

# North West and North East Africa: a different perspective from mtDNA haplogroups distribution

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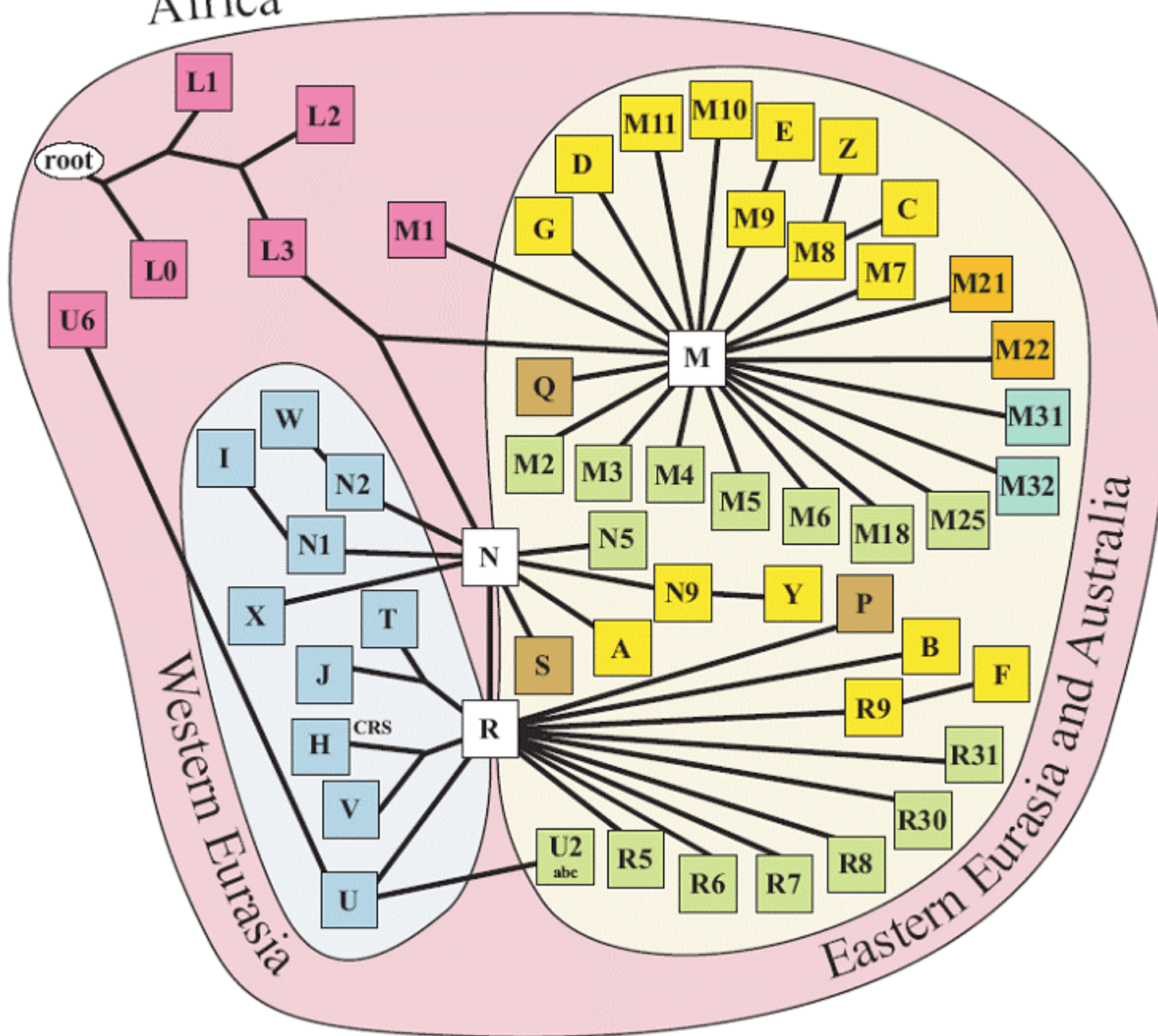
Laboratoire d'Étude des Polymorphismes de L'ADN, Nantes

Aussois,  
23/09/2005

# Mitochondrial DNA

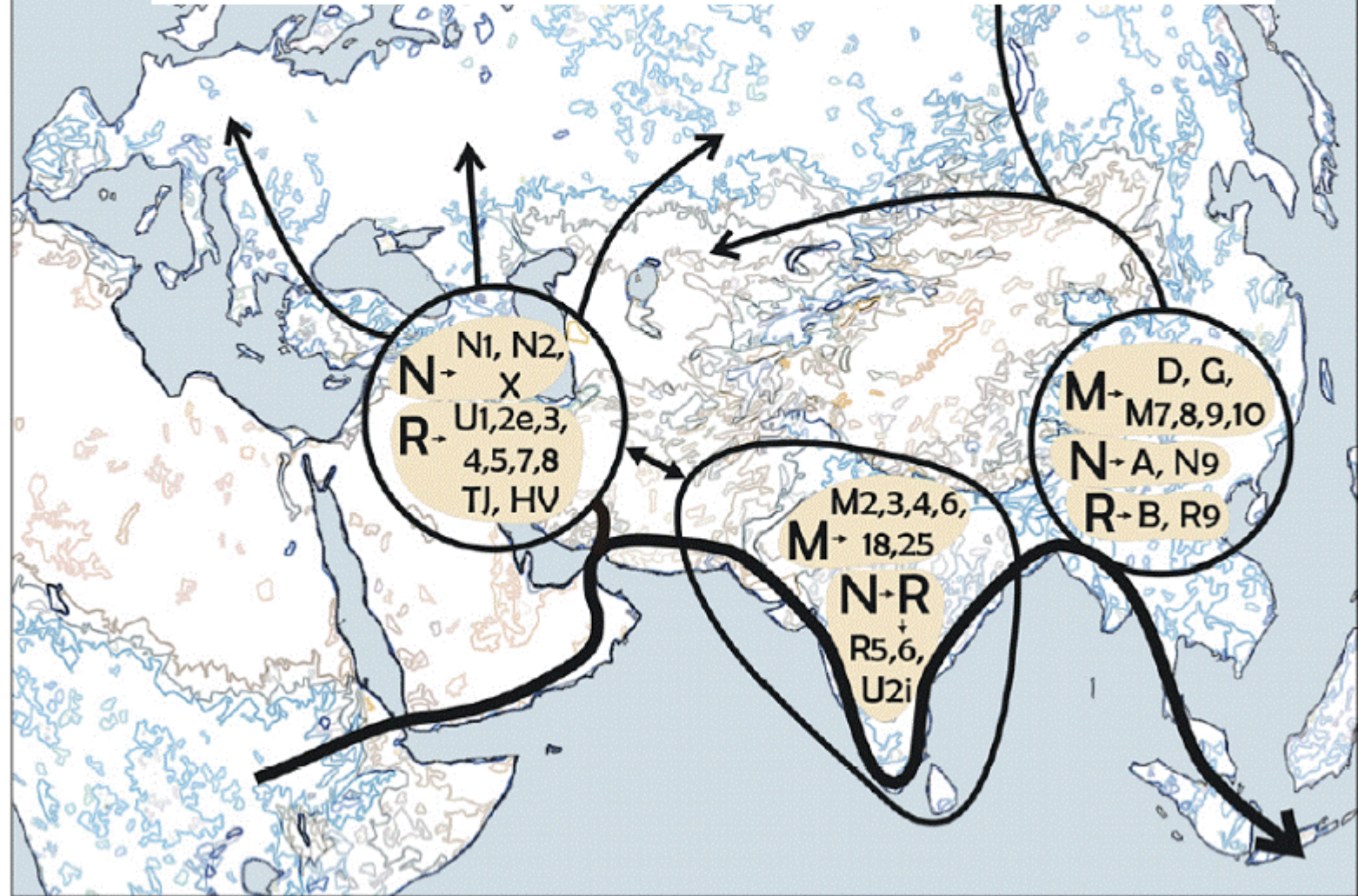
- Maternally Inherited
- No recombination
- Higher mutation rate than autosomes
- HVS I (and II) sequencing + RFLP/SNP typing allows unambiguous classification into haplogroups

# Africa



# Most of the extant mtDNA boundaries in South and Southwest Asia were likely shaped during the initial settlement of Eurasia by anatomically modern humans

Mait Metspalu<sup>\*1</sup>, Toomas Kivisild<sup>1</sup>, Ene Metspalu<sup>1</sup>, Jüri Parik<sup>1</sup>,  
Georgi Hudjashov<sup>1</sup>, Katrin Kaldma<sup>1</sup>, Piia Serk<sup>1</sup>, Monika Karmin<sup>1</sup>,  
Doron M Behar<sup>2</sup>, M Thomas P Gilbert<sup>6</sup>, Phillip Endicott<sup>7</sup>, Sarabjit Mastana<sup>4</sup>,  
Surinder S Papiha<sup>5</sup>, Karl Skorecki<sup>2</sup>, Antonio Torroni<sup>3</sup> and Richard Villems<sup>1</sup>



# North Africa

- Inhabited as far as 40 kya
- Ibero-maurusian industry 22-9 kya
- Berbers are thought to be the aboriginal North African
- Neolithic time: spread of farming alongside the Northern and Southern shore of Mediterranean sea
- Muslim ruling

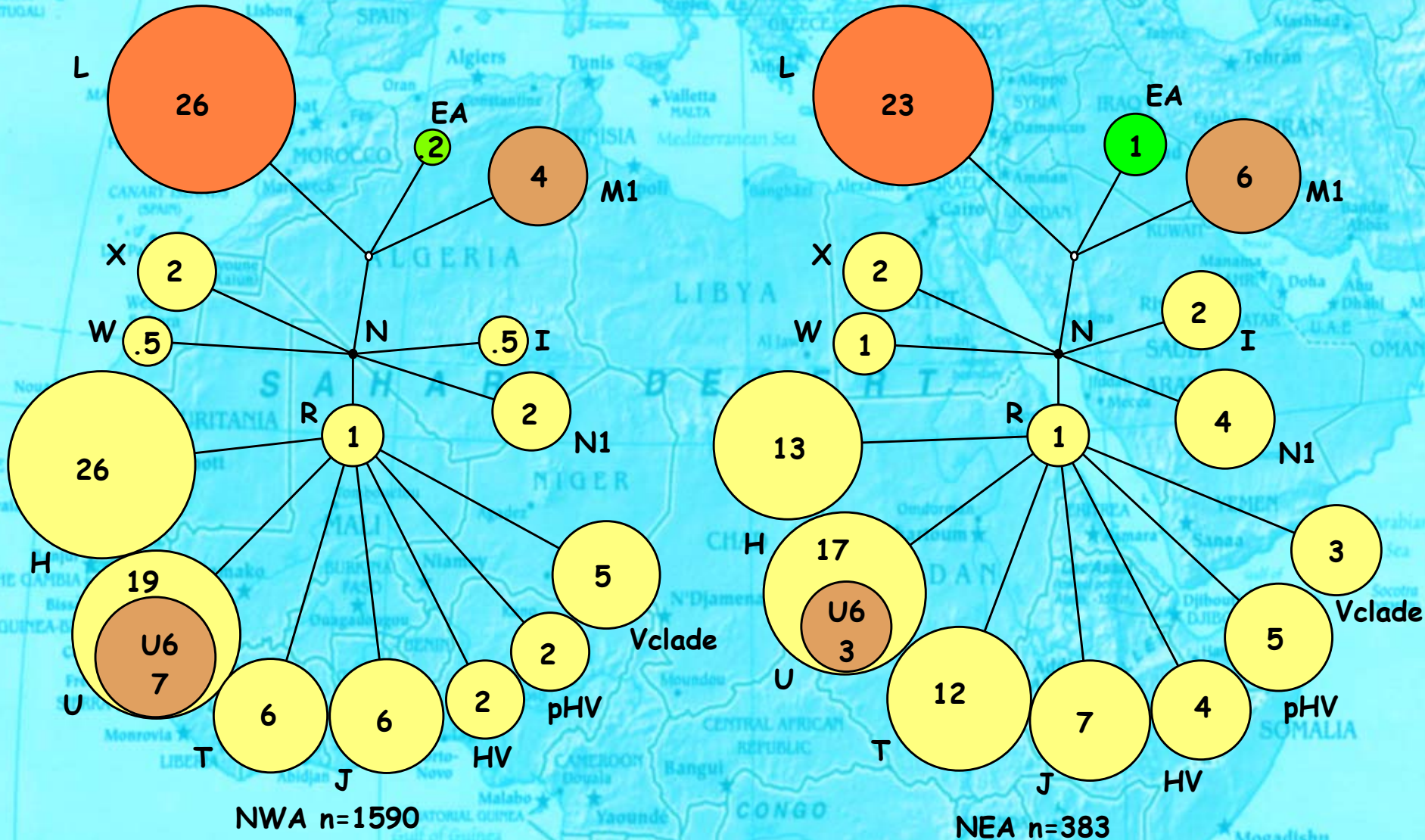
# Populations studied

- 24 different populations
- Total number: 5107 DNAs
- Coverage of the different regions of interest
  - West Africa (Waf)
  - NW Africa (NWaf)
  - NE Africa (NEAf)
  - E Africa (EAf)
  - Near East (NE/ME)
  - "North Mediterranean" (NM)

# Populations studied

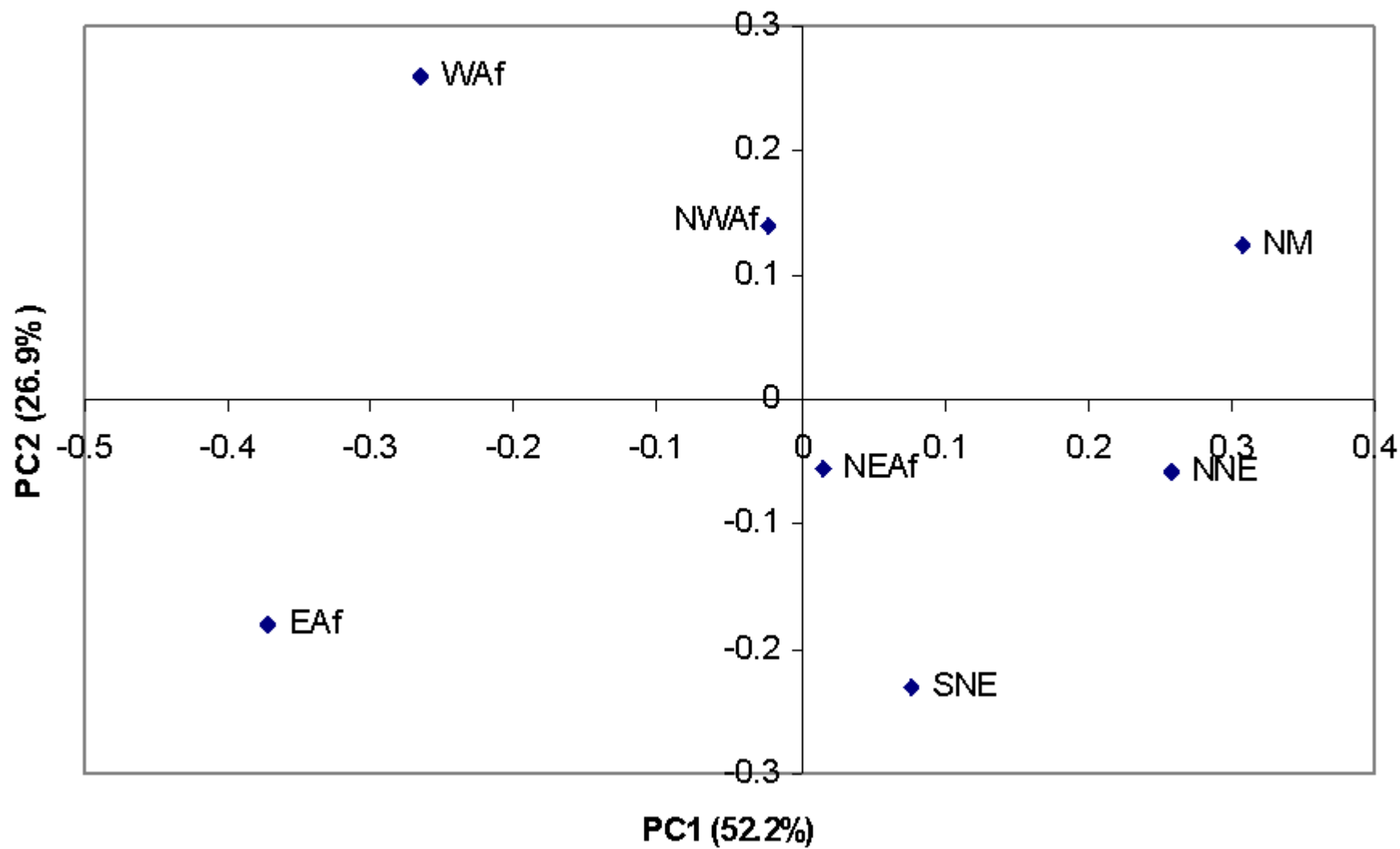


# Frequencies in NWA & NEA

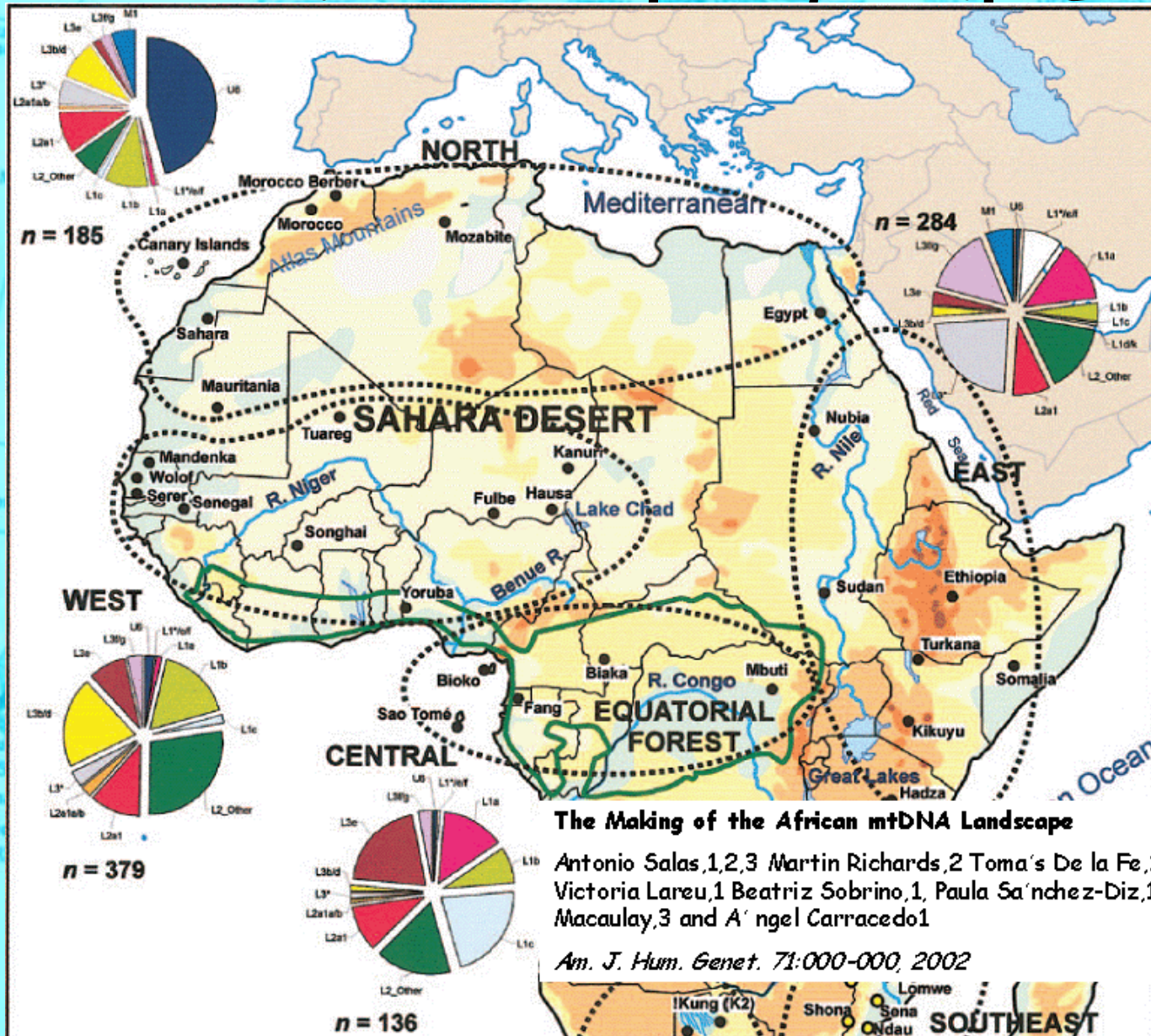




# NW\_NEsim8



# Macrohaplogroup L



## The Making of the African mtDNA Landscape

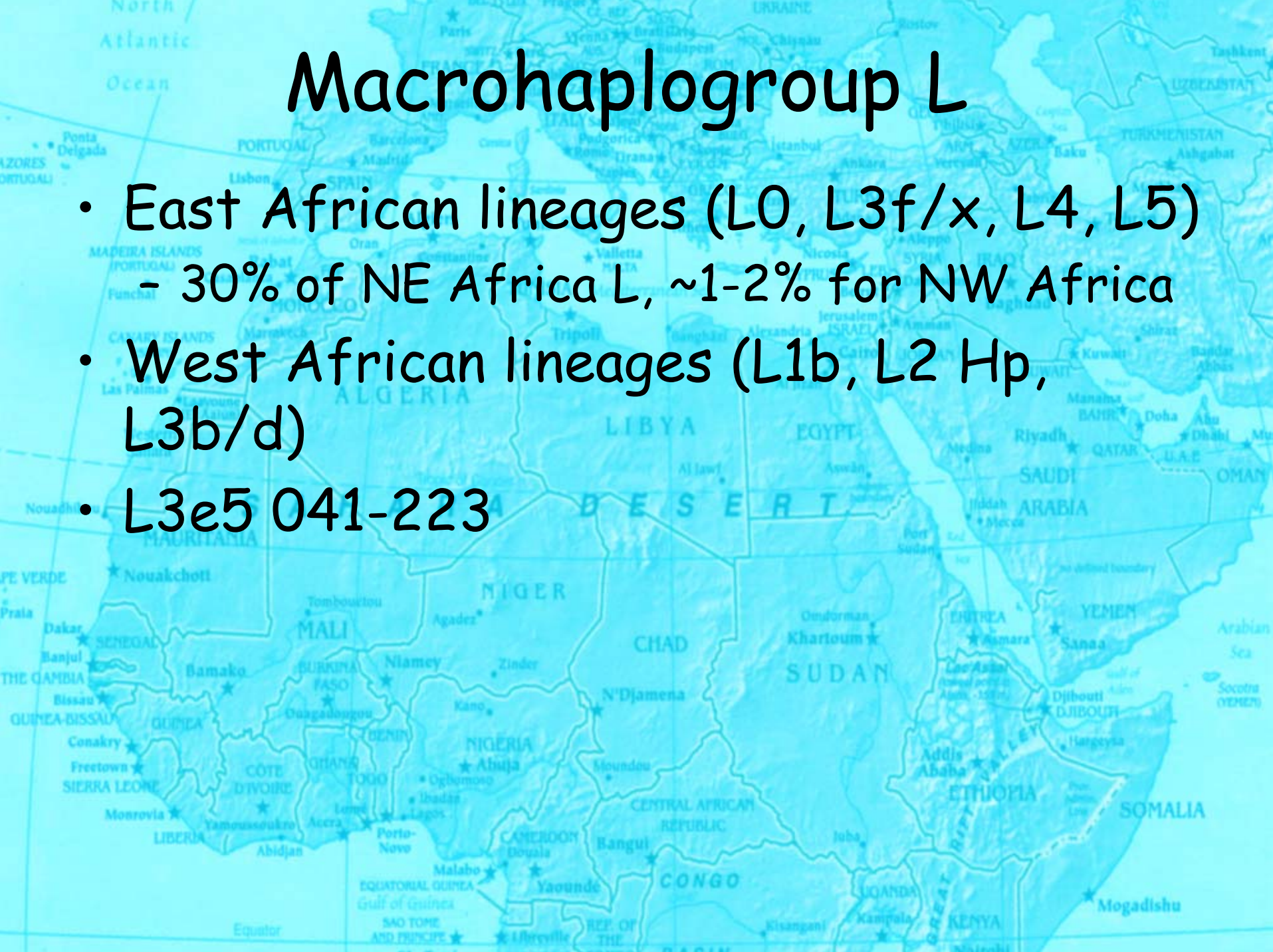
Antonio Salas,1,2,3 Martin Richards,2 Toma's De la Fe,1 Mari'a-Victoria Lareu,1 Beatriz Sobrino,1, Paula Sa'nchez-Diz,1 Vincent Macaulay,3 and A'ngel Carracedo1

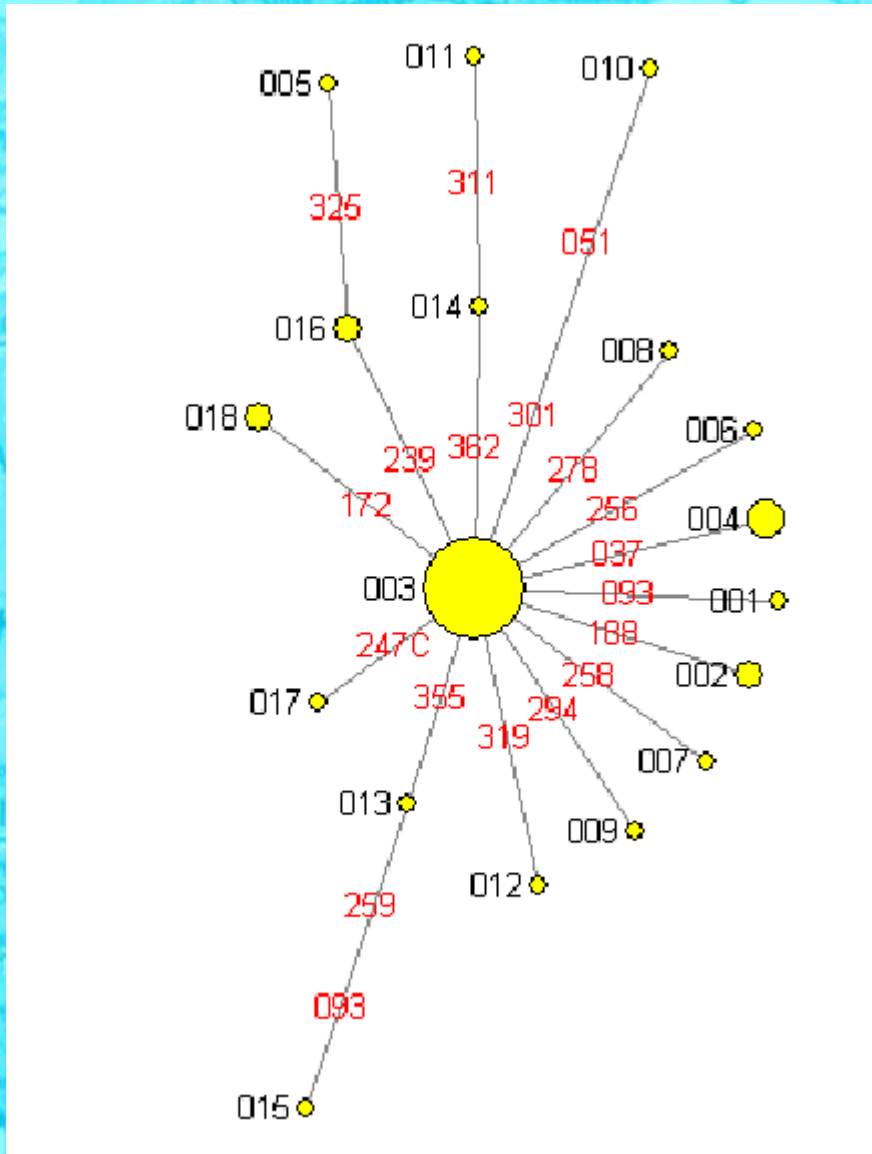
*Am. J. Hum. Genet.* 71:000-000, 2002



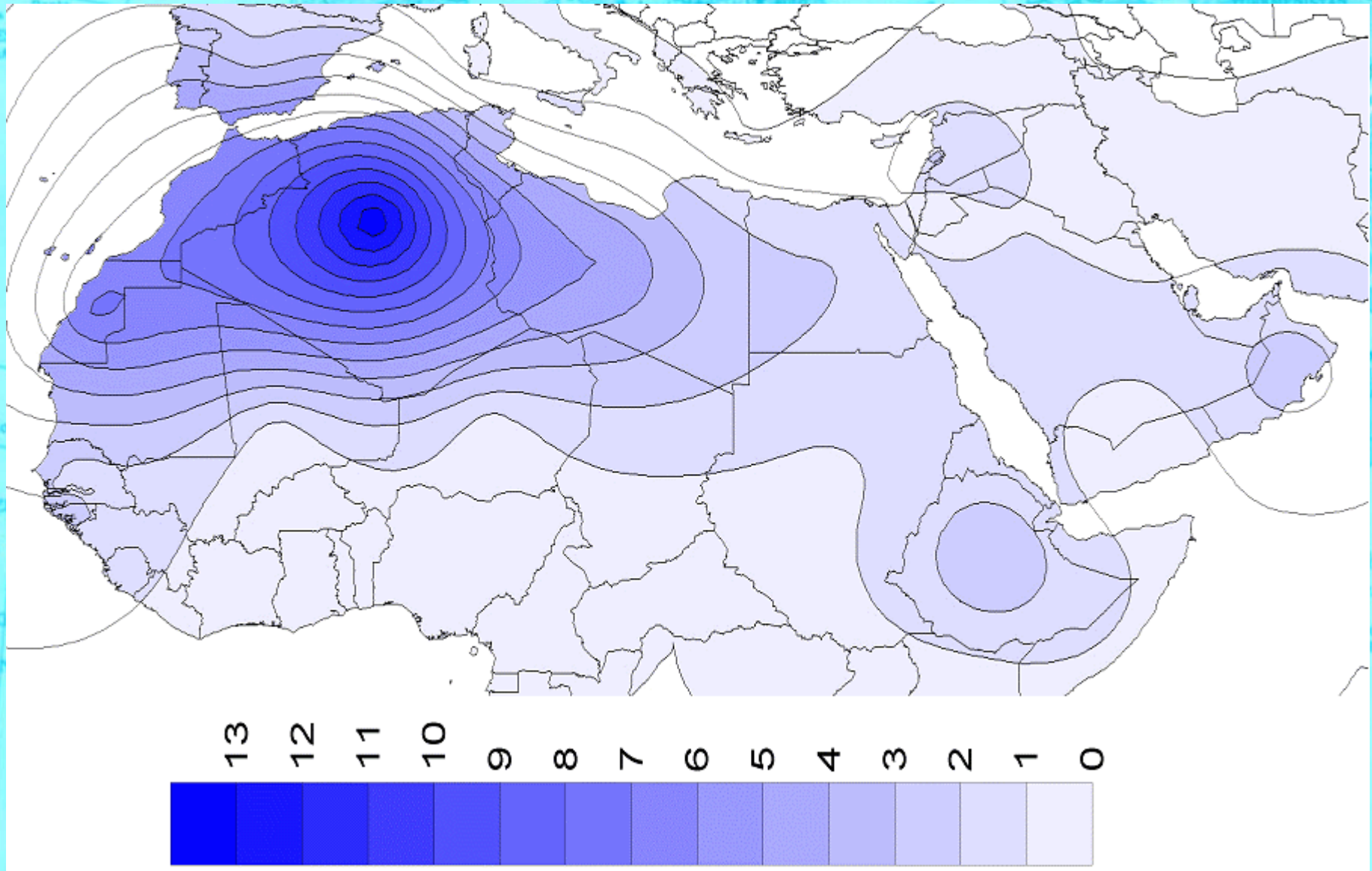
# Macrohaplogroup L

- East African lineages (L0, L3f/x, L4, L5)
  - 30% of NE Africa L, ~1-2% for NW Africa
- West African lineages (L1b, L2 Hp, L3b/d)
- L3e5 041-223

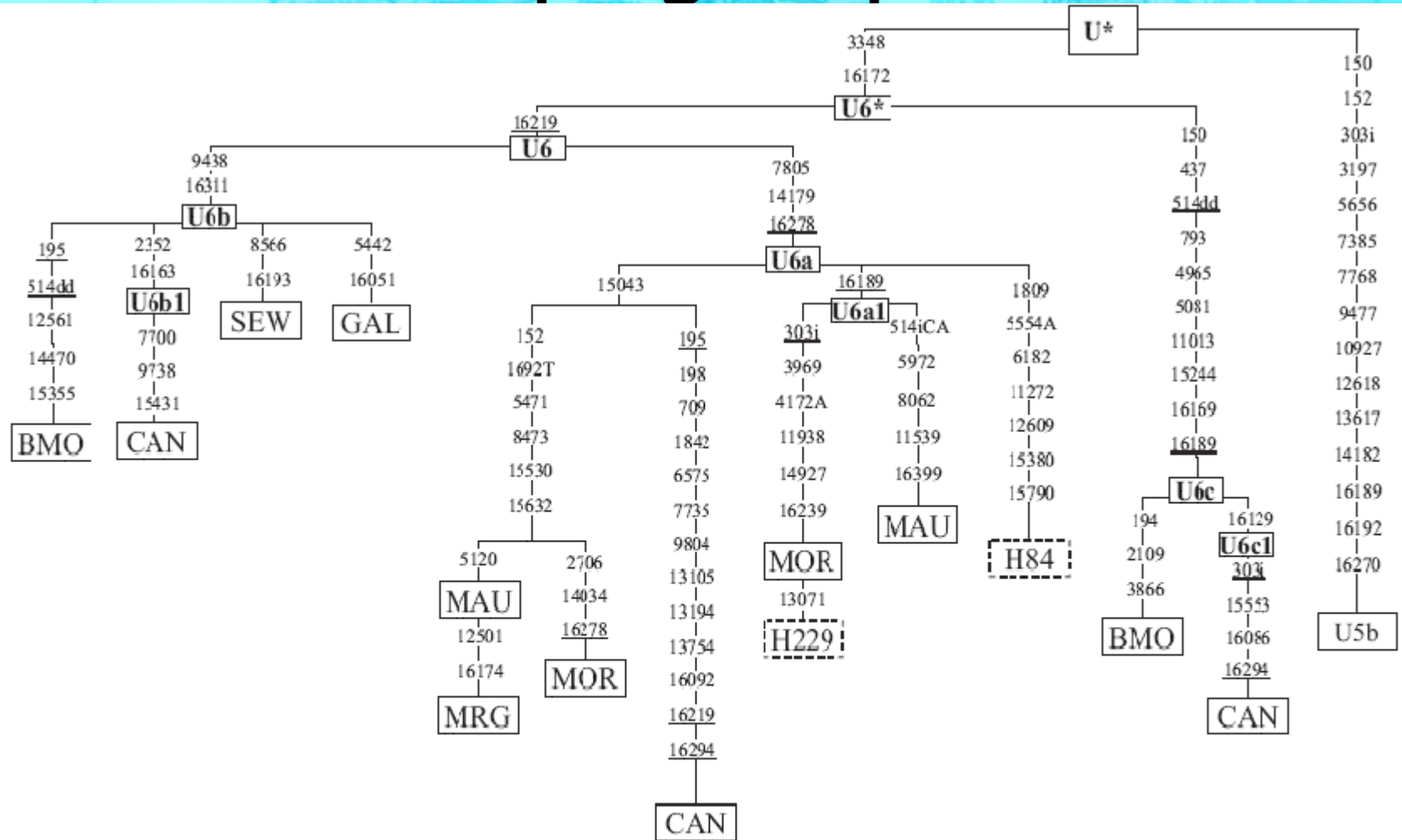




# Haplogroup U6



# Haplogroup U6

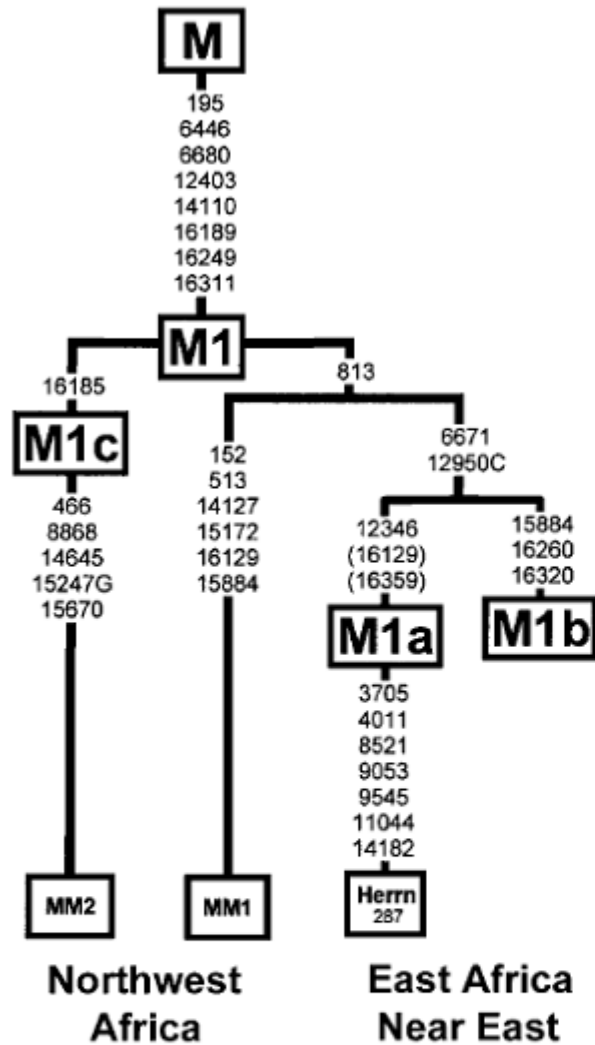


## Mitochondrial DNA transit between West Asia and North Africa inferred from U6 phylogeography

Nicole Maca-Meyer<sup>1</sup>, Ana M González<sup>1</sup>, José Pestano<sup>2</sup>, Carlos Flores<sup>1</sup>, José M Larruga<sup>1</sup> and Vicente M Cabrera\*<sup>1</sup>

*BMC Genetics* 2003, **4**:15

# Haplogroup M1



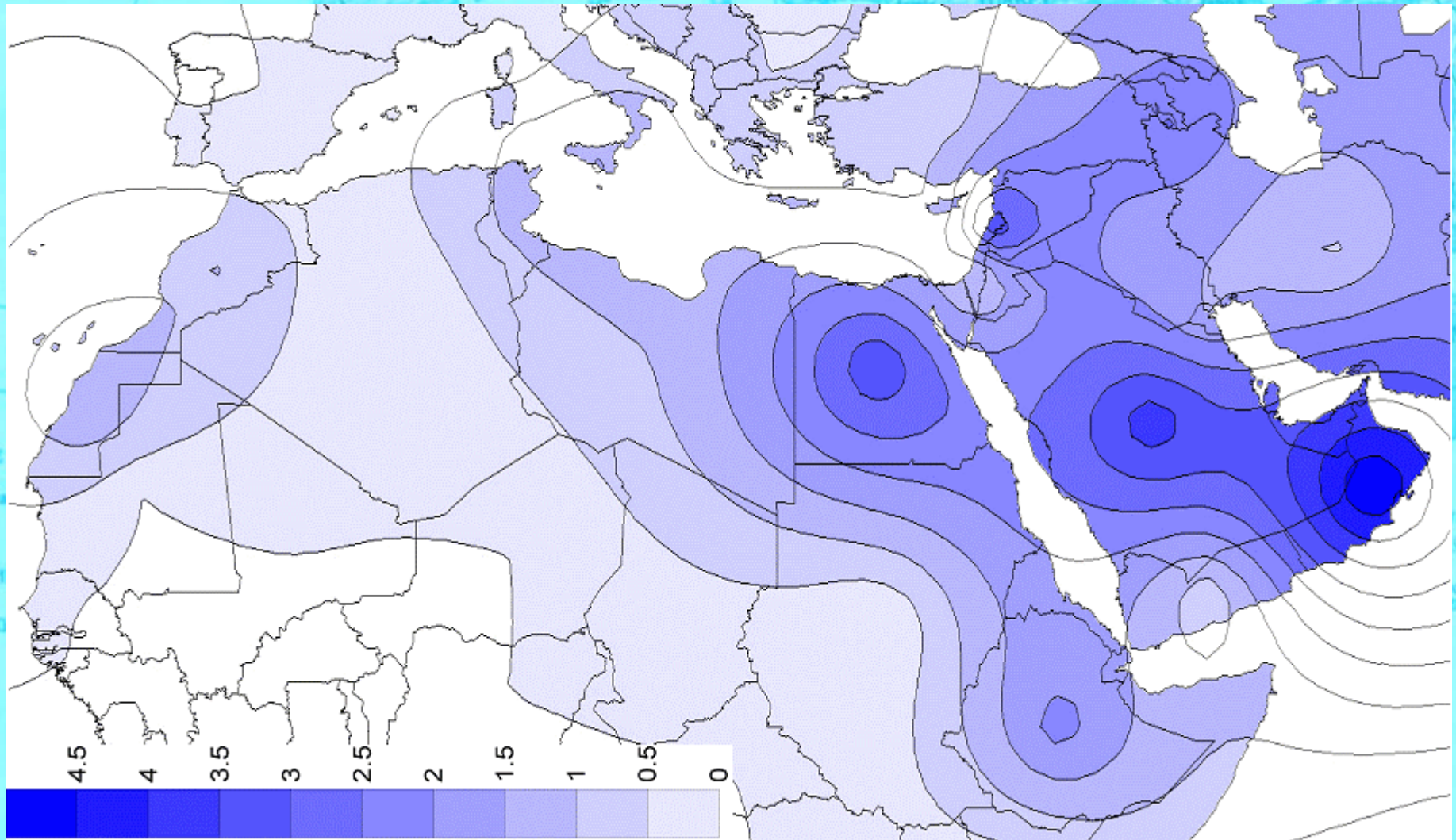
## Ethiopian Mitochondrial DNA Heritage: Tracking Gene Flow Across and Around the Gate of Tears

Toomas Kivisild,<sup>1</sup> Maere Reidla,<sup>1</sup> Ene Metspalu,<sup>1</sup> Alexandra Rosa,<sup>1</sup> Antonio Brehm,<sup>2</sup> Erwan Pennarun,<sup>1</sup> Ju`ri Parik,<sup>1</sup> Tarekegn Geberhiwot,<sup>3</sup> Esien Usanga,<sup>4</sup> and Richard Villems<sup>1</sup>

*Am. J. Hum. Genet.* 75:752-770, 2004

# Middle East haplogroups

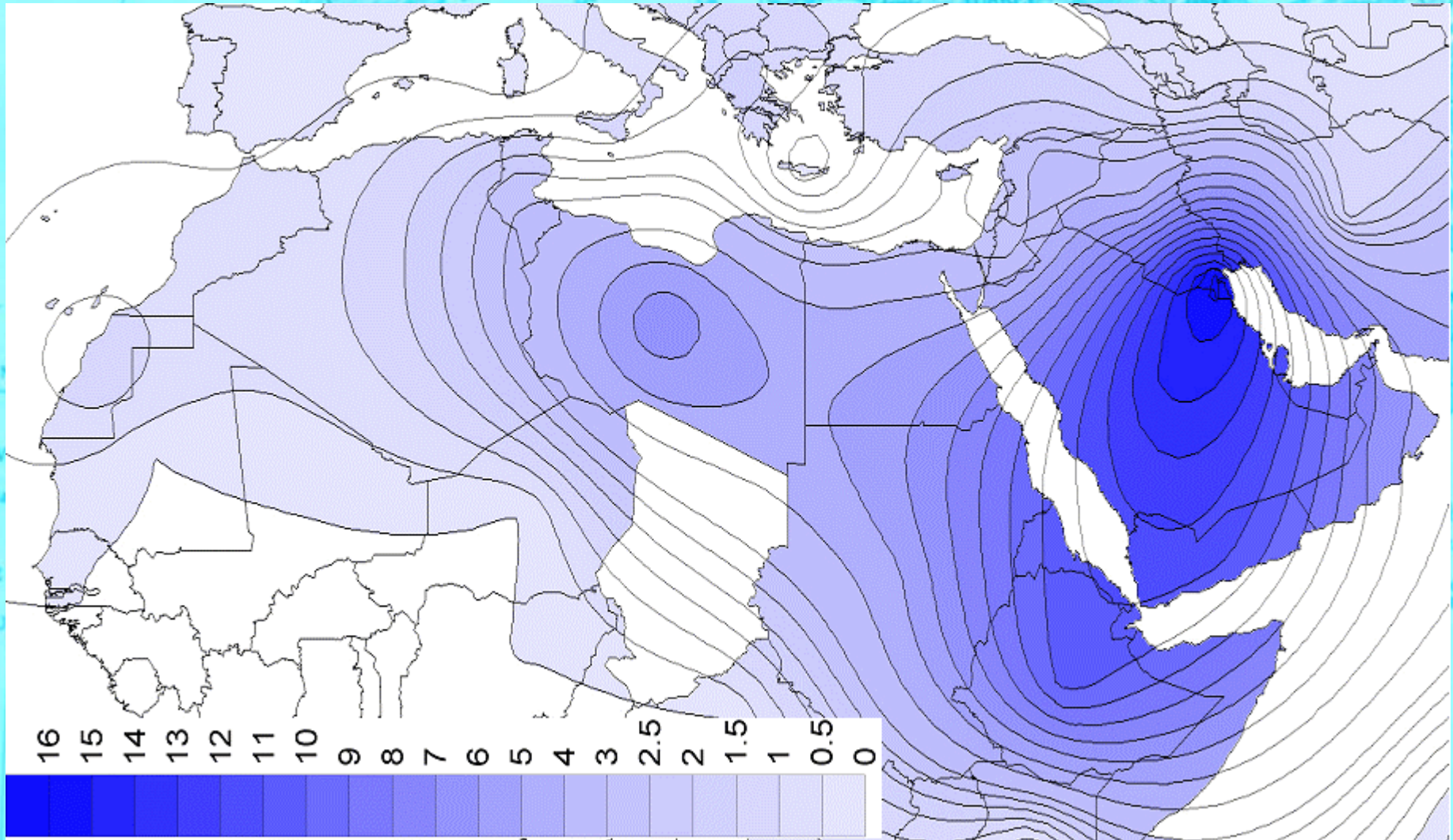
- (pre-HV)1





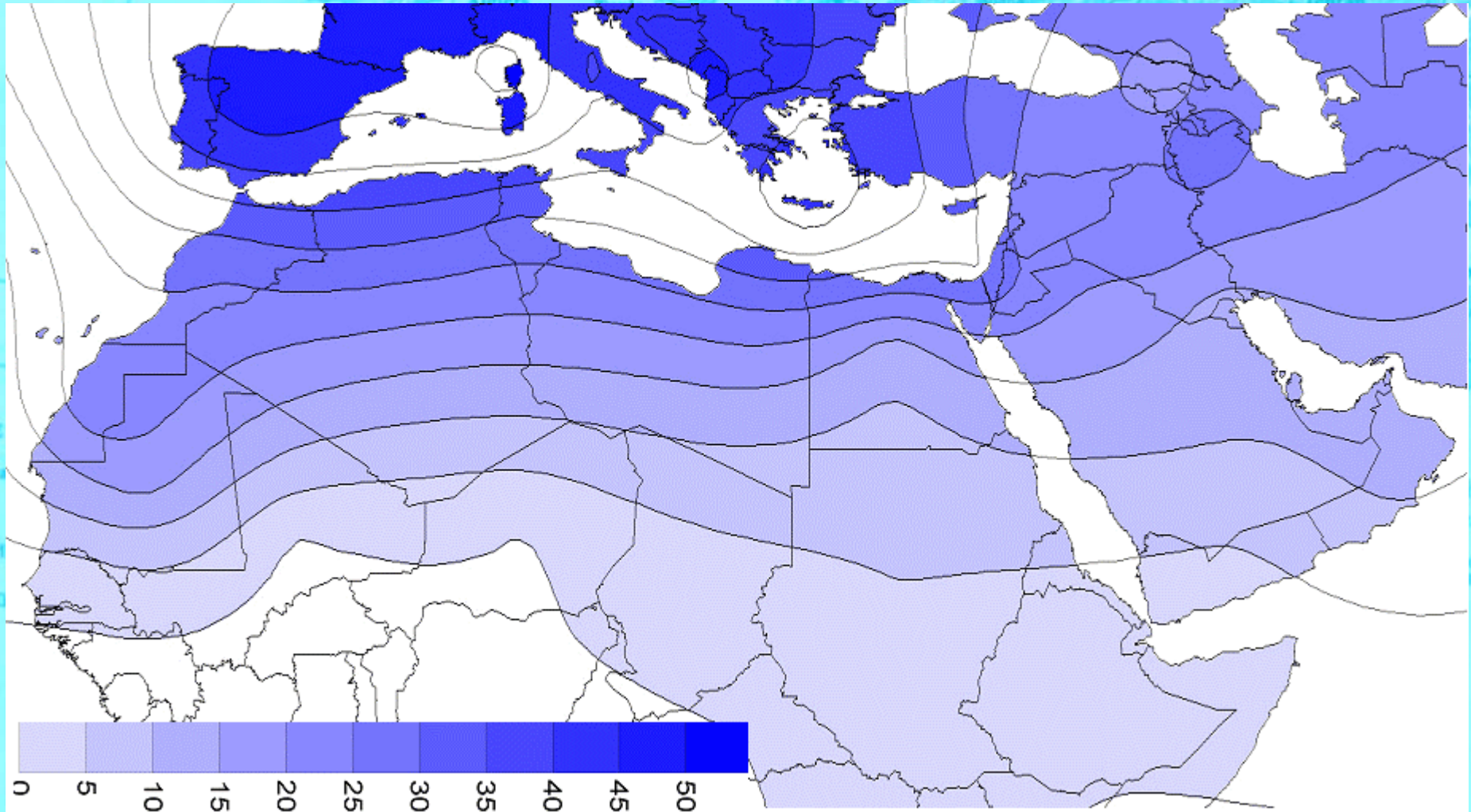
# Middle East haplogroups

- N1b



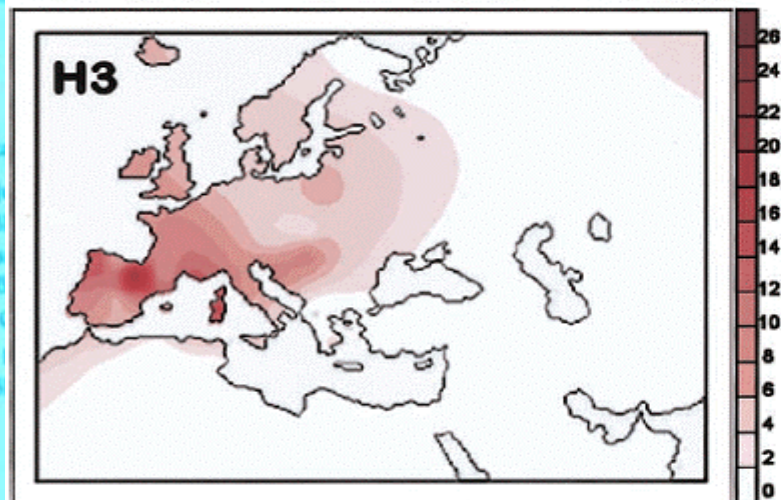
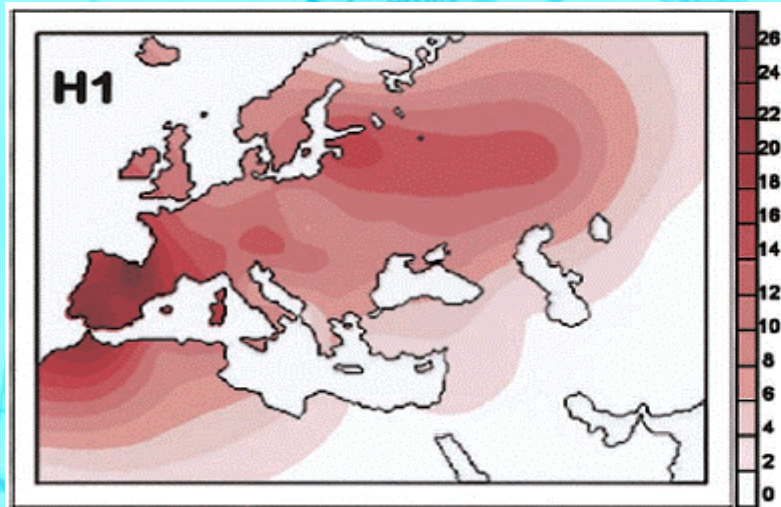
# West Eurasian haplogroups

- Haplogroup H



# West Eurasian haplogroups

- Haplogroup H



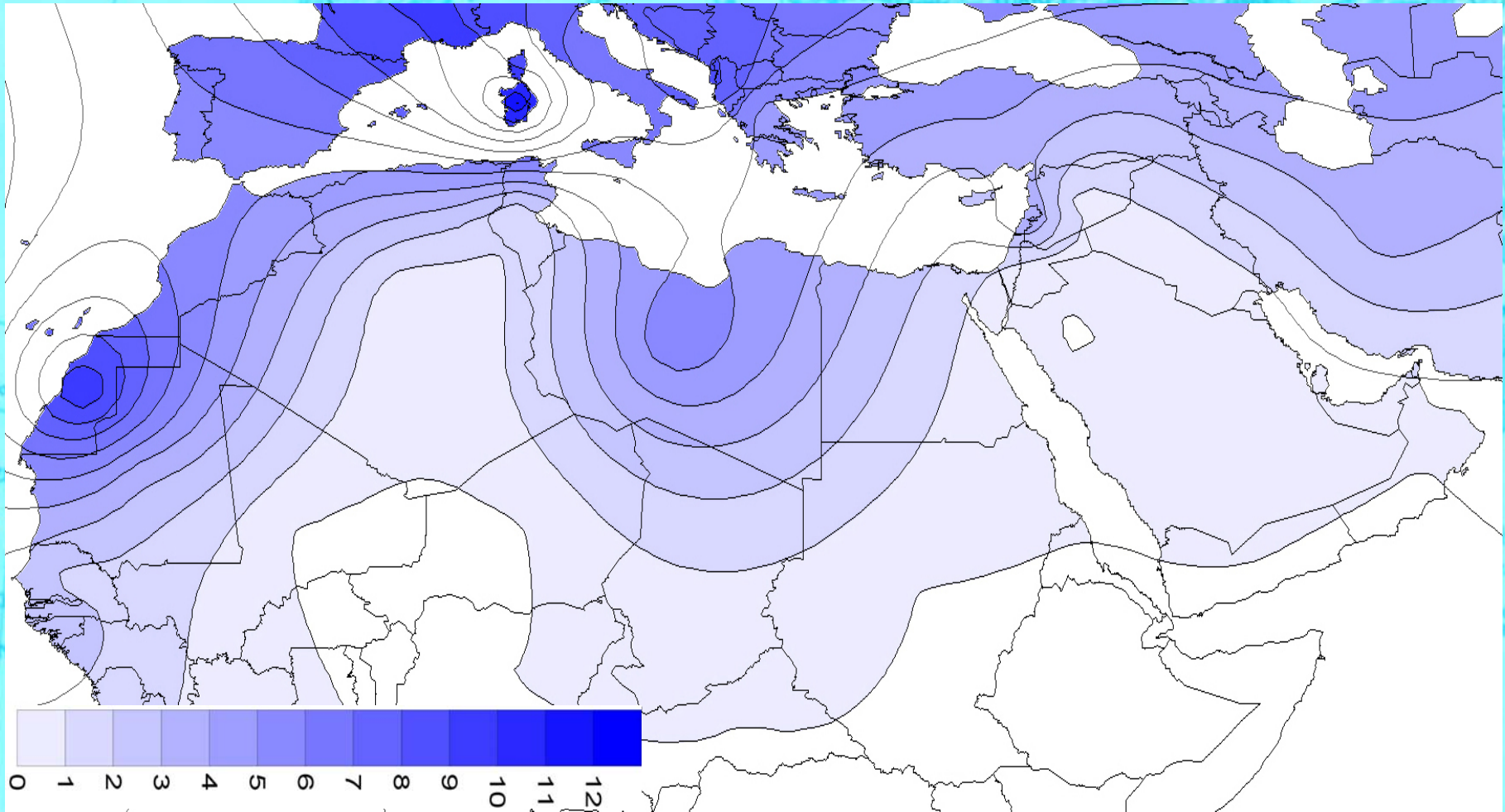
**The Molecular Dissection of mtDNA Haplogroup H Confirms That the Franco-Cantabrian Glacial Refuge Was a Major Source for the European Gene Pool**

*Achilli et al.*

*Am. J. Hum. Genet.* 75:910-918, 2004

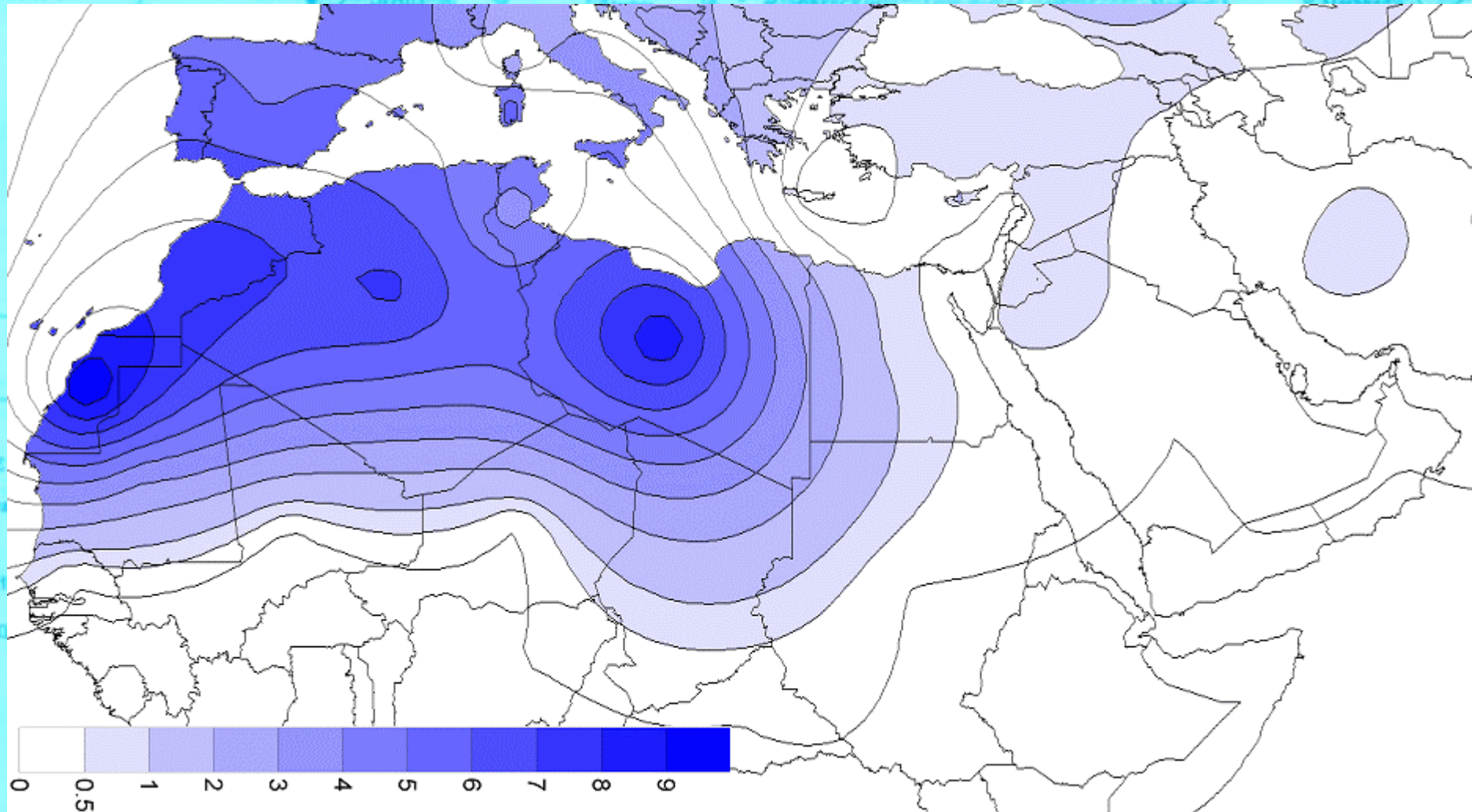
# West Eurasian haplogroups

- Haplogroup U5



# West Eurasian haplogroups

- V clade



# Conclusions

- NWA Vs. NEA:
  - Contrast in haplogroups frequencies
  - Contrast in lineages composition
- → Common ground but different influences during pre-/History

