# Language and Genes of the Greater Himalayan Region

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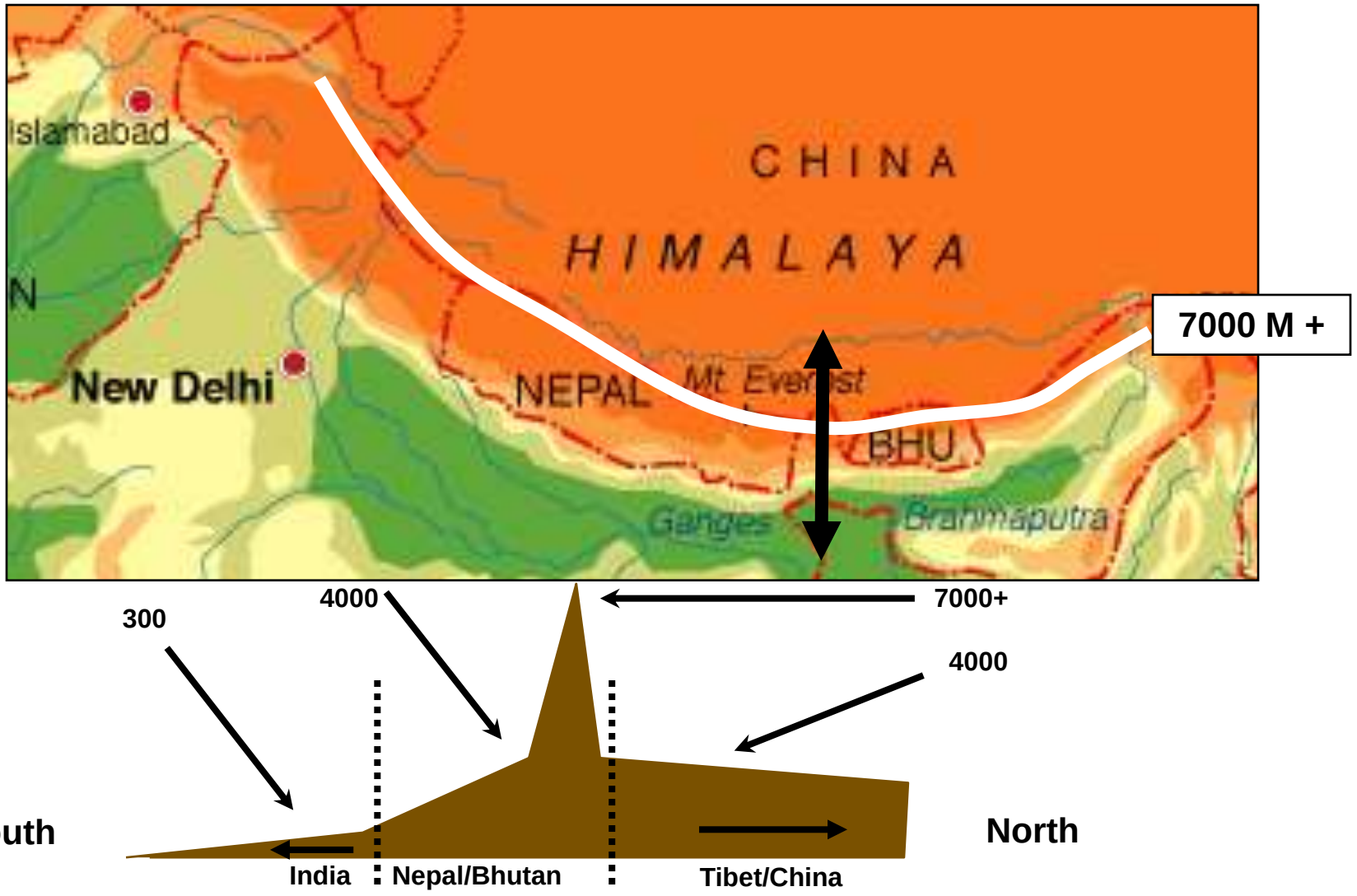
# Outline

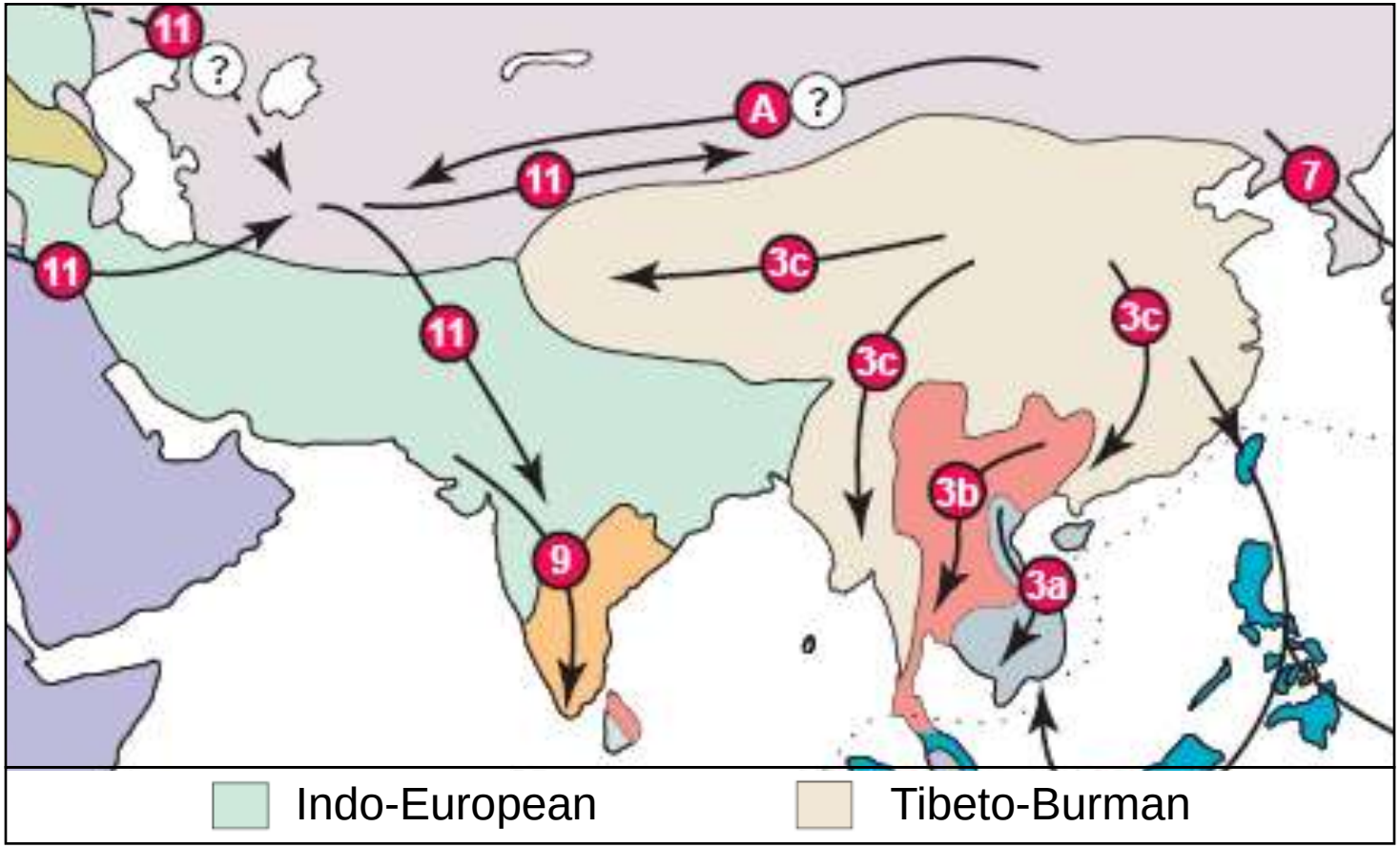
- Why the Himalayan region?
- The project
- Location of genetic boundaries
- The peopling of the area
- Patterning of diversity within Bhutan

# The geographical barrier is obvious

# The Himalaya

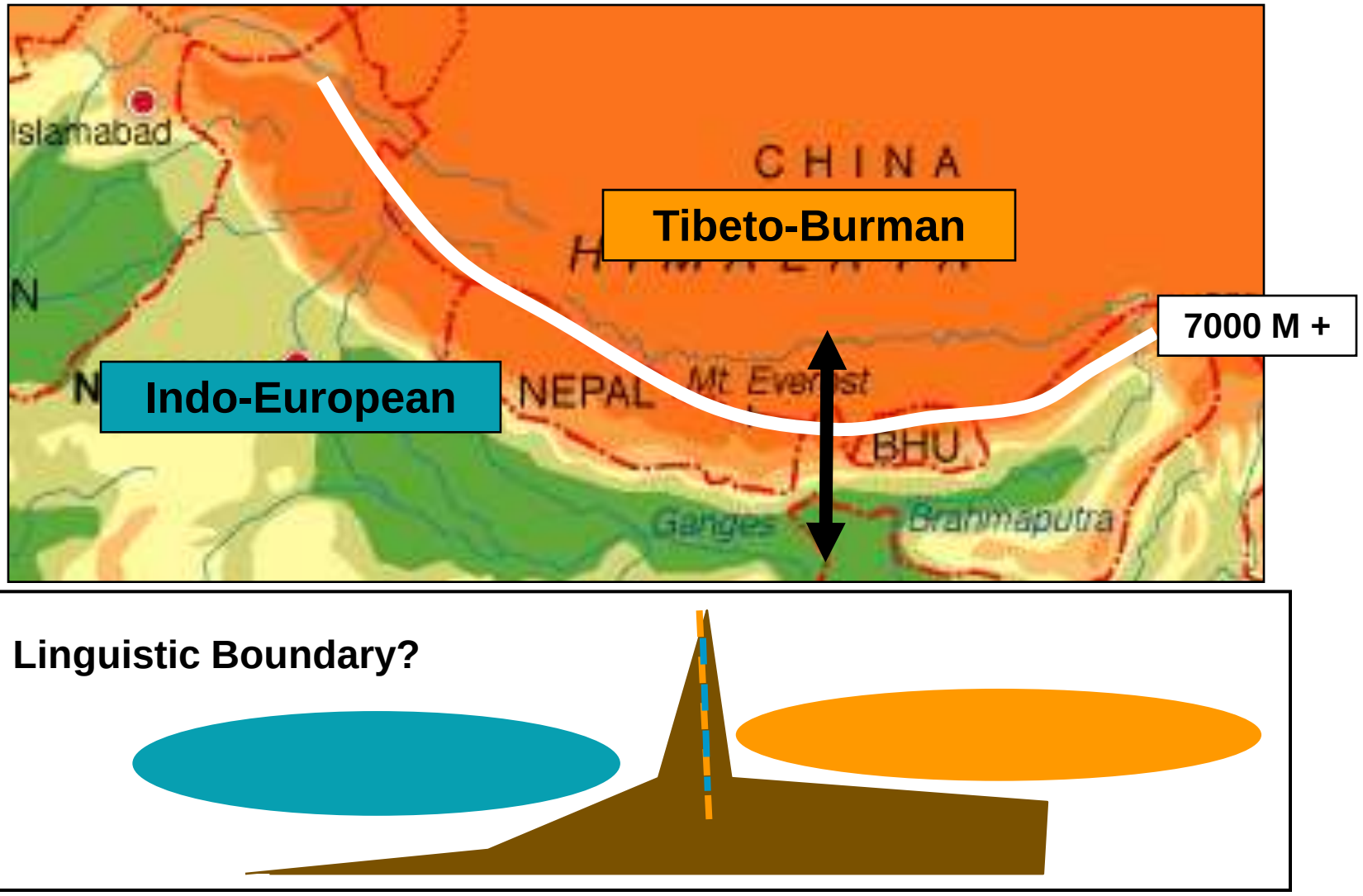


# There is also a linguistic boundary …

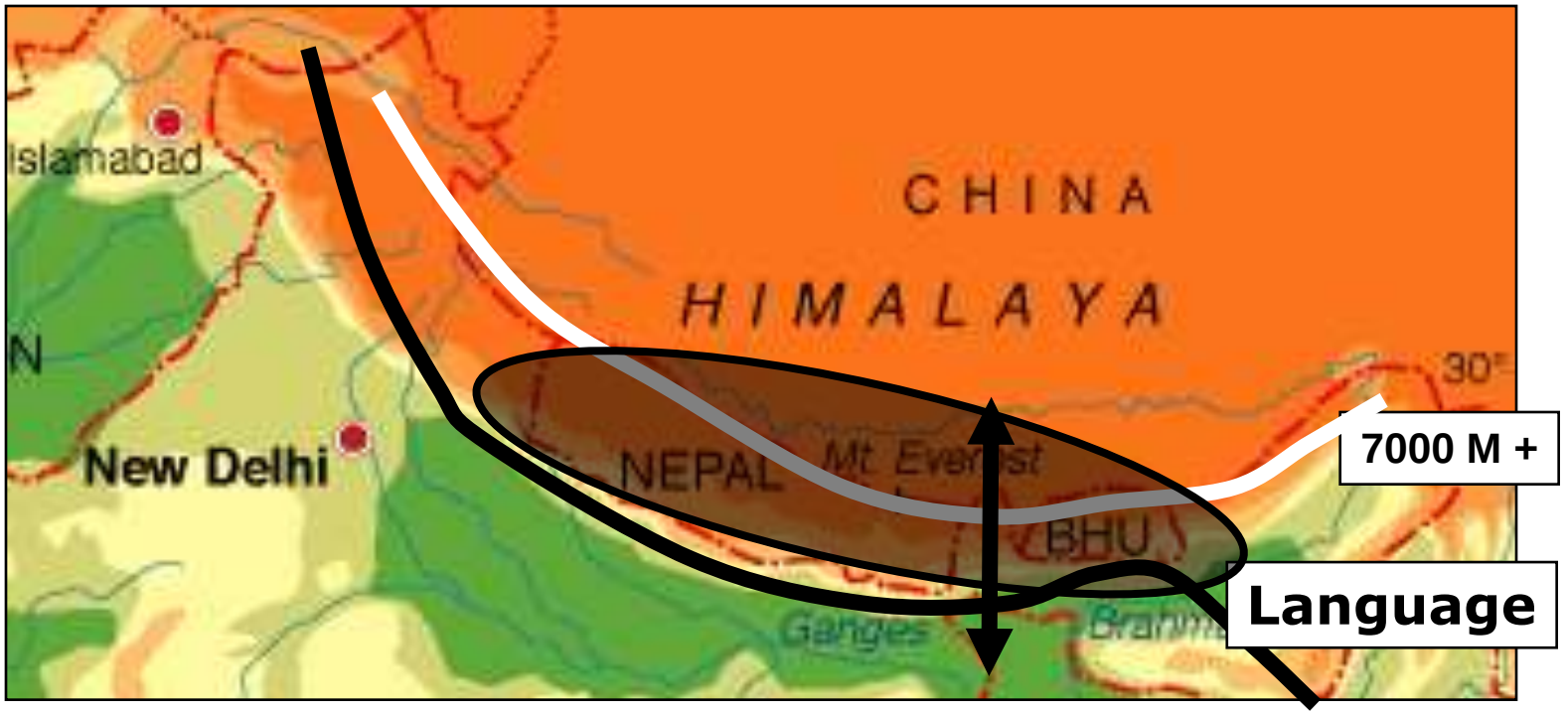


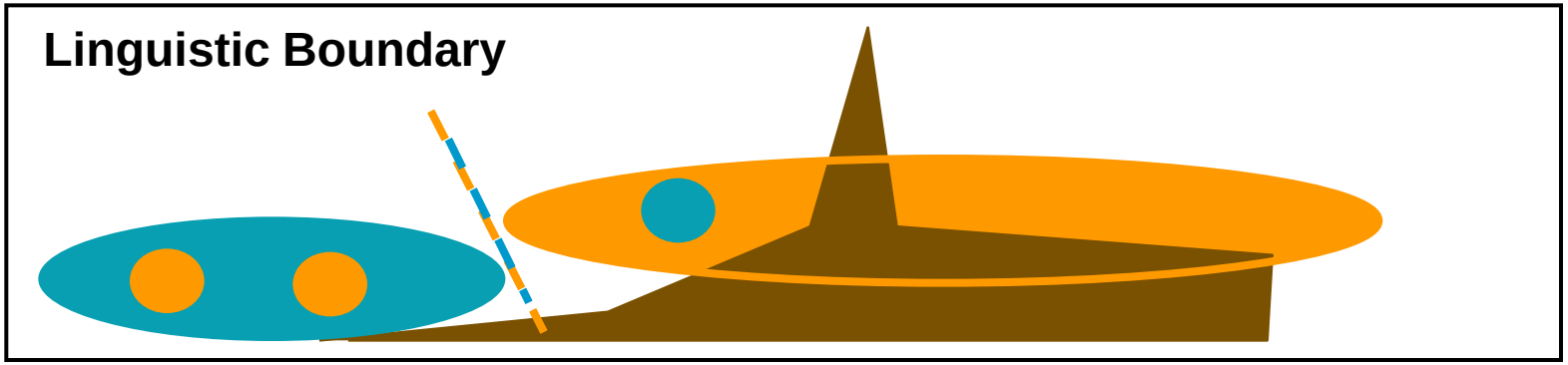

Diamond and Bellwood (2003) *Science* **300**, 597-603

# Where exactly is it?



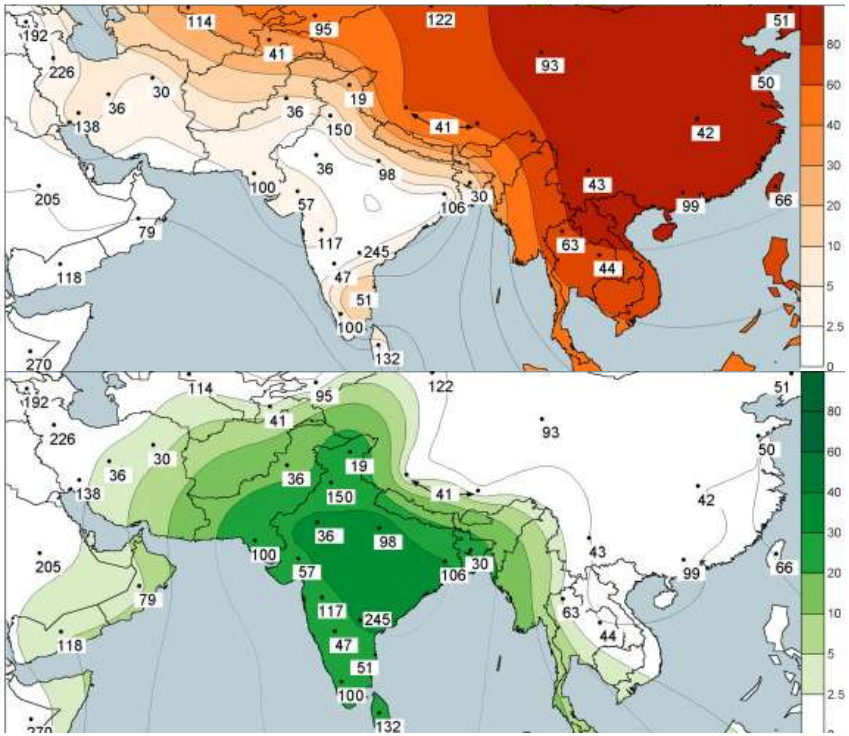
# Actually like this…



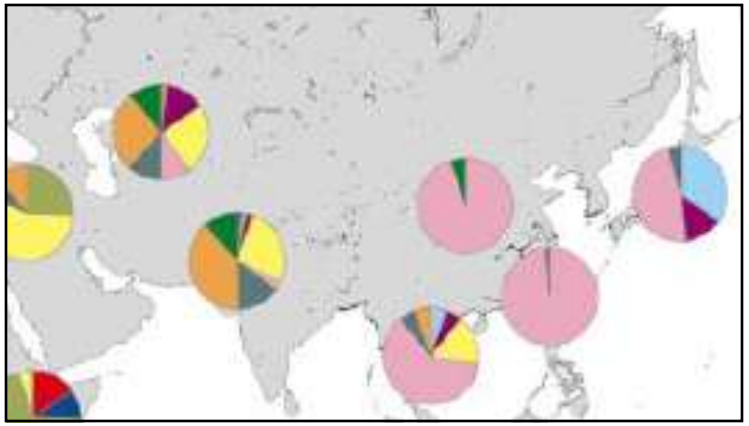
# Previous evidence for a genetic boundary

mtDNA



Metspalu *et al*. (2004) *BMC Genetics* **5**, 26

Y chromosome



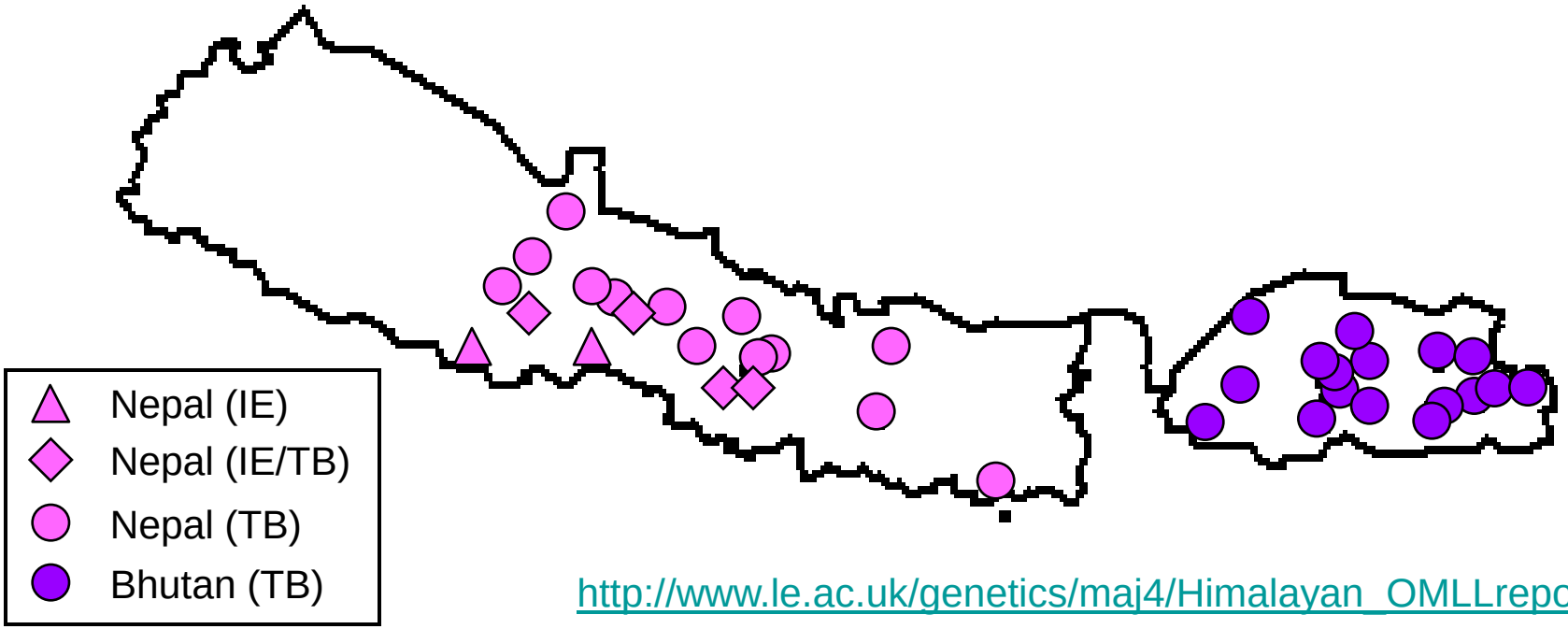
Underhill *et al*. (2001) *Ann. Hum. Genet*. **65**, 43-62

# Questions

- How precisely do the geographical, linguistic and genetic boundaries correspond?
- What can we say about the origins of the people now living in the Himalayan region?
- How has the extreme landscape patterned diversity at the local level? Is language important?

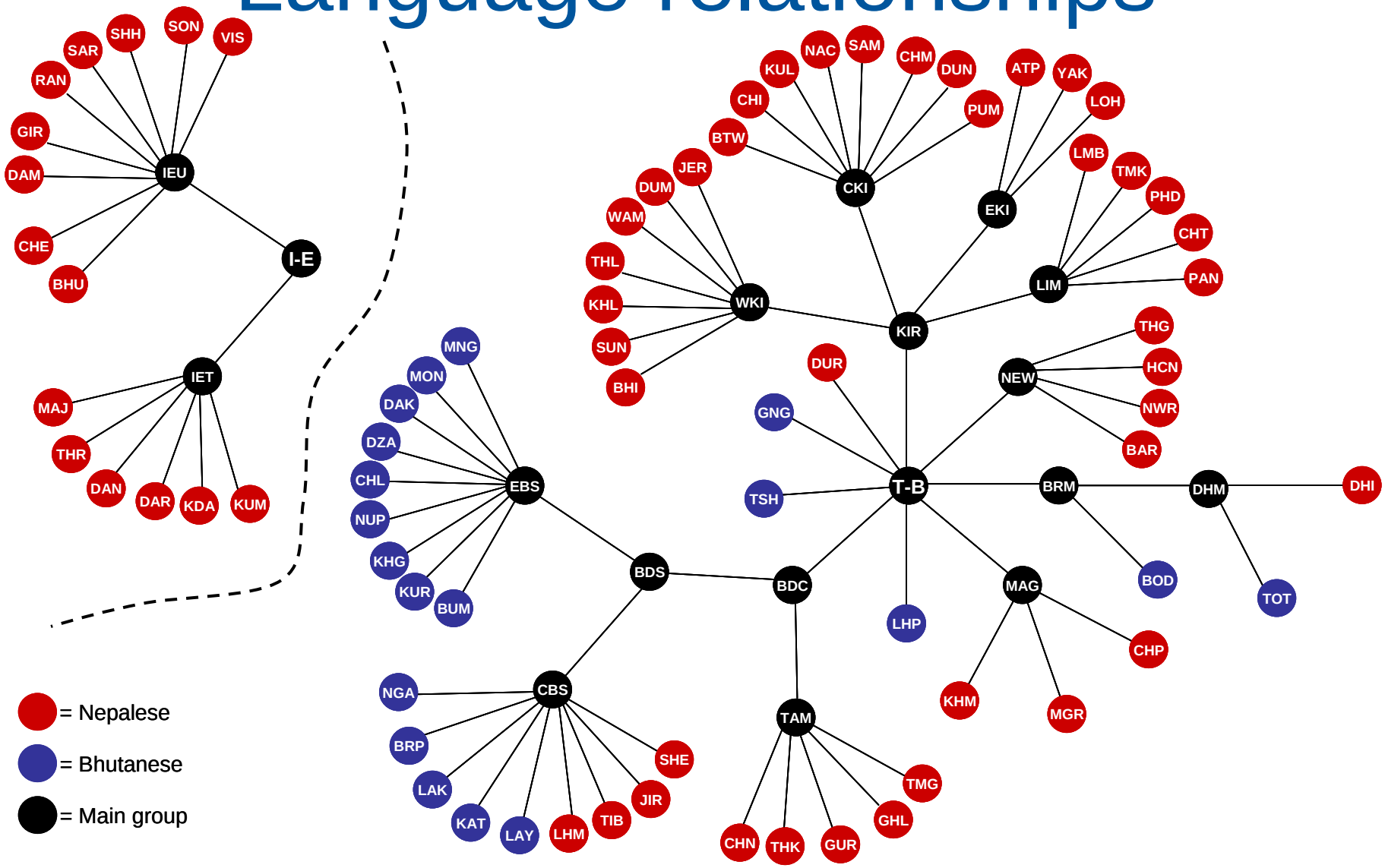
# Samples collected (2002-4)

- Nepal: 955 (764 males), 19 ethnolinguistic groups
- Bhutan: 941 (839 males) 17 ethnolinguistic groups
- Plus data from the literature on 5000-10,000 individuals from neighbouring populations

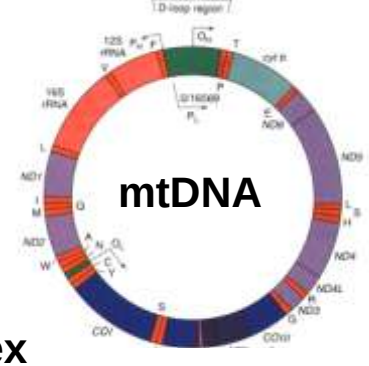


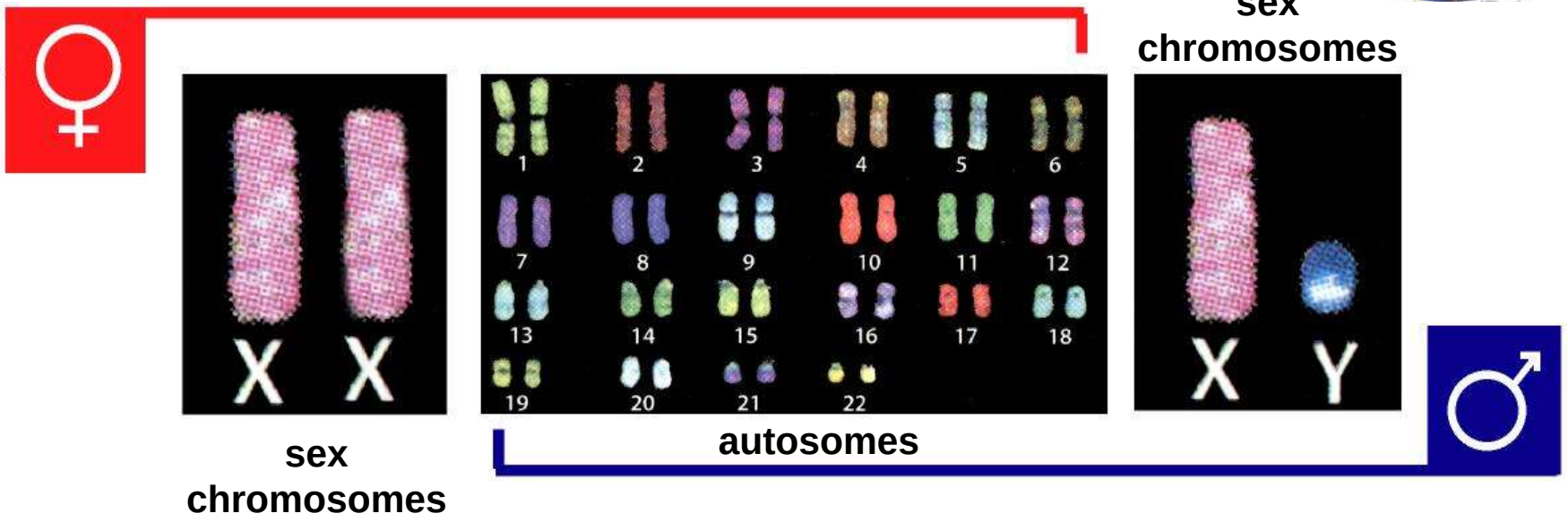

http://www.le.ac.uk/genetics/maj4/Himalayan_OMLLreport.pdf

# Language relationships



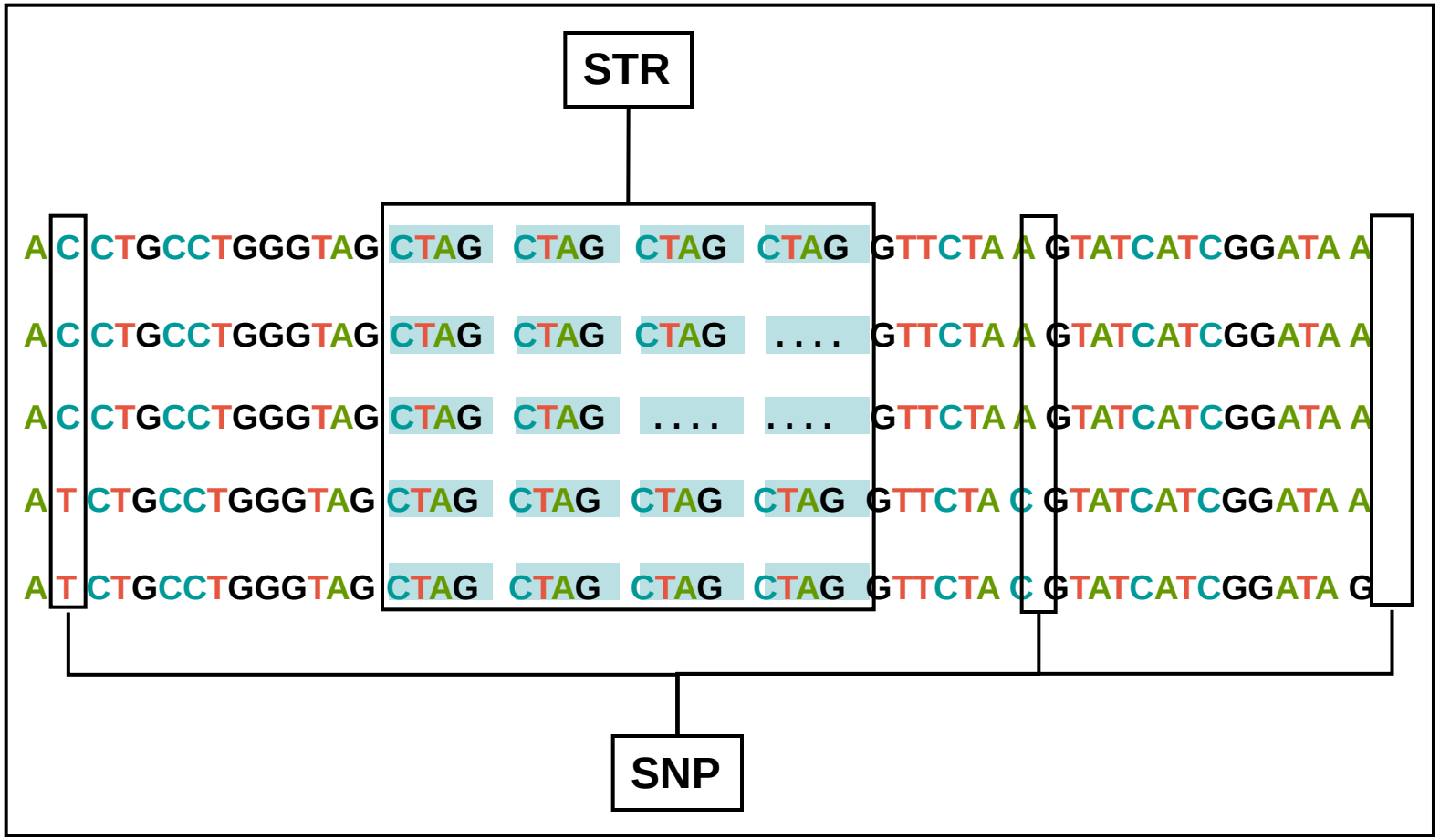
# Inheritance




- Y chromosome sex-determining - father to son
- Both sexes have mtDNA, but inherited only from mothers to children
- Autosomes inherited from both parents

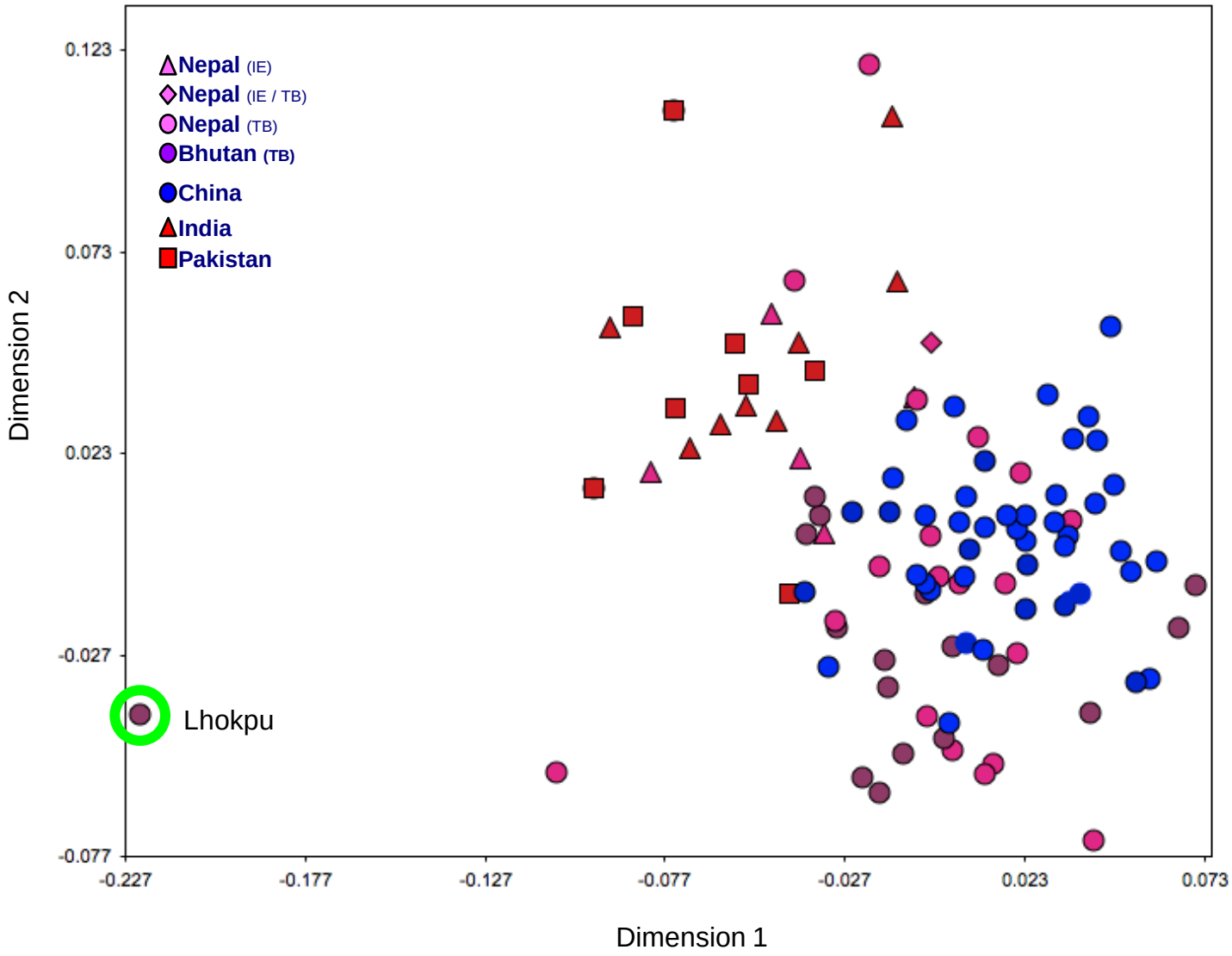
# STRs and SNPs



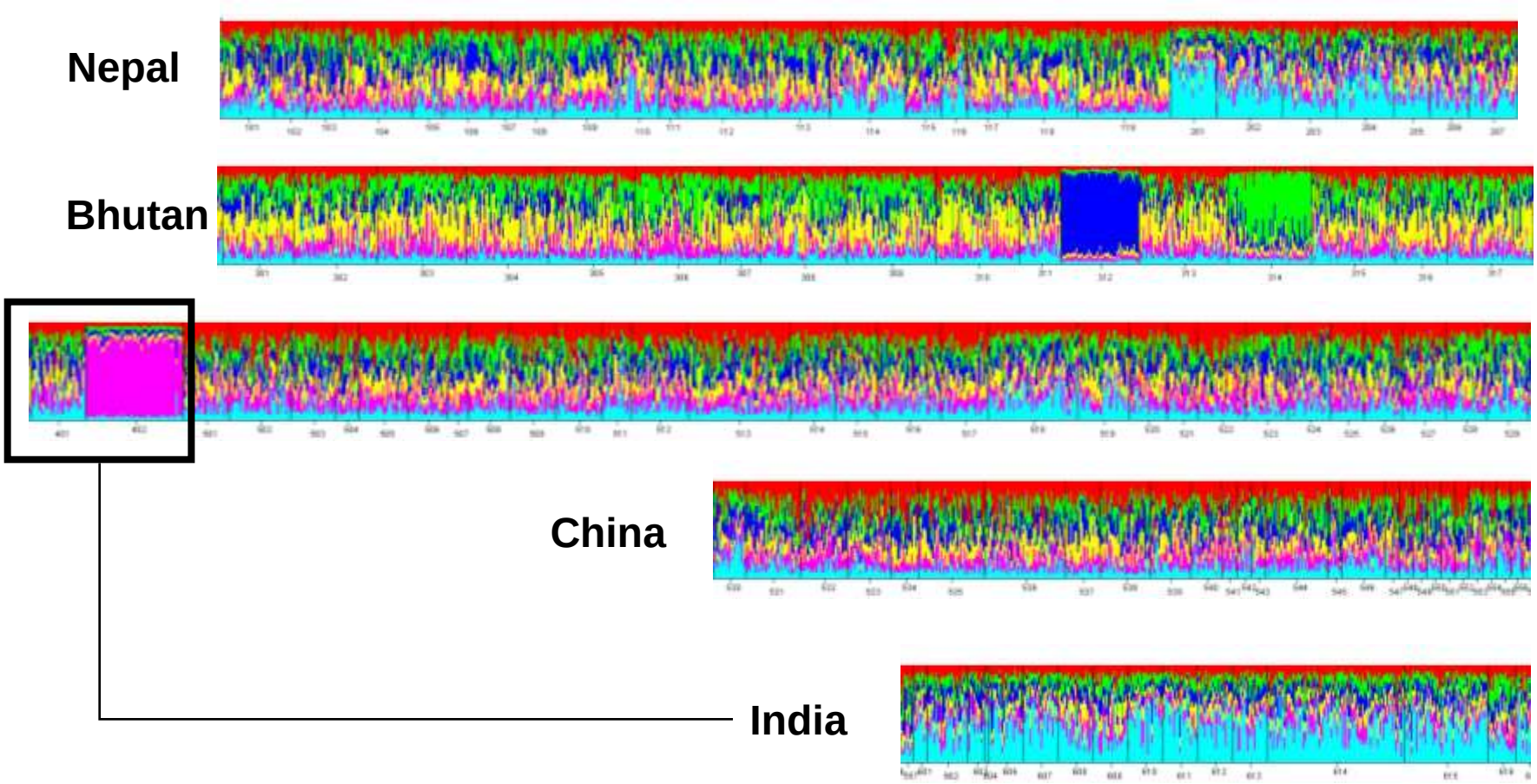
# Markers typed

- Autosomal
  - 21 forensic STRs
  - 13 high-differentiation SNPs
- Y-chromosomal
  - 26 Y-STRs
  - 60 Y-SNPs in hierarchical mode
- mtDNA
  - HVSI
  - 20 coding-region SNPs

# 15 autosomal STRs - MDS

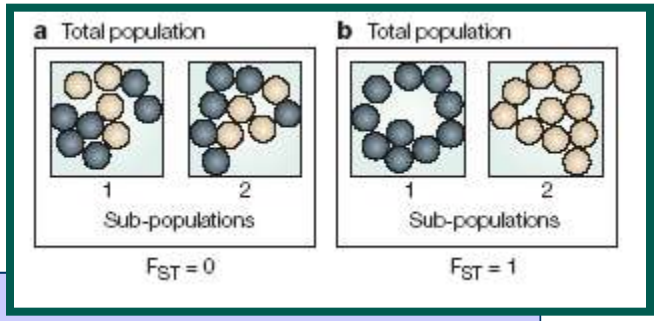


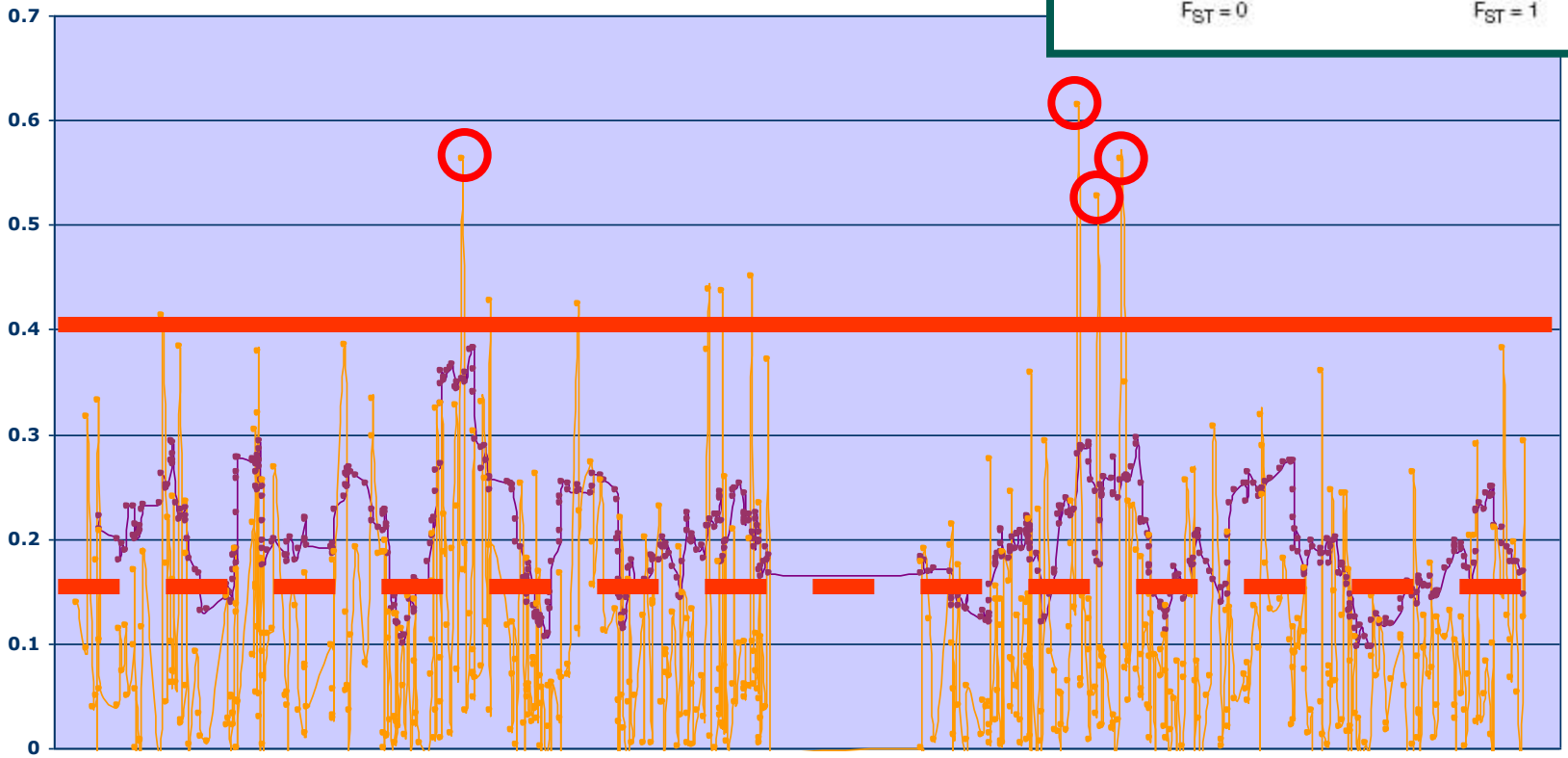
# STRUCTURE: 15 autosomal STRs



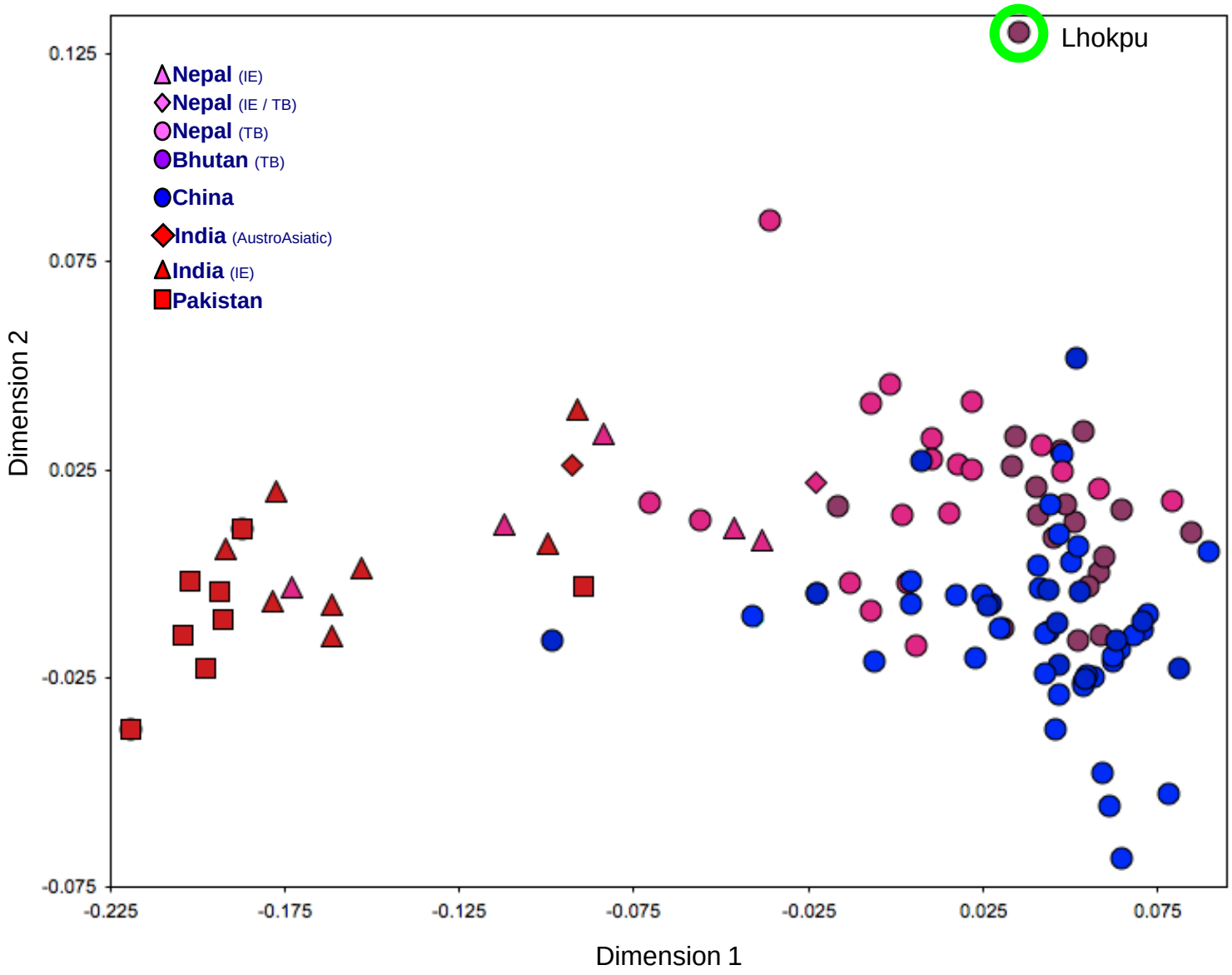
# Identifying informative autosomal SNPs

- $F_{ST}$ between Chinese and Pakistani samples in CEPH-HGDP panel

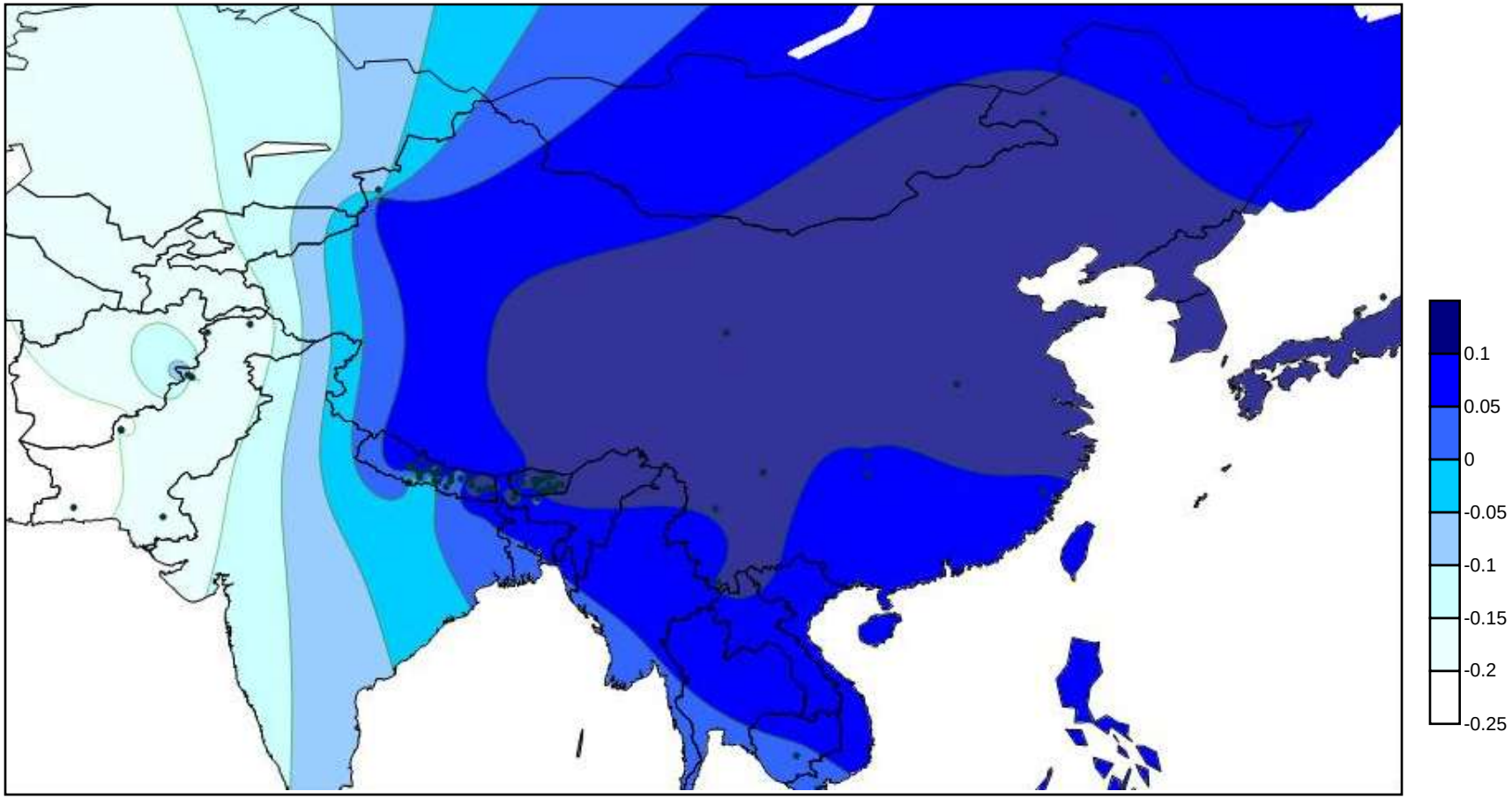


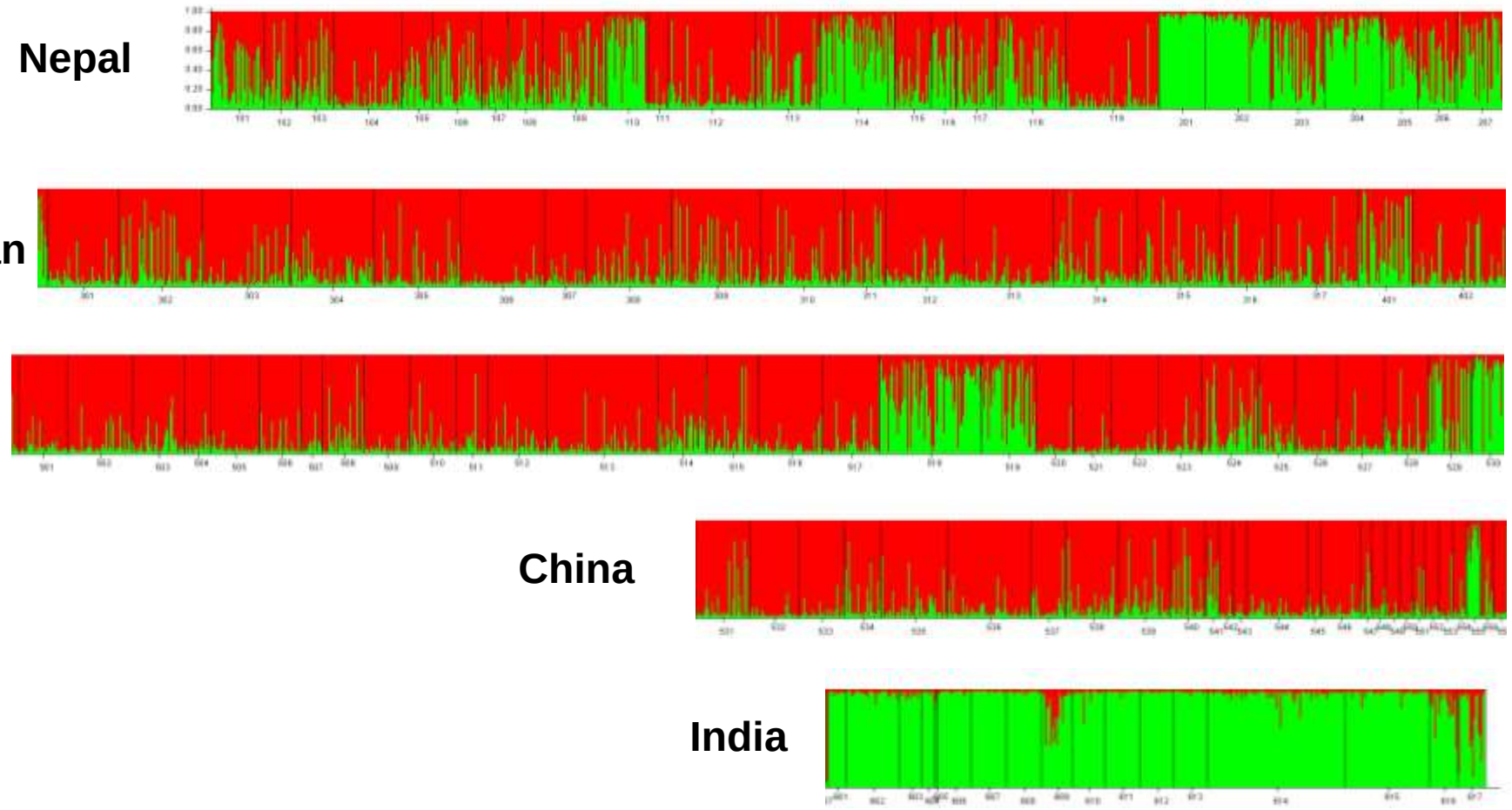
# 13 autosomal SNPs - MDS



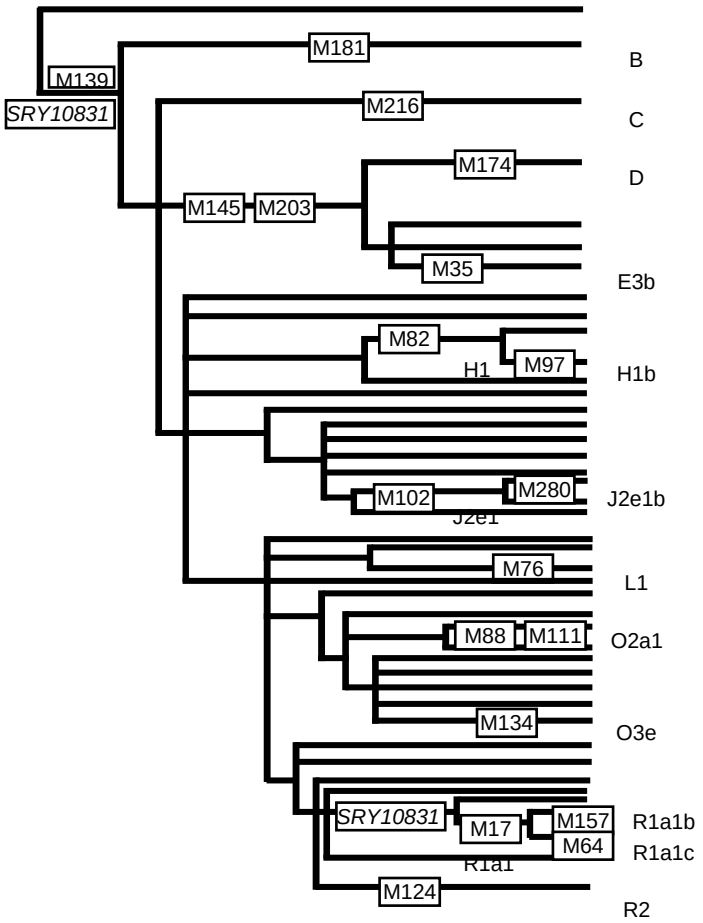
# Contour map of SNP MDS dimension 1

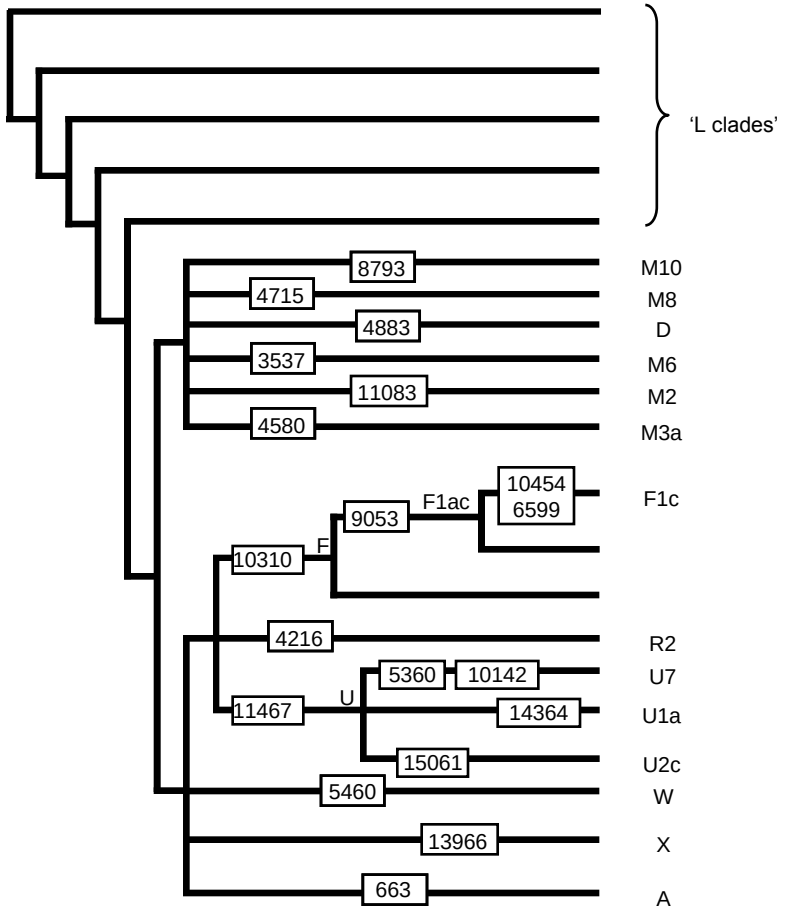


# STRUCTURE: 13 autosomal SNPs

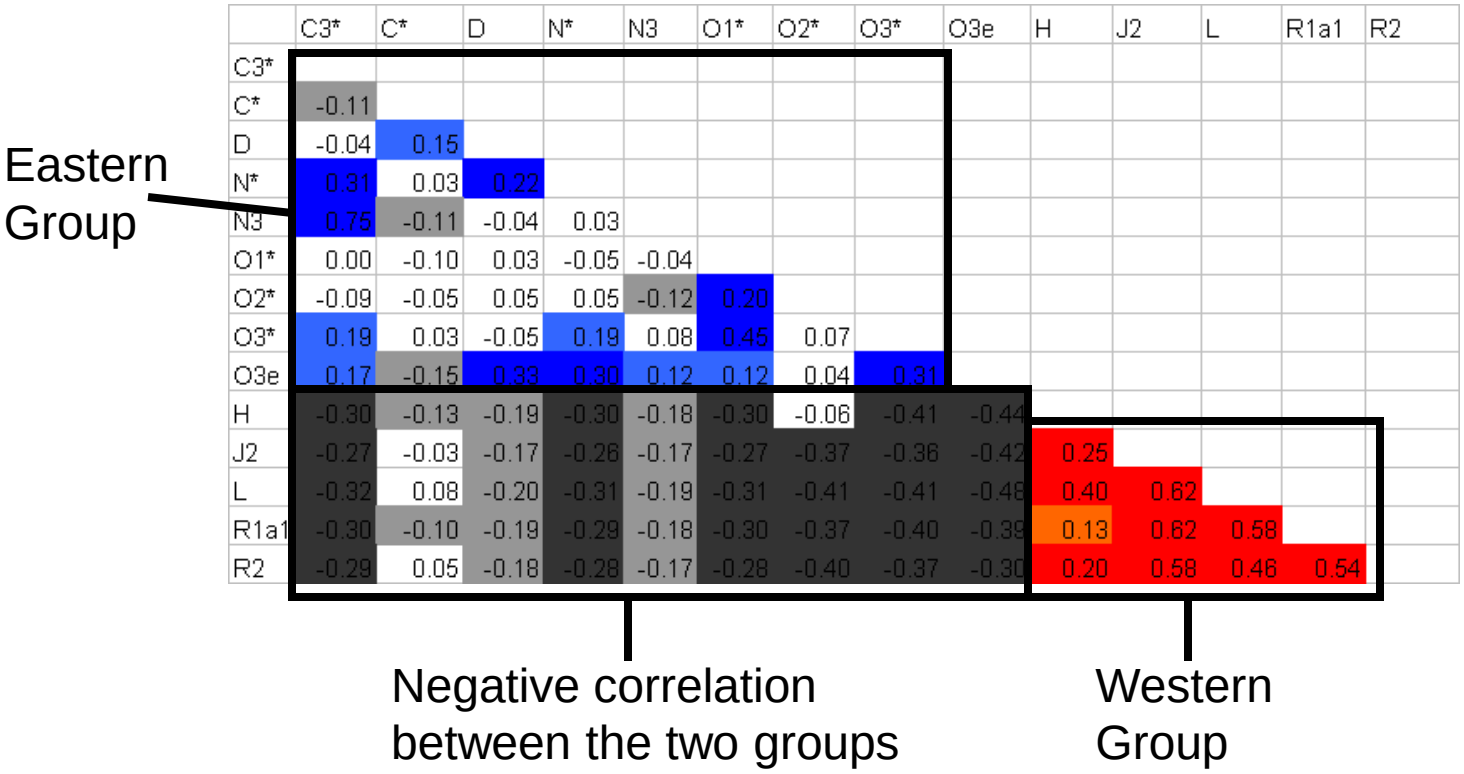


# Y and mtDNA


M181
M139
SRY10831
M216
M174
M145
M203
M35
M82
H1
M97
M102
M280
J2e1
M76
M88
M111
M134
SRY10831
M17
M157
M64
R1a1
M124
B
C
D
E3b
H1b
J2e1b
L1
O2a1
O3e
R1a1b
R1a1c
R2

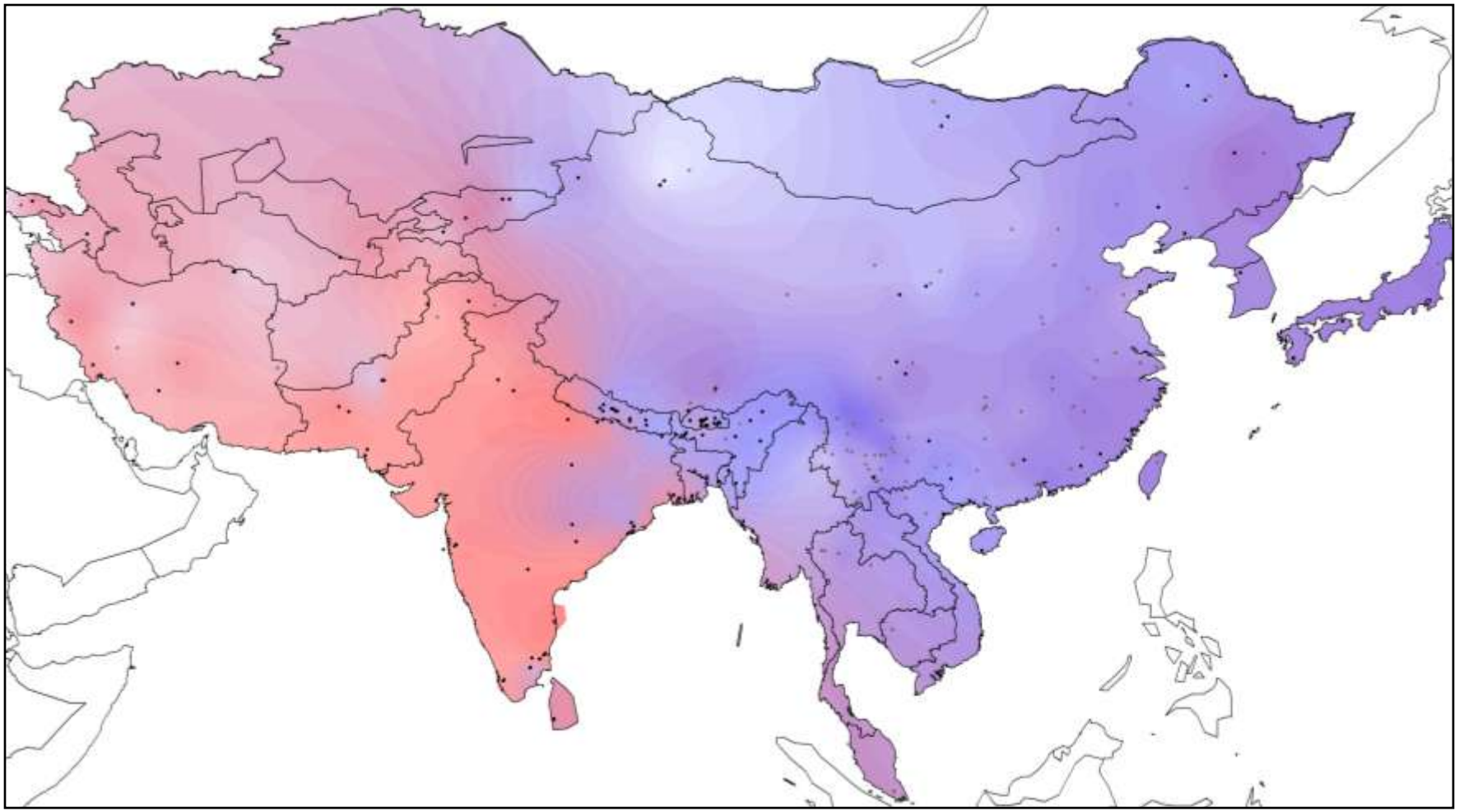

'L clades'
8793
4715
4883
3537
11083
4580
9053
F1ac
10454
6599
F
10310
4216
5360
10142
U
11467
14364
15061
5460
13966
663
M10
M8
D
M6
M2
M3a
F1c
R2
U7
U1a
U2c
W
X
A

# Identification of geographically-correlated Y-haplogroups

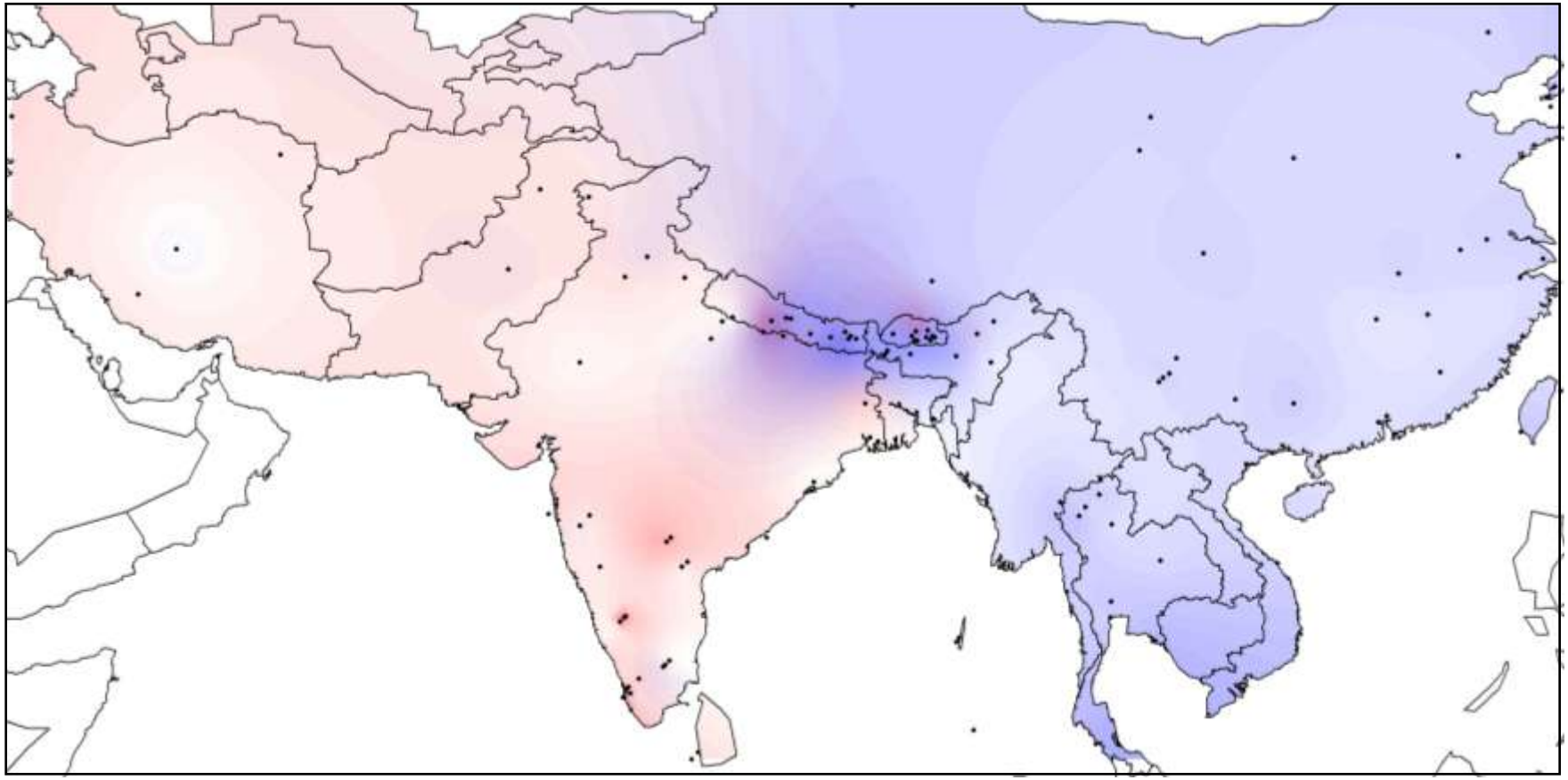


| | C3* | C* | D | N* | N3 | O1* | O2* | O3* | O3e | H | J2 | L | R1a1 | R2 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| C3* | | | | | | | | | | | | | | |
| C* | -0.11 | | | | | | | | | | | | | |
| D | -0.04 | 0.15 | | | | | | | | | | | | |
| N* | 0.31 | 0.03 | 0.22 | | | | | | | | | | | |
| N3 | 0.75 | -0.11 | -0.04 | 0.03 | | | | | | | | | | |
| O1* | 0.00 | -0.10 | 0.03 | -0.05 | -0.04 | | | | | | | | | |
| O2* | -0.09 | -0.05 | 0.05 | 0.05 | -0.12 | 0.20 | | | | | | | | |
| O3* | 0.19 | 0.03 | -0.05 | 0.19 | 0.08 | 0.45 | 0.07 | | | | | | | |
| O3e | 0.17 | -0.15 | 0.33 | 0.30 | 0.12 | 0.12 | 0.04 | 0.31 | | | | | | |
| H | -0.30 | -0.13 | -0.19 | -0.30 | -0.18 | -0.30 | -0.06 | -0.41 | -0.44 | | | | | |
| J2 | -0.27 | -0.03 | -0.17 | -0.26 | -0.17 | -0.27 | -0.37 | -0.36 | -0.42 | 0.25 | | | | |
| L | -0.32 | 0.08 | -0.20 | -0.31 | -0.19 | -0.31 | -0.41 | -0.41 | -0.48 | 0.40 | 0.62 | | | |
| R1a1 | -0.30 | -0.10 | -0.19 | -0.29 | -0.18 | -0.30 | -0.37 | -0.40 | -0.39 | 0.13 | 0.62 | 0.58 | | |
| R2 | -0.29 | 0.05 | -0.18 | -0.28 | -0.17 | -0.28 | -0.40 | -0.37 | -0.30 | 0.20 | 0.58 | 0.46 | 0.54 | |

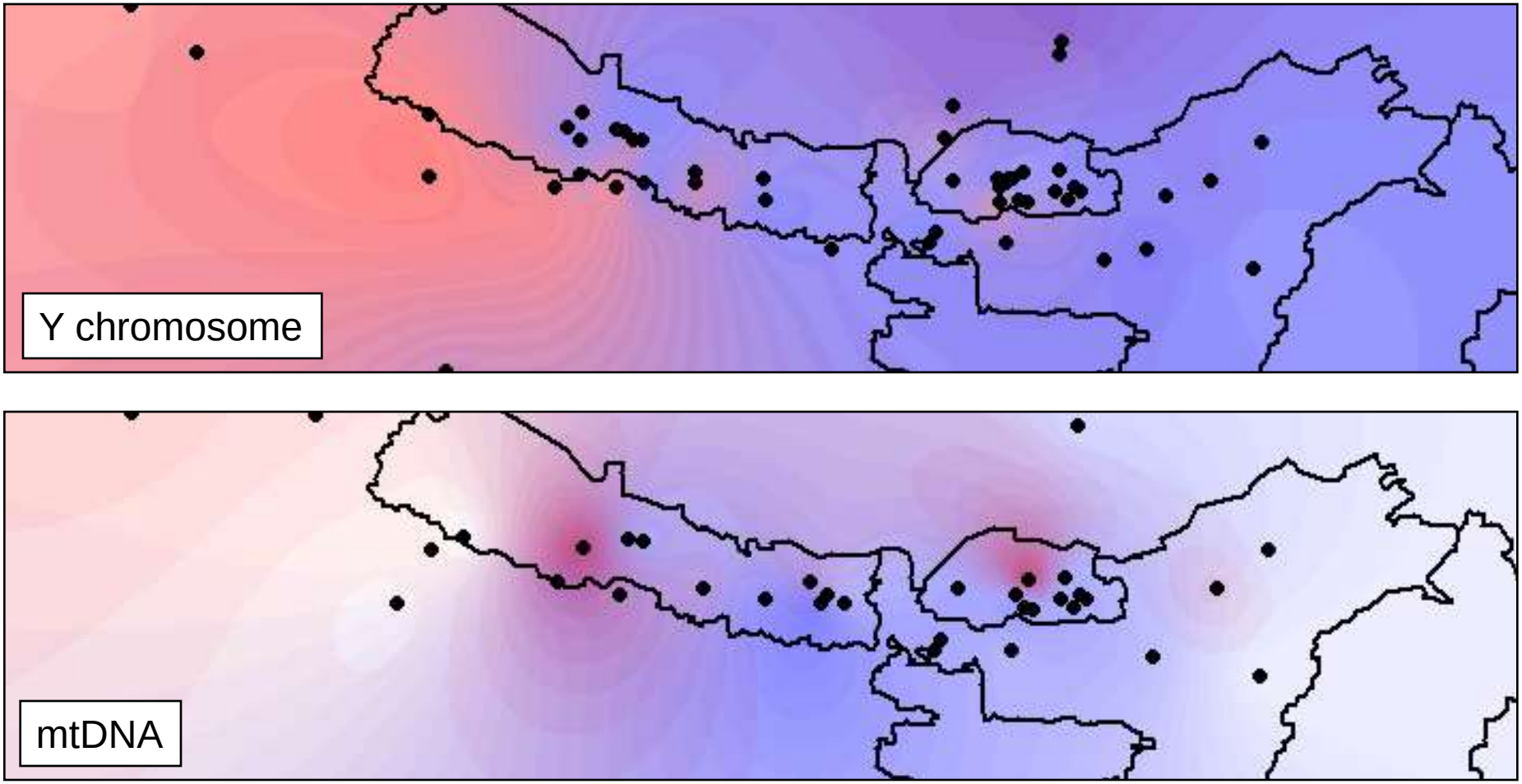
# The Y-chromosomal landscape



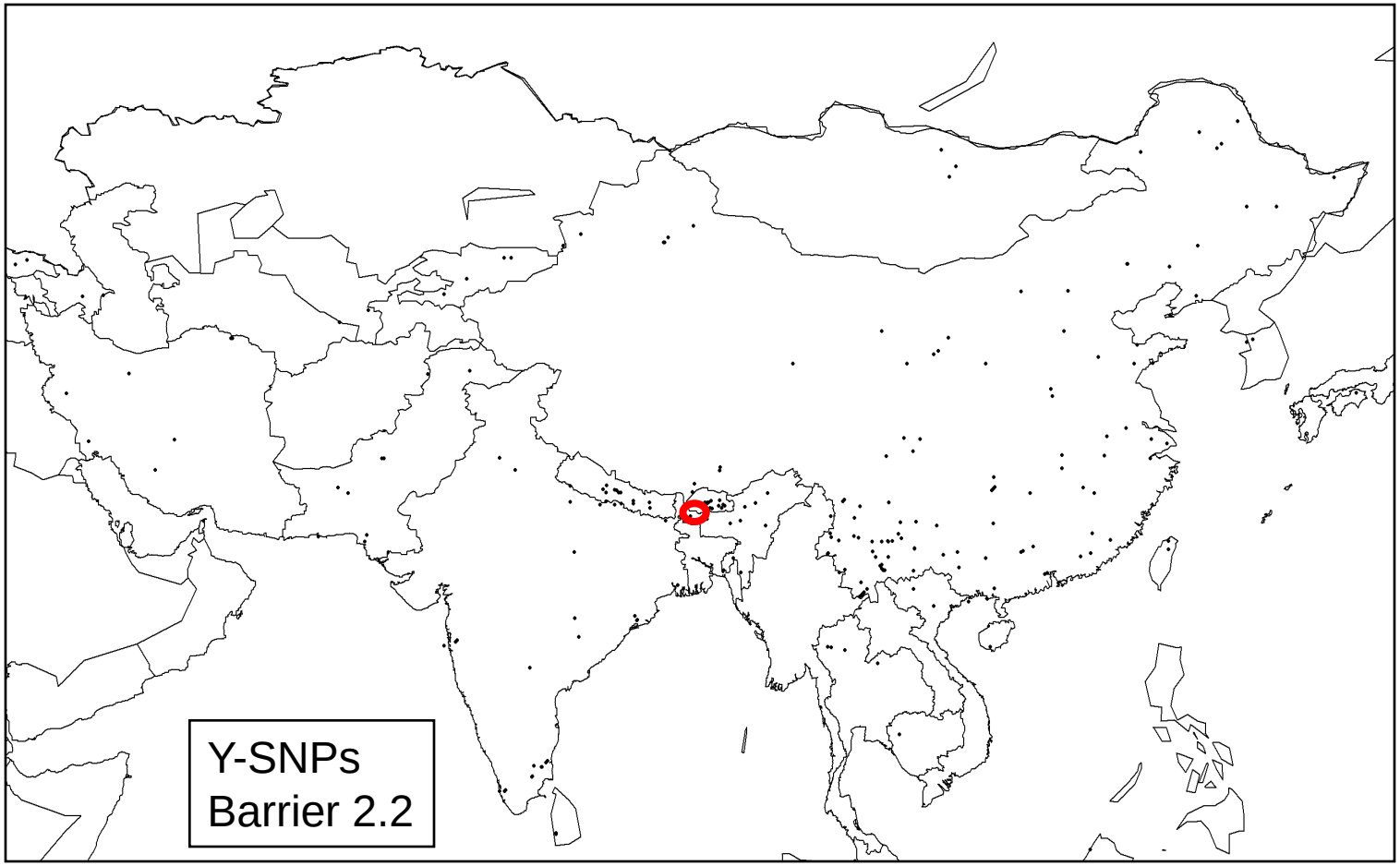
# The mtDNA landscape



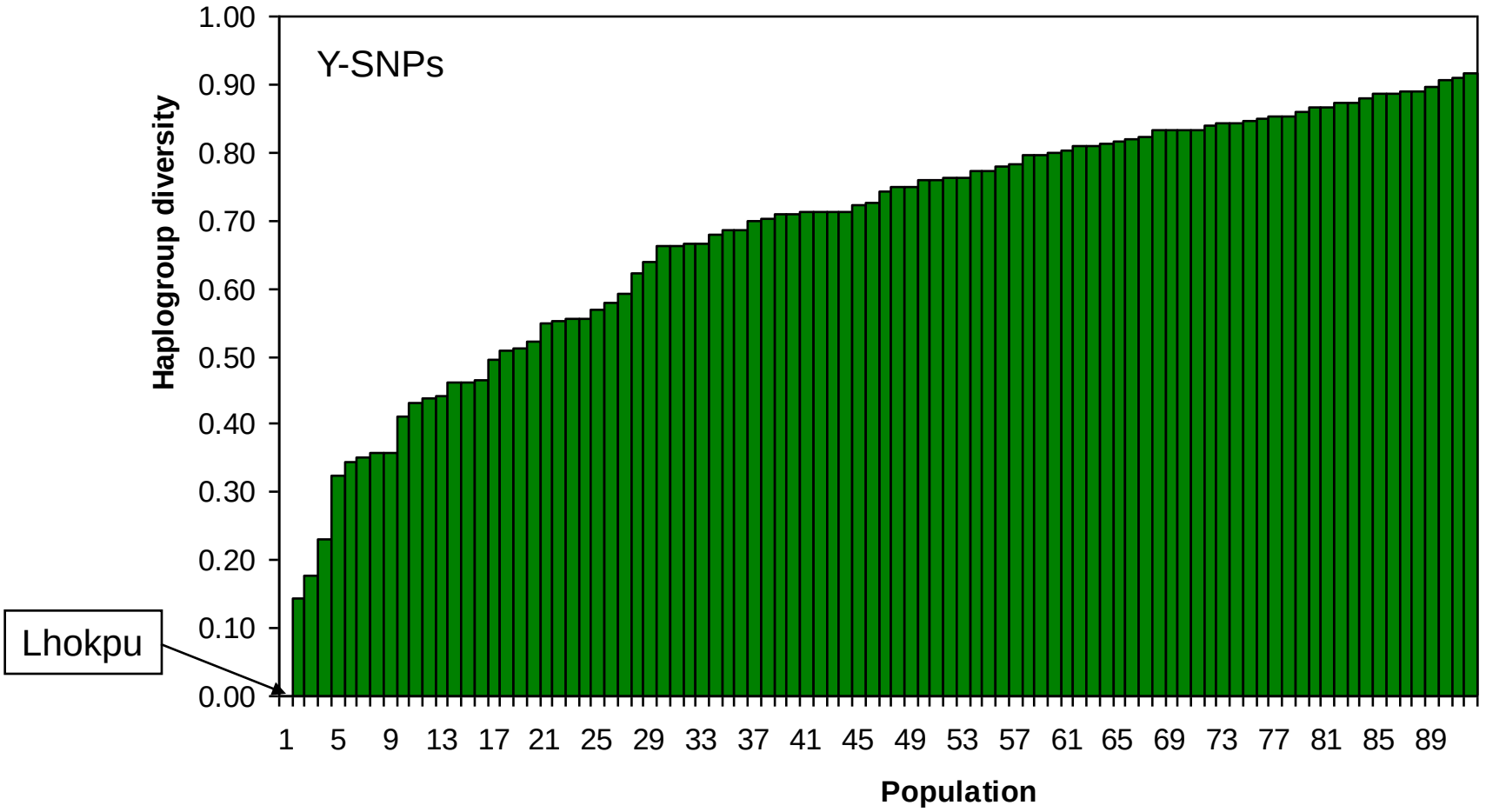
# The Himalayan region



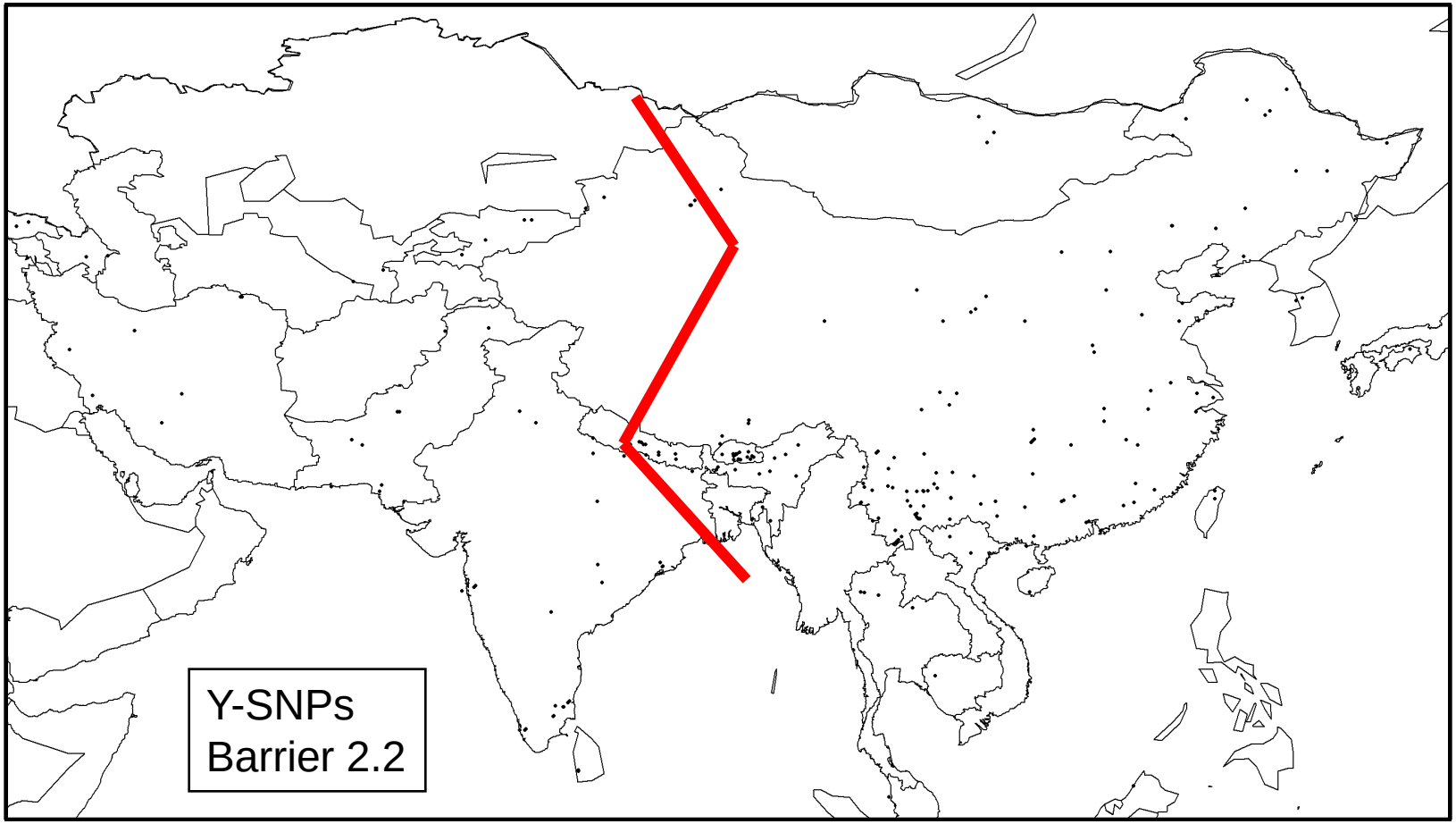
# Searching for boundaries with Barrier



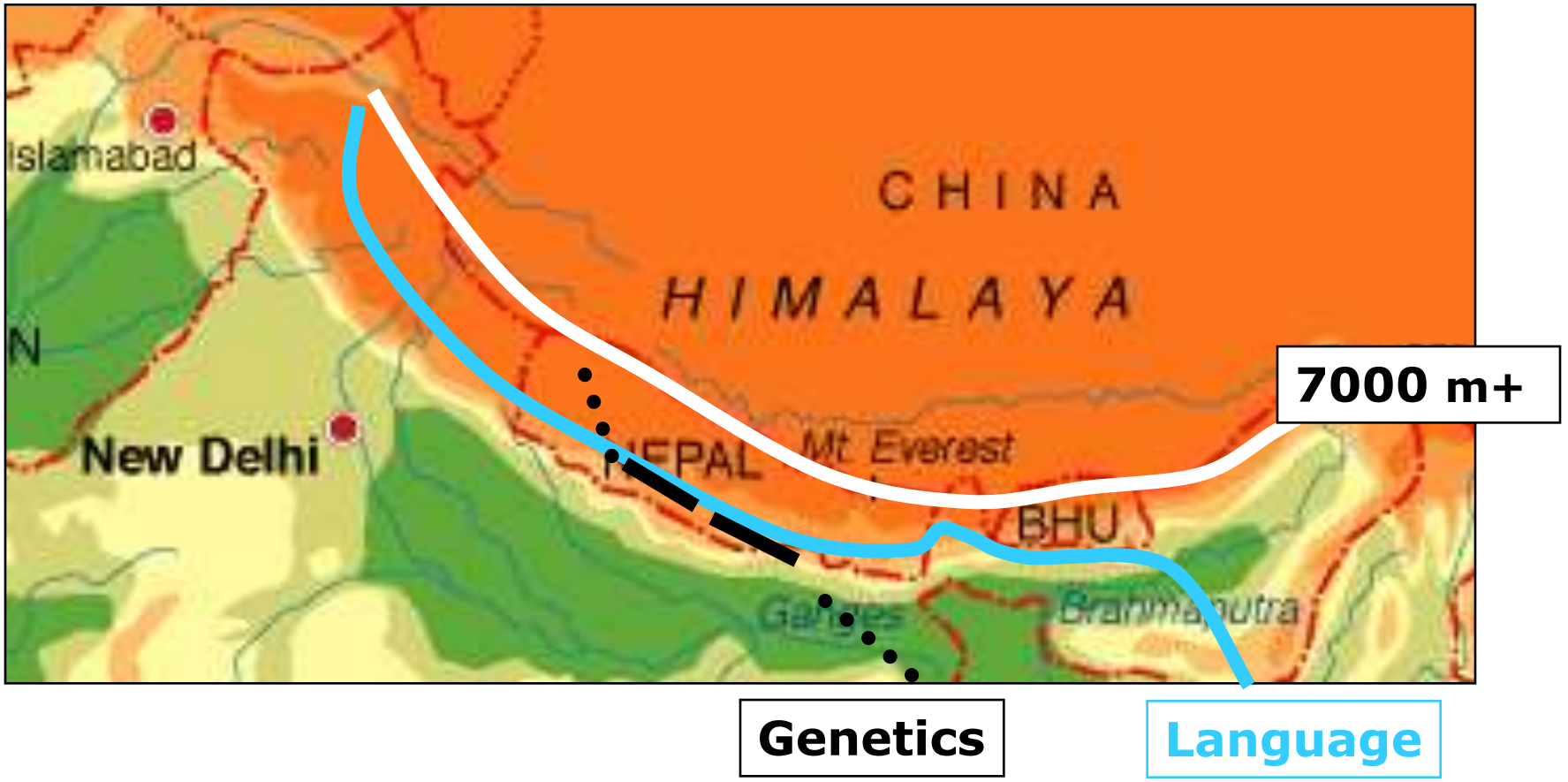
# Diversity varies between populations



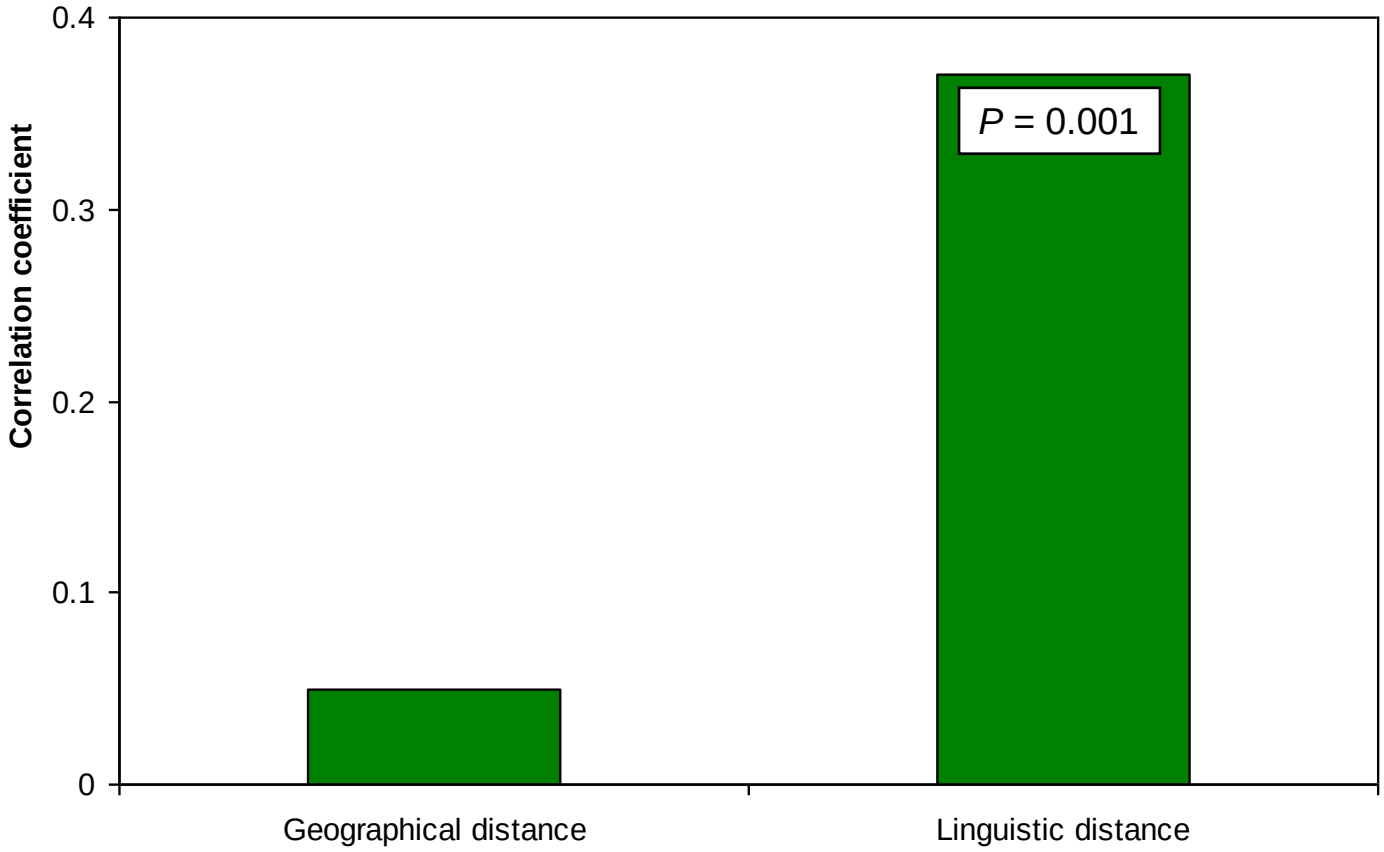
# Boundary after removing low-diversity populations



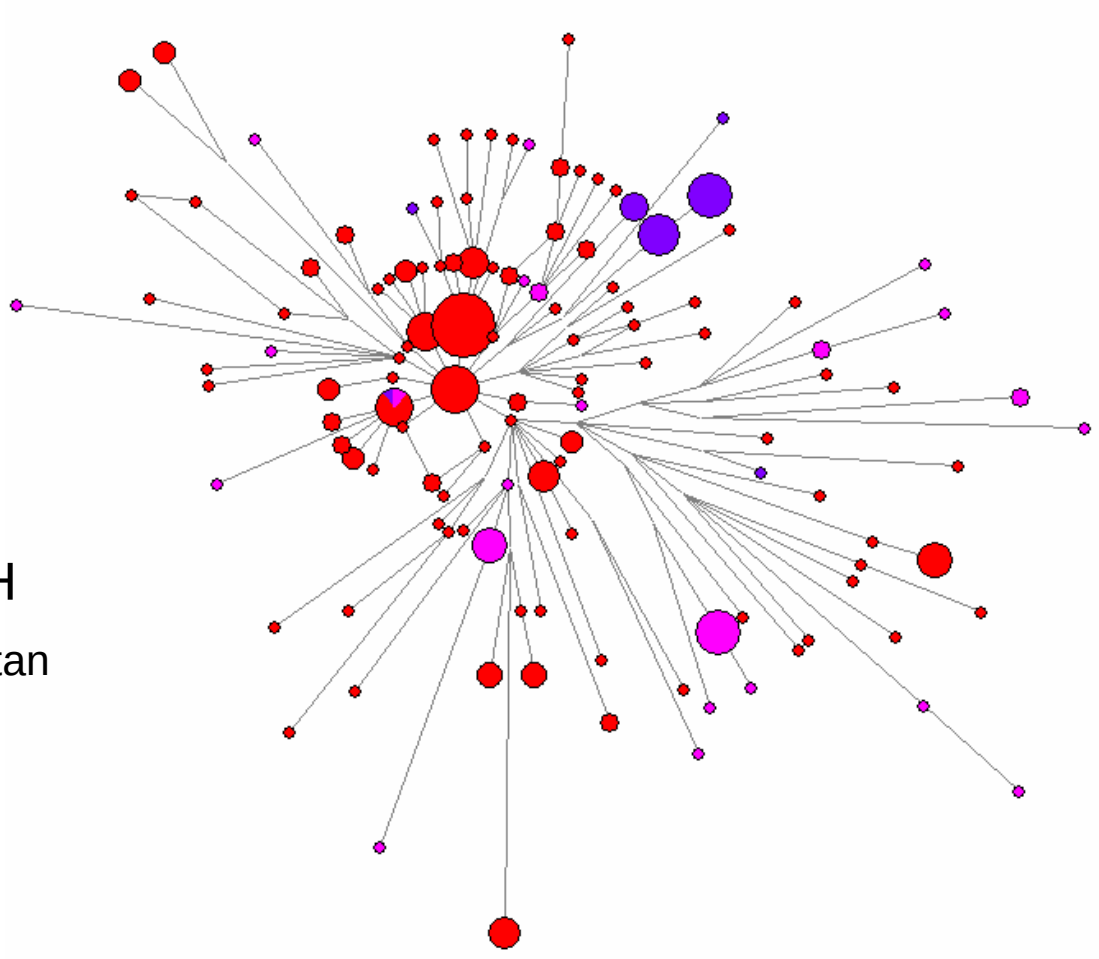
# Comparison of boundaries



# Mantel tests of genetic distance correlation (Y data)



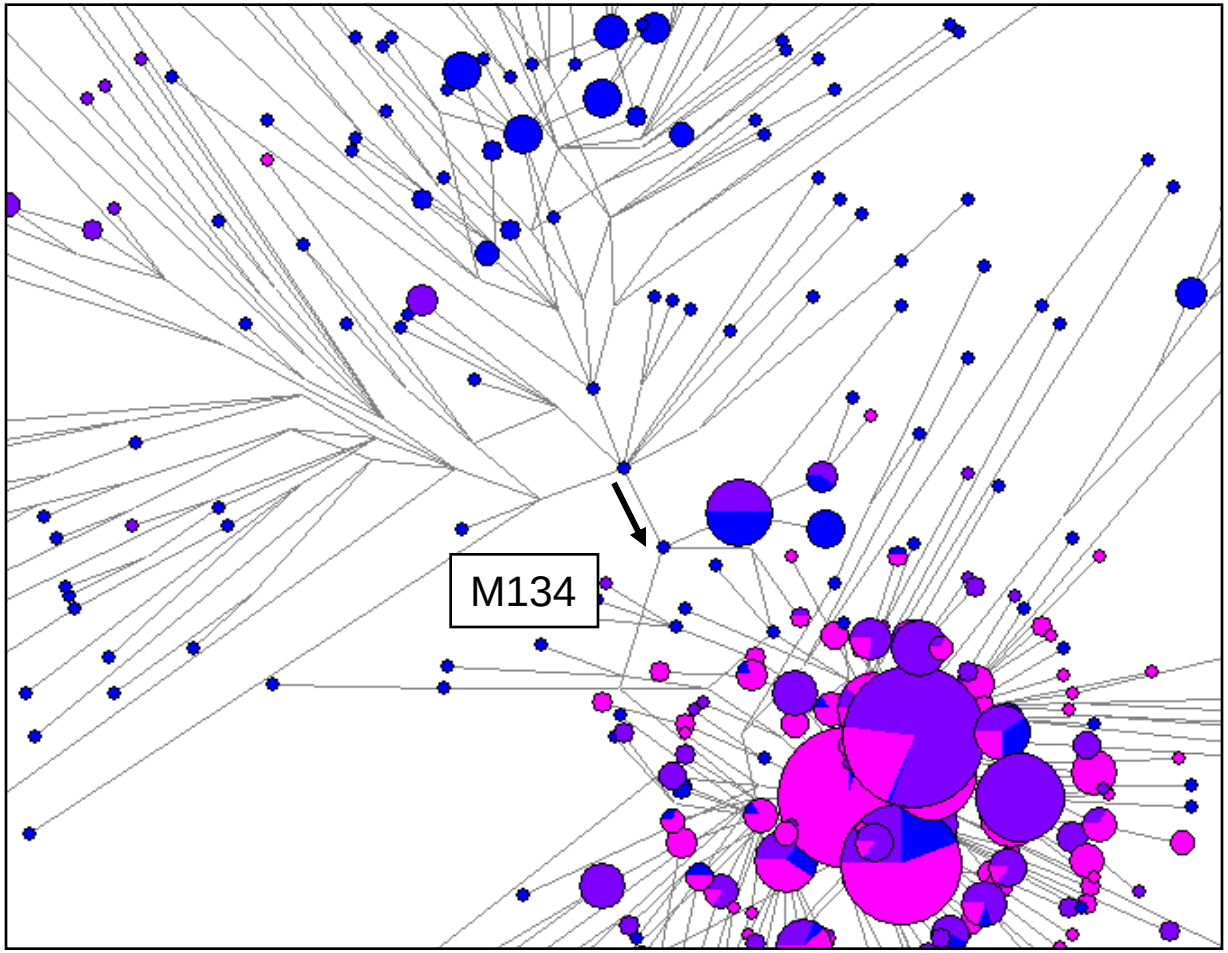
# Direction of gene flow?



Haplogroup H

- India, Pakistan
- Nepal
- Bhutan

# Direction of gene flow?




Haplogroup O3

- East Asia
- Nepal
- Bhutan

# Peopling of the region

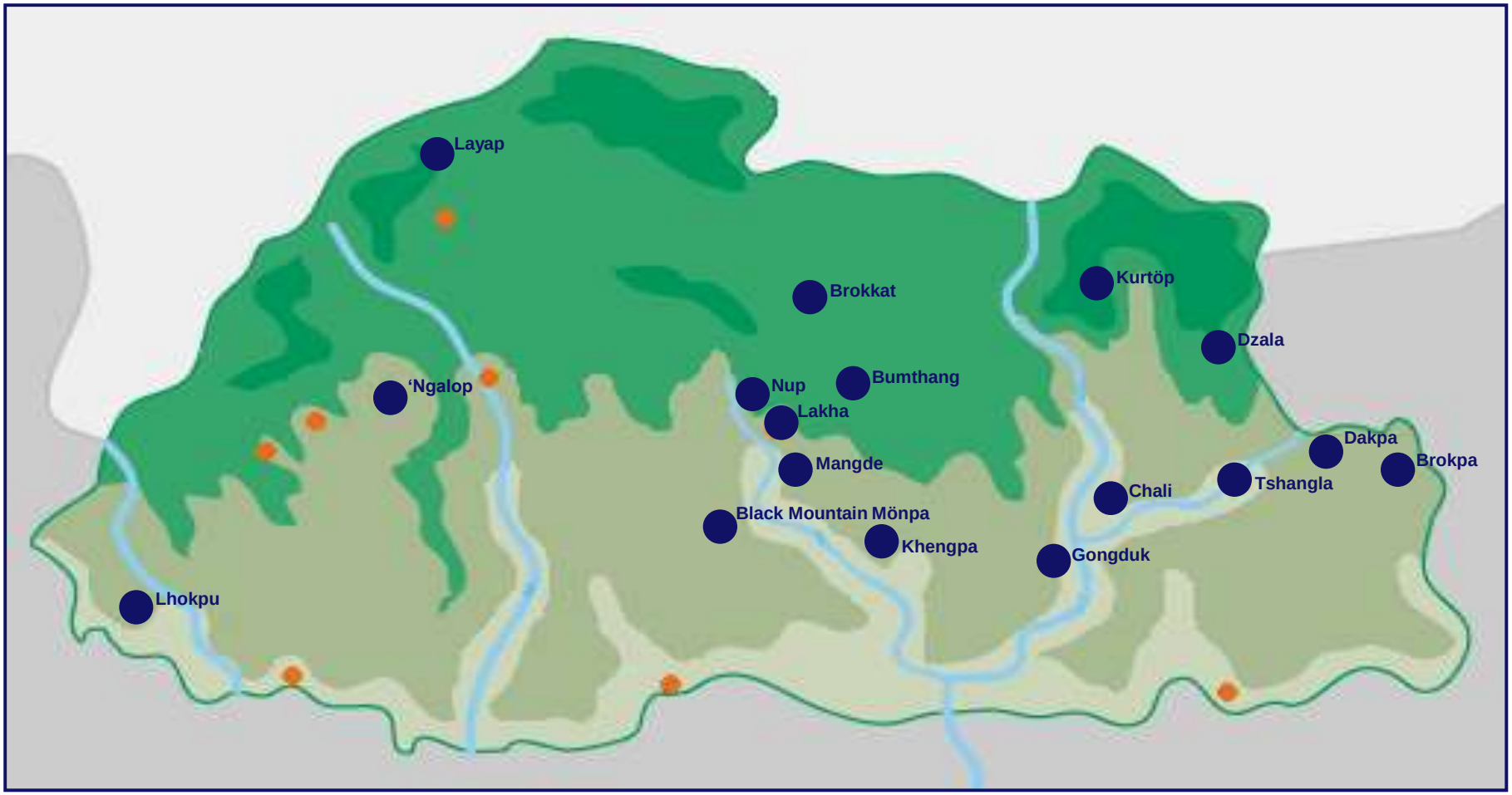

- When? Archaeology; palaeoclimatology; linguistics; genetics…

# Conclusions

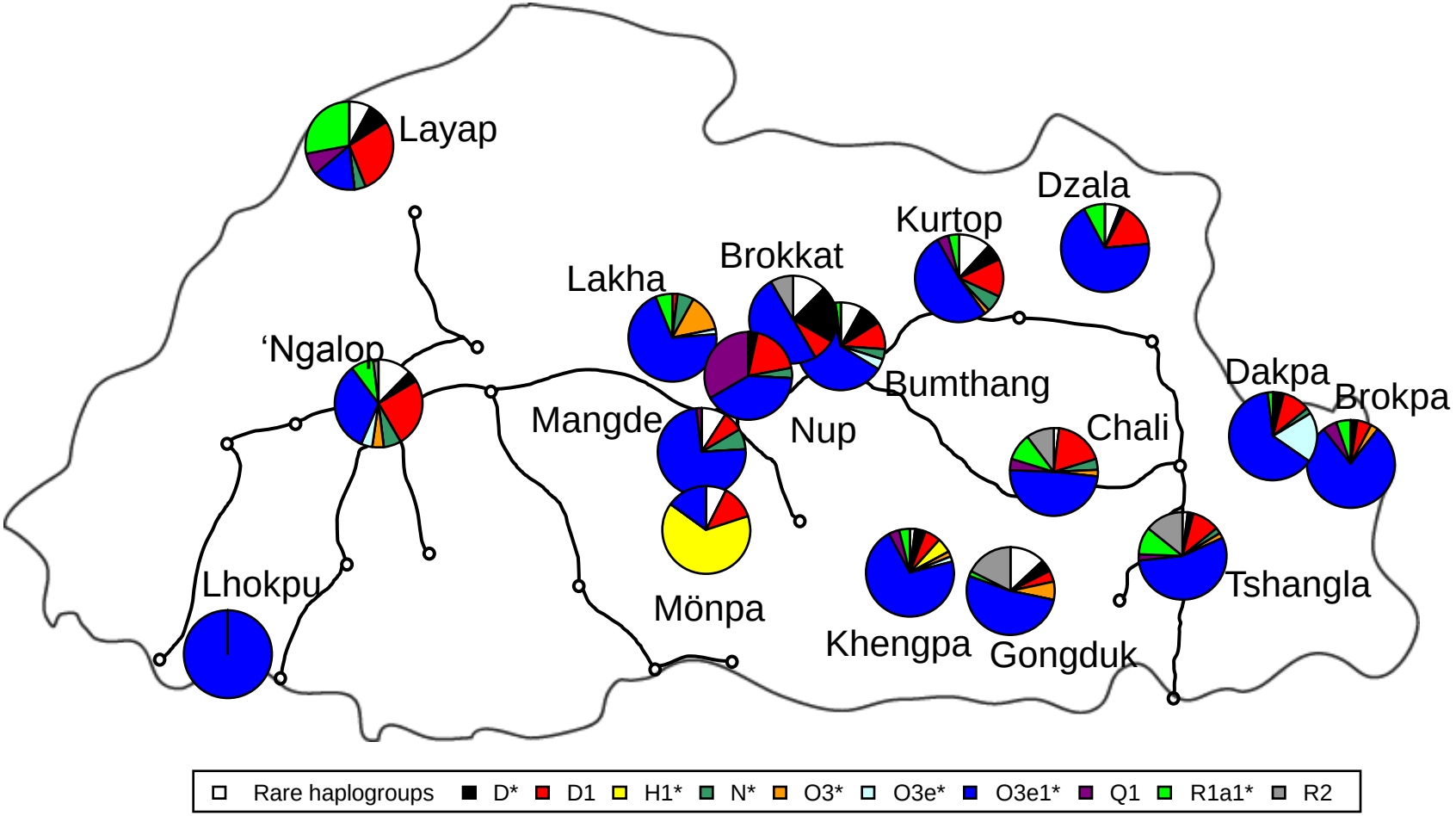
- Genetic variation correlates better with language than with geography
- Almost perfect fit between linguistic and genetic boundaries
- Consistent with TB-speaker origins from the east and IE-speaker origins from the west within the last few thousand years
- High diversity within Nepal and Bhutan

# Extreme geography in Bhutan

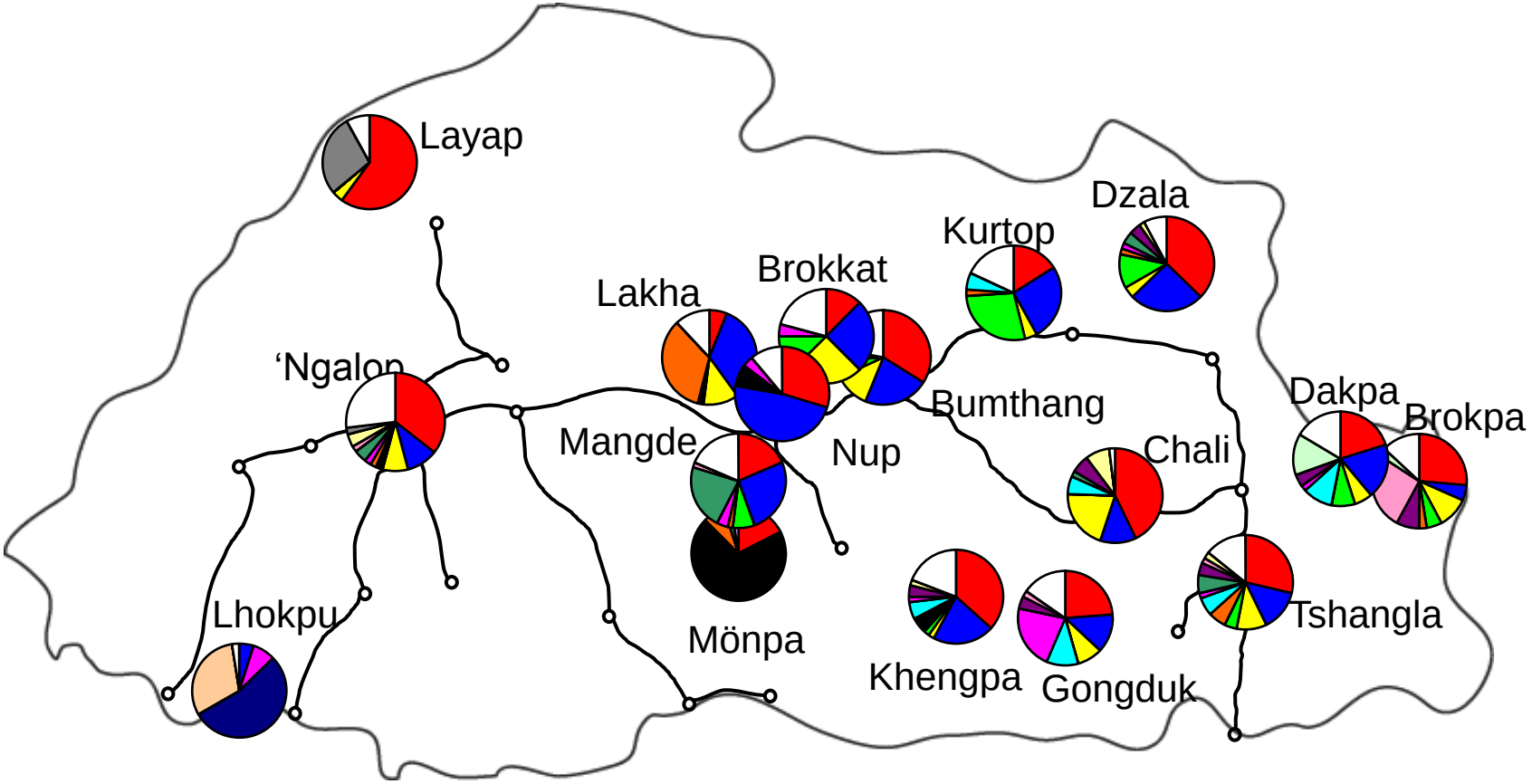
# Ethnolinguistic centres in Bhutan



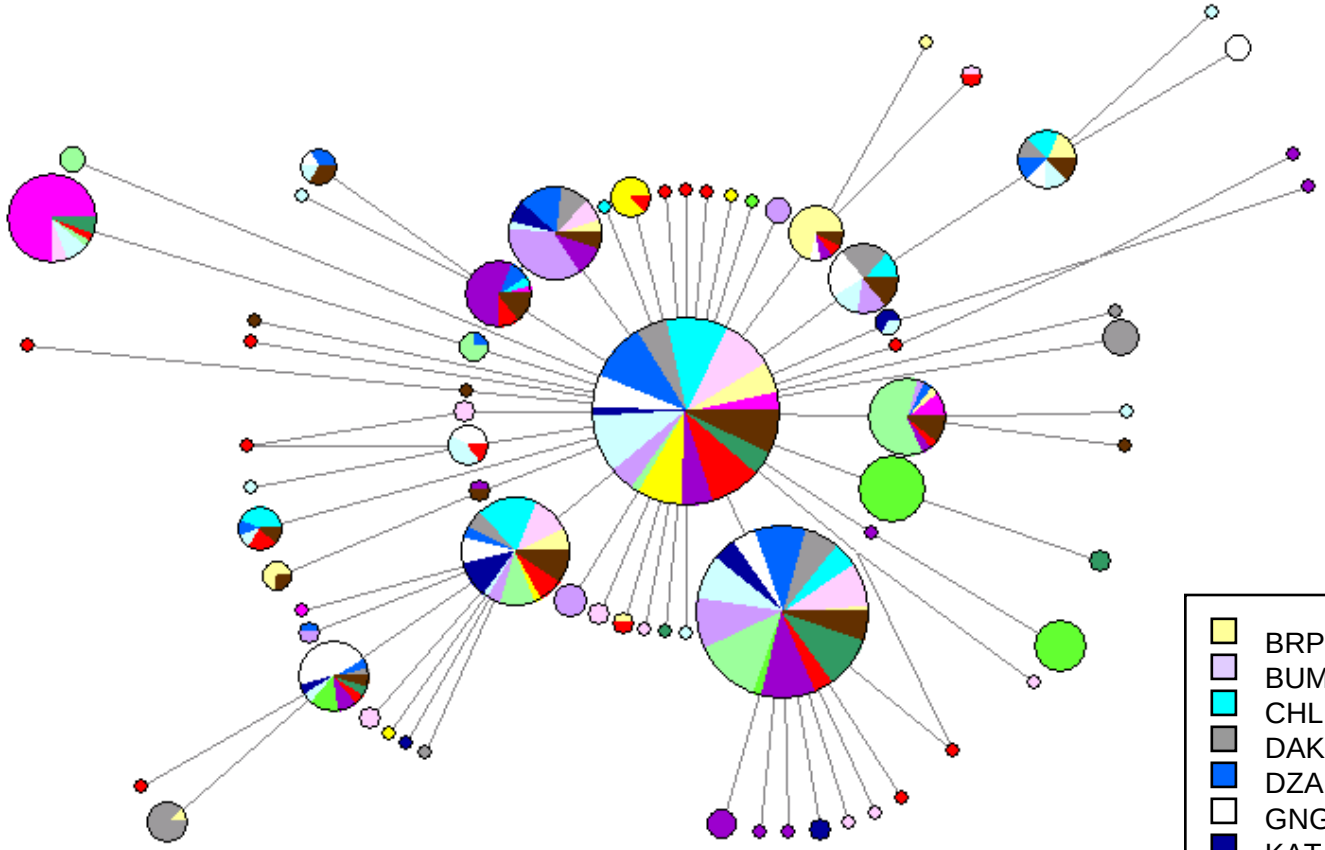
# Y haplogroups - high diversity, much population structure



# mtDNA haplotypes - high diversity, much population structure



# mtDNA haplotypes



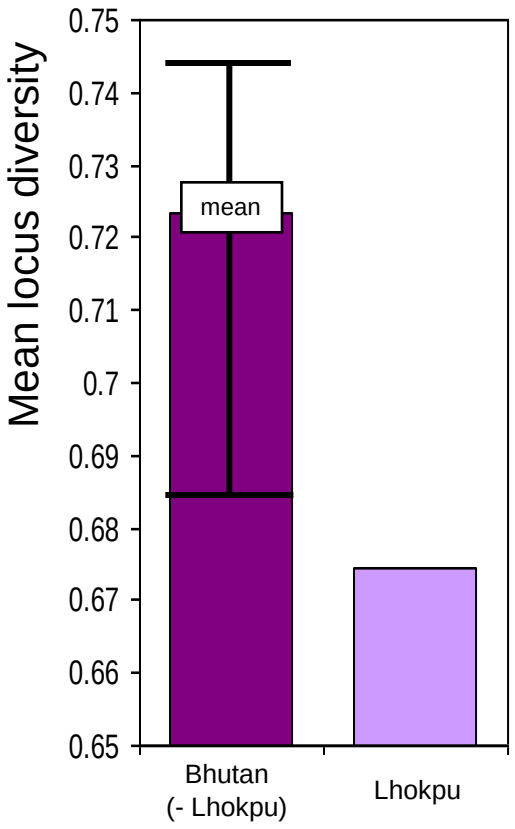

Median-Joining Network of HVSI sequences

All nucleotide sites with a recurrence greater than 2 removed

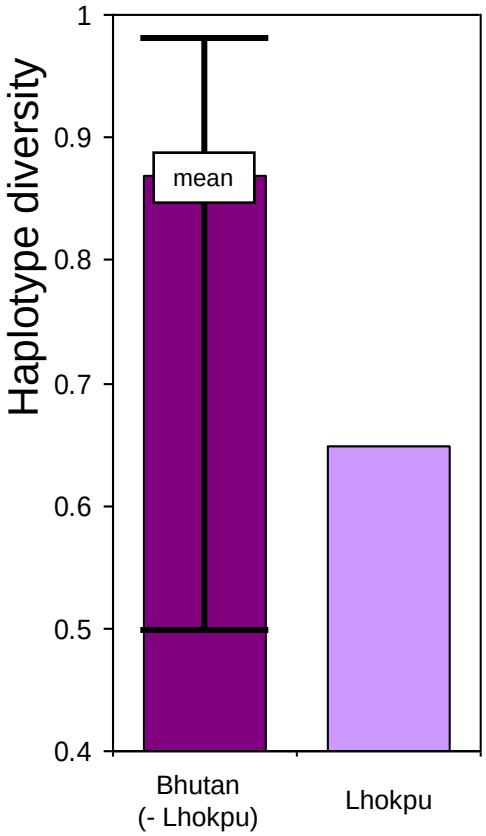
Network weighted with ts:tv of 1:15

# The Lhokpu: low diversity

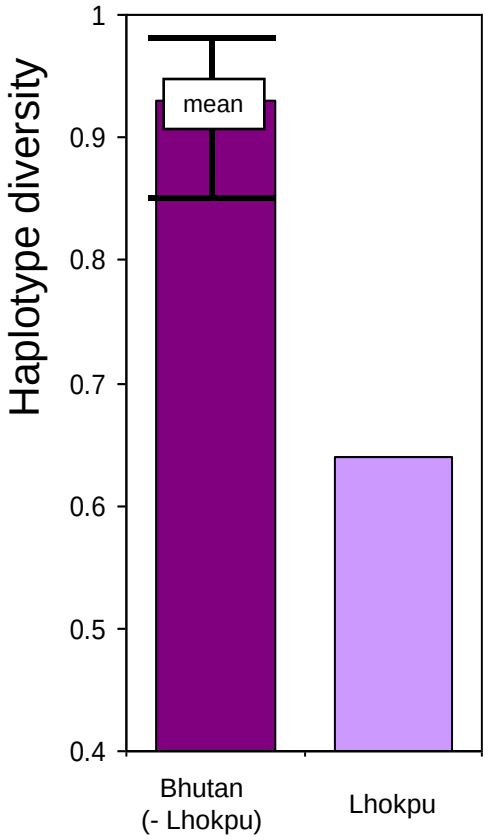
**Autosomal STRs**




**mtDNA HVSI**

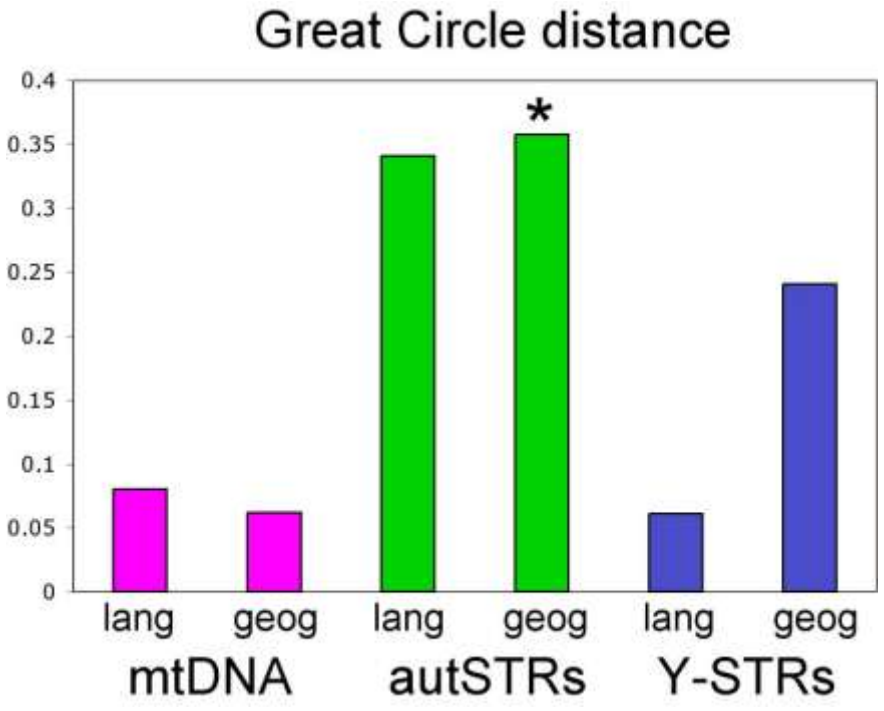


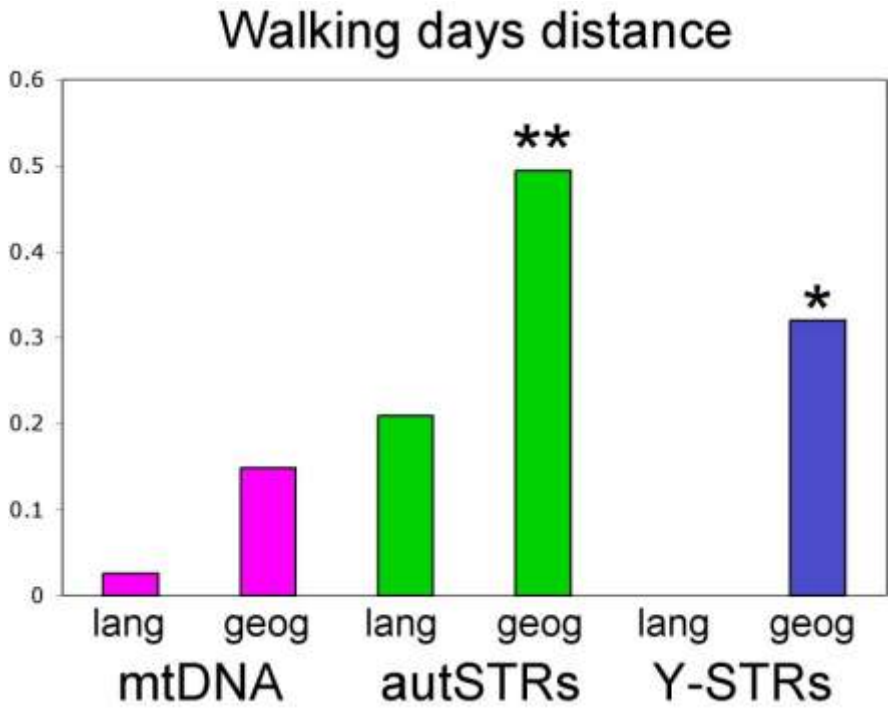

**Y-STRs**



# Diversity patterned more by geography than language

- Mantel testing, using distances:
  - Genetic: $F_{ST}$, $R_{ST}$
  - Geographic: Great Circle, or walking days
  - Linguistic: phylogenetic units within language tree



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