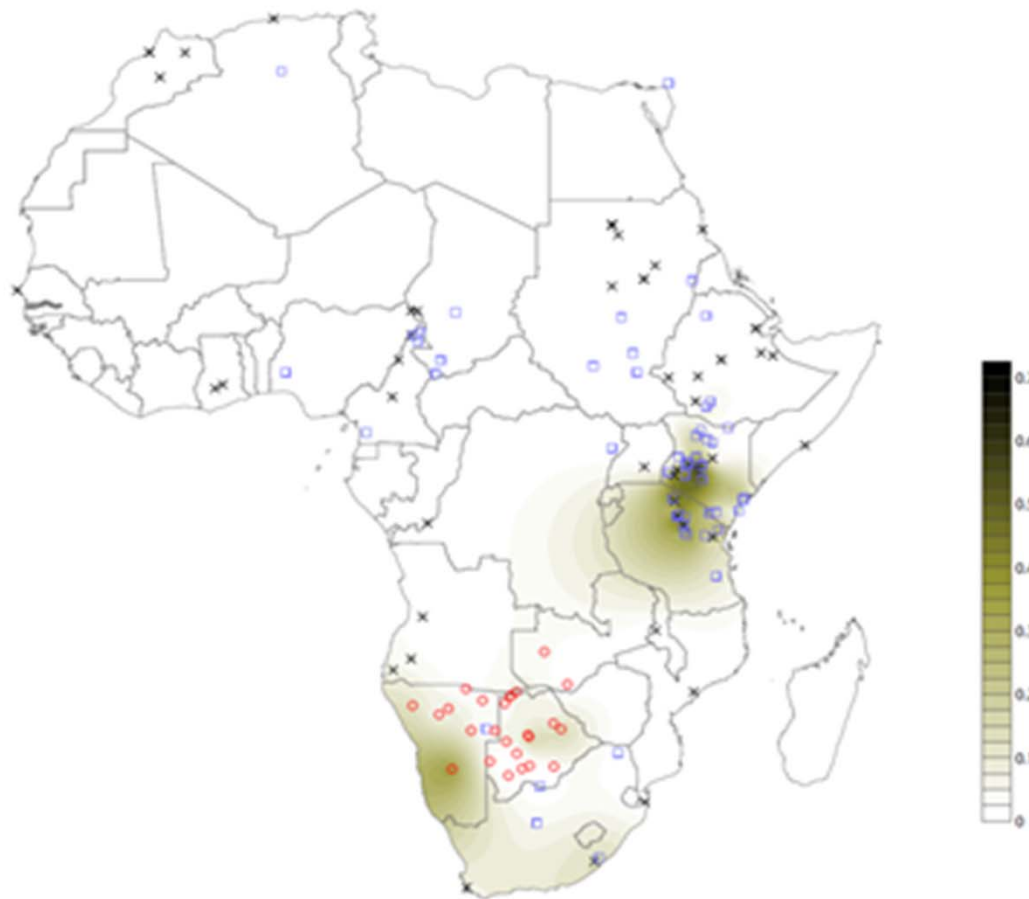


Lactase persistence

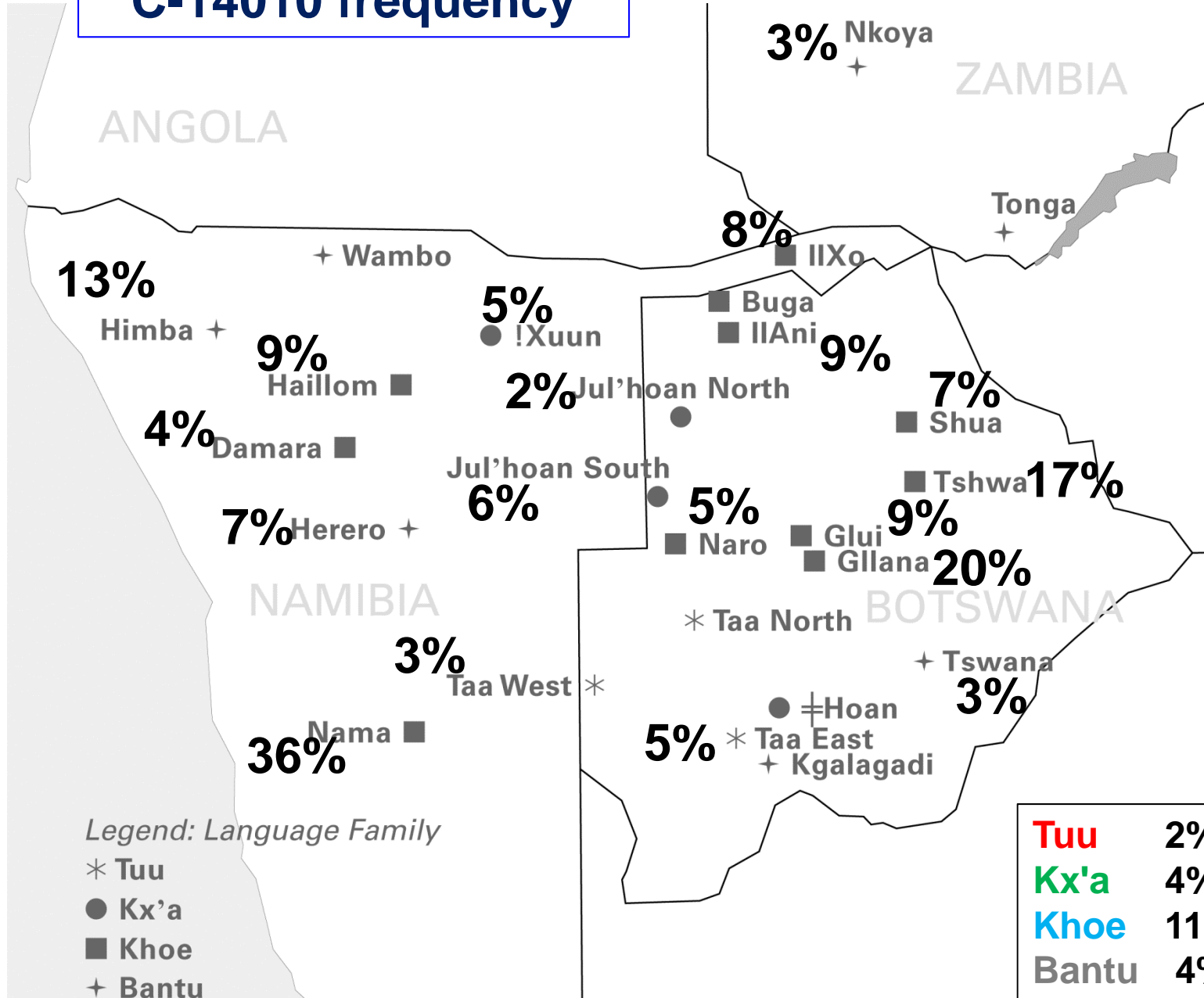
- ability to digest milk into adulthood (**lactase persistence** or **lactose tolerance**) is conferred by several mutations which show geographic specificity and have been subject to positive selection in pastoralist populations
- one particular mutation (C-14010) that originated in eastern Africa is prevalent in southern Africa



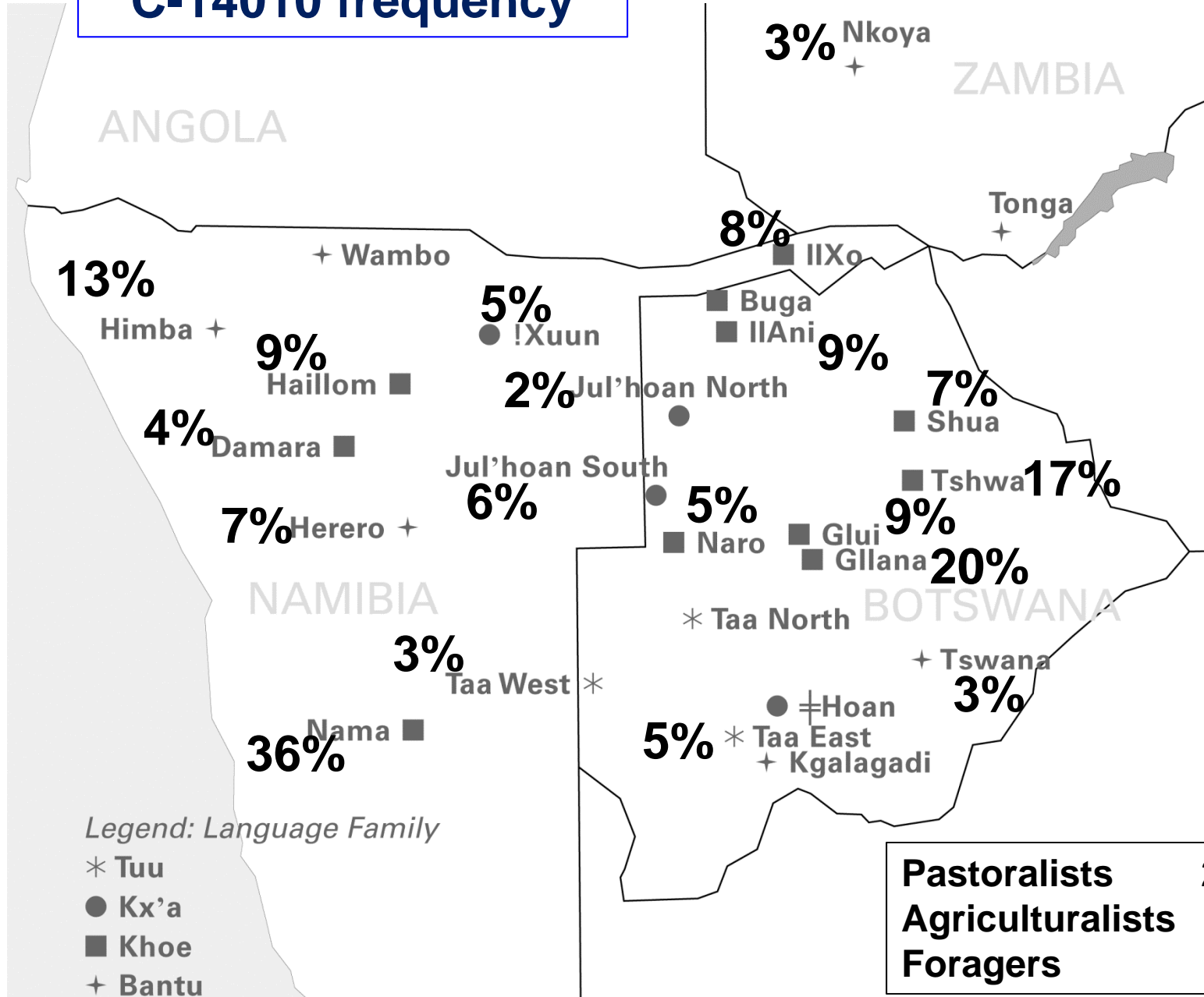
C-14010 frequency



C-14010 frequency



C-14010 frequency

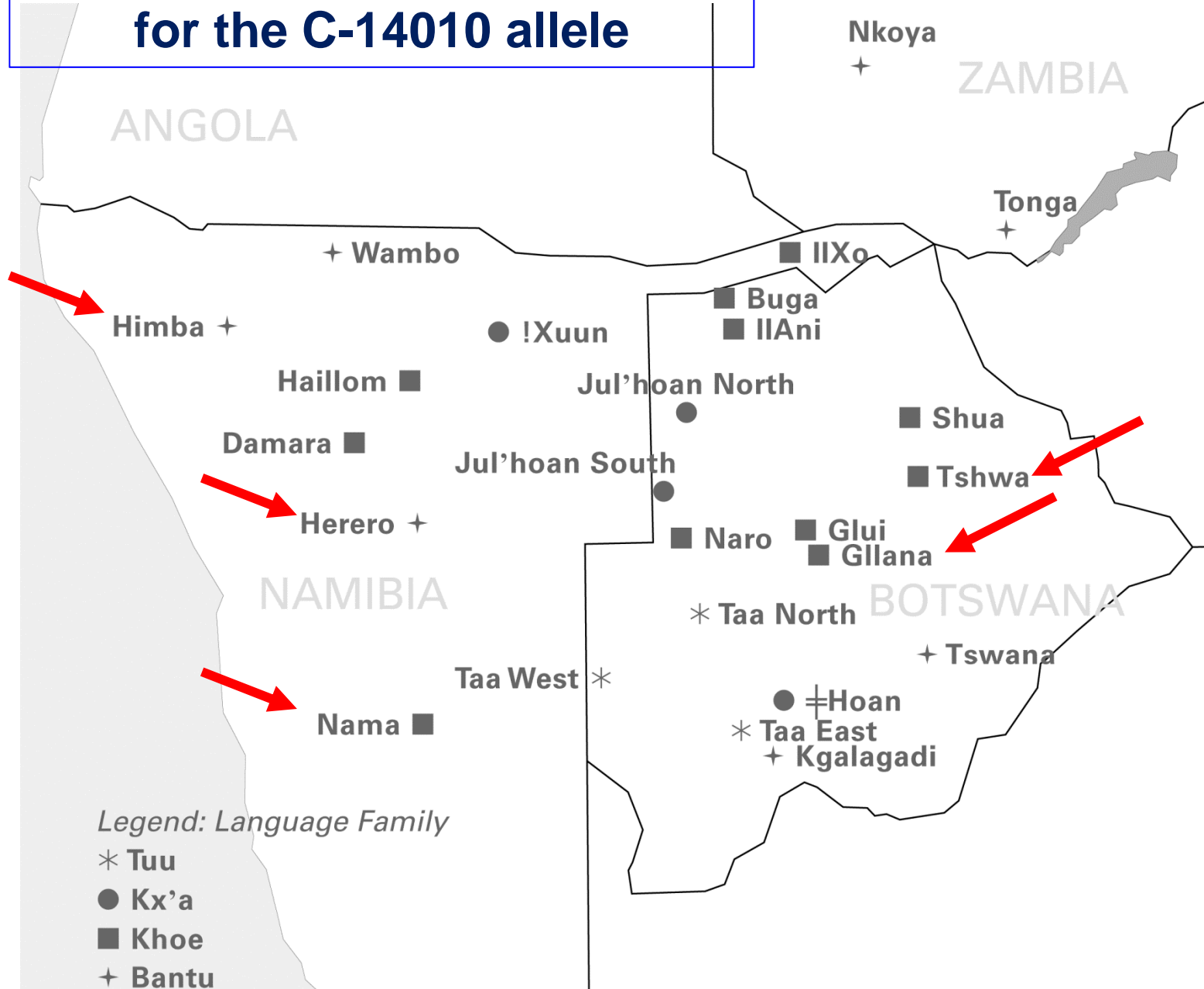


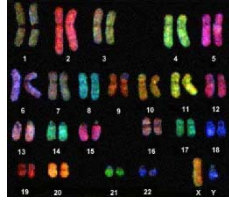
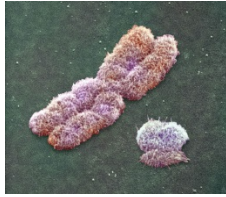
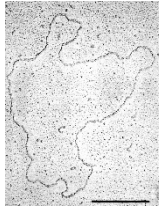
Legend: Language Family

- * Tuu
- Kx'a
- Khoe
- + Bantu

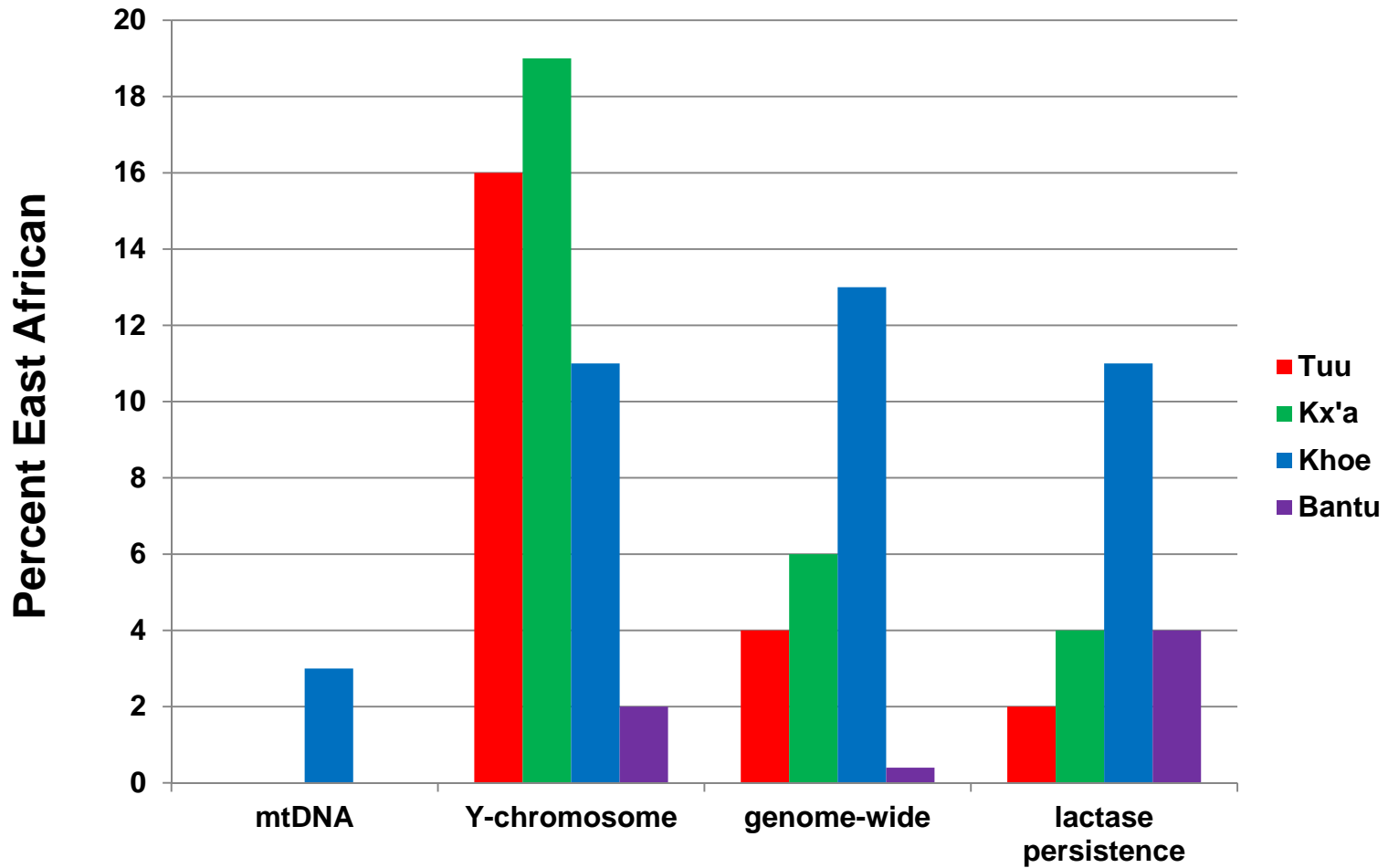
Pastoralists	20%
Agriculturalists	1%
Foragers	7%

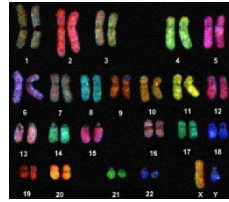
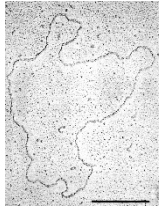
Ongoing positive selection for the C-14010 allele





Summary





Conclusions

- **Y-chromosome, genome-wide, and lactase persistence data all show a strong signal of a migration from eastern to southern Africa that had the strongest influence on Khoe speakers**
- **mtDNA data are more equivocal**
- **whether this is due to a lack of sufficient comparative mtDNA data from eastern Africa, or rather indicates that the migration was primarily male-mediated, remains to be seen**