

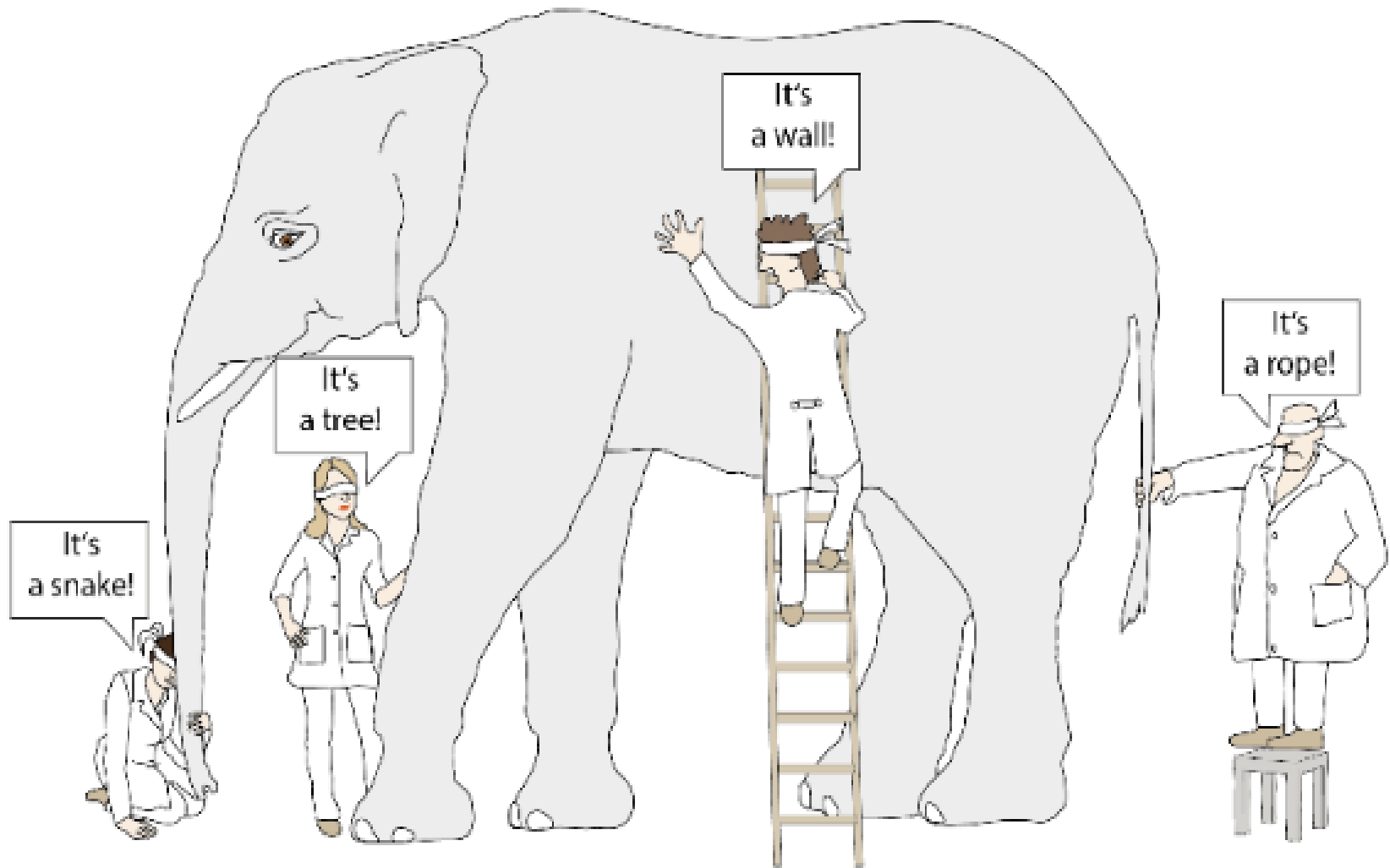
# **Kalahari Basin Prehistory: Genetic divergence**

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

**Kalahari Basin Area**

**Endangered Language & Population History Research**

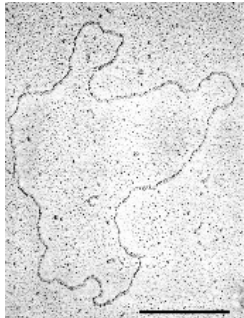
# The blind researchers and the elephant



(with thanks to B. Pakendorf)

# Outline

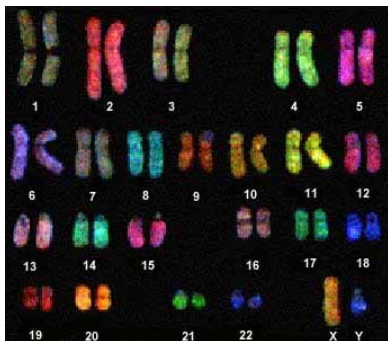
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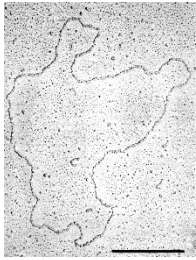
mtDNA: **maternal** history



Y chromosome: **paternal** history



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



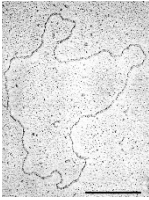
# MtDNA

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## Unraveling the Complex Maternal History of Southern African Khoisan Populations

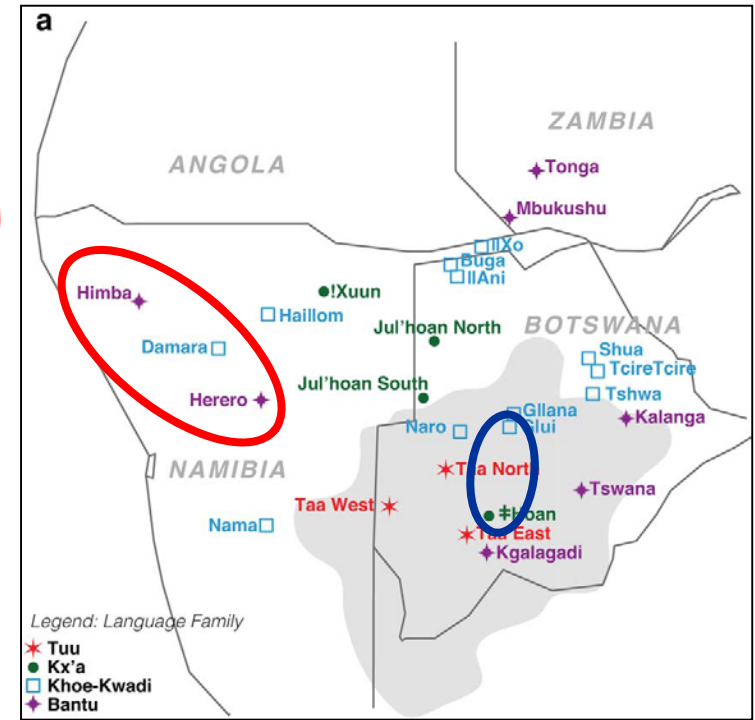
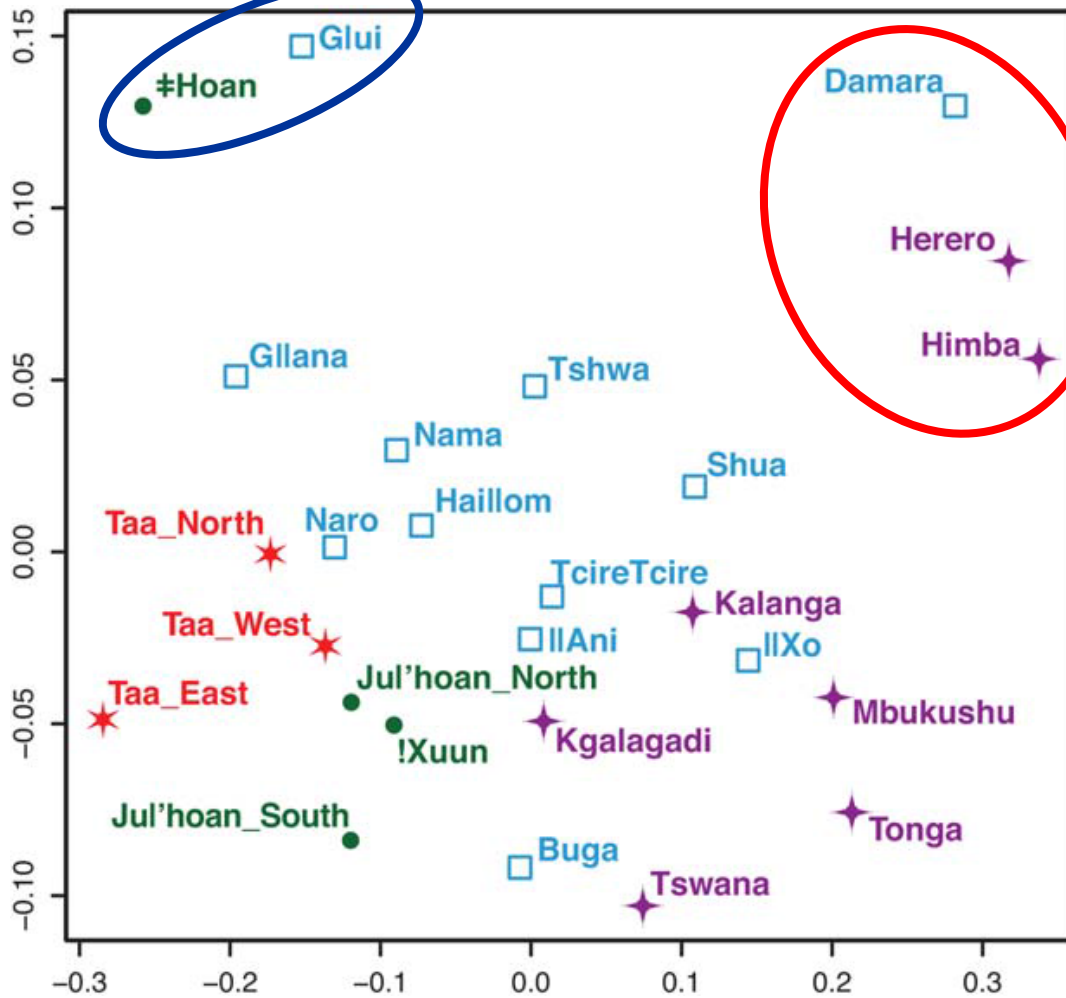
Chiara Barbieri,<sup>1\*</sup> Tom Güldemann,<sup>2,3</sup> Christfried Naumann,<sup>2,3</sup> Linda Gerlach,<sup>1</sup> Falko Berthold,<sup>1</sup> Hiroshi Nakagawa,<sup>4</sup> Sununguko W. Mpoloka,<sup>5</sup> Mark Stoneking,<sup>6</sup> and Brigitte Pakendorf<sup>1\*</sup>

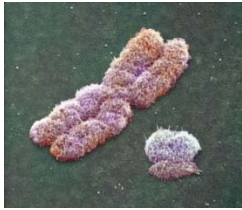
AMERICAN JOURNAL OF PHYSICAL ANTHROPOLOGY 153:435–448 (2014)



# MtDNA

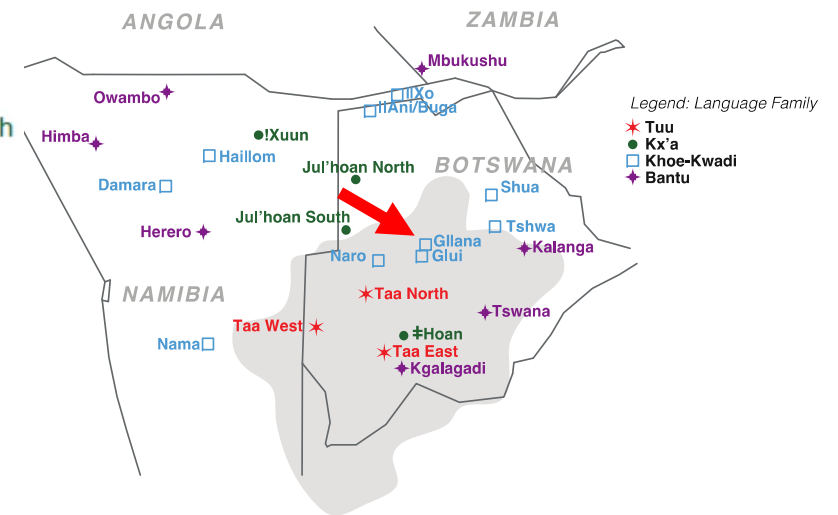
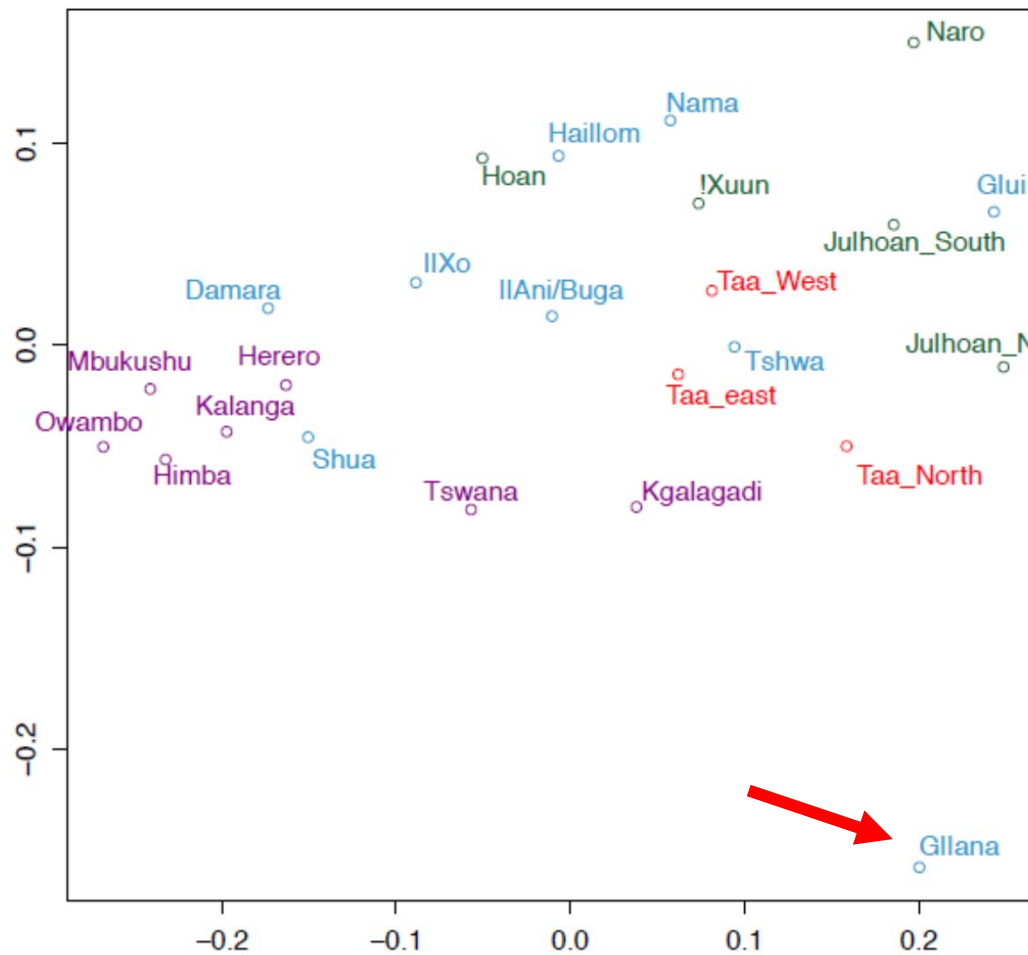
## Multidimensional scaling (MDS) plot



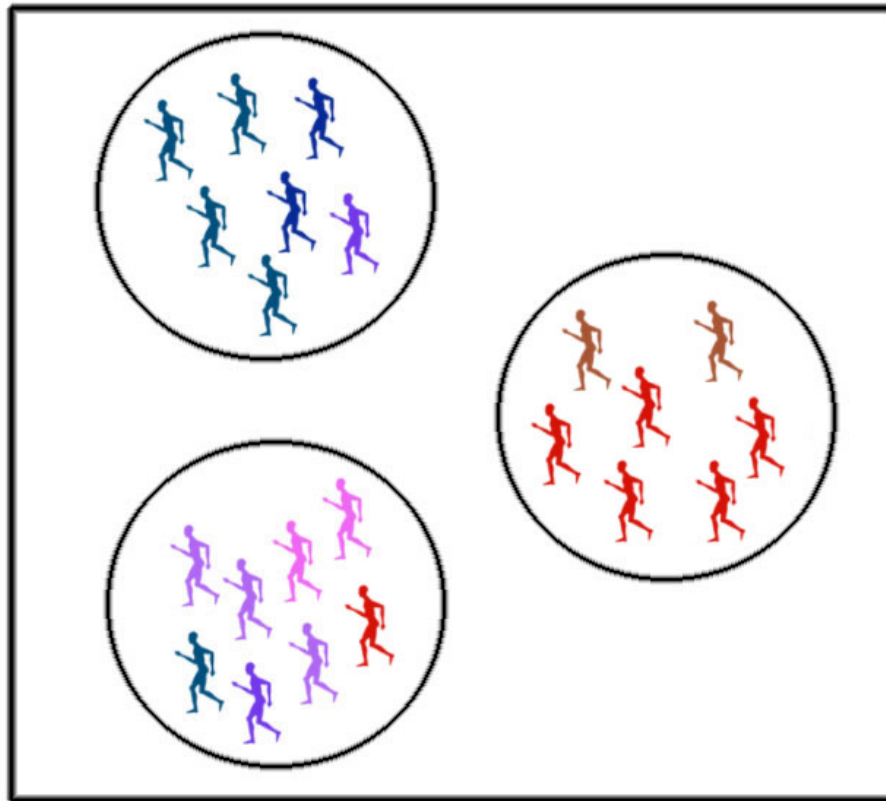


# Y-chromosome

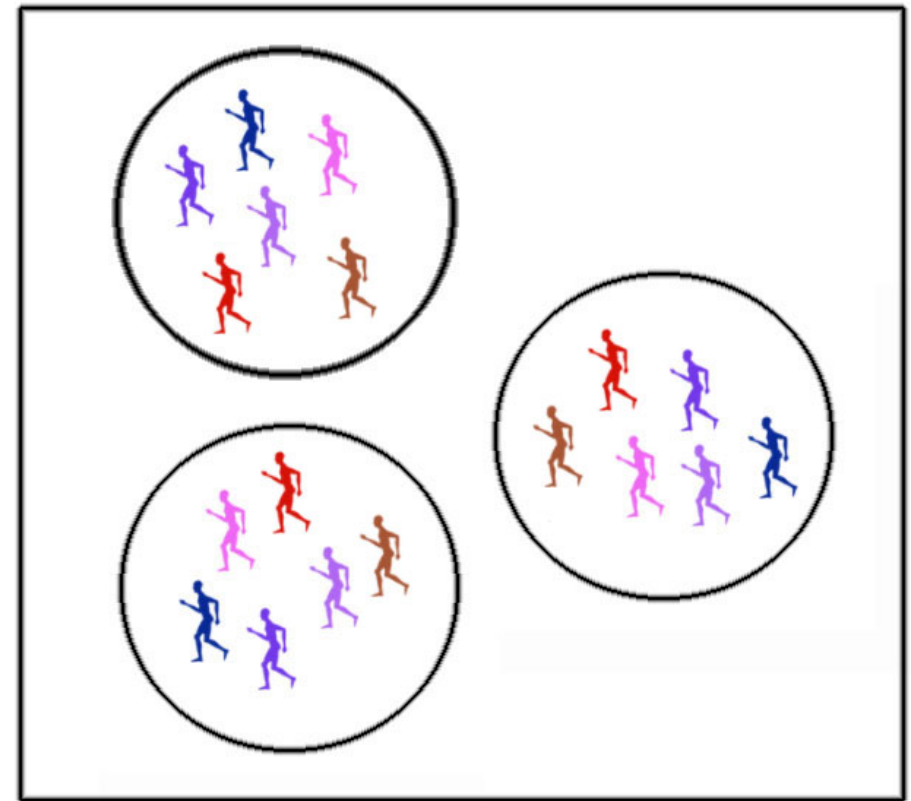
## MDS plot



# Analysis of Molecular Variance: AMOVA



**Larger** genetic differences  
**between** groups

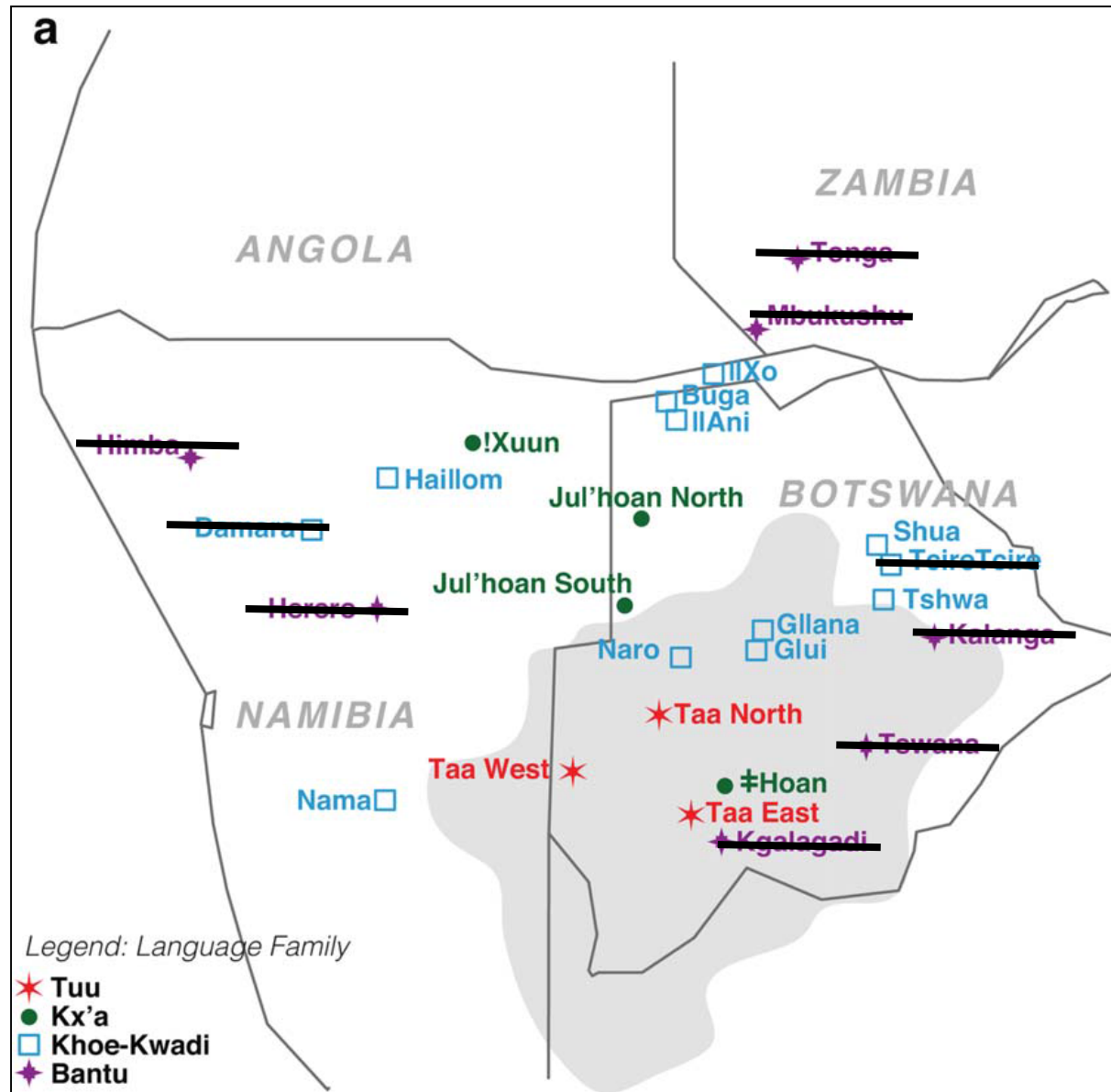


**Smaller** genetic differences  
**between** groups (and so  
more of the variance is  
**within** groups)

(with thanks to C. Barbieri)

# AMOVA – populations excluded

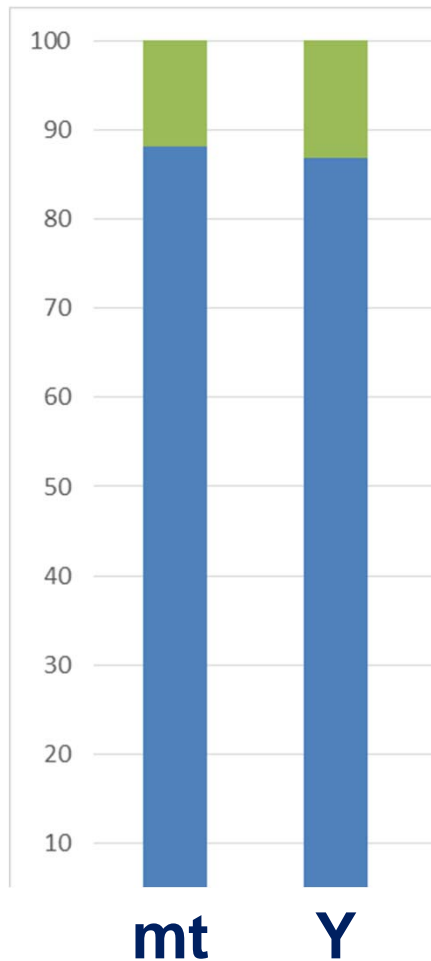
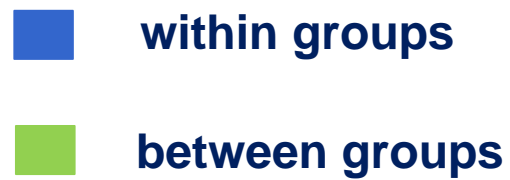
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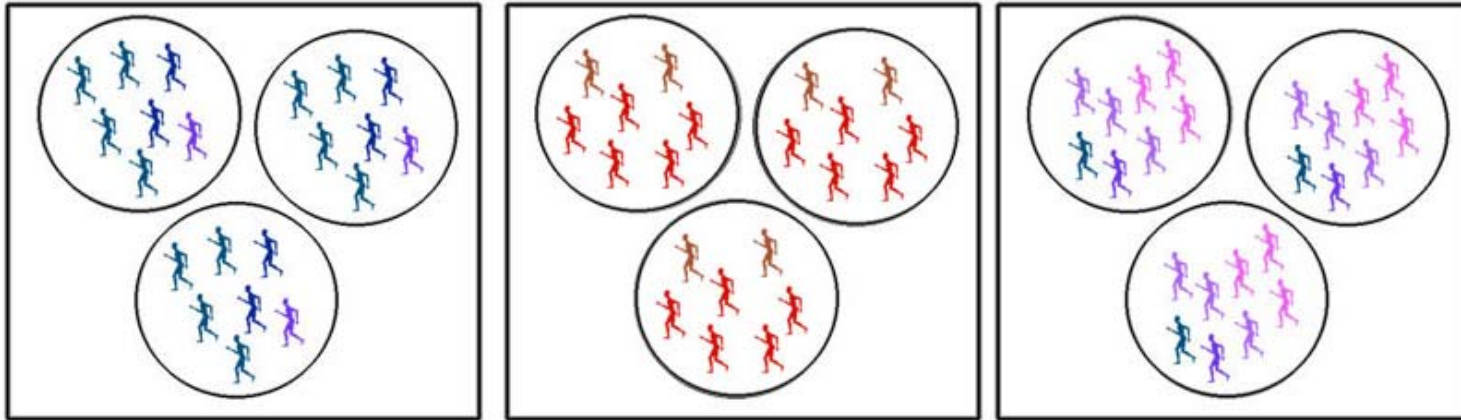
# AMOVA – Khoisan groups

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# AMOVA to test groupings

1

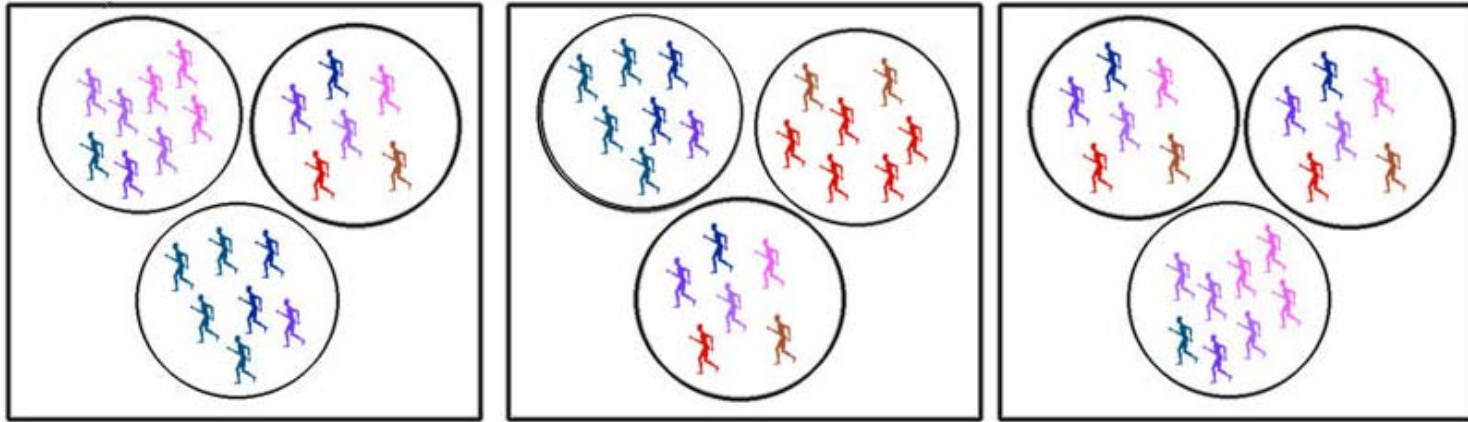


**Larger genetic differences between groups**  
**Smaller genetic differences among populations**  
**from the same group**

**Genetic differences are mostly between groups**  
**=> groupings correspond to genetic structure**

# AMOVA to test groupings

2



**Larger genetic differences among populations from the same group**

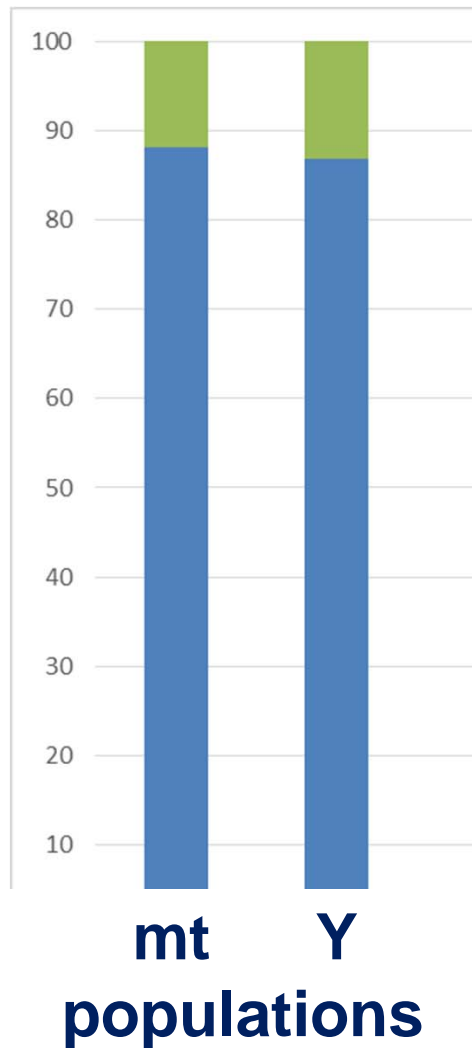
**Smaller genetic differences between groups**

**Genetic differences are mostly among populations from the same group**

**=> groupings DO NOT correspond to genetic structure**

# AMOVA – Khoisan populations

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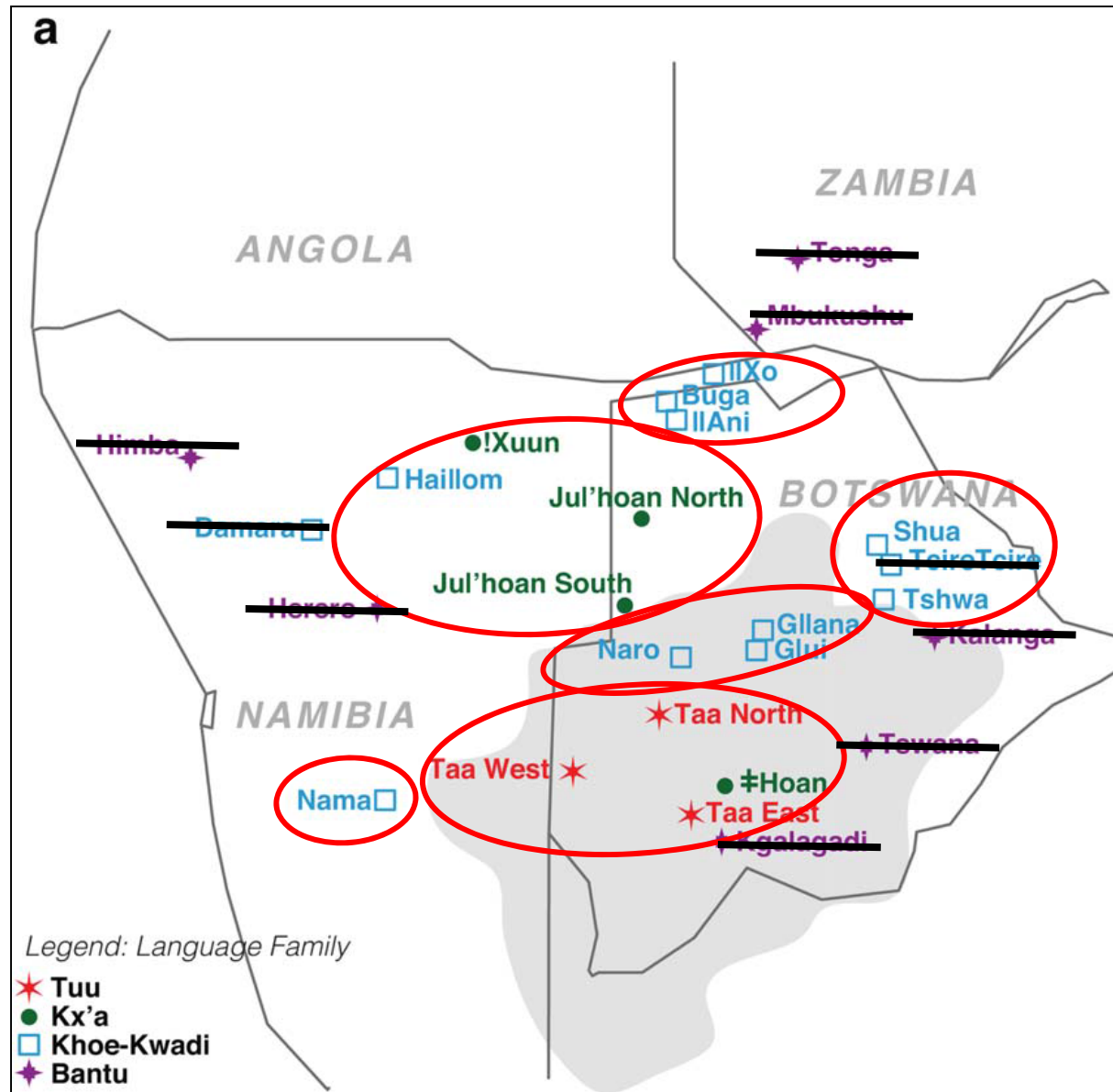


■ within groups

■ among populations  
within groups

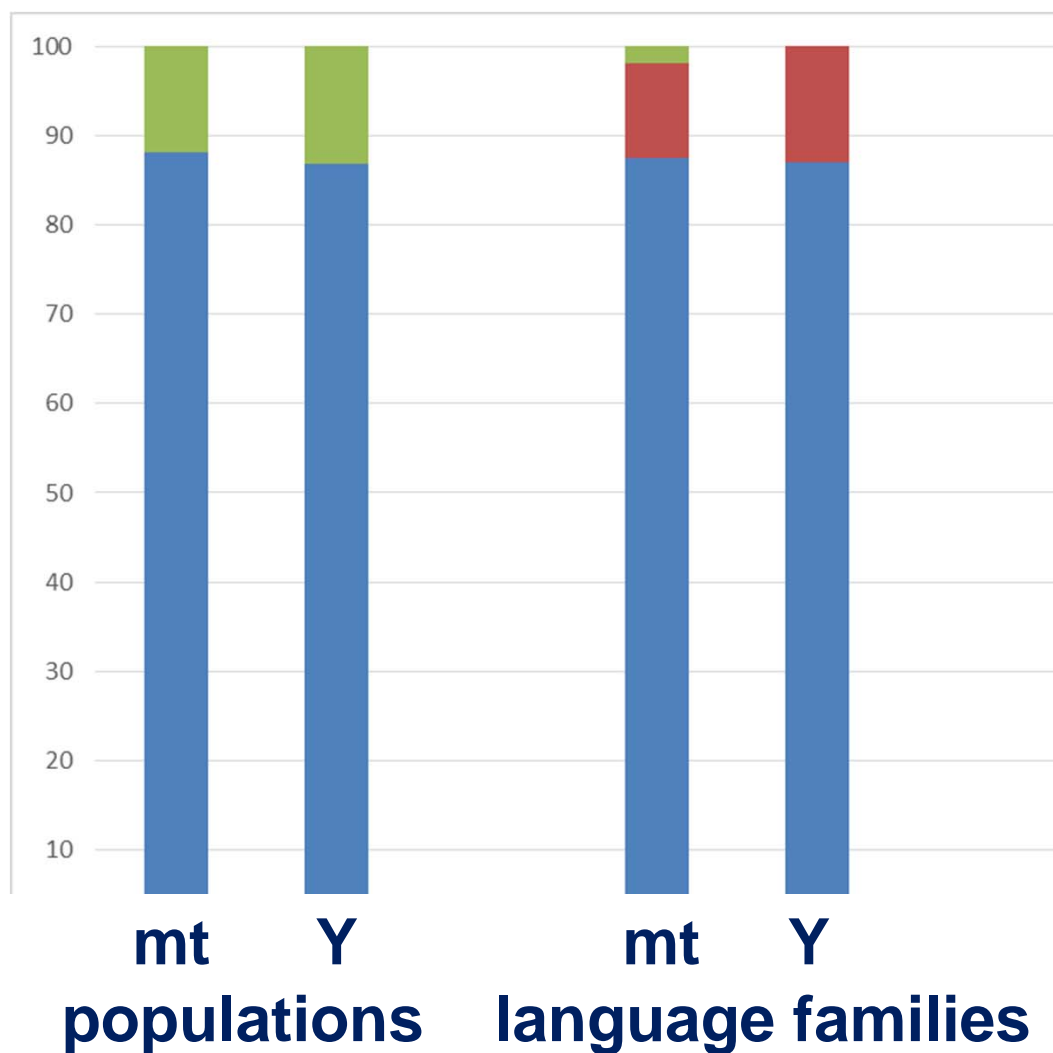
■ between groups

# Geographic clusters (Khoisan only!)



# AMOVA – Khoisan groups

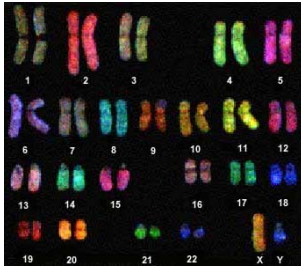
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■ within groups

■ among populations  
within groups

■ between groups



# Genome-wide data

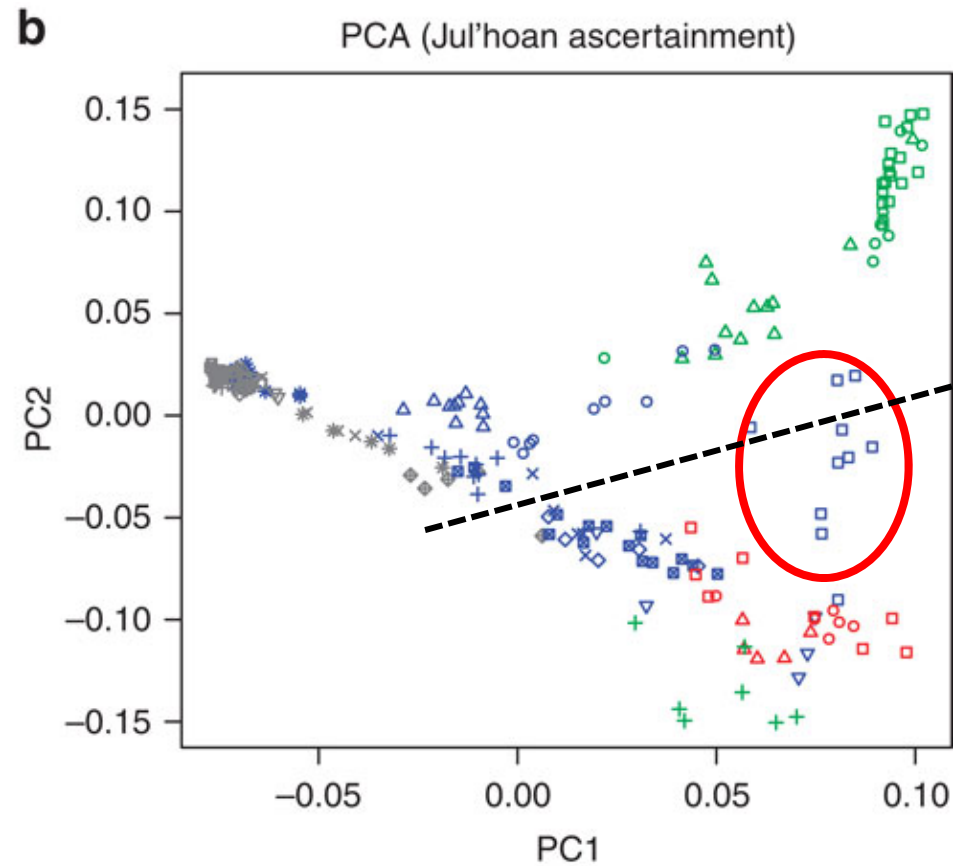
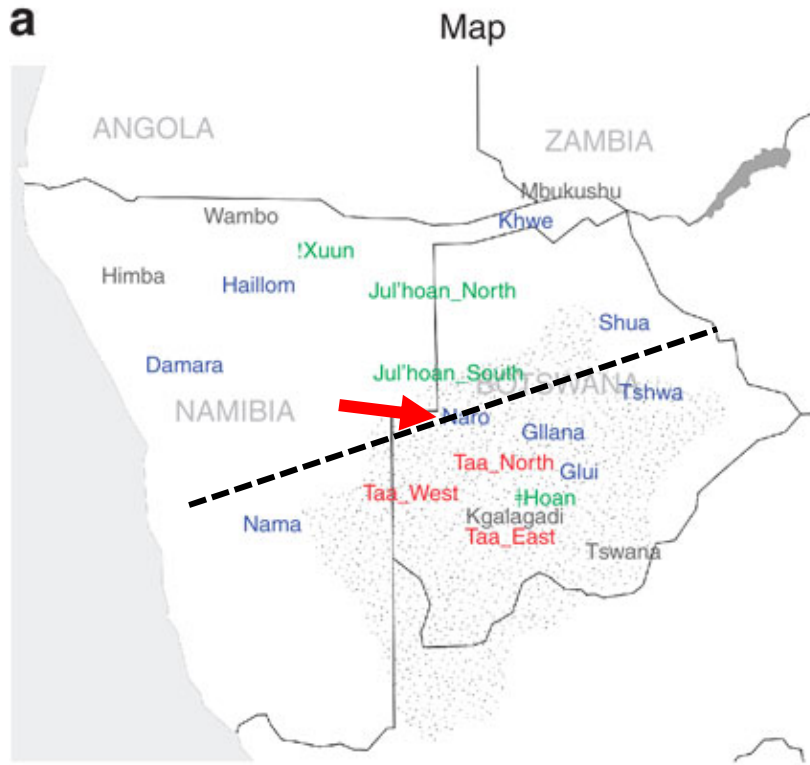
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## The genetic prehistory of southern Africa

Joseph K. Pickrell<sup>1</sup>, Nick Patterson<sup>2</sup>, Chiara Barbieri<sup>3,†</sup>, Falko Berthold<sup>3,†</sup>, Linda Gerlach<sup>3,†</sup>, Tom Güldemann<sup>4,5</sup>, Blesswell Kure<sup>6</sup>, Sununguko Wata Mpoloka<sup>7</sup>, Hiroshi Nakagawa<sup>8</sup>, Christfried Naumann<sup>4,5</sup>, Mark Lipson<sup>9,10</sup>, Po-Ru Loh<sup>9,10</sup>, Joseph Lachance<sup>11,12</sup>, Joanna Mountain<sup>13</sup>, Carlos D. Bustamante<sup>14</sup>, Bonnie Berger<sup>9,10</sup>, Sarah A. Tishkoff<sup>11,12</sup>, Brenna M. Henn<sup>14</sup>, Mark Stoneking<sup>15</sup>, David Reich<sup>1,2</sup> & Brigitte Pakendorf<sup>3,†</sup>

NATURE COMMUNICATIONS | 3:1143 | DOI: 10.1038/ncomms2140 | [www.nature.com/naturecommunications](http://www.nature.com/naturecommunications)

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Non-Khoisan

- Himba
- Wambo
- △ Dinka
- + Yoruba
- × BantuSouthAfrica
- ◇ BantuKenya
- ▽ Mbukushu
- Mandenka
- \* Tswana
- ◆ Kgalagadi

Khoe-Kwadi

- Naro
- Haillom
- △ Khwe
- + Shua
- × Tshwa
- ◇ Gilana
- ▽ Glui
- Nama
- \* Damara

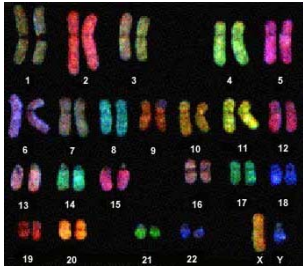
Kx'a

- Jul'hoan\_North
- Jul'hoan\_South
- △ !Xuun
- + #Hoan

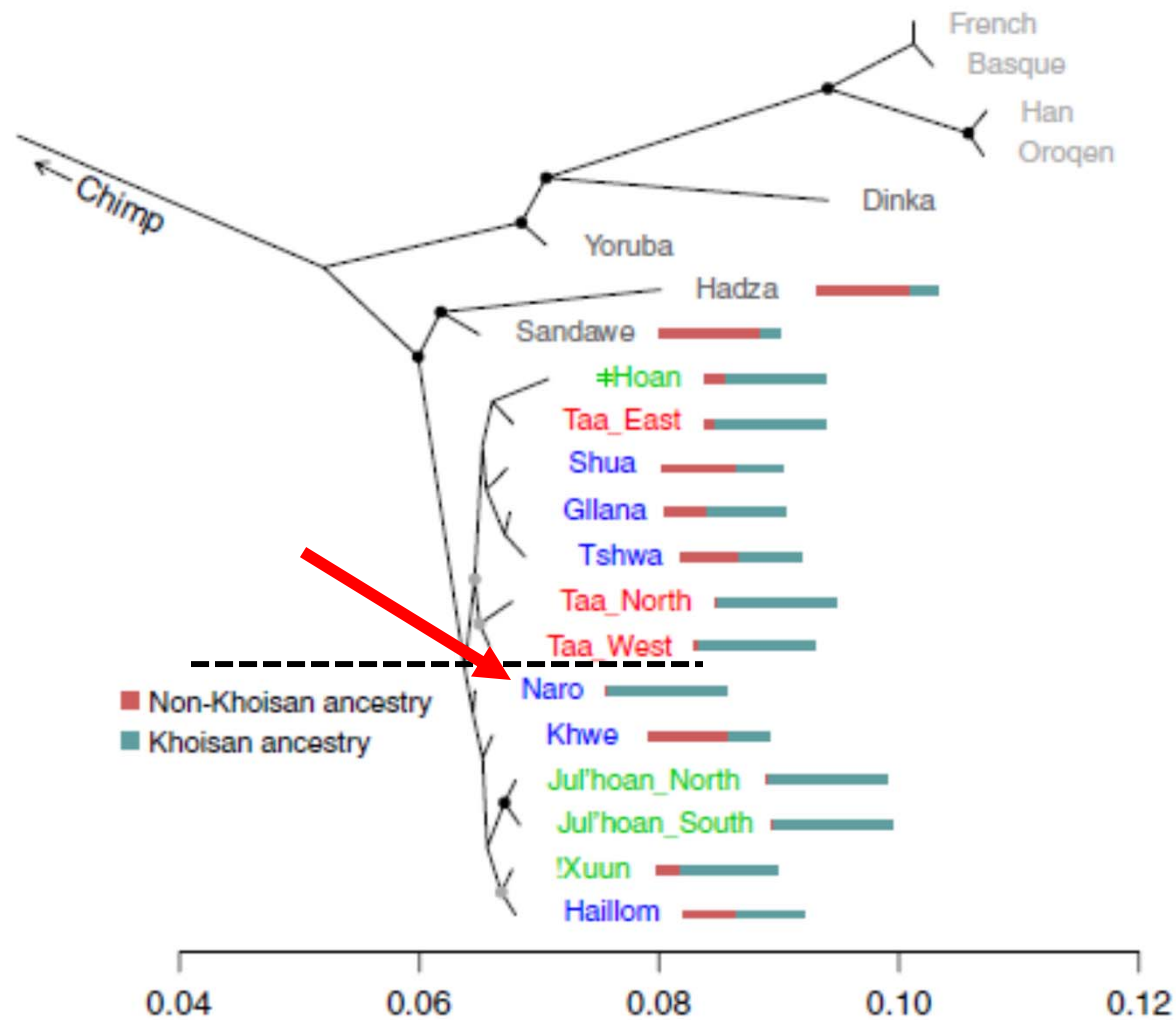
Tuu

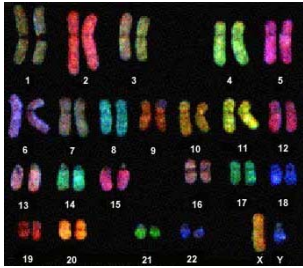
- Taa\_West
- Taa\_North
- △ Taa\_East



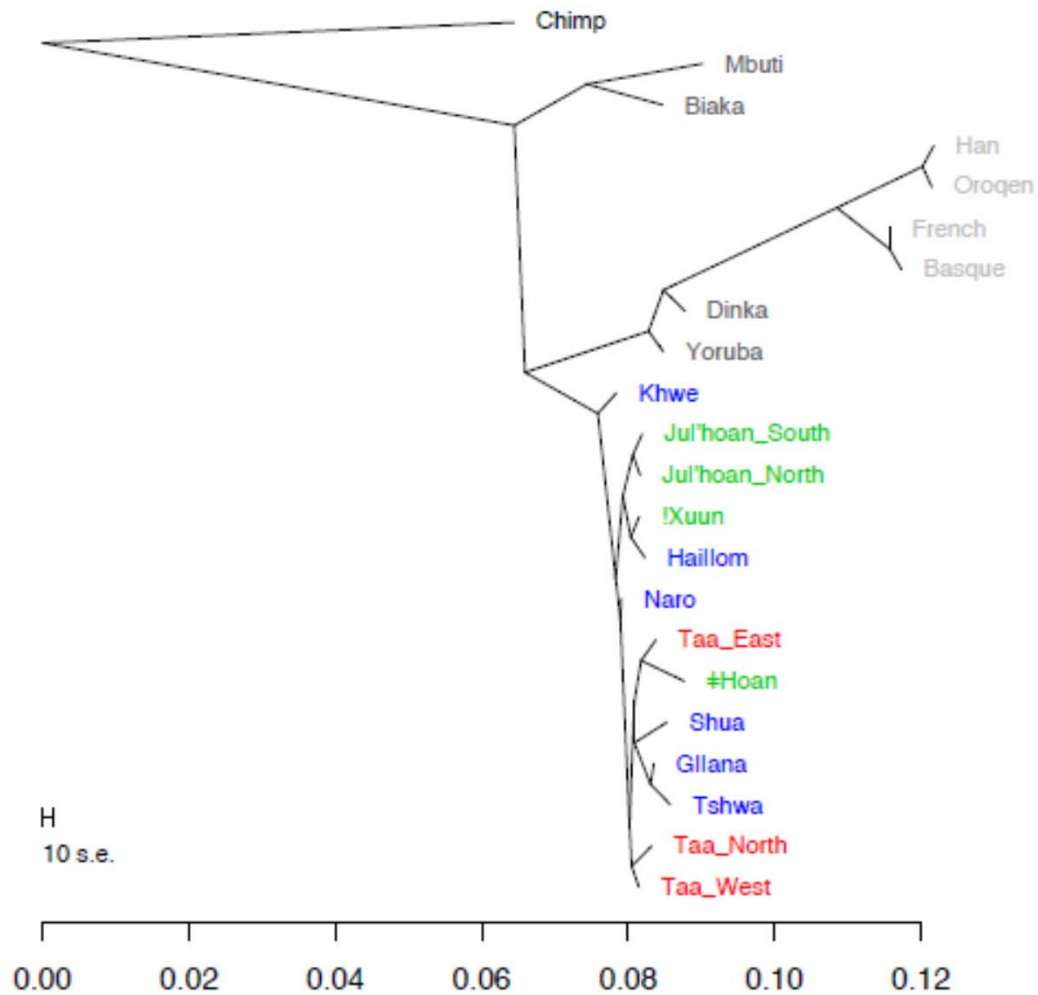


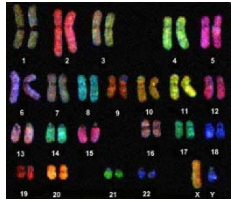
# Tree based on “Khoisan” ancestry





# Tree based on “non-Bantu” ancestry

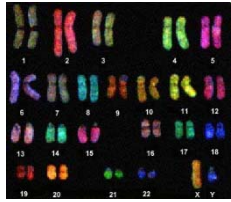




# How old is this divergence?

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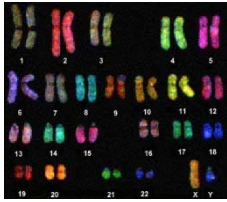
- Developed an approach based on new mutations that occurred after the divergence of NW and SE Kalahari
- Result: divergence **~30,000** years ago
- many assumptions involved in the method that mean it is probably an upper bound
- however, a different method applied to a different dataset gives a similar date (**~35,000** years ago; Schlebusch et al. 2012)
- the existence of further substructure within mtDNA lineages suggests divergence could be even older



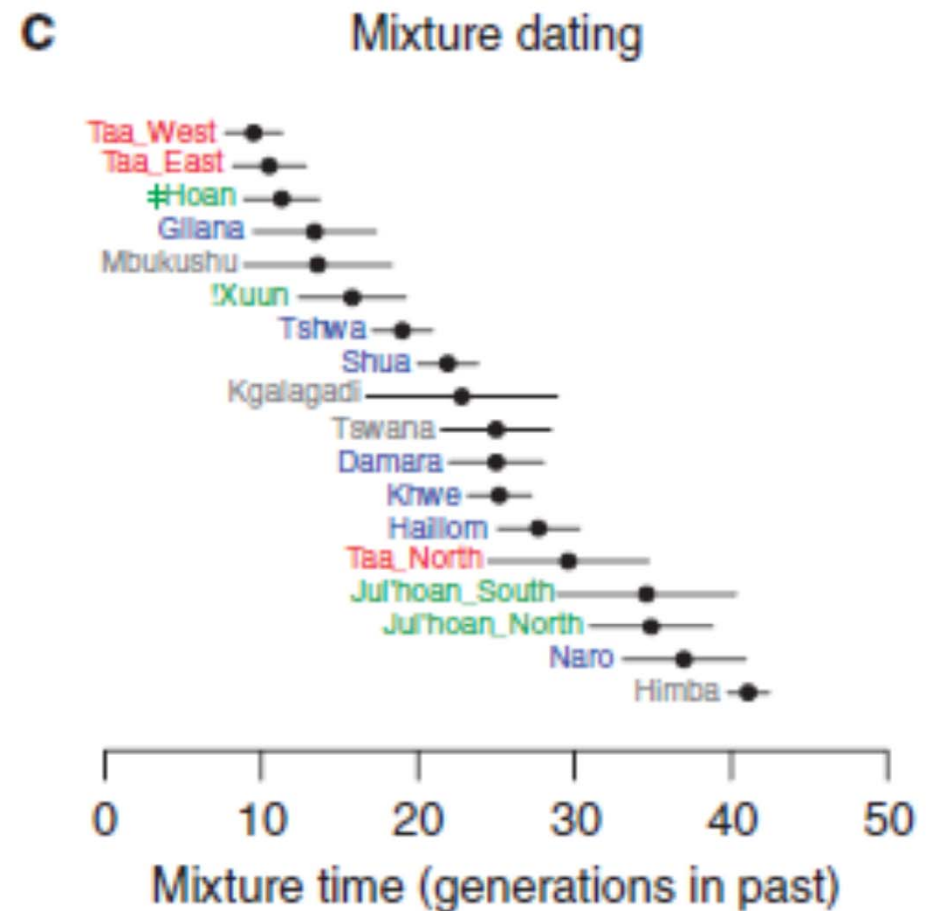
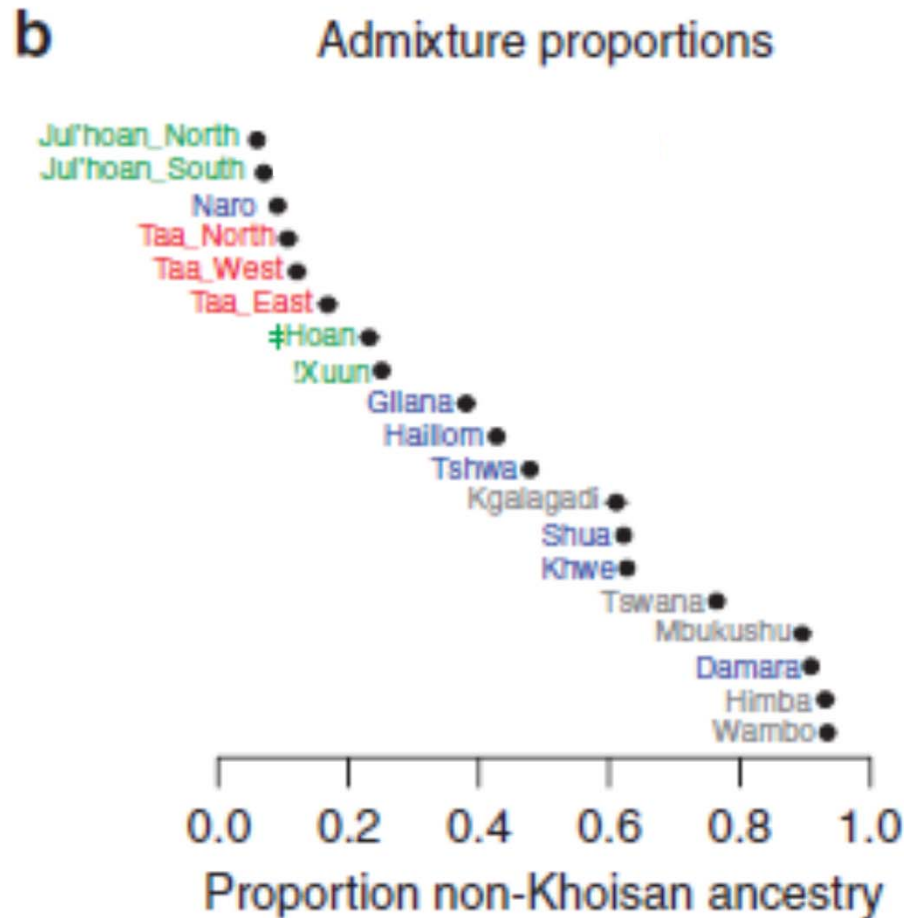
# Was there contact after divergence?

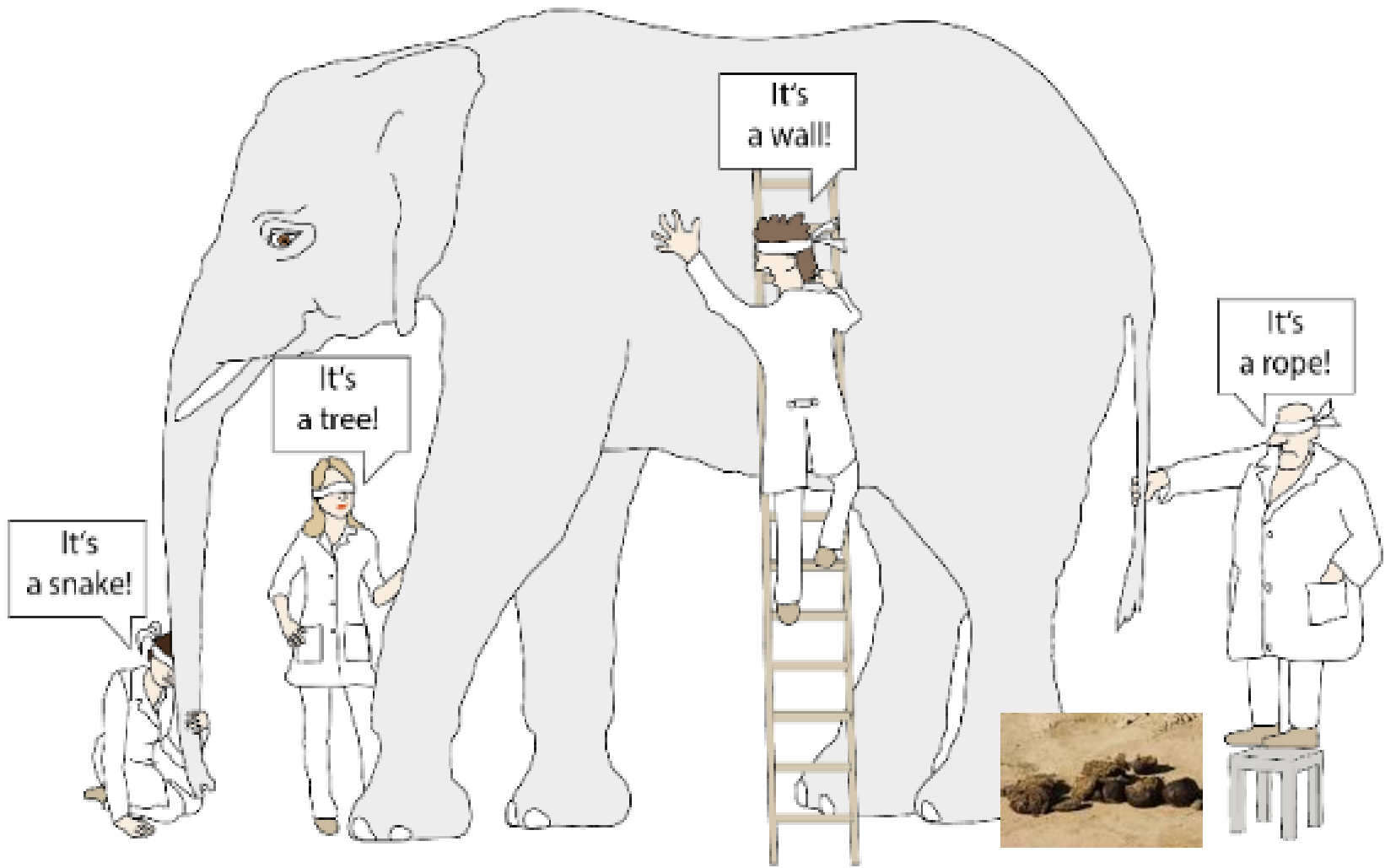
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- **YES: identical mtDNA sequences** shared between NW and SE Kalahari groups
- Modeling shows that such sharing reflects contact within the past **7,500 years ago** (if it were any older, mtDNA sequences would no longer be identical)
- Overall, the results suggest **deep divergence** between NW and SE Kalahari groups (possibly due to the presence of Makgadikgadi Lake?) followed by more recent contact (possibly due to drying of the lake?)



# And don't forget: extensive contact with other groups!





It's a snake!

It's a tree!

It's a wall!

It's a rope!