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Genetics 453

Evolutionary Genetics

Coalescents

Joe Felsenstein

Department of Genome Sciences
University of Washington, Seattle

email: `joe@genetics.washington.edu`

Cann, R. L., M. Stoneking, and A. C. Wilson. 1987. Mitochondrial DNA and human evolution. *Nature* **325:a** 31-36.

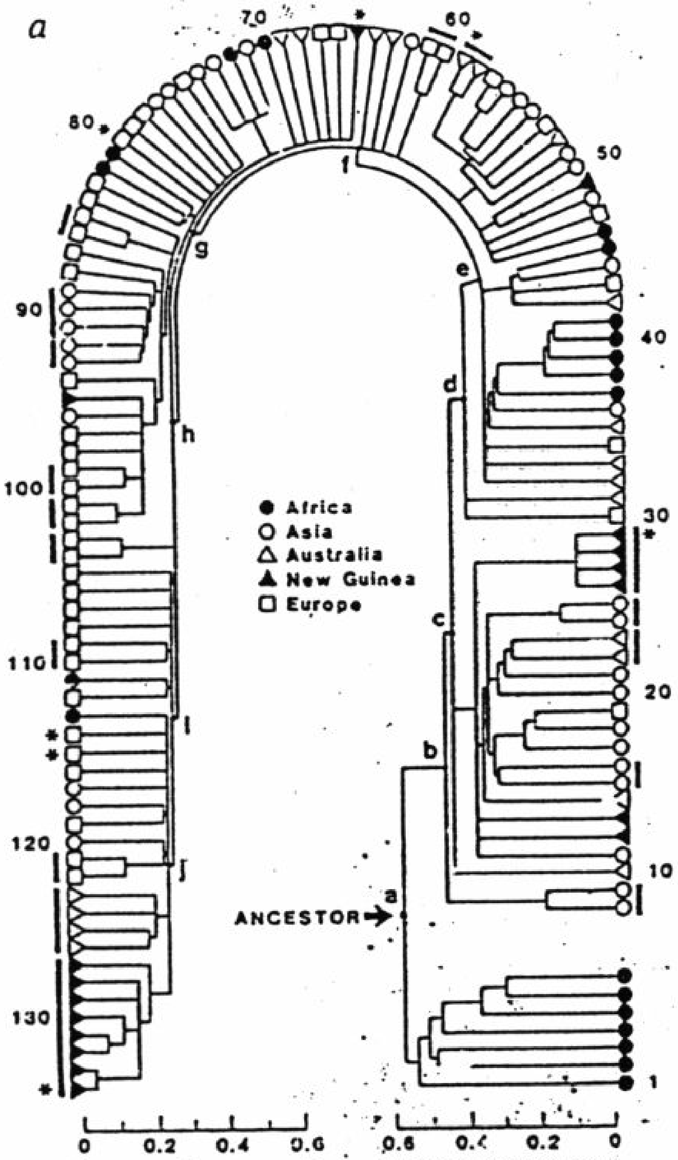
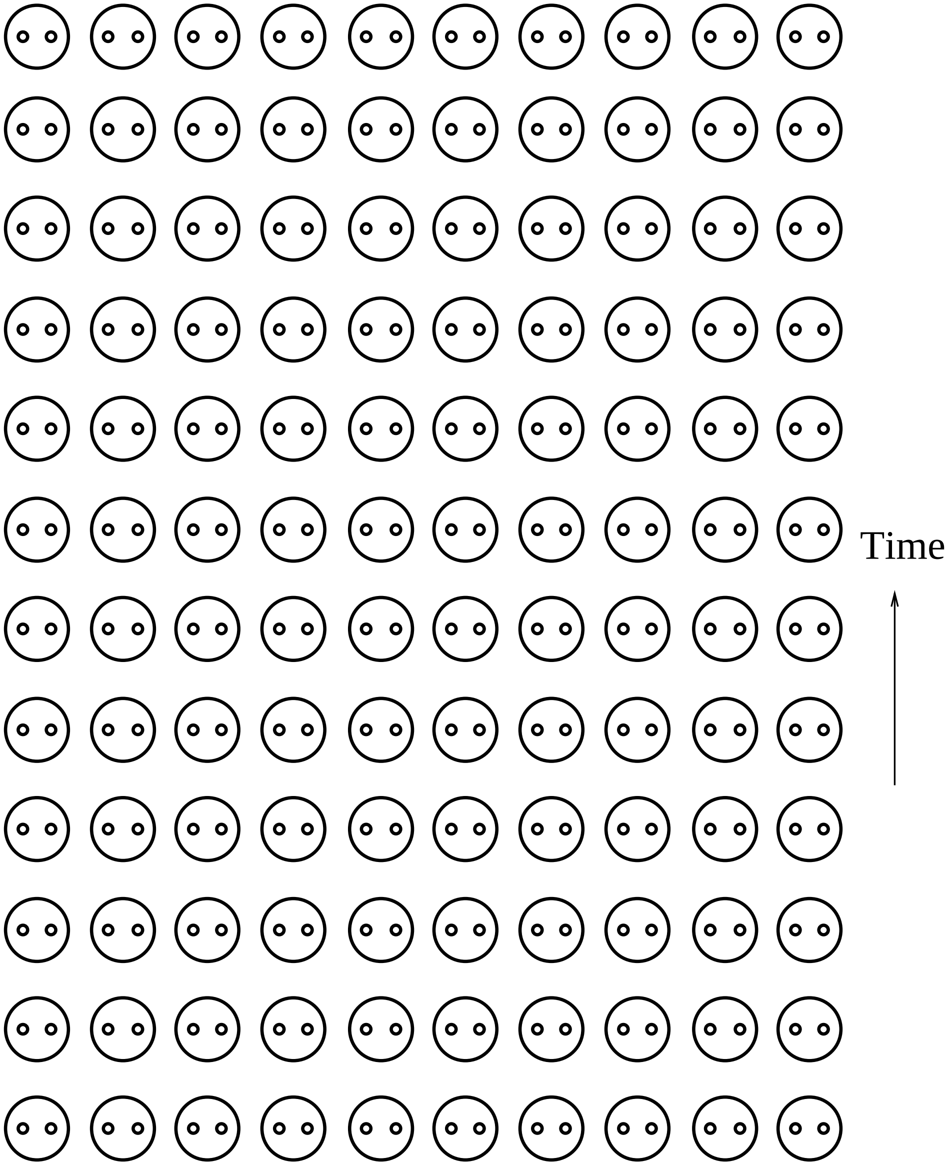
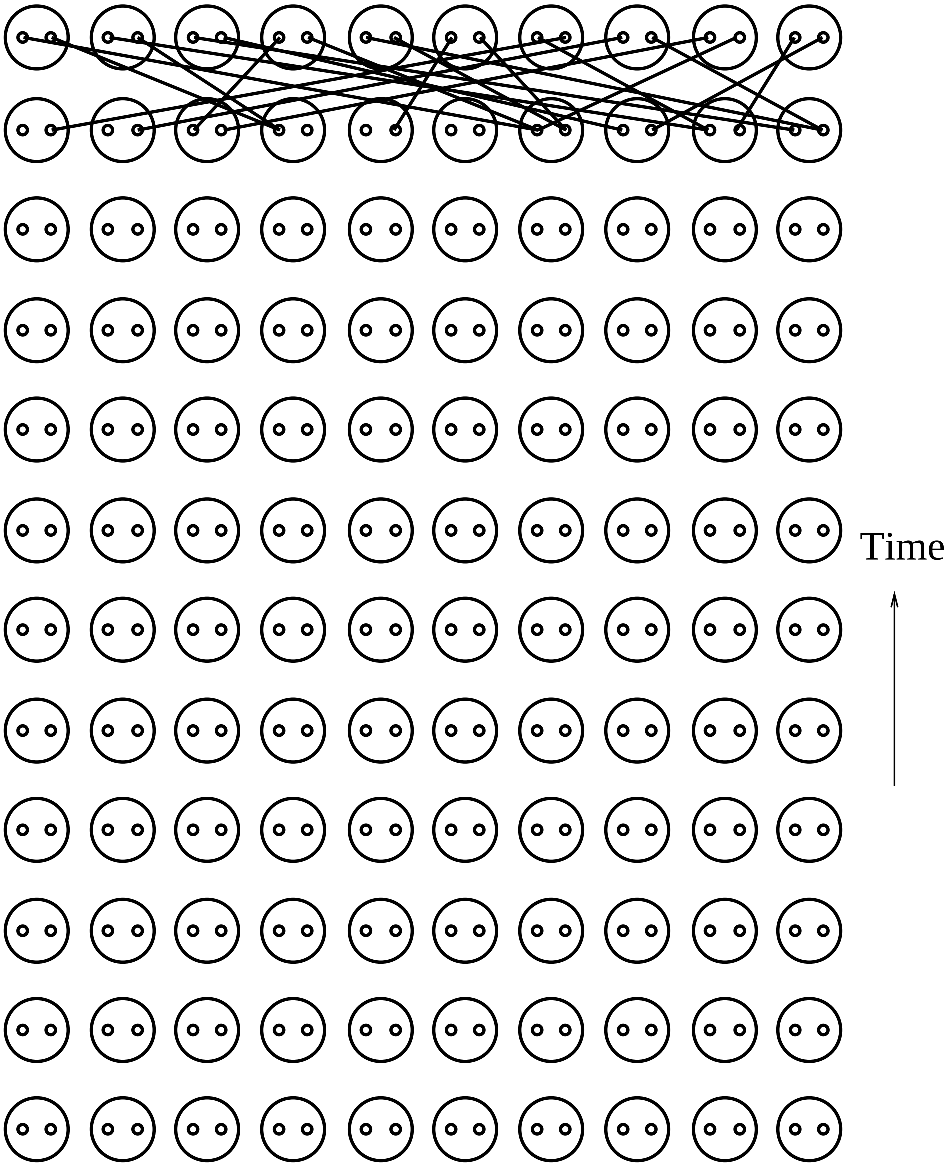


Fig. 3 *a*, Genealogical tree for 134 types of human mtDNA (133 restric
restriction sites used. The tree accounts for the site differences observ

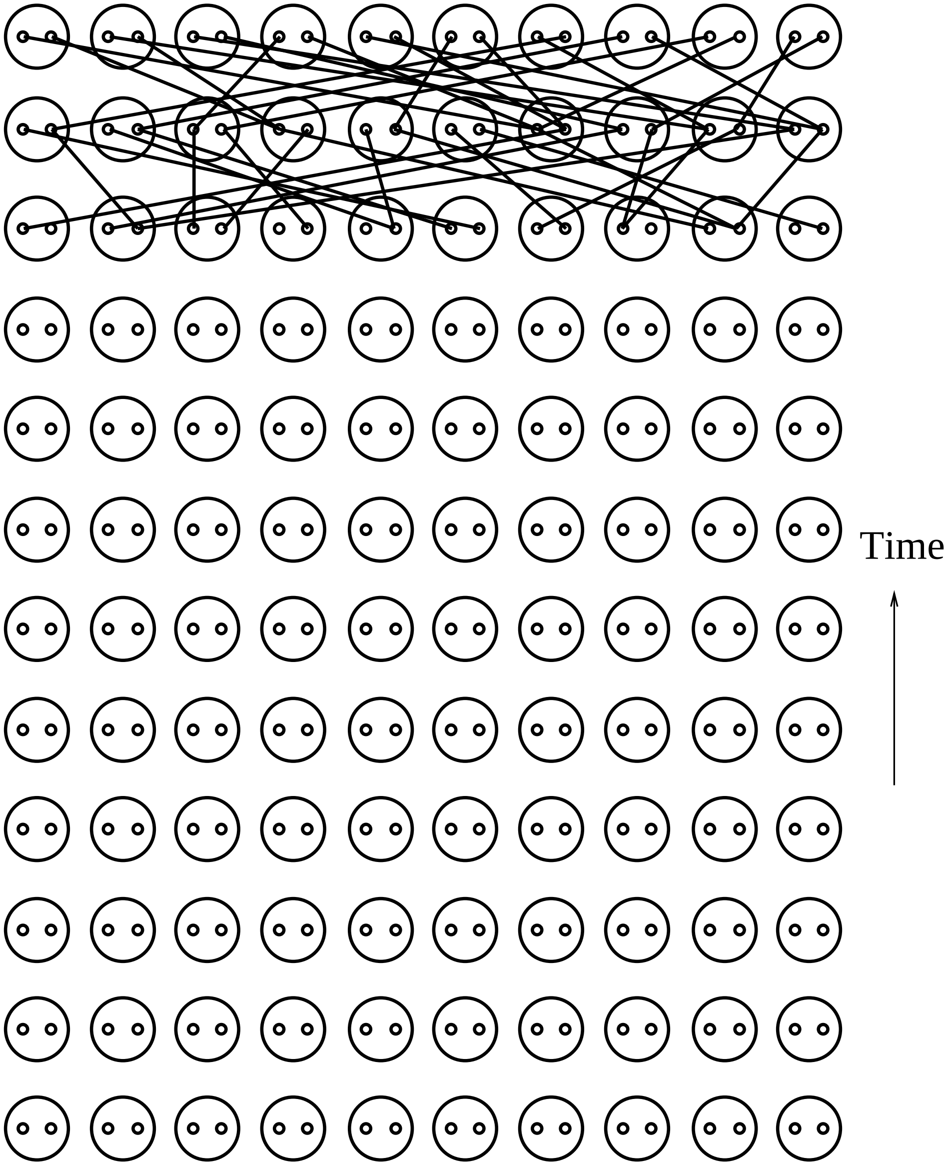
A random−mating population



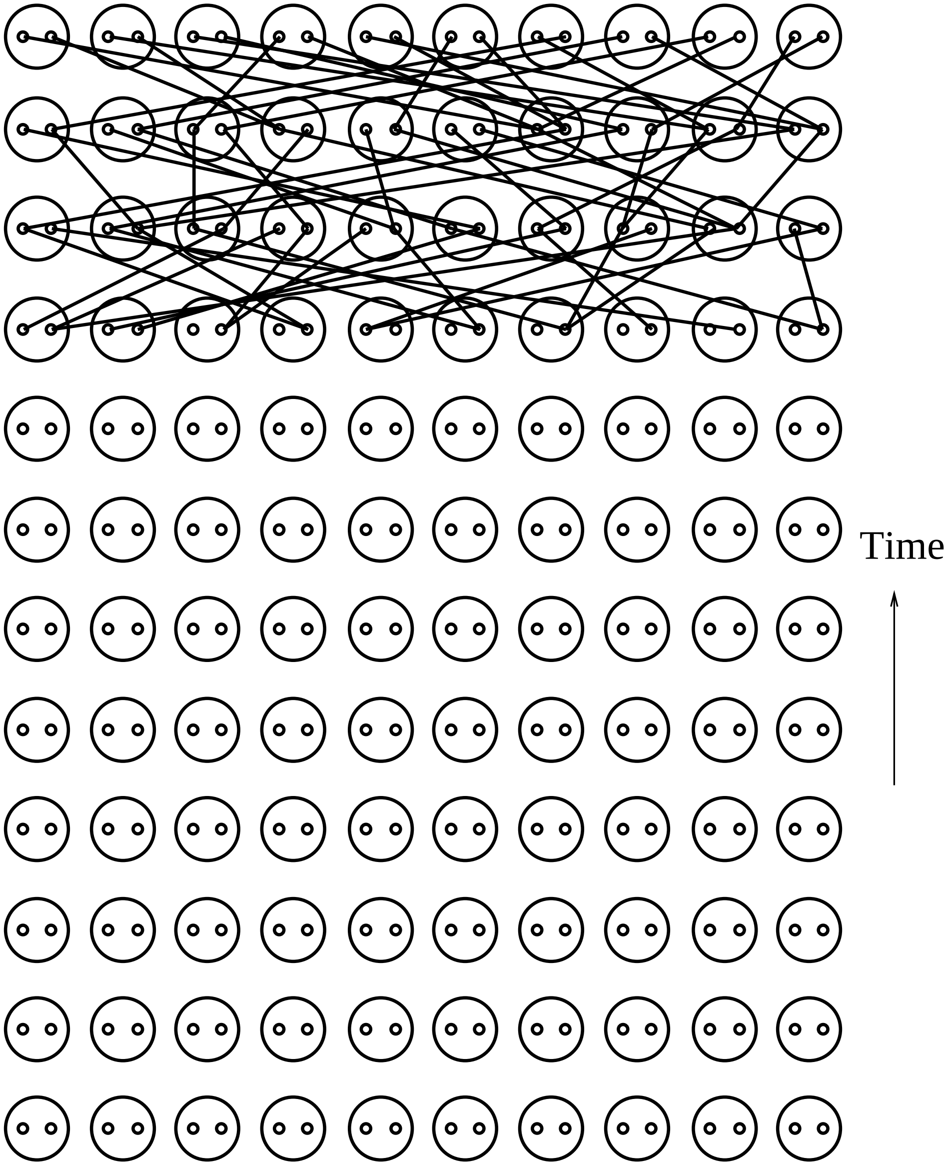
A random−mating population



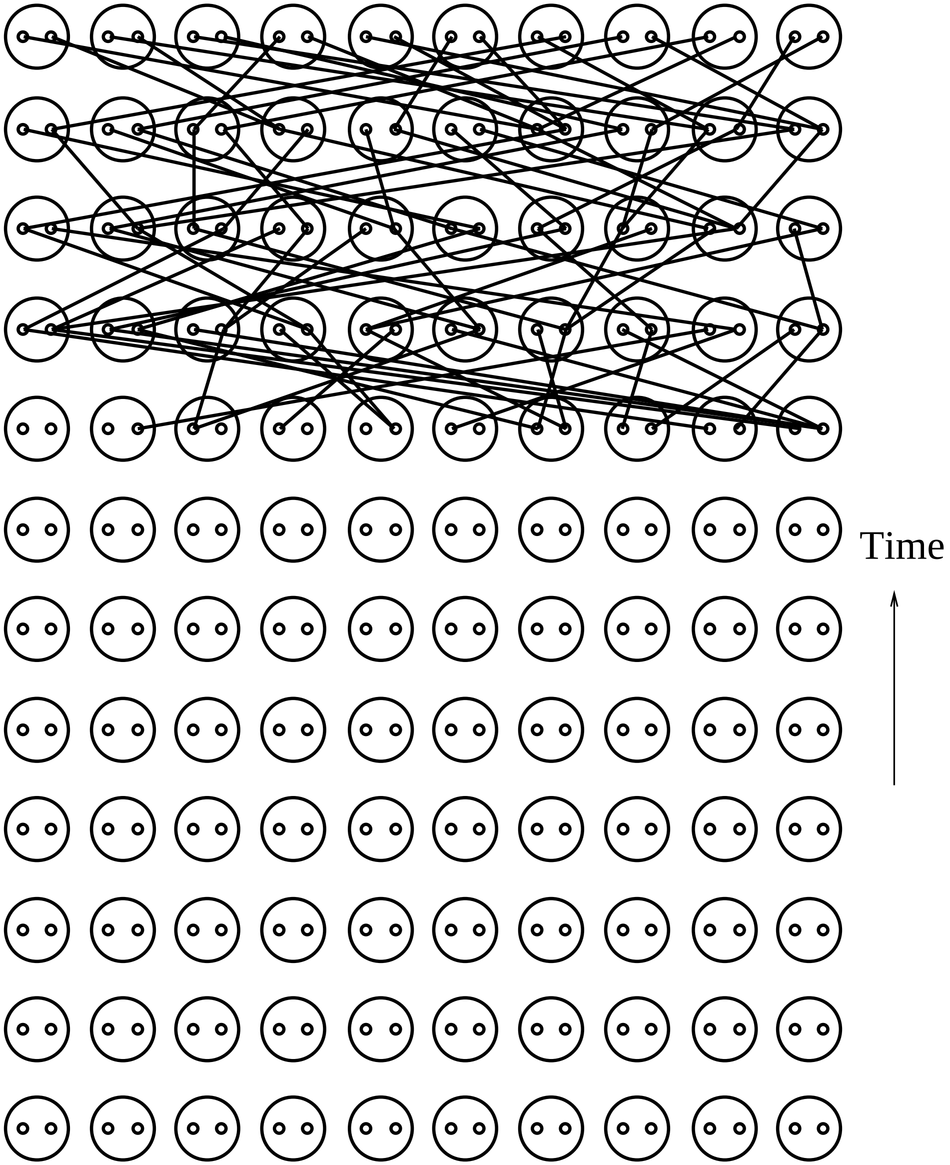
A random−mating population



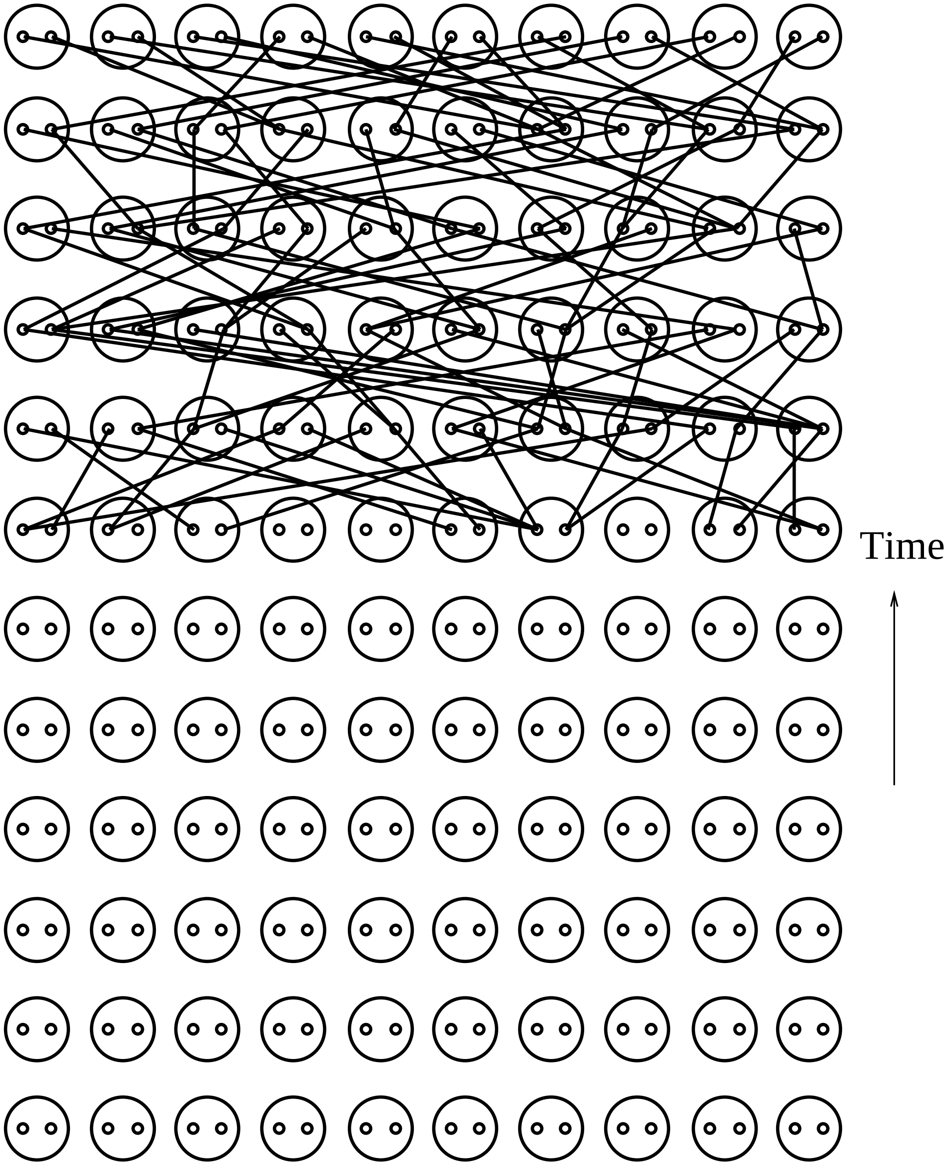
A random−mating population



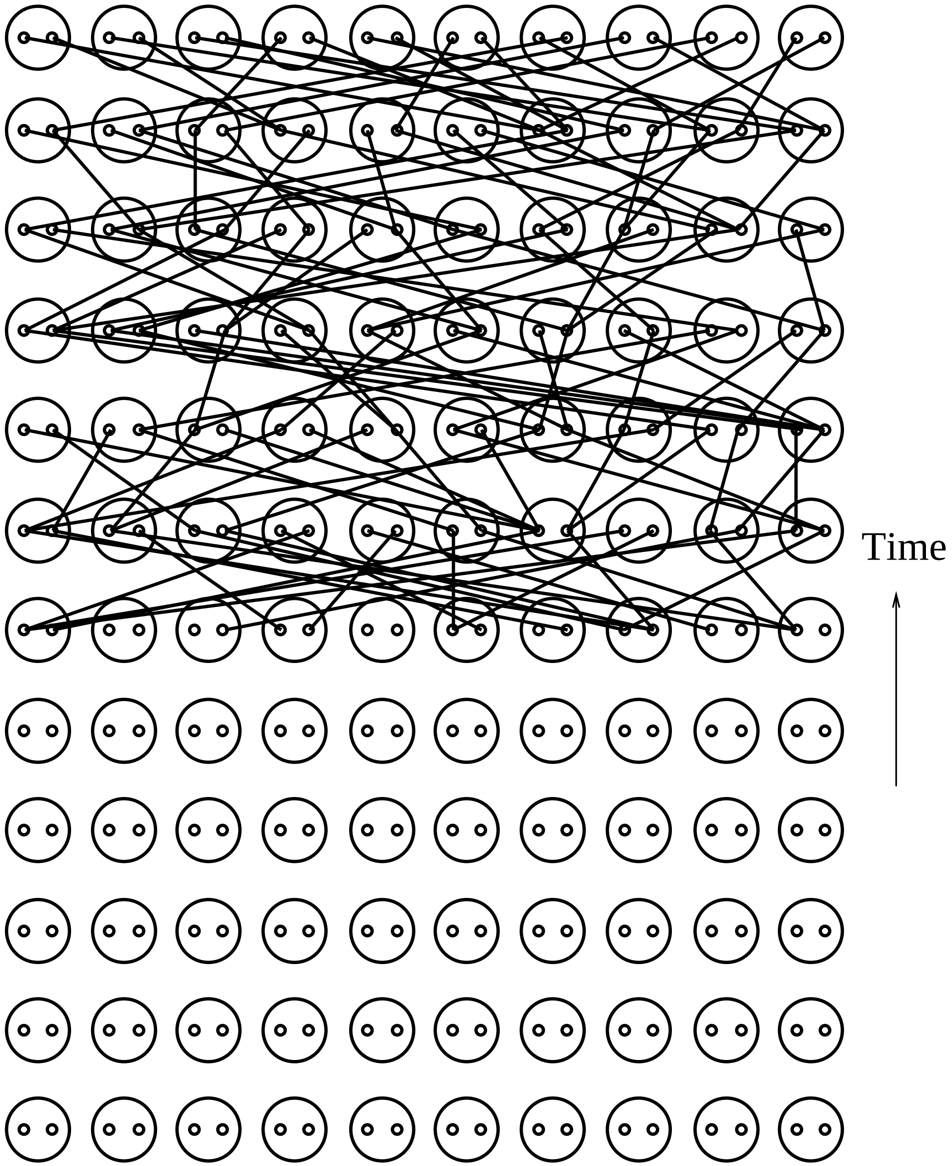
A random−mating population



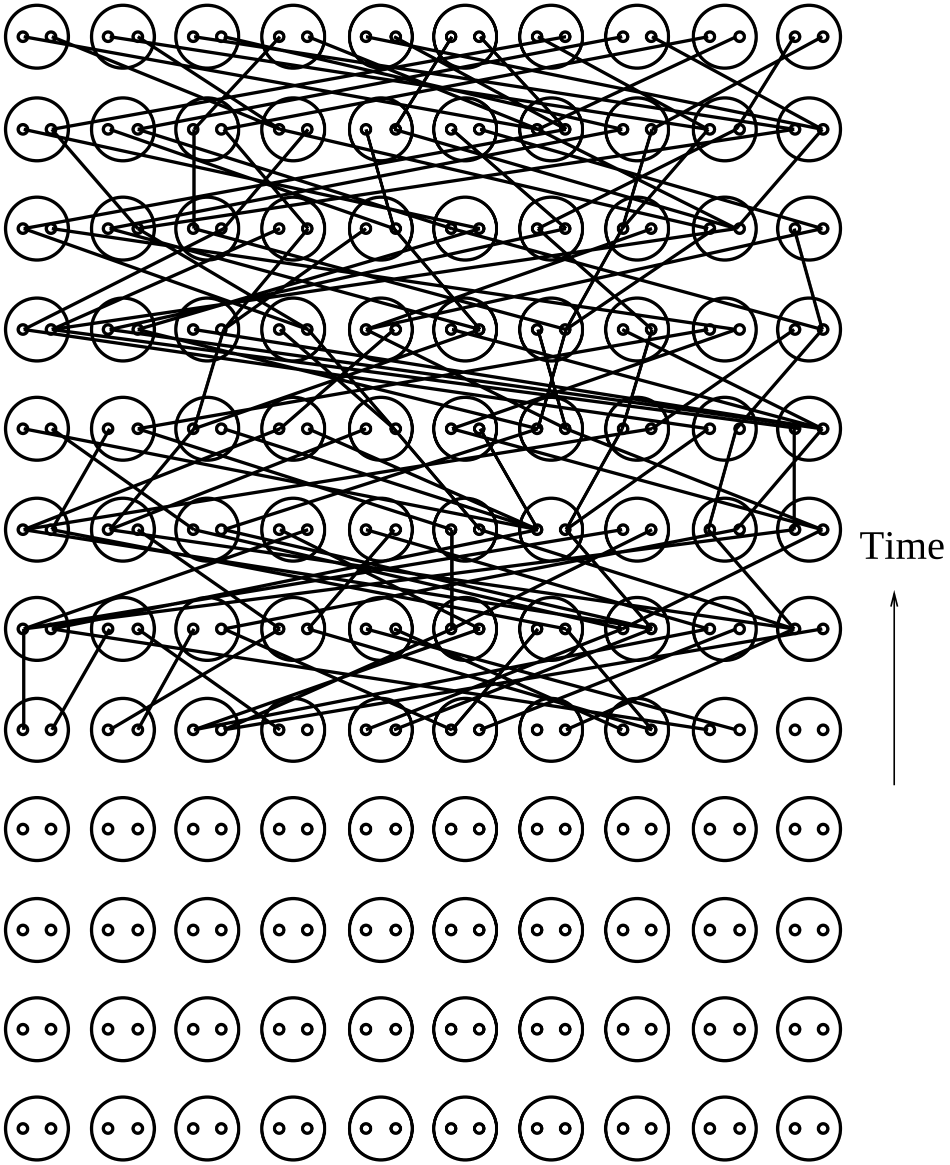
A random−mating population



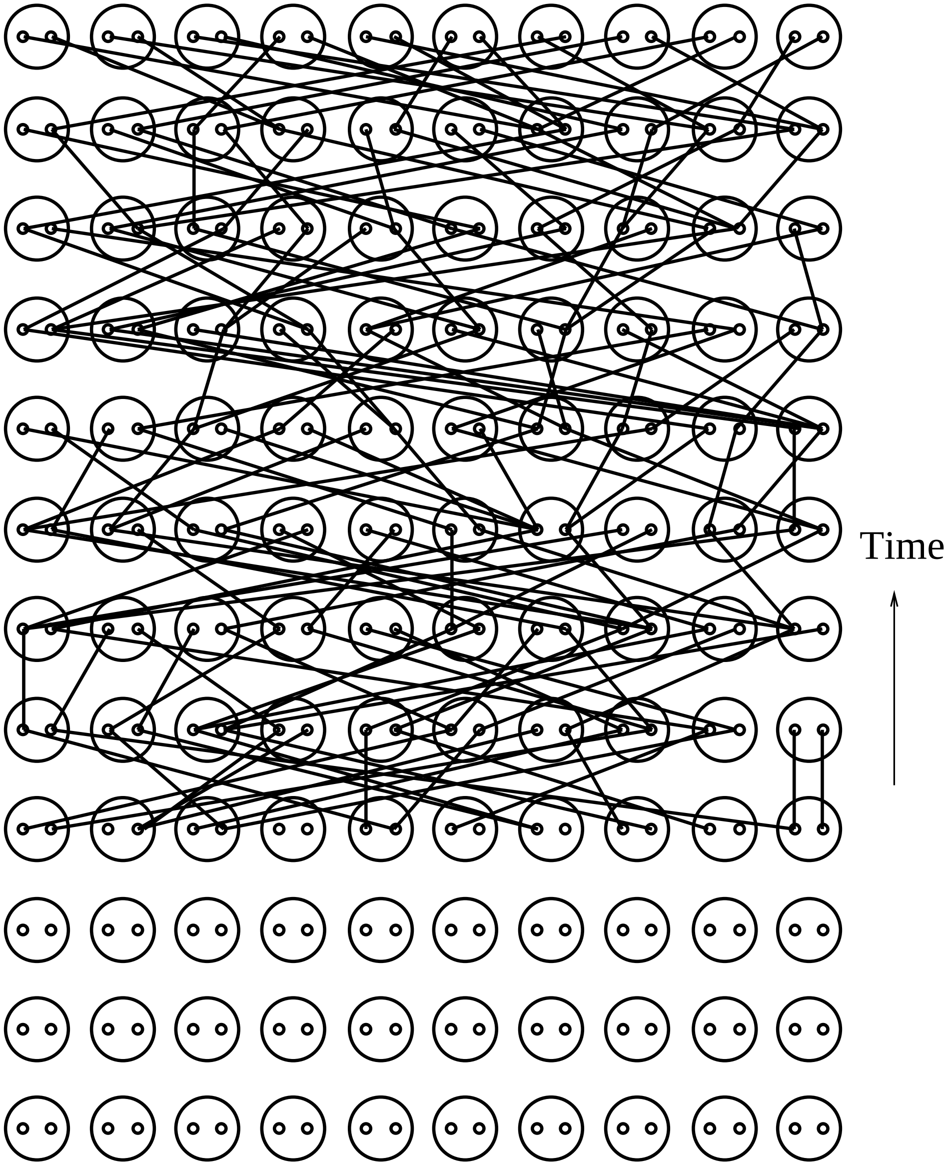
A random−mating population



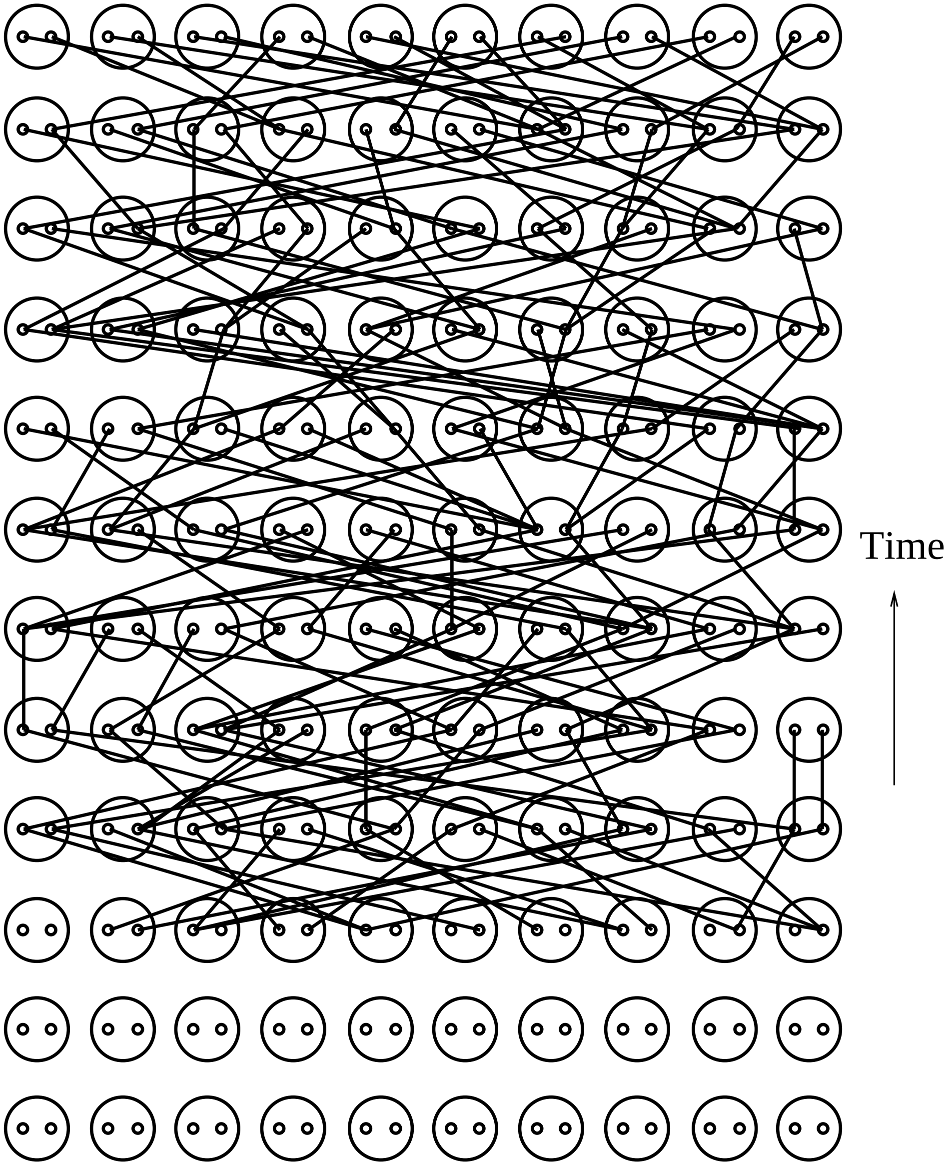
A random−mating population



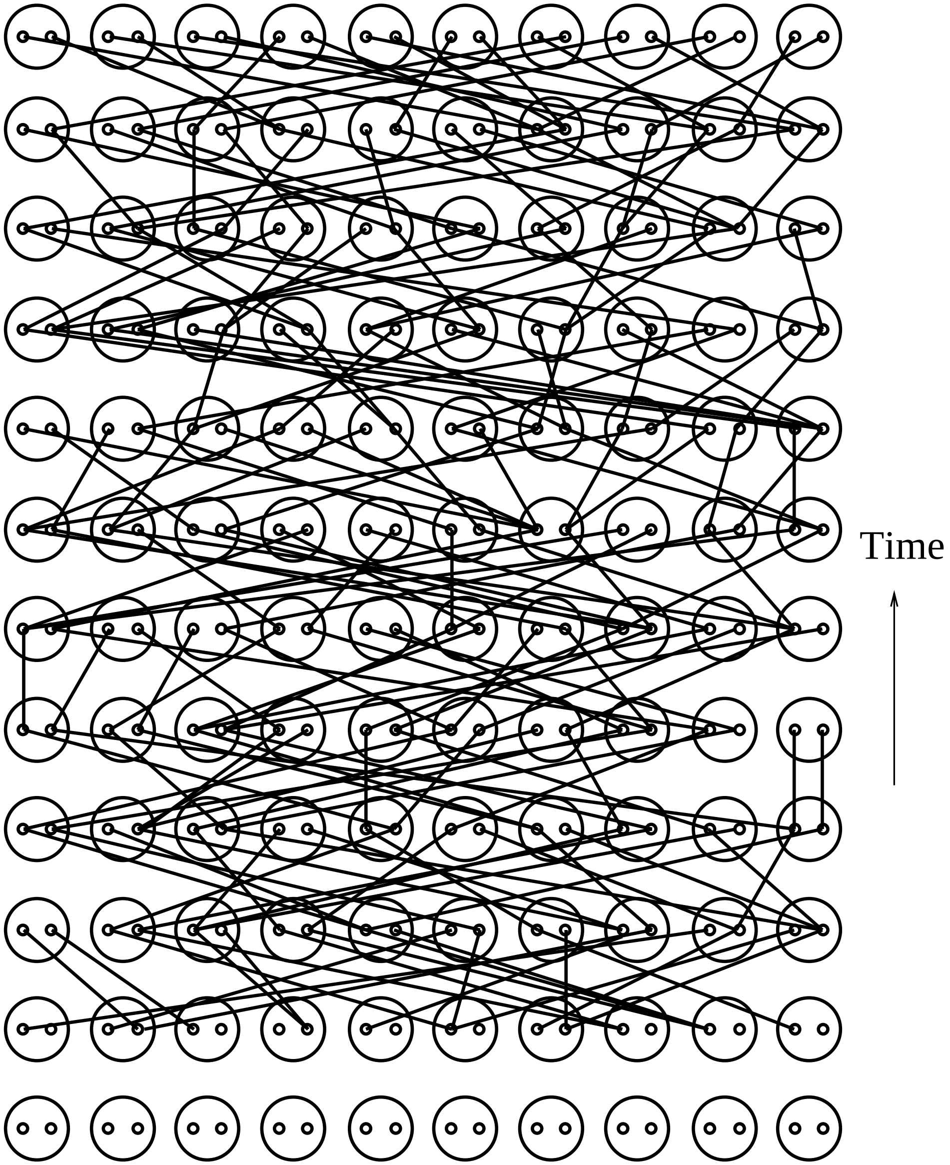
A random−mating population



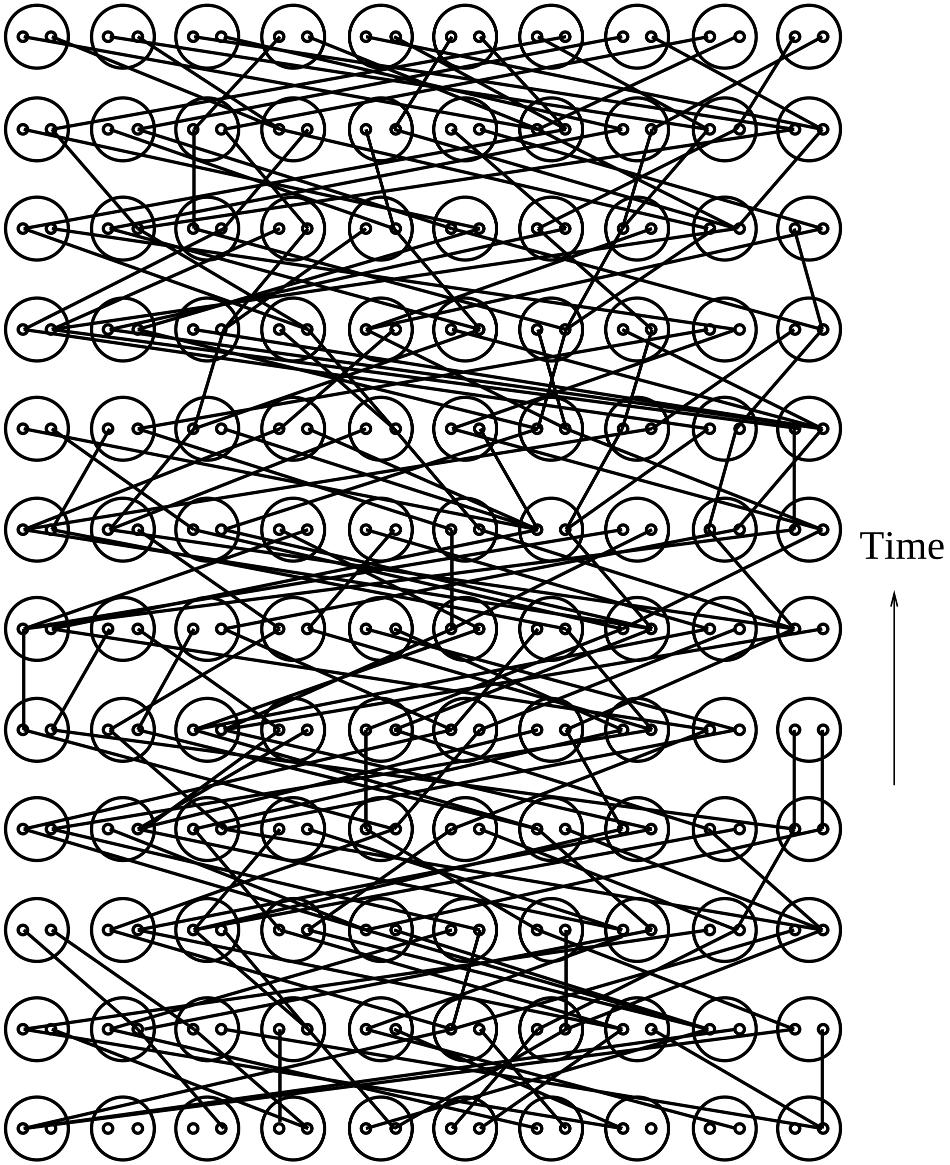
A random−mating population



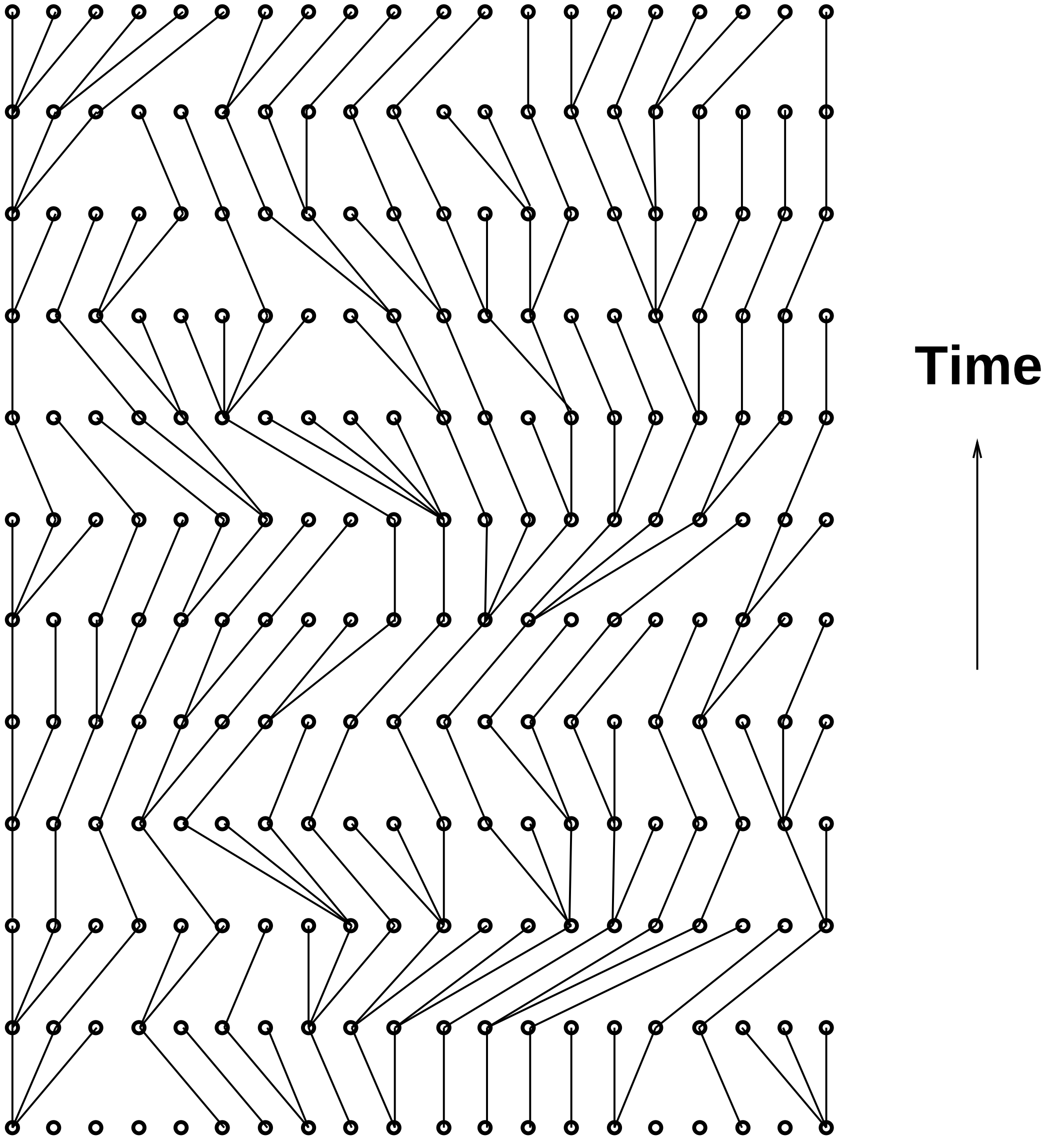
A random−mating population



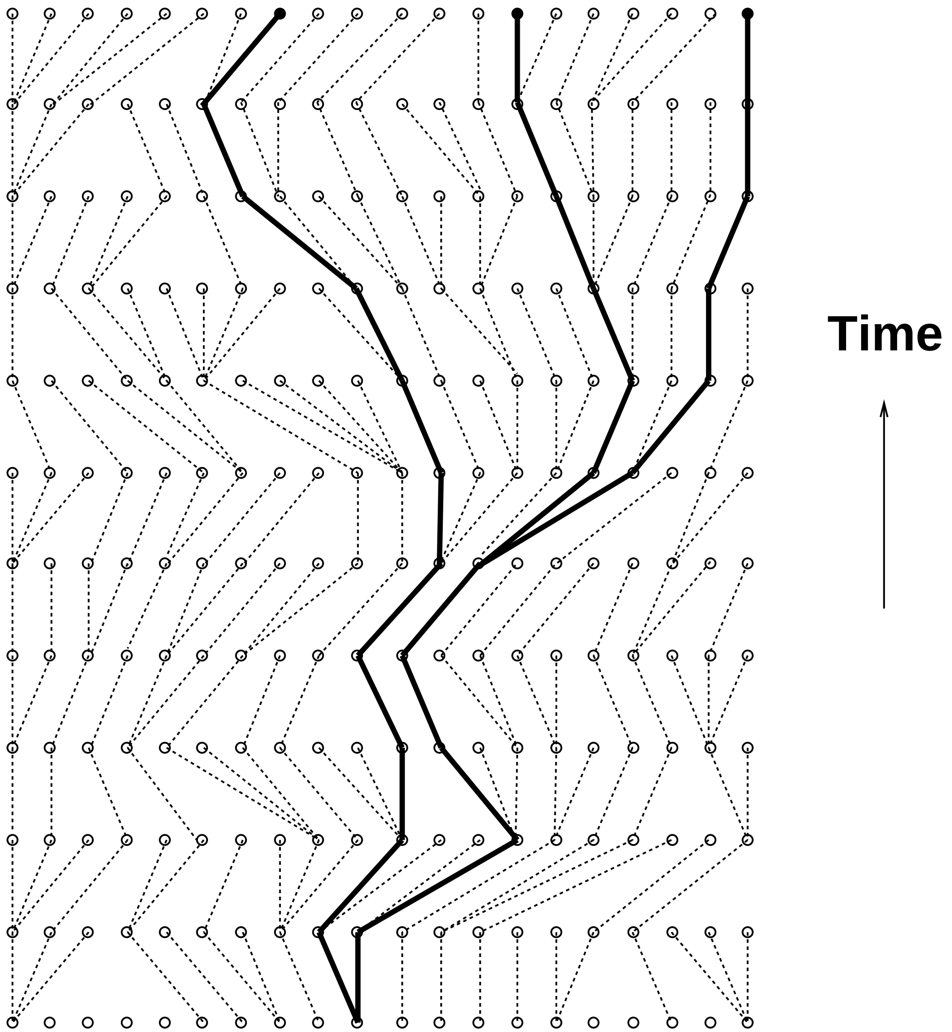
A random−mating population

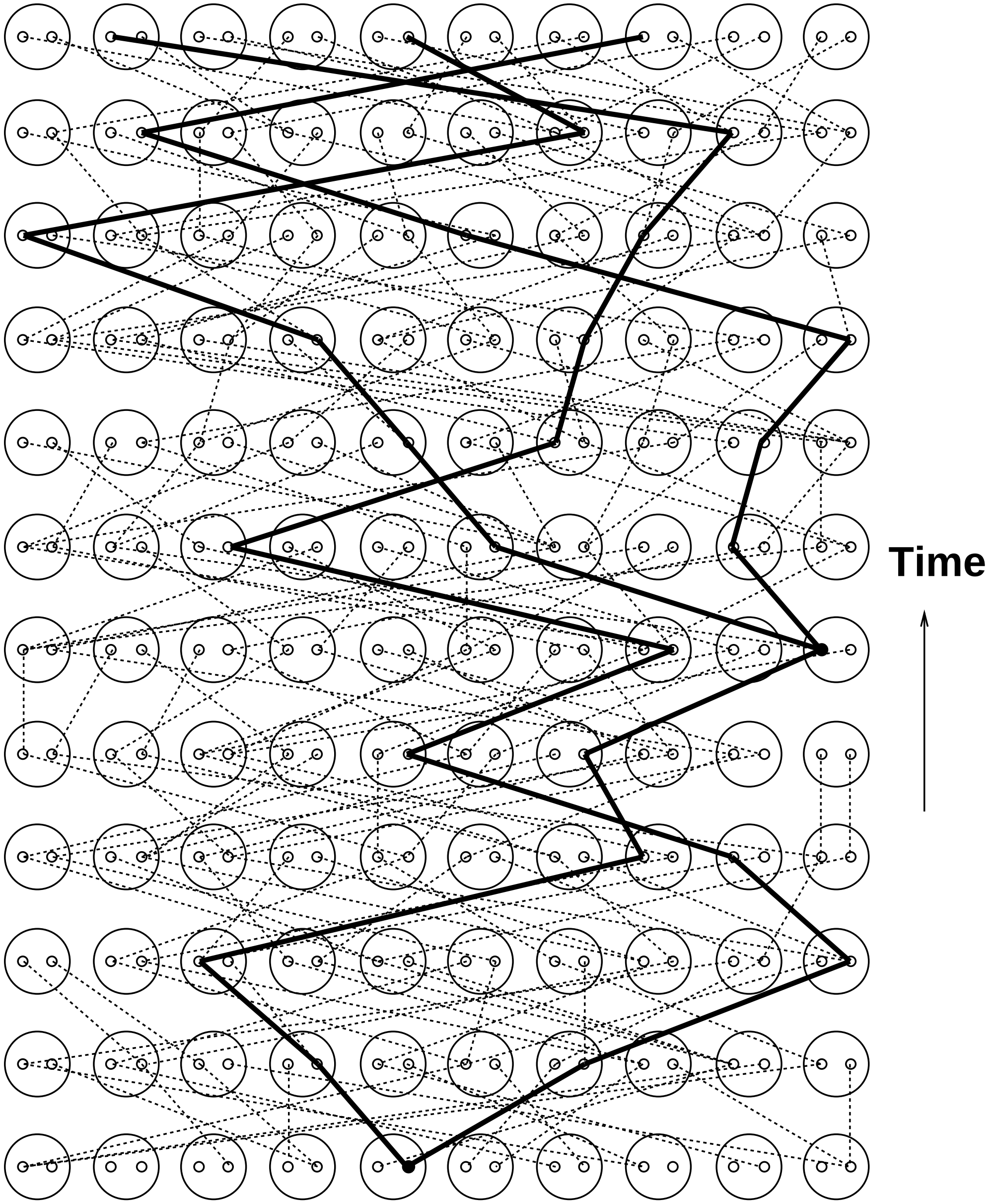


Genealogy of gene copies, after reordering the copies



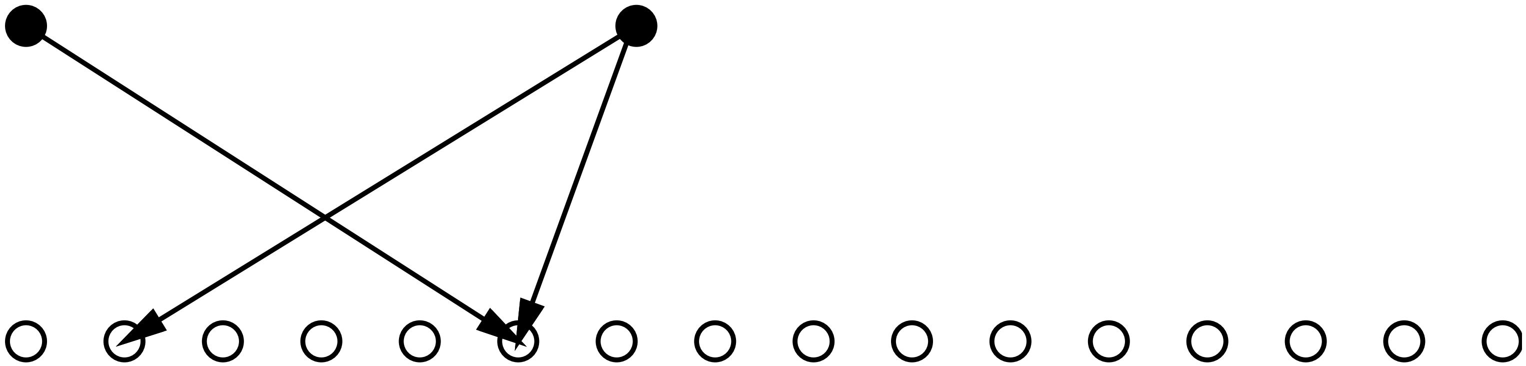
Genealogy of a small sample of genes from the population




Time

under the Wright−Fisher model

each gene comes from a random copy
in the previous generation



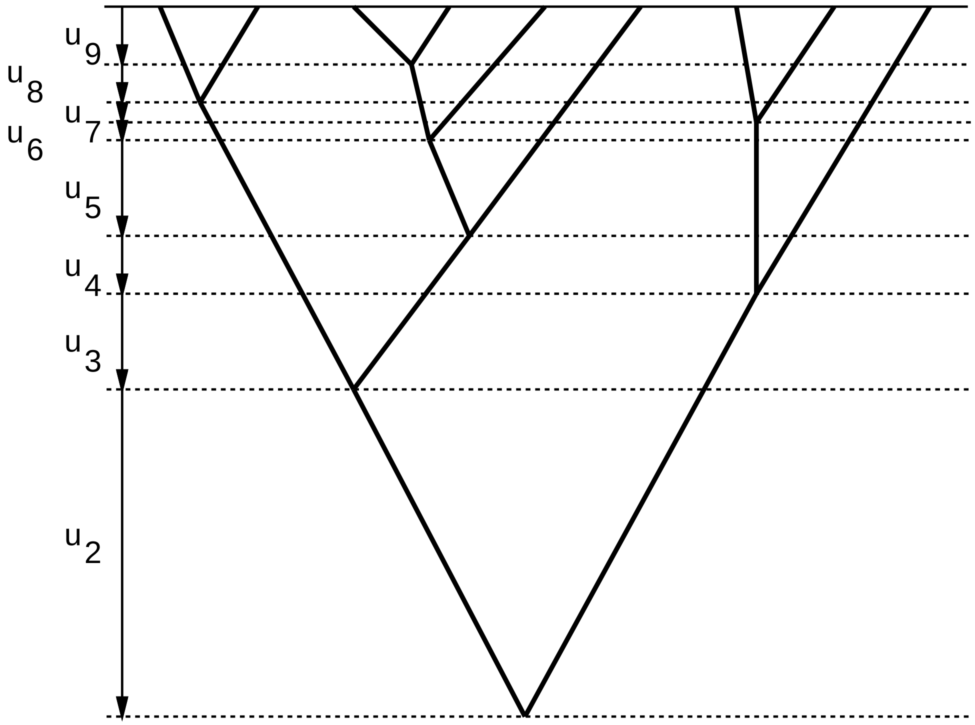
a chance of 1 out of 2N that another
one comes from the same copy

hence it takes about 2N generations for
two lineages to coalesce

Coalescent trees of gene copies within species (Kingman, 1982)

Random collision of lineages as go back in time (sans recombination)

Collision is faster the smaller the effective population size



In a diploid population of effective population size N_e

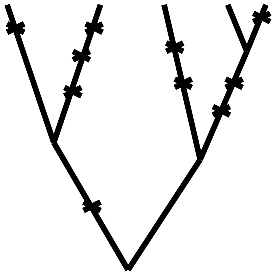
Average time for k copies to coalesce to $k-1$ $= \dfrac{4N_e}{k(k-1)}$

Average time for two copies to coalesce = $2N_e$ generations

Average time for n copies to coalesce = $4N_e \left(1 - \dfrac{1}{n}\right)$ generations

Two levels of variability

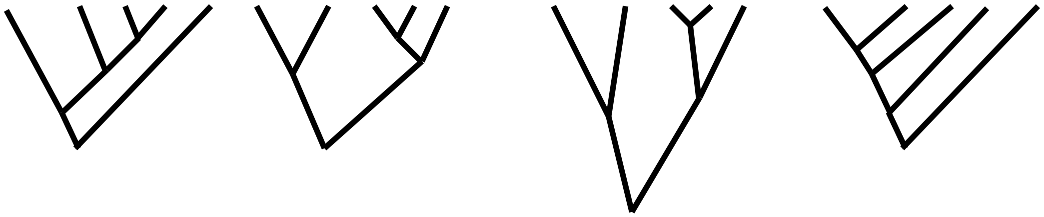
(1) Randomness of mutation



affected by the mutation rate u

can reduce variance of number of mutations per site per branch by examining more sites

(2) Randomness of coalescence of lineages



affected by effective population size N_e

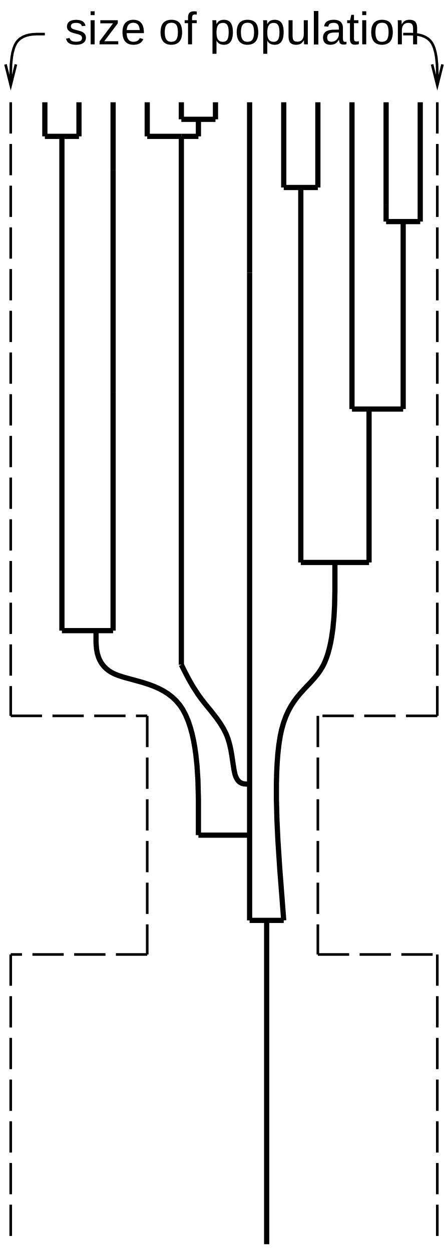
coalescence times allow estimation of N_e

can reduce variability by looking at

(i) more gene copies, or

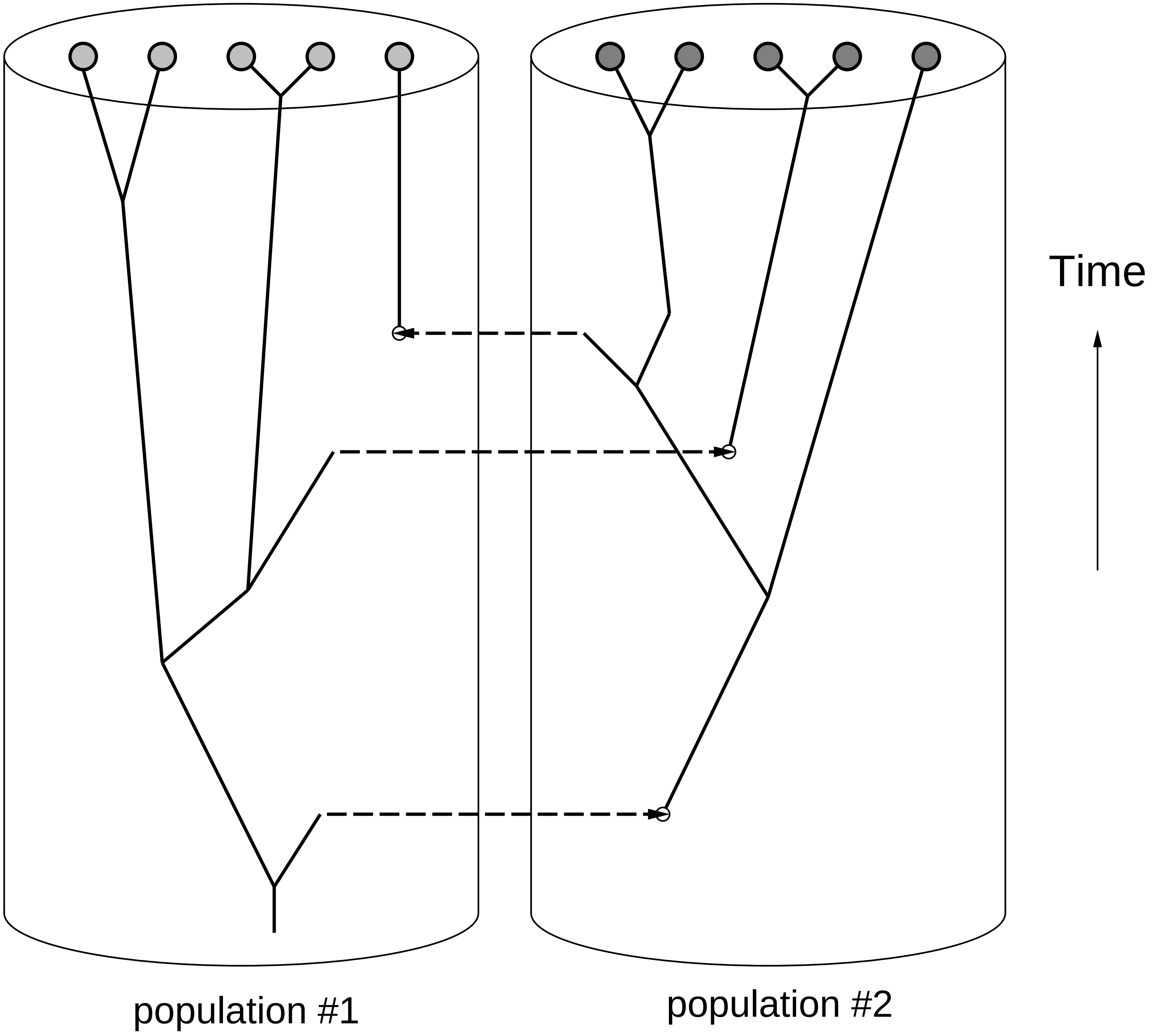
(ii) more loci

Coalescent with population size changing through time



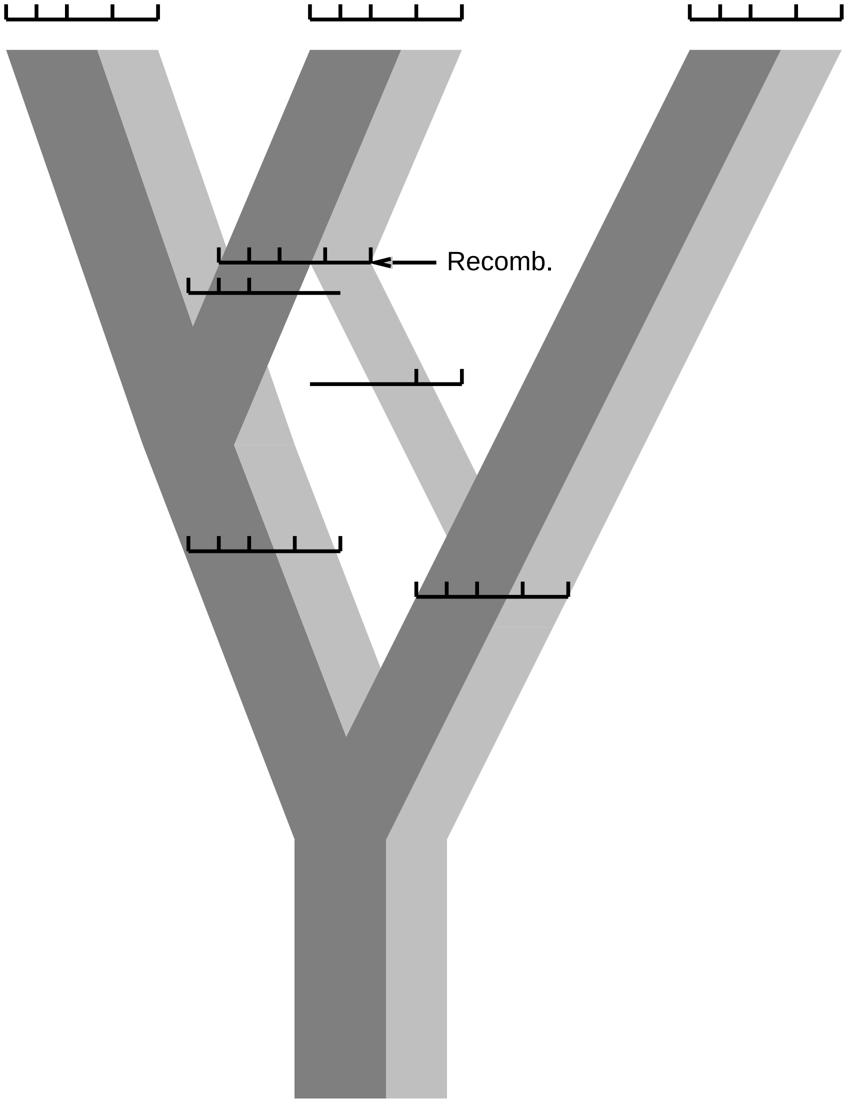
During a population bottleneck there is expected to be a burst of coalescence

Down near the root of the tree effects of population size become difficult to see

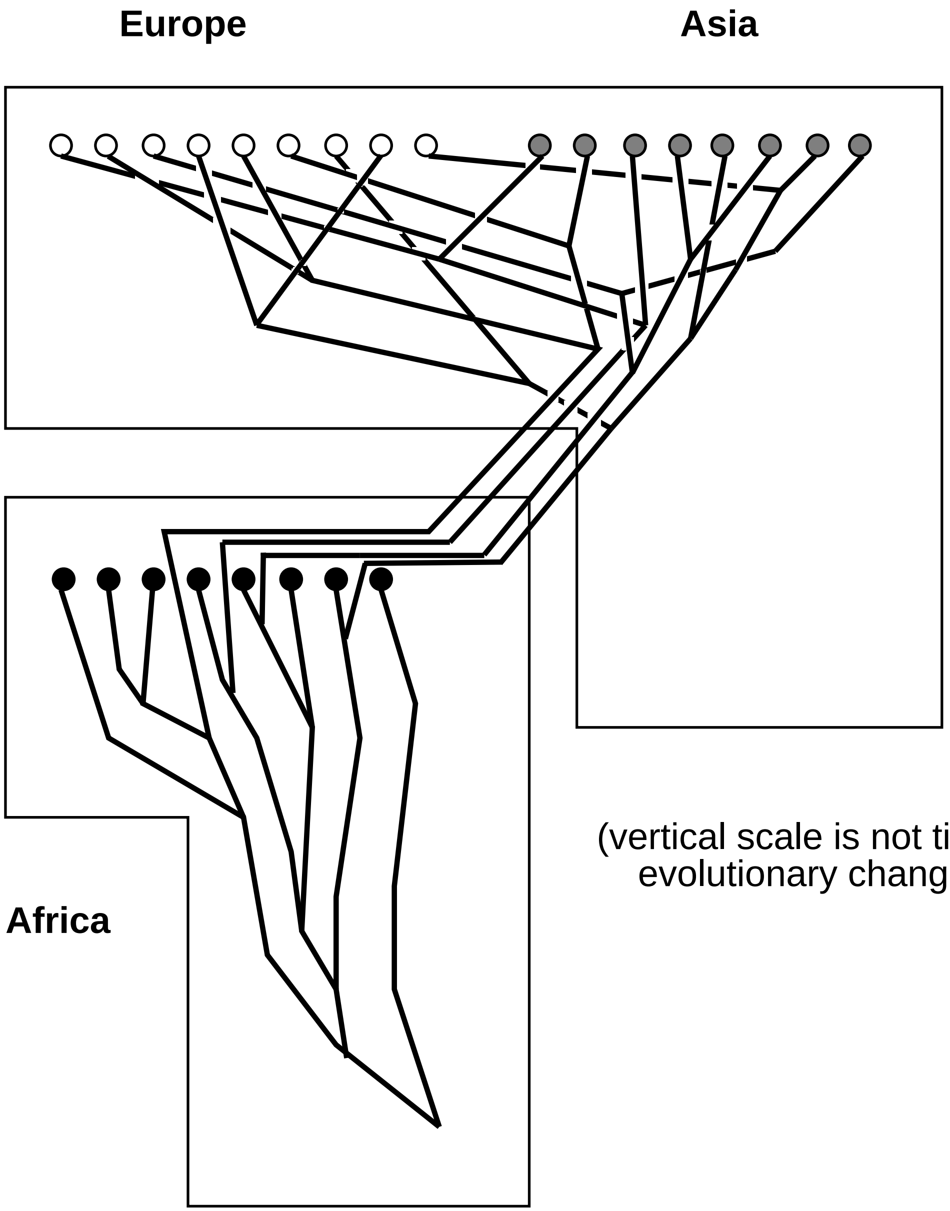

Time
population #1
population #2

A recombining coalescent

Different markers have slightly different coalescent trees



"Out of Africa" hypothesis

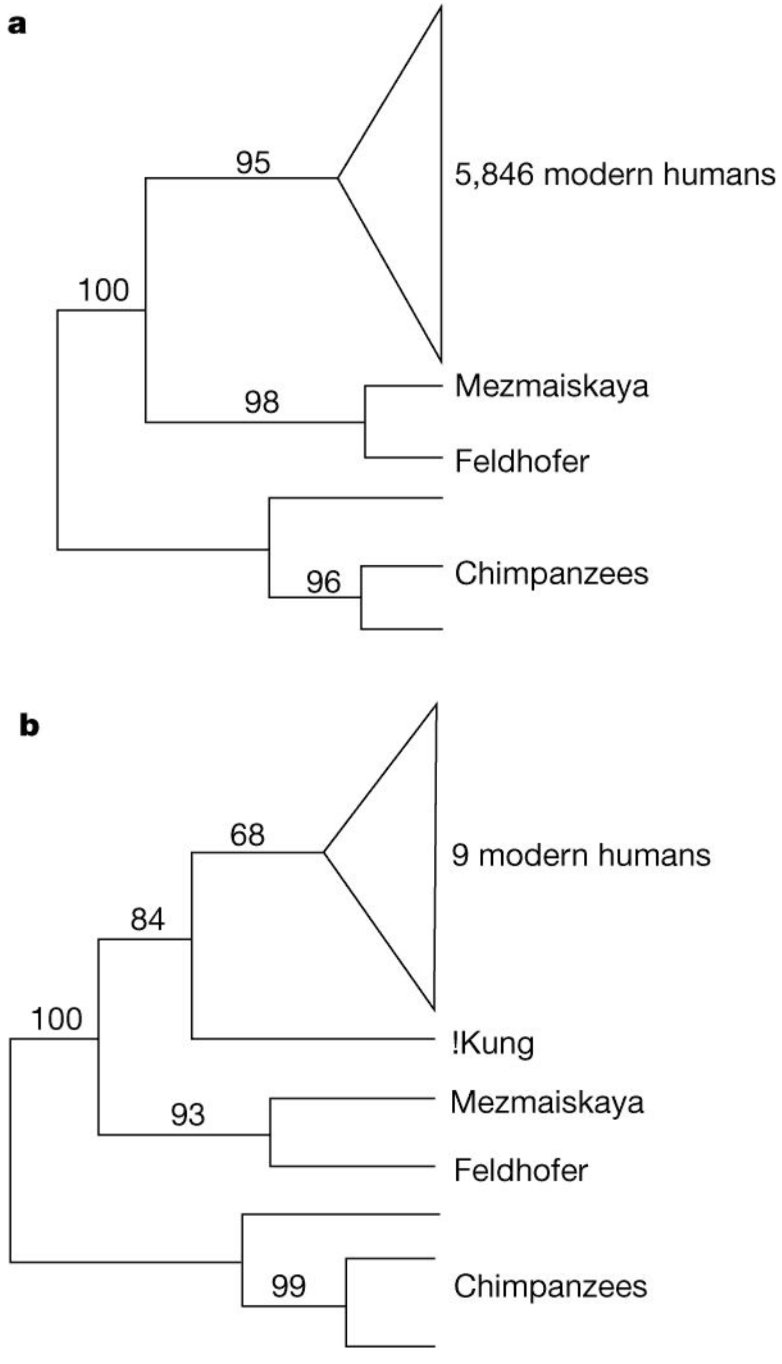


(vertical scale is not time or evolutionary change)

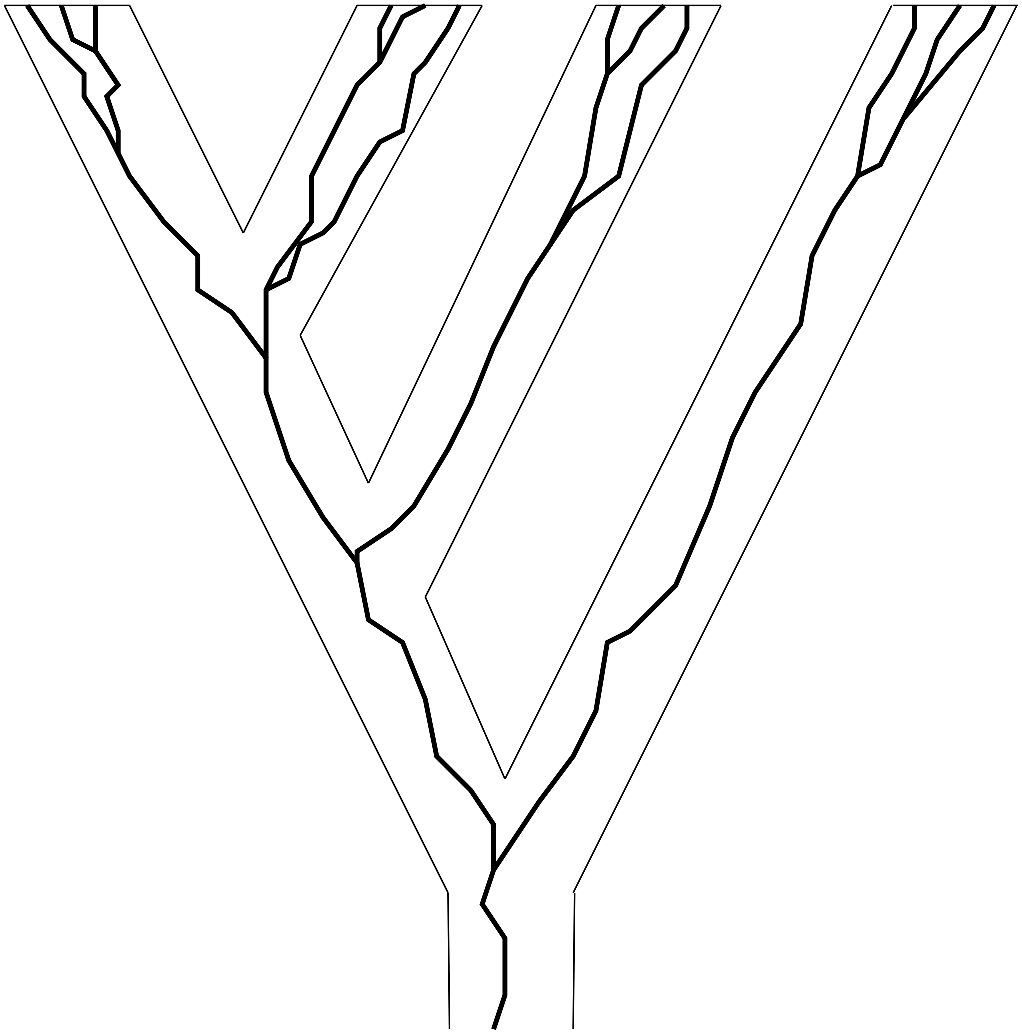
Svante Pääbo

Krings, M., H. Geisert, R. W. Schmitz, H. Krainitzki, and S. Paabo. 1999. DNA sequence of the mitochondrial hypervariable region II from the neandertal type specimen. *Proceedings of the Natonal Academy of Sciences USA* **96:** 5581-5585.

Ovchinnikov, I. V., A. Gotherstrom, G. P. Romanova, V. M. Kharitonov, K. Liden, and W. Goodwin. 2000. Molecular analysis of Neanderthal DNA from the northern Caucasus. *Nature* **404:** 490-493.

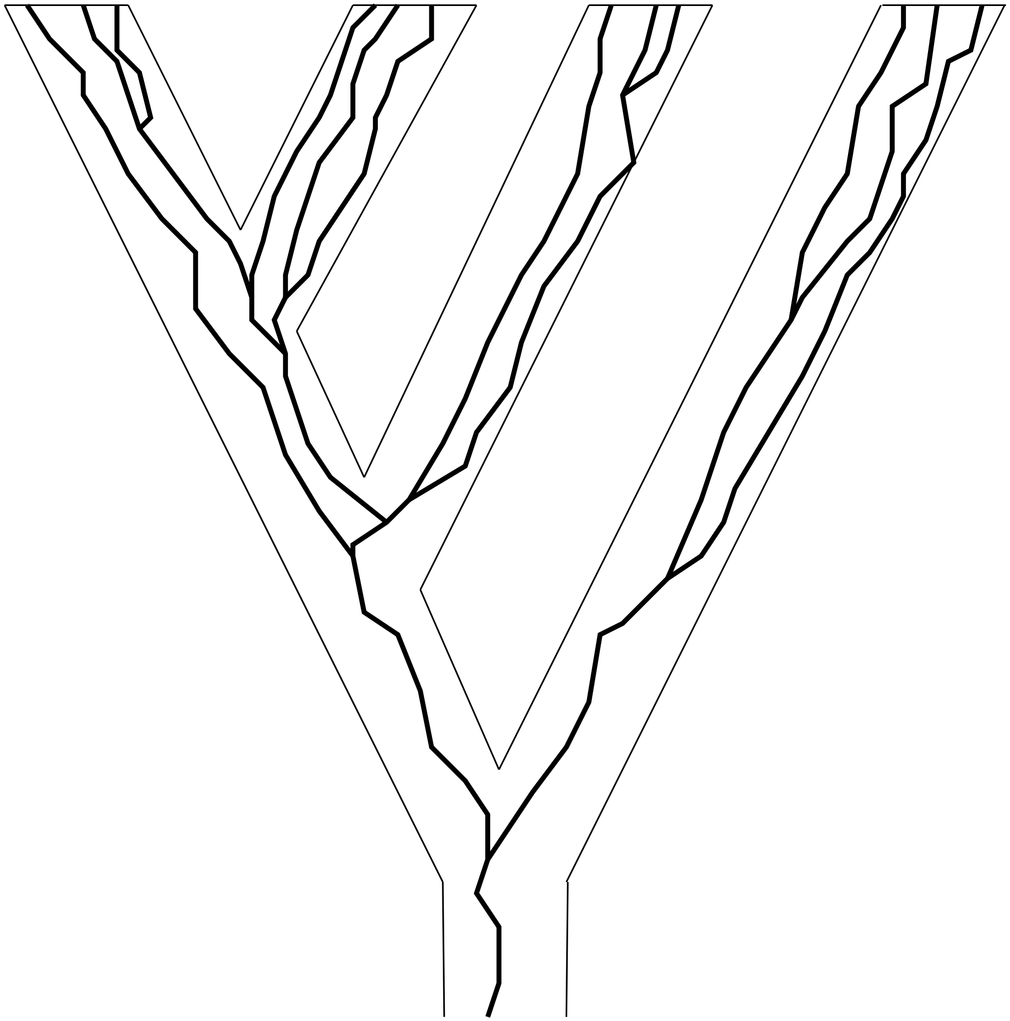

a
95
5,846 modern humans
100
98
Mezmaiskaya
Feldhofer
96
Chimpanzees
b
68
9 modern humans
84
100
!Kung
93
Mezmaiskaya
Feldhofer
99
Chimpanzees

Tree of gene copies, compared with the phylogeny of the species
for the case in which effective population size is small
compared to the number of generations between speciations

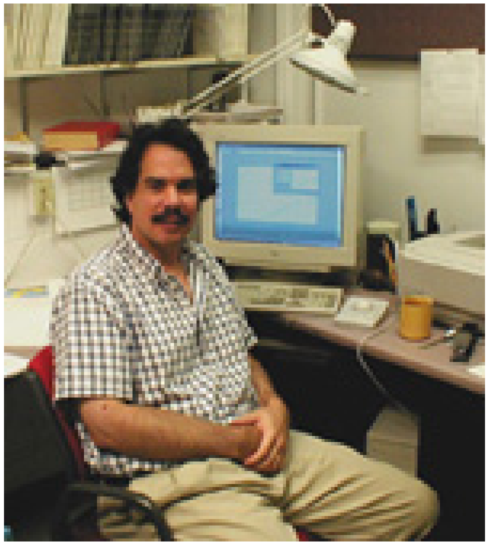


The tree of gene copies has topology consistent with the phylogeny

Tree of gene copies, compared with the phylogeny of the species
for the case in which effective population size is large
compared to the number of generations between speciations



The tree of gene copies has topology inconsistent with the phylogeny



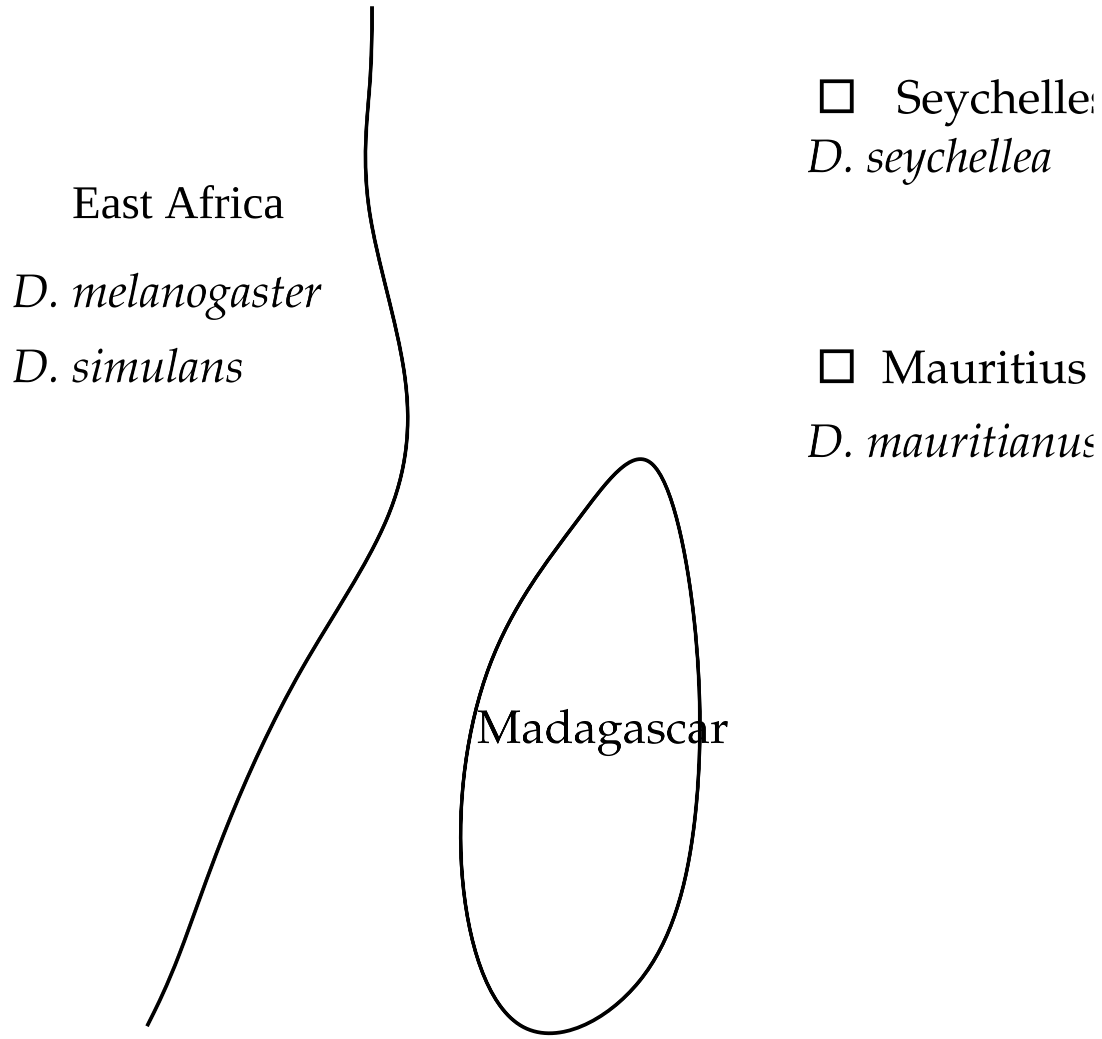
Jody Hey

Rich Kliman and friend

Kliman, R. M., and J. Hey. 1993. DNA sequence variation at the period locus within and among species of the *Drosophila melanogaster* complex. *Genetics* **133:** 375-87

Hey, J. and R. M. Kliman. 1993. Population genetics and phylogenetics of DNA sequence variation at multiple loci within the Drosophila melanogaster species complex. *Molecular Biology Evolution* **10:** 804-822.

East African species related to *D. melanogaster*



East Africa

D. melanogaster

D. simulans

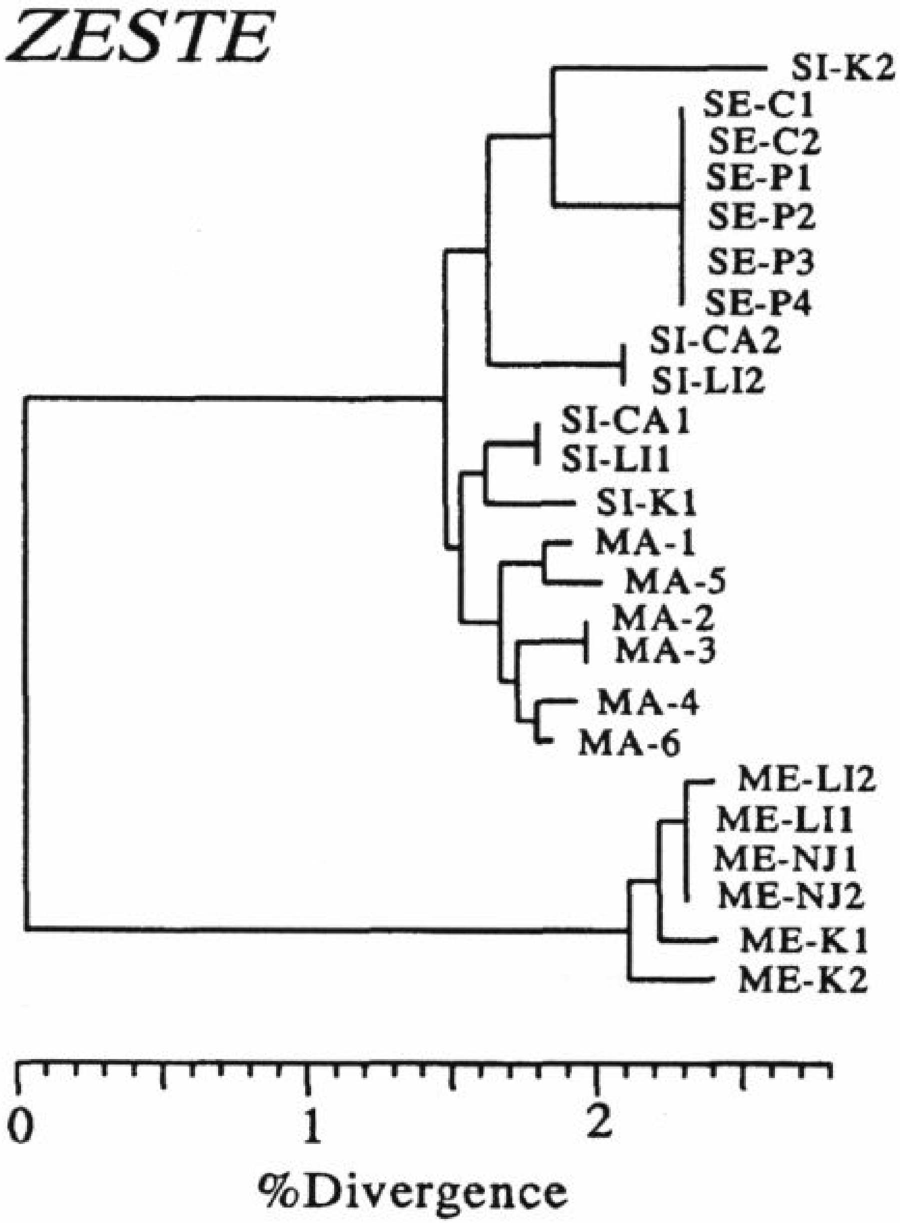
☐ Seychelles

D. seychellea

☐ Mauritius

D. mauritianus

Madagascar


ZESTE
SI-K2
SE-C1
SE-C2
SE-P1
SE-P2
SE-P3
SE-P4
SI-CA2
SI-LI2
SI-CA1
SI-LI1
SI-K1
MA-1
MA-5
MA-2
MA-3
MA-4
MA-6
ME-LI2
ME-LI1
ME-NJ1
ME-NJ2
ME-K1
ME-K2
0
1
2
%Divergence

This freeware-friendly presentation prepared with

- Linux (operating system)
- PDFLaTeX (mathematical typesetting and PDF preparation)
- Idraw (drawing program to modify plots and draw figures)
- Adobe Acrobat Reader (to display the PDF in full-screen mode)

(except that we had to use Microsoft Windows to project this as the X server I have in Linux is not too great)