



# **BIOL 502 Population Genetics Spring 2017**

## **Week 14 Human Population Genomics**

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# Peoples and Cultures

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Table 1. Genetic distances among major continents or continental areas, based on 120 classical polymorphisms

	Africa	Oceania	East Asia	Europe
Oceania	24.7			
East Asia	20.6	10.0		
Europe	16.6	13.5	9.7	
America	22.6	14.6	8.9	9.5

Information for this table was adapted from refs. 1 and 2.

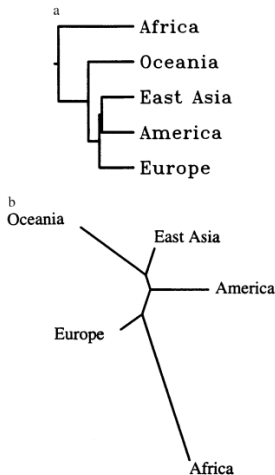
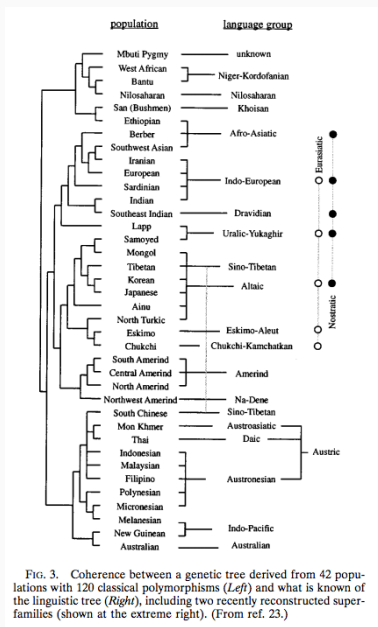
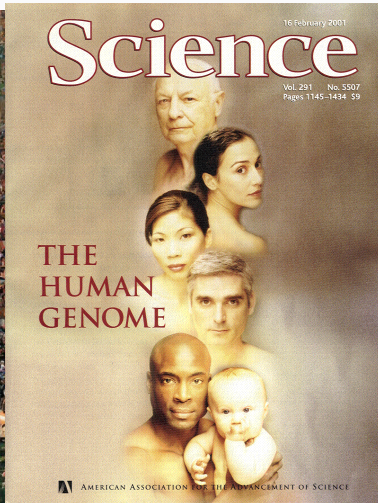
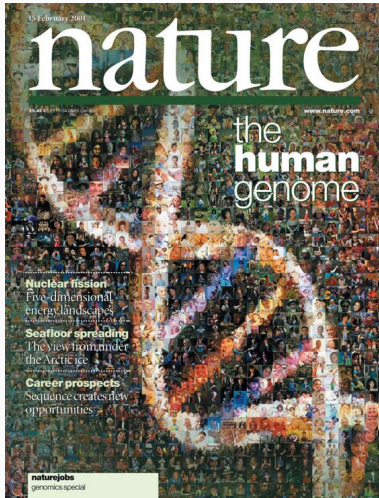


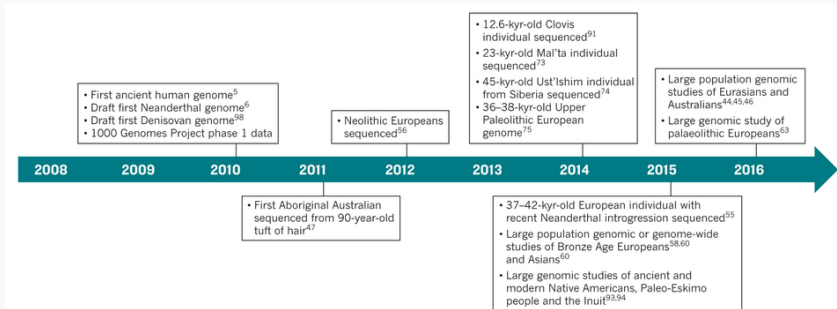
FIG. 1. (a) Tree derived from data in Table 1 by unweighted pair-group method with arithmetic mean. (b) Tree from same data by neighbor joining. Note the difference in the location of the branch leading to Europe.



# The Human Genome Project - 2001



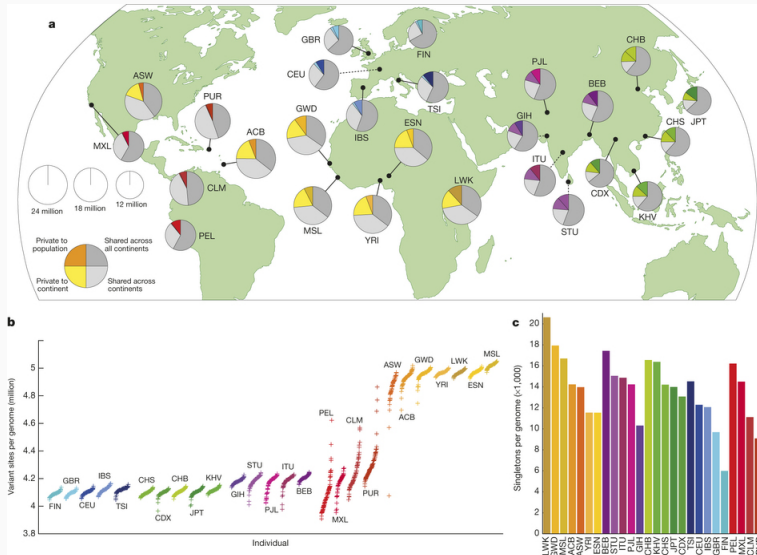
# A timeline - Nielsen et al. 2017 DOI:10.1038/nature21347



# Human Genomic Variation

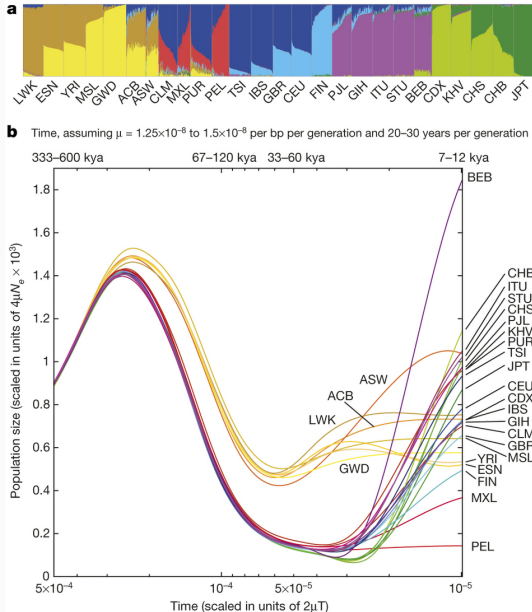
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# The 1000 Genome Project - 2015 DOI:10.1038/nature15393

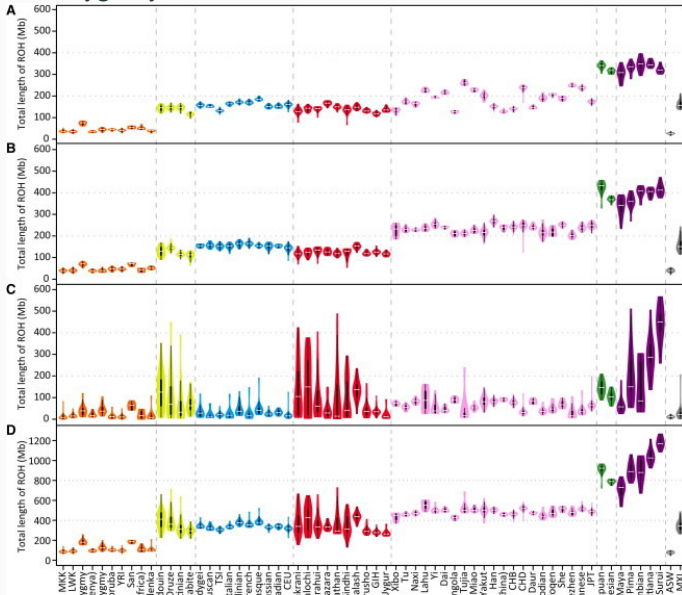


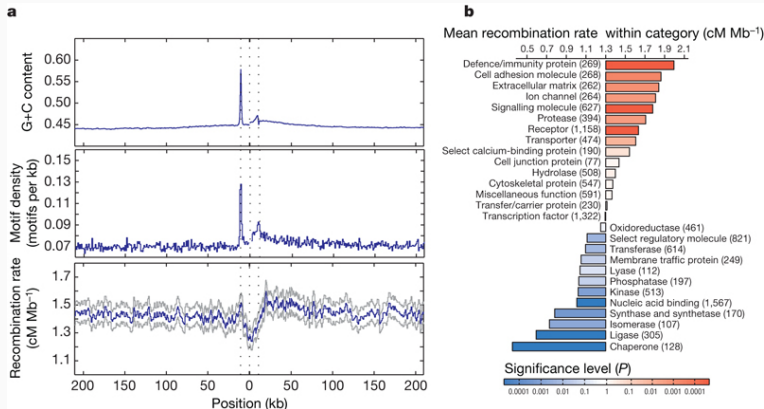


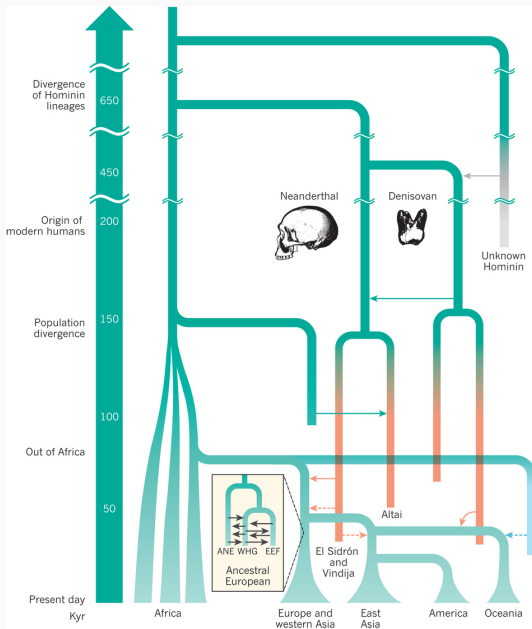
# The 1000 Genome Project - 2015

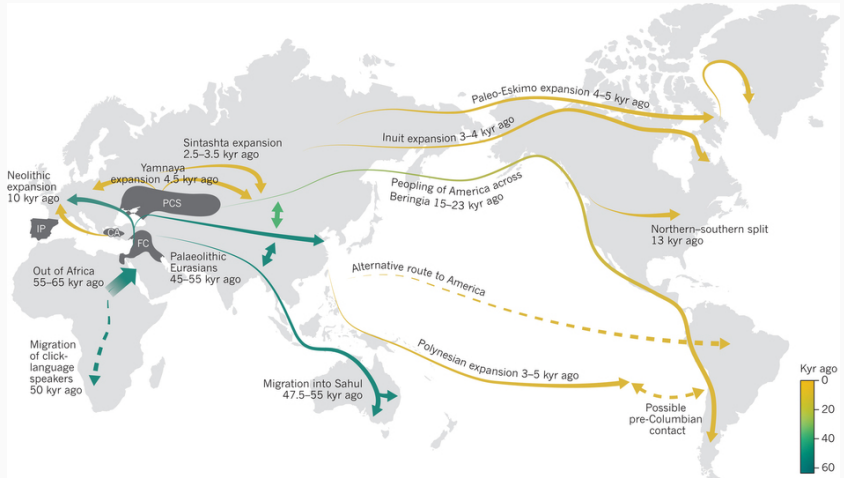


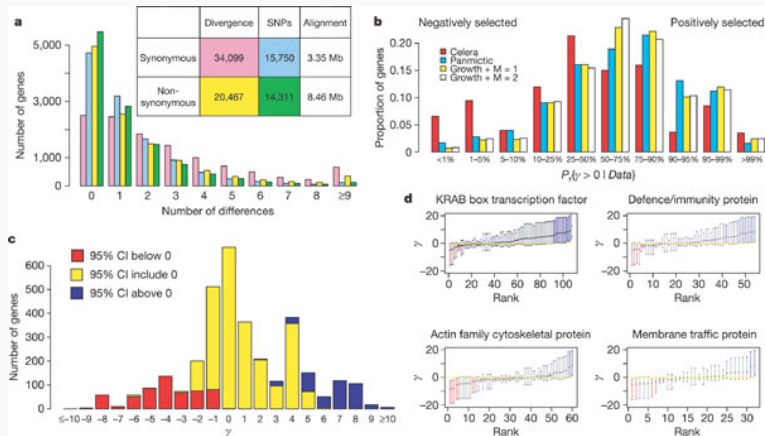
## Homozygosity











## Where to access data:

- 1000 Genome Project - <http://www.internationalgenome.org/>
- UK10K Project - <https://www.uk10k.org/>
- DECODE Project - Genome of Iceland - <https://www.decode.com/>
- Complete Genomics Data -  
<http://www.completegenomics.com/public-data/69-genomes/>
- Private databases - 23andMe, ancestry.com, etc.

# Statistical Issues

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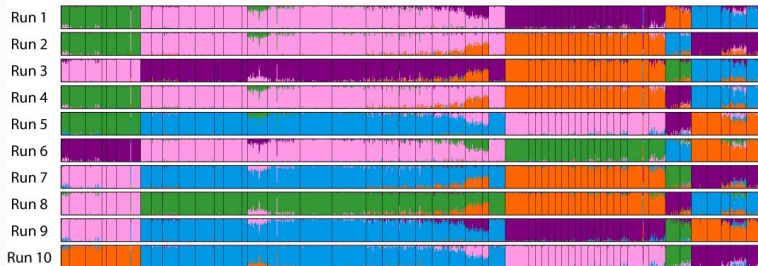


- Sample size
- Multiple testing and corrections
- Missing data/unsampled variation
- Genotyping/sequencing errors
- Quality control
- Bioinformatics errors
- Identifiability
- Confounding signals
- Label switching

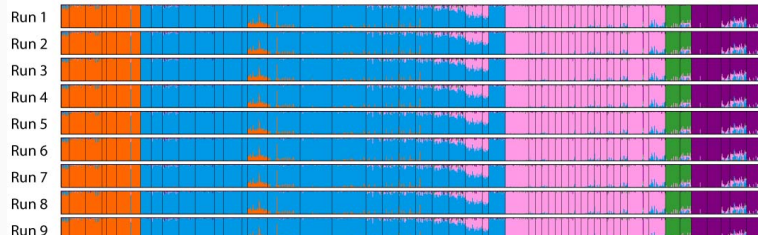
## e.x. Label switching

Jakobsson and Rosenberg 2007 DOI:10.1093/bioinformatics/btm233

Pre-CLUMPP

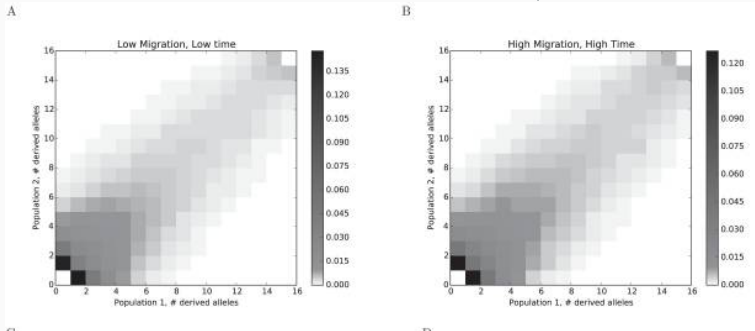


Post-CLUMPP



## e.x. Identifiability

Hey, Chung and Sethuraman 2016 DOI:10.1111/mec.13381



## e.x. Confounding effects

Falush et al. 2017 DOI:

<http://biorxiv.org/content/biorxiv/early/2016/07/28/066431.full.pdf>

