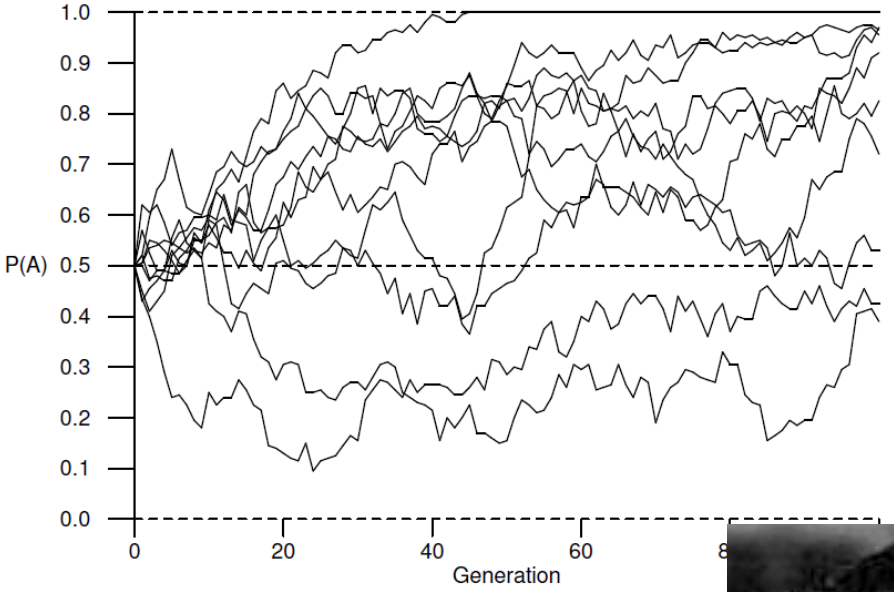
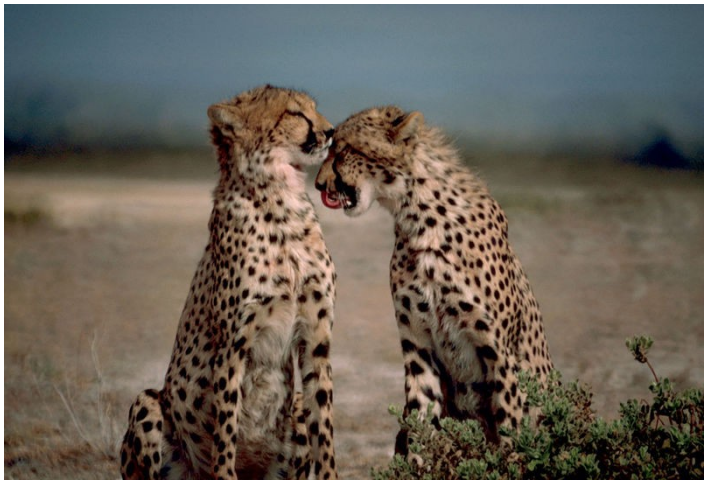
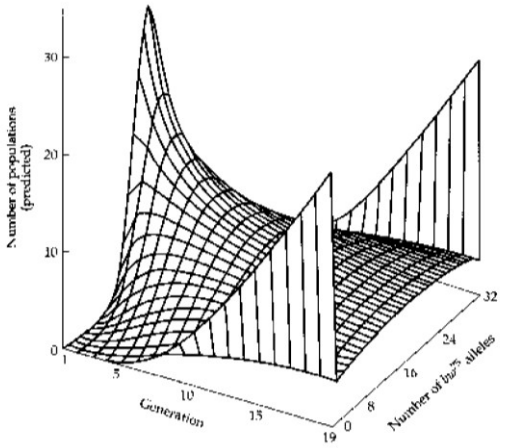
RANDOM GENETIC DRIFT







HW: infinite population but in real world population sizes finite
$\Rightarrow$ random processes, nonadaptive evolution

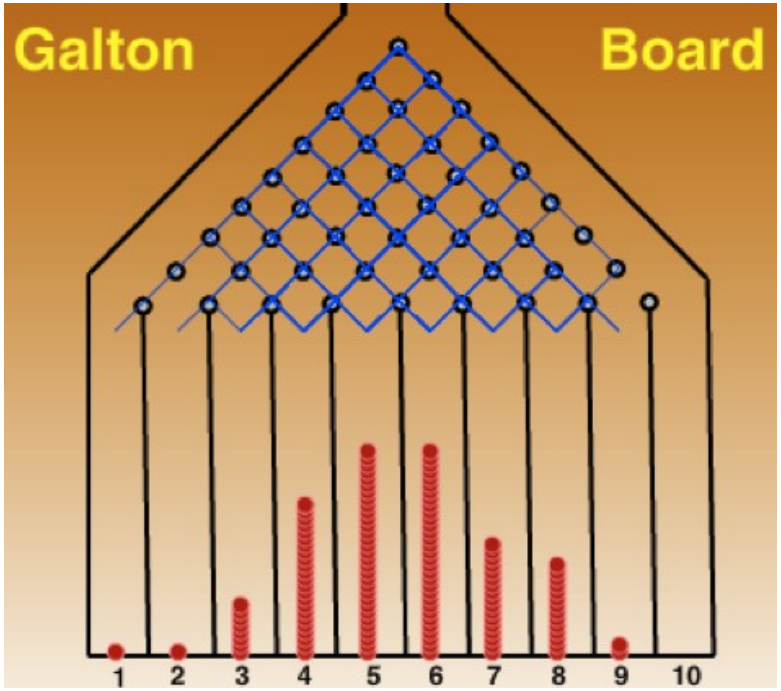
Why randomness?

when number of repetitions finite probability of an event $\neq$ its frequency
(cf. H-W principle)

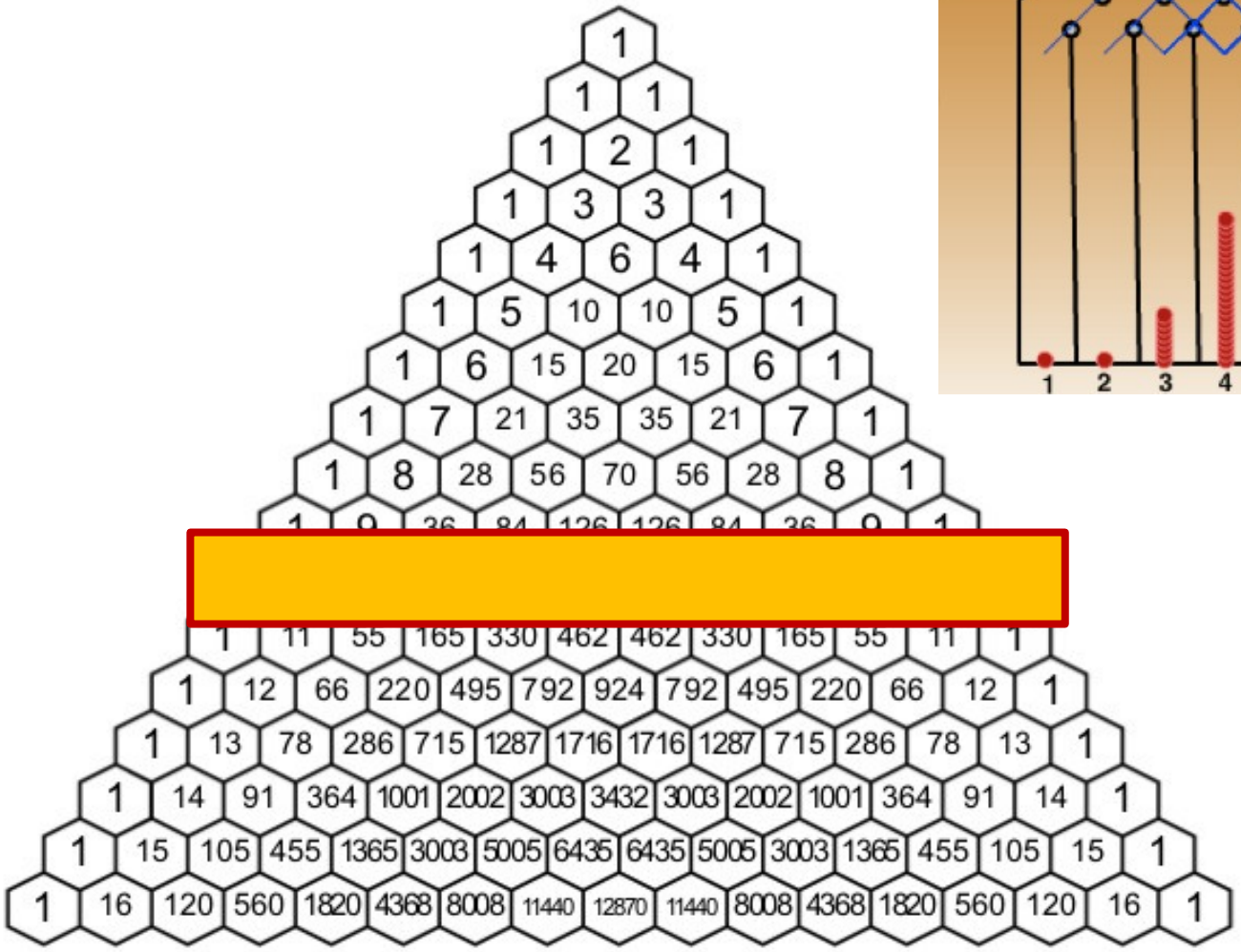
10 coins $\rightarrow$ in more than 75 % cases the ratio differs from 1 : 1

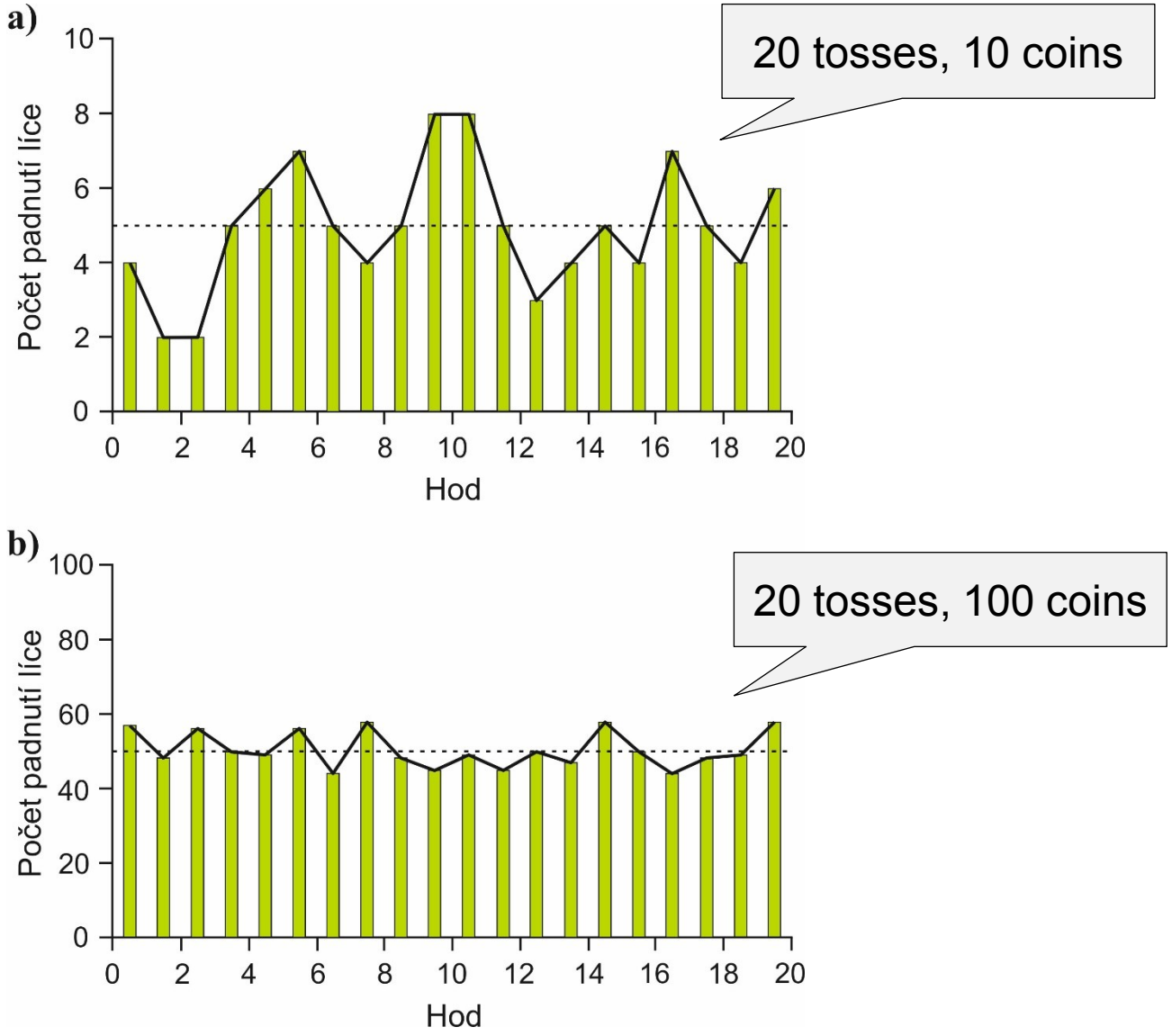
Pascal s triangle:

11 possible results



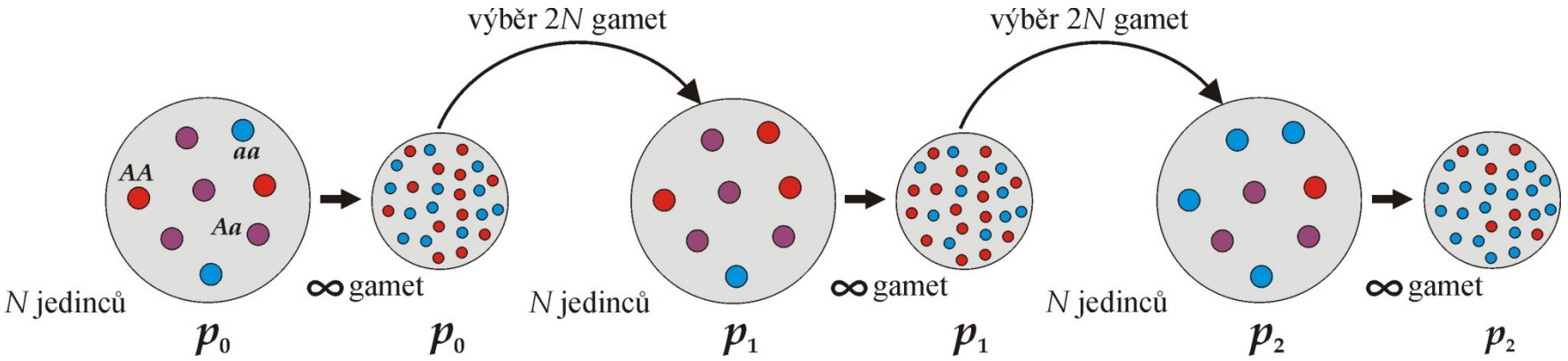
quincunx





With more coins lower variance around expected value

Random sampling from gene pool (sampling error):

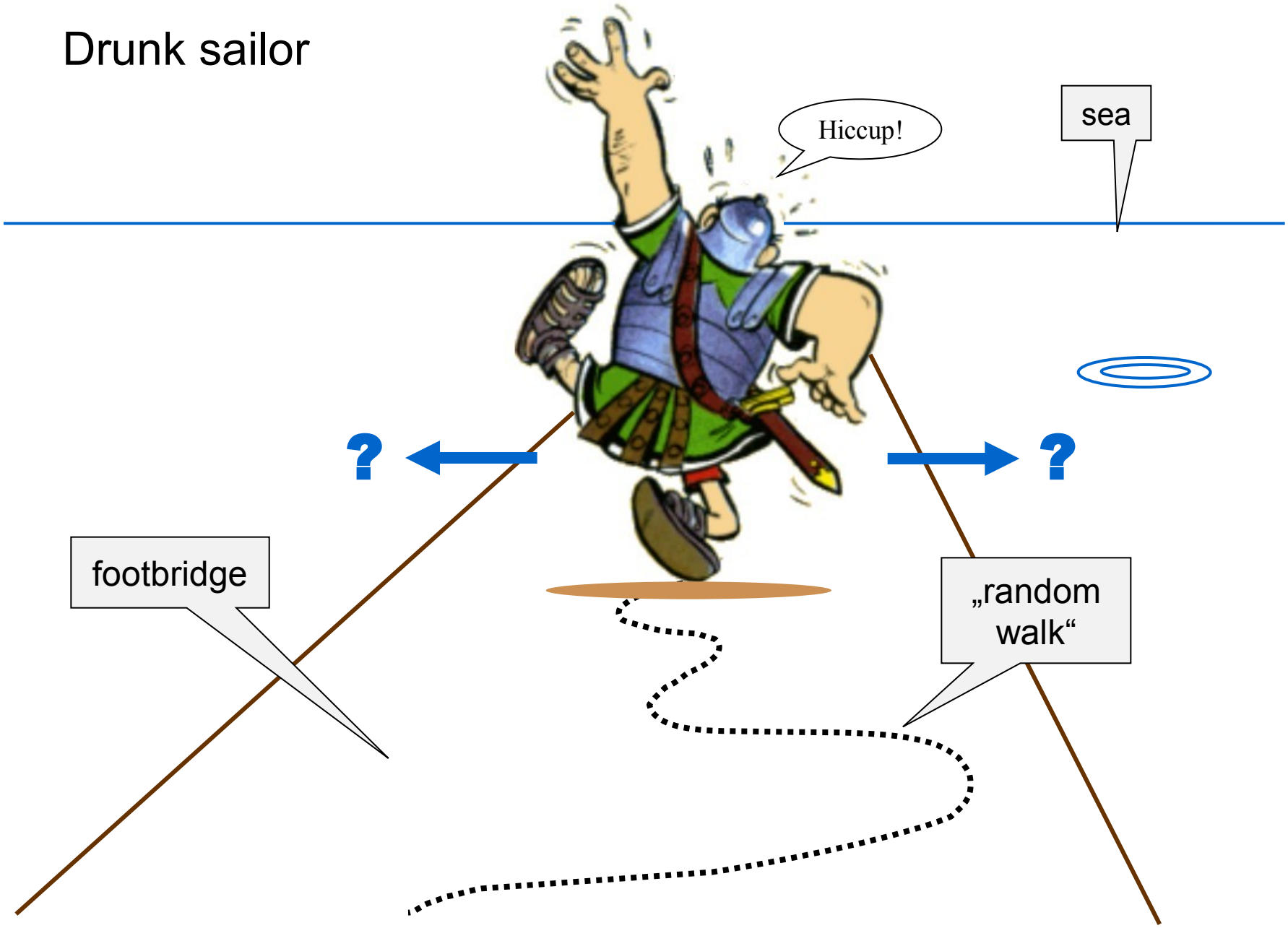


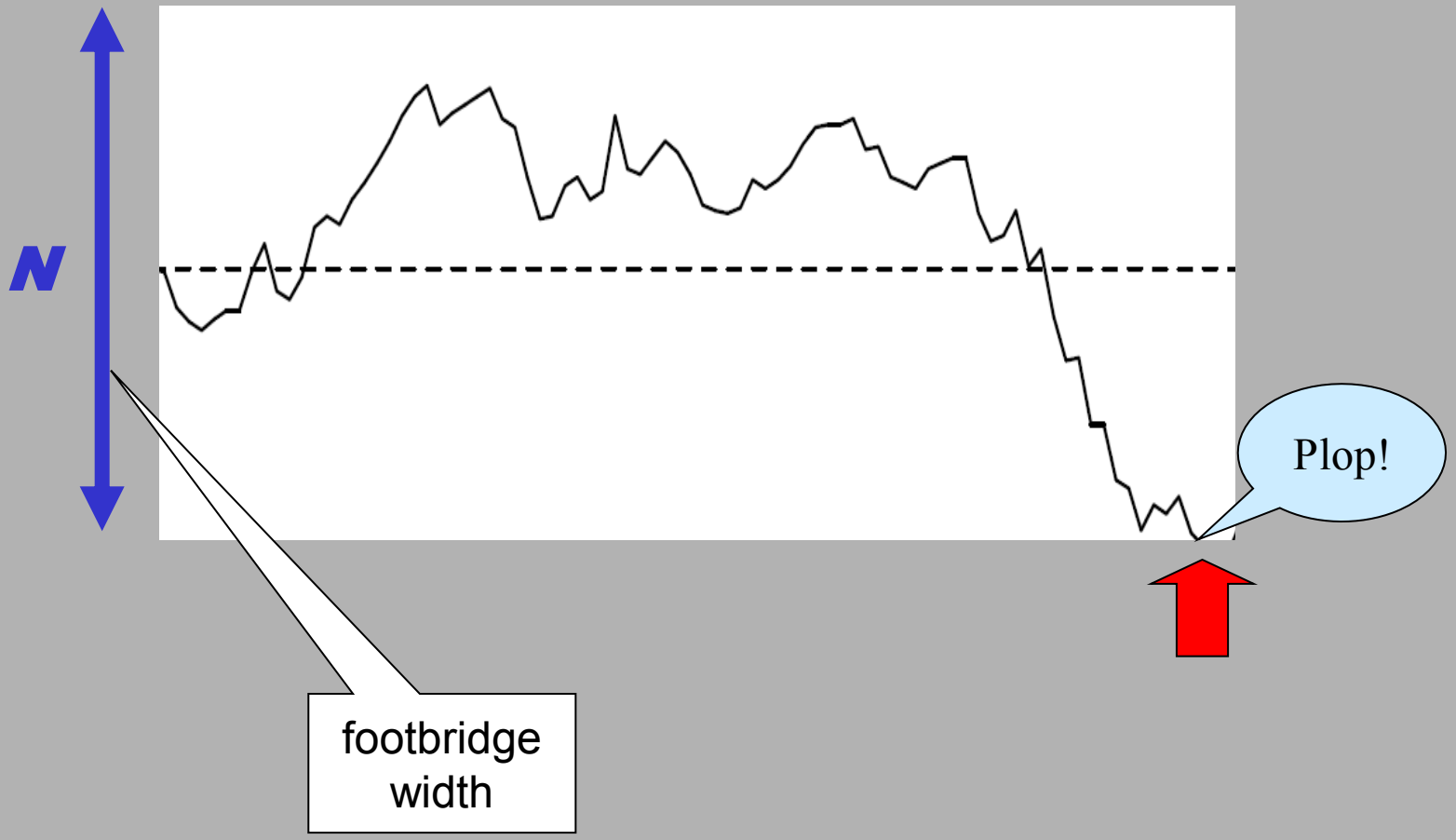
Random sampling results in fluctuations of allele frequencies across generations = „random walk“

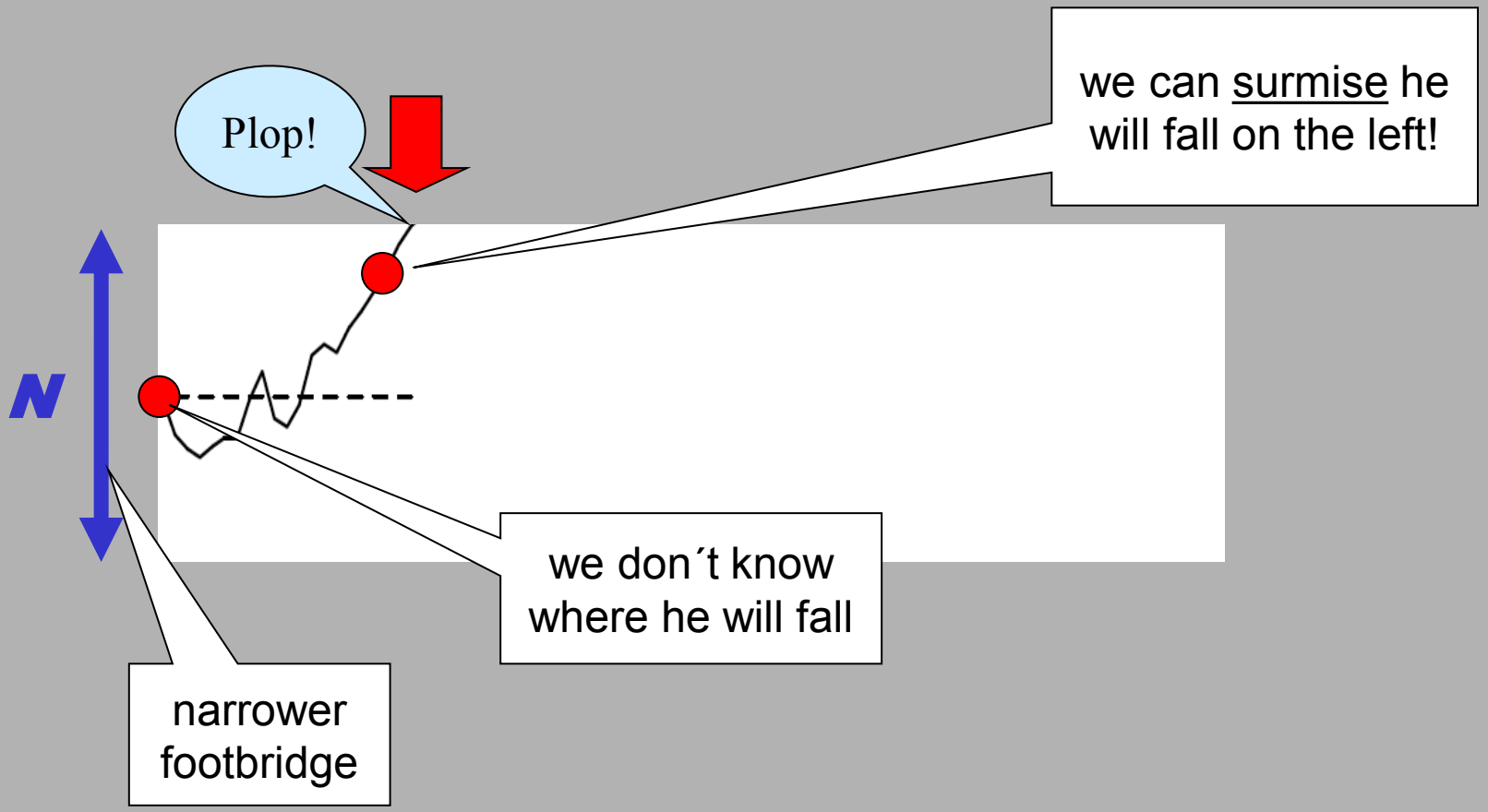
Wright-Fisher model

≈ Hardy-Weinberg model for finite populations

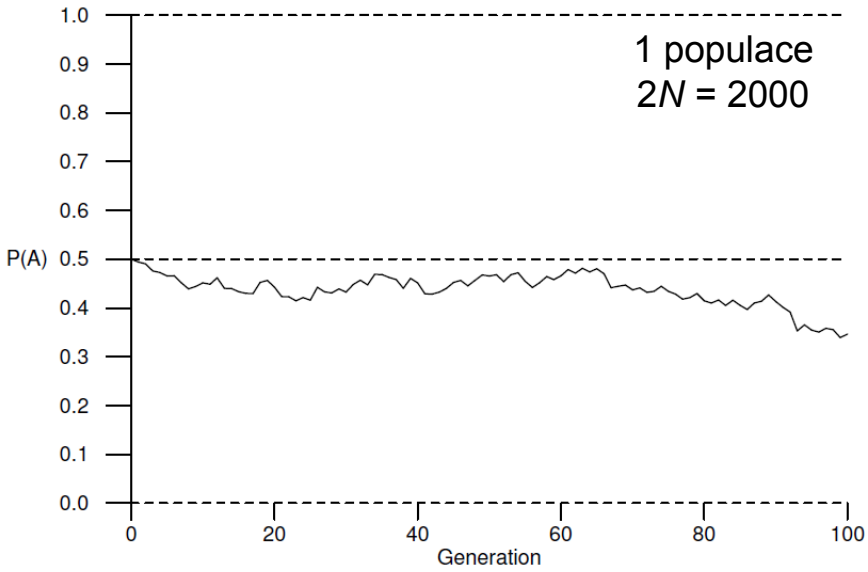
Drunk sailor

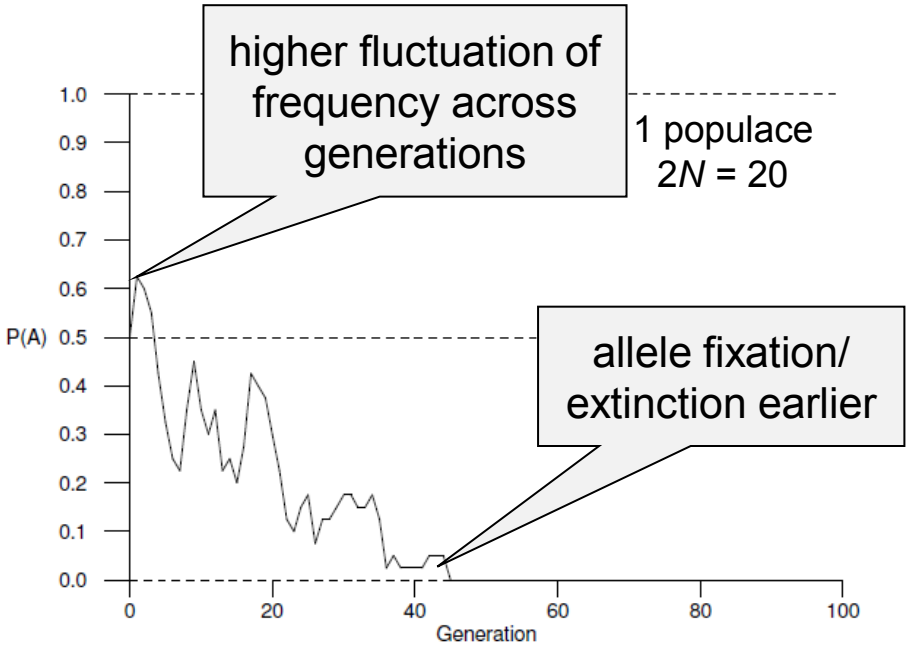



N
Plop!
footbridge
width


Plop!
we can surmise he will fall on the left!
N
we don´t know where he will fall
narrower footbridge

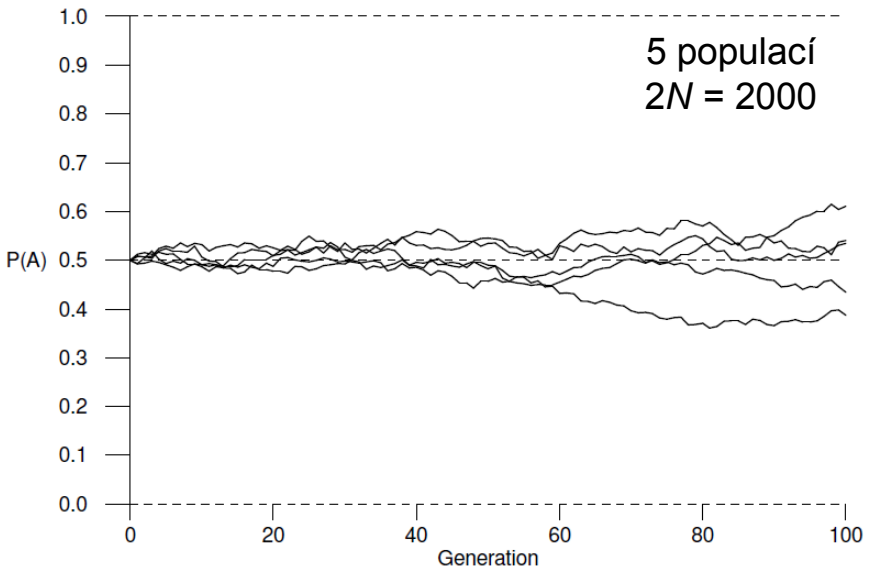
Modelling drift:

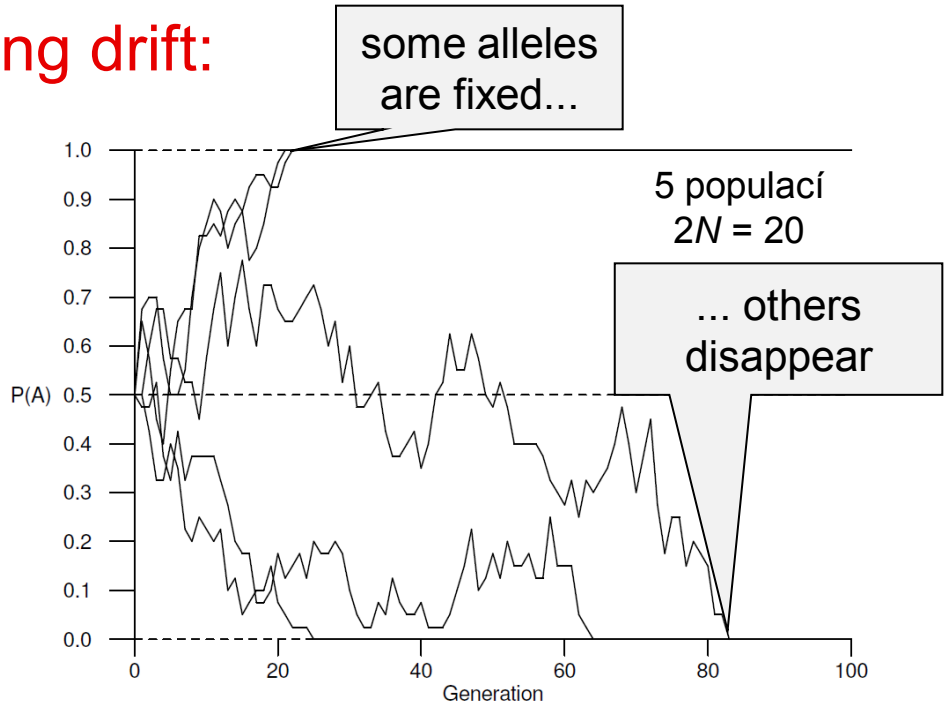




Fluctuation of frequencies across generations stronger in small populations (~ drunker sailor).

Modelling drift:





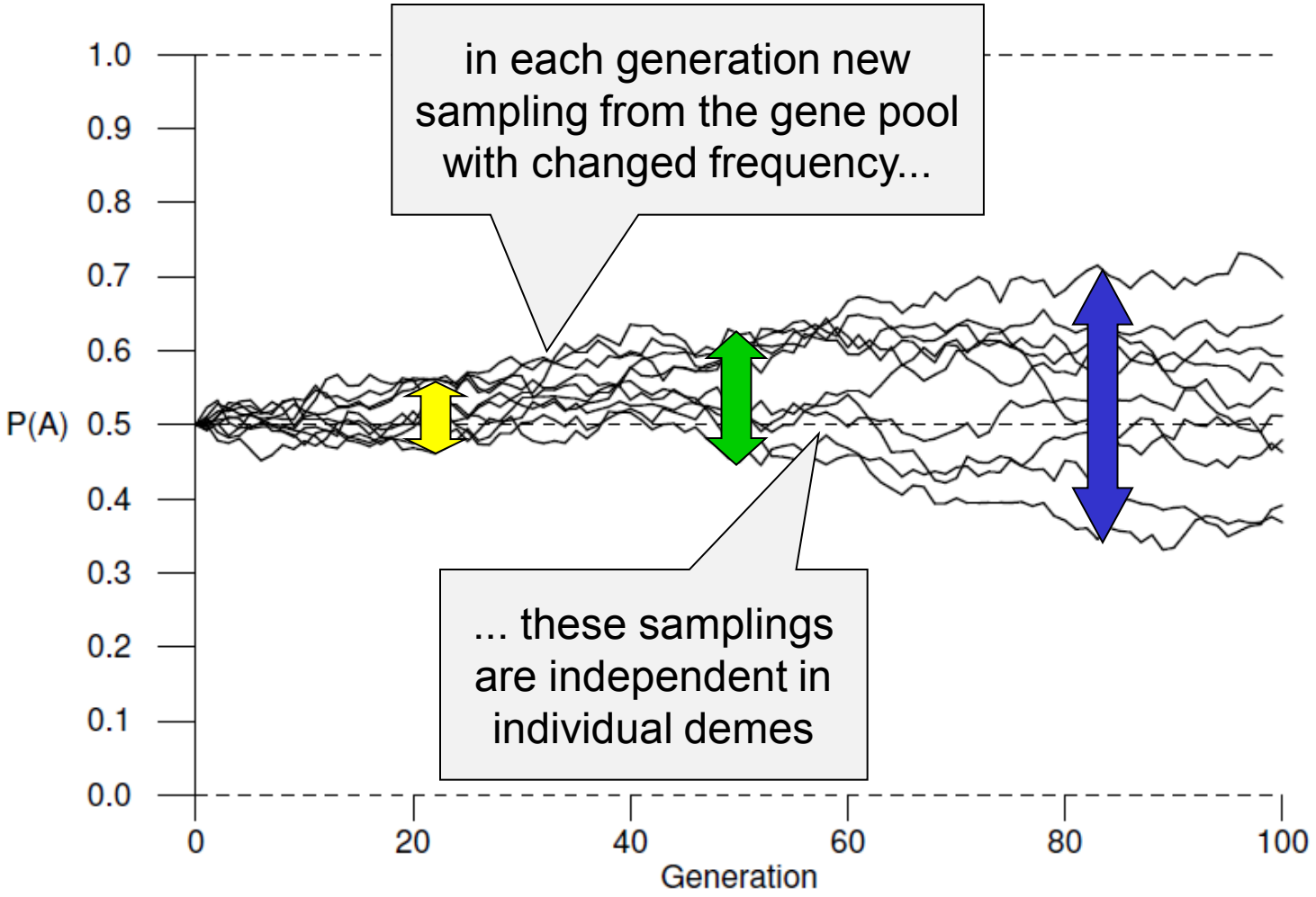
Conclusion 1: Drift results either in allele fixation or allele extinction.

Conclusion 2: Drift results in loss of variation in demes.

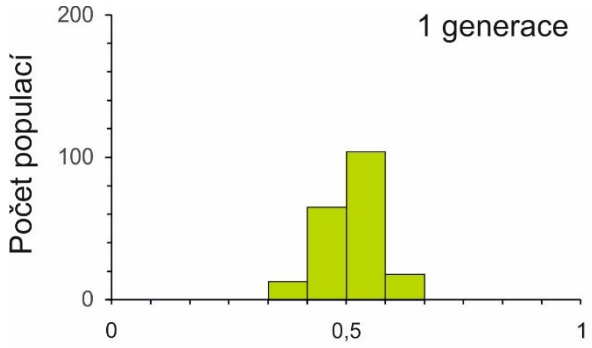
Conclusion 3: Probability of allele fixation equals its frequency.

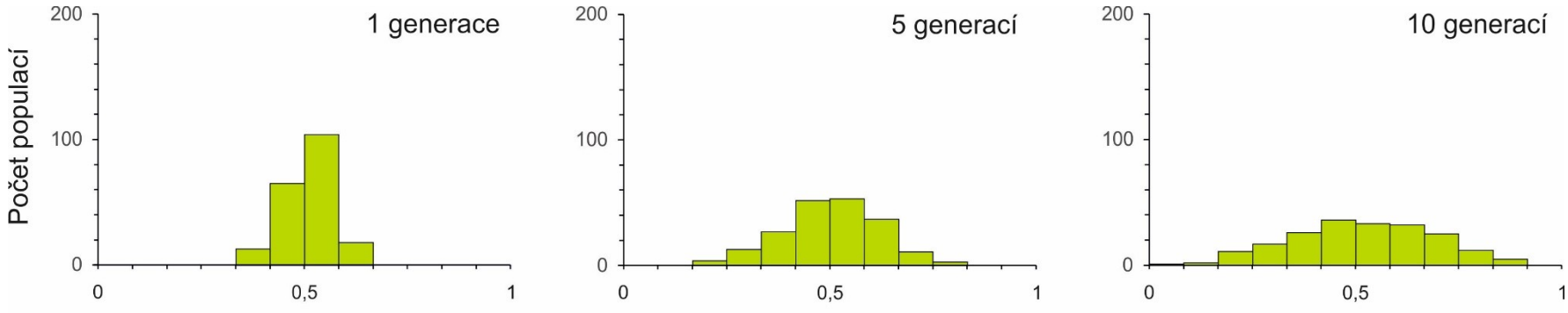
Probability of fixation of a new allele in diploids = $1/(2N)$

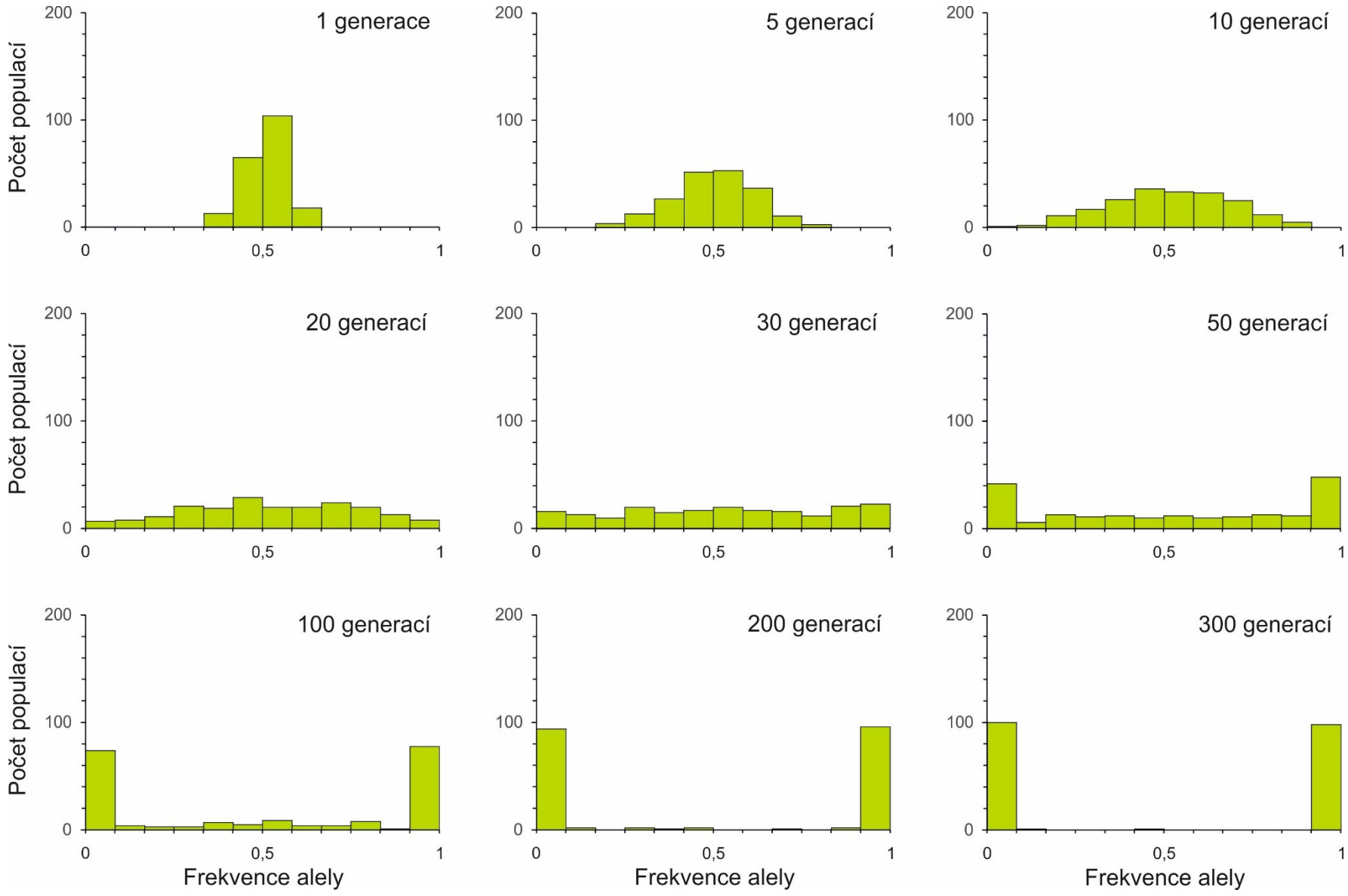
Conclusion 4: Mean time to fixation of a new allele $\approx 4N$.



Conclusion 5: Drift results in divergence among demes.


1 generace
Počet populací
200
100
0
0
0,5
1


Počet populací
200
100
0
0
0,5
1
1 generace
5 generací
10 generací


1 generace
5 generací
10 generací
20 generací
30 generací
50 generací
100 generací
200 generací
300 generací
Počet populací
Frekvence alely
0
100
200
0,5
1

Peter Buri (1956):

107 populations of *D. melanogaster*

zeroth generation: 16 heterozygous bw^{75}/bw individuals in each population

in each generation random sampling of 8 males and 8 females

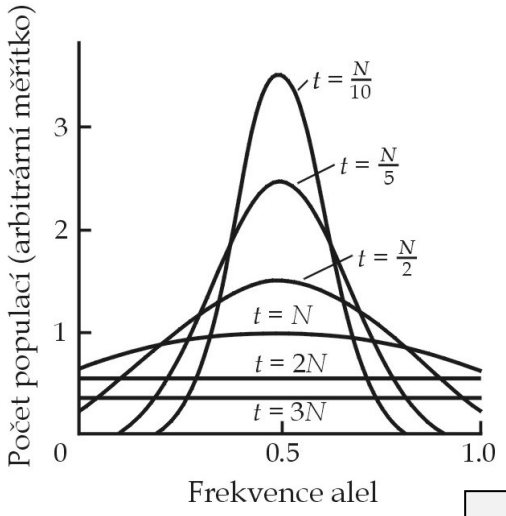
19 generations

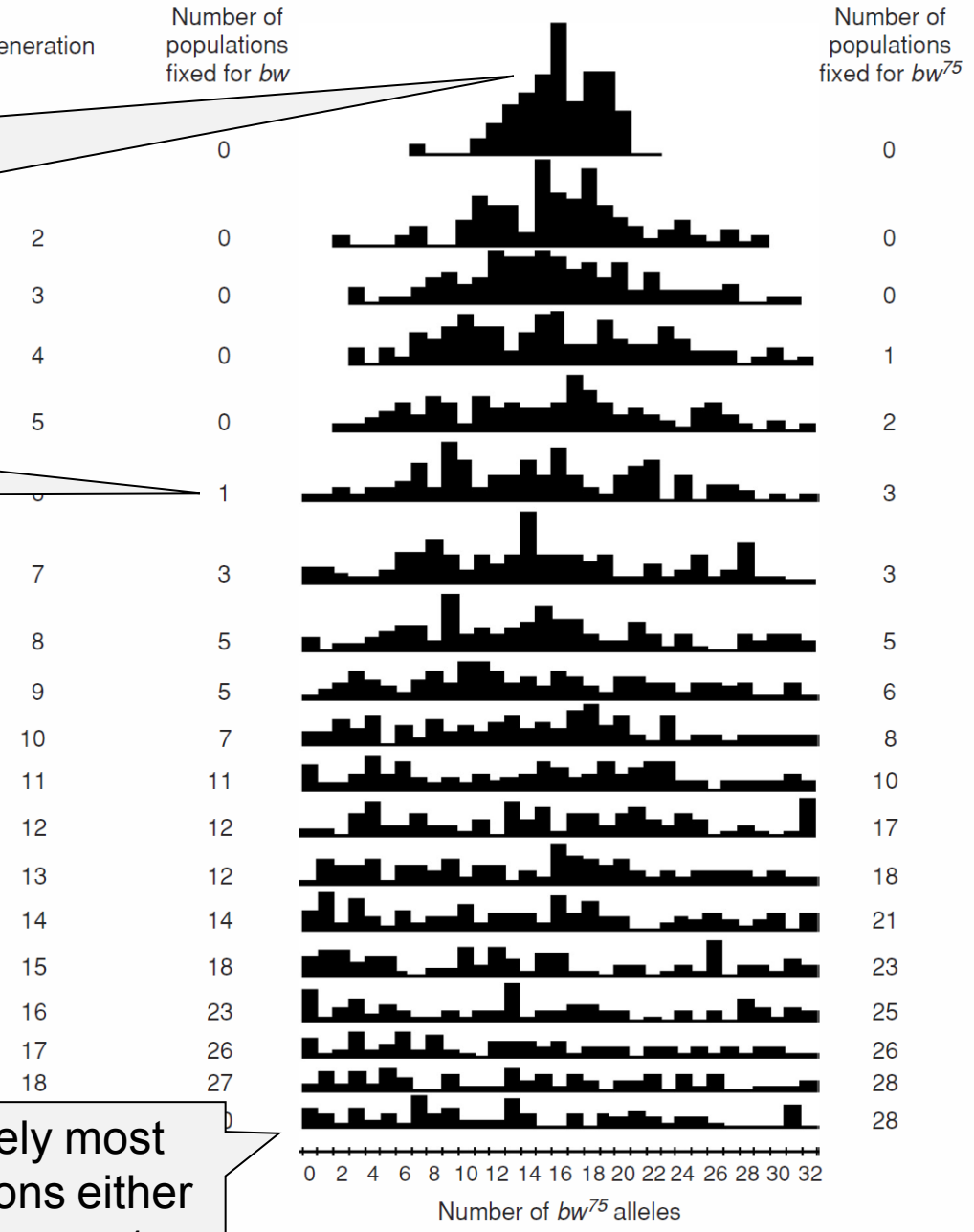
Buri (1956):

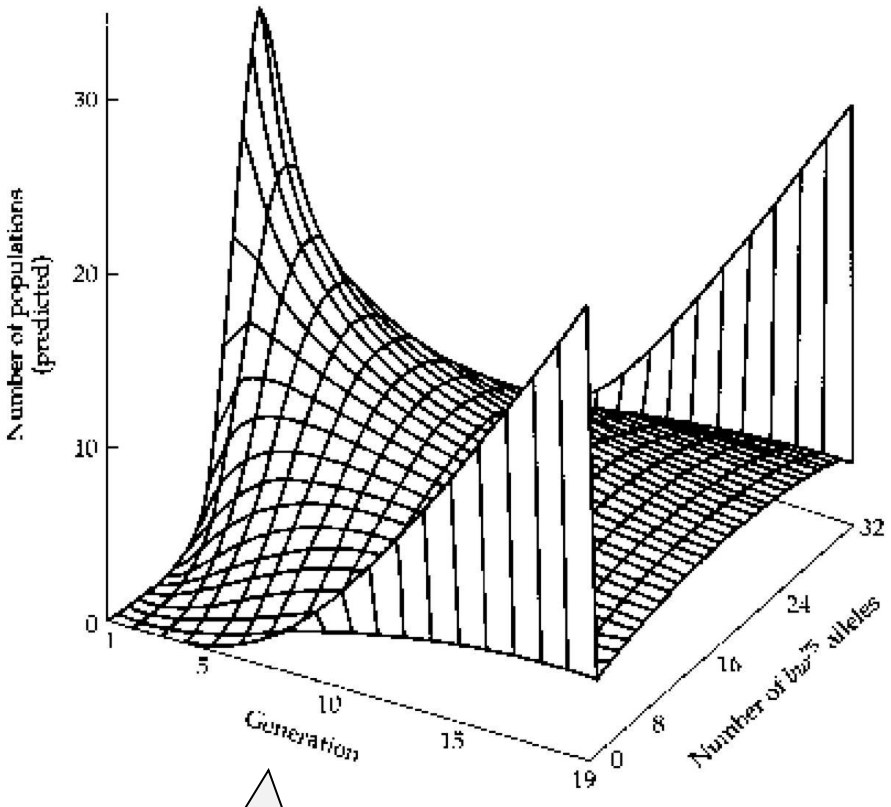
in the first generation most populations around $p = 0{,}5$

population divergence

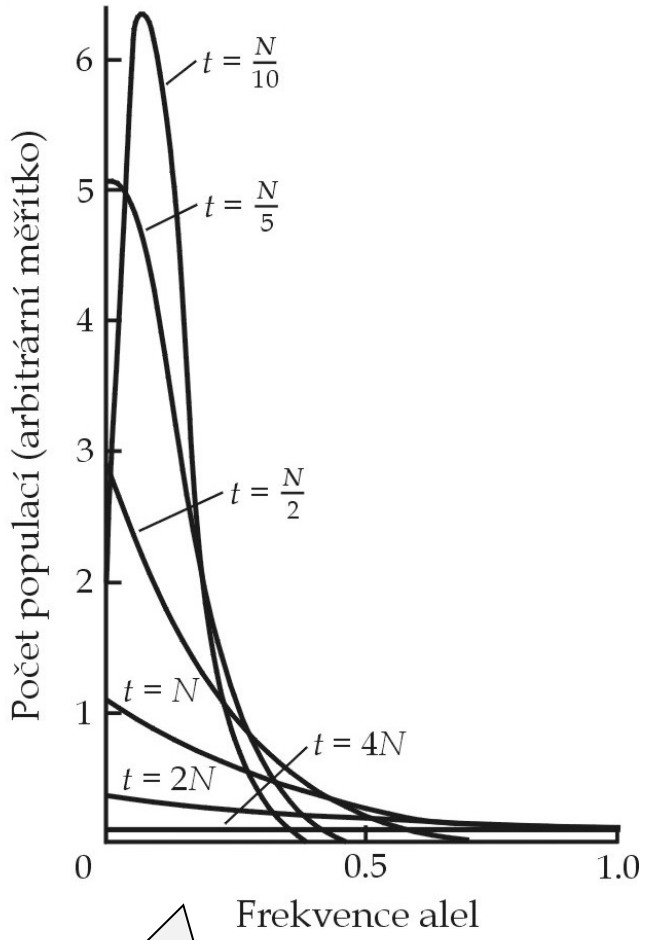
ultimately most populations either $p = 0$ or $p = 1$







mathematical simulation (difussion aproximation)

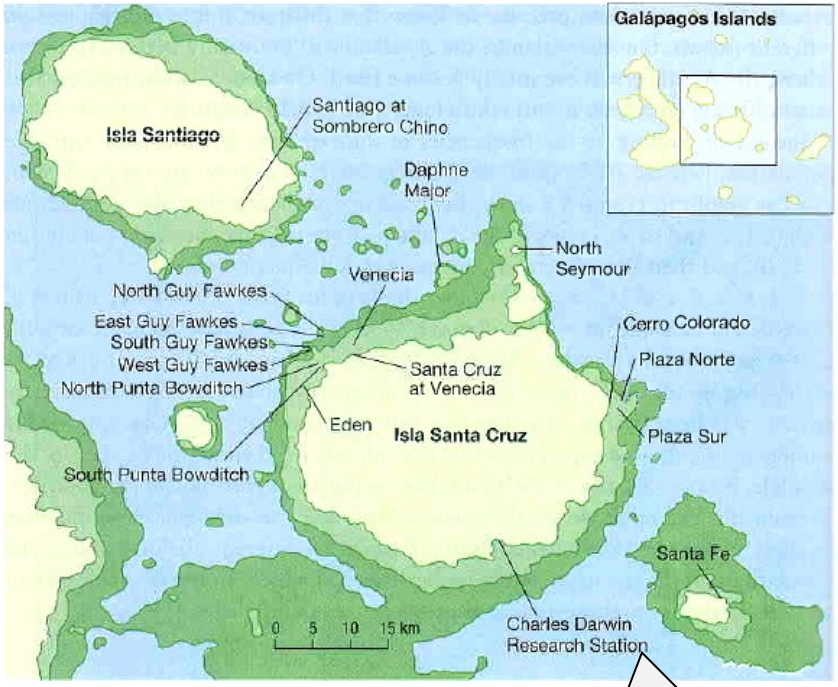


simulation with initial frequency p = 0,1

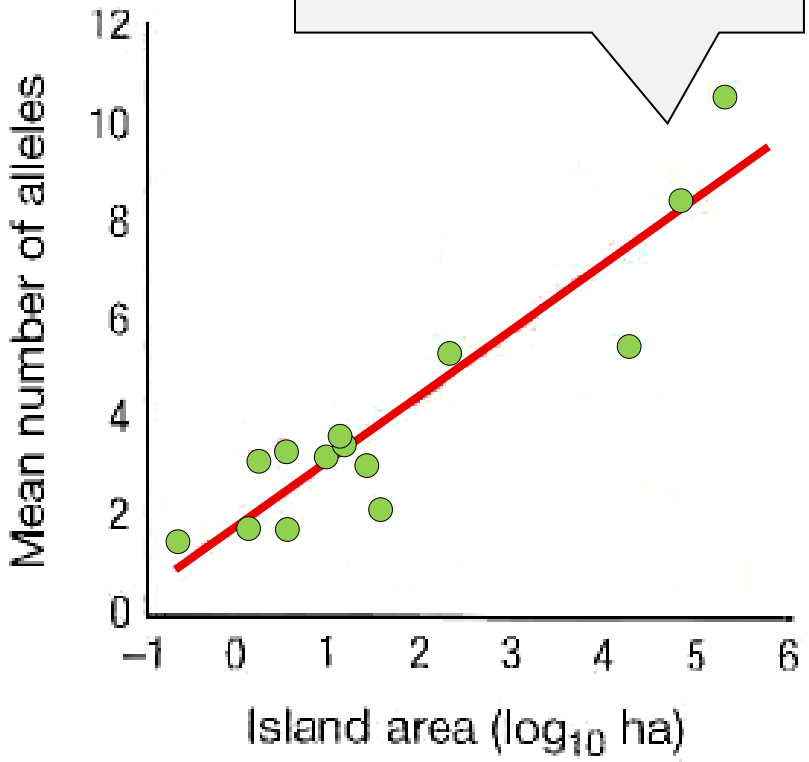
Eg.: Galapágos lava lizard (*Microlophus albemarlensis*)

M. Jordan, H. Snell (2002):

17 populations
11 microsatellite loci

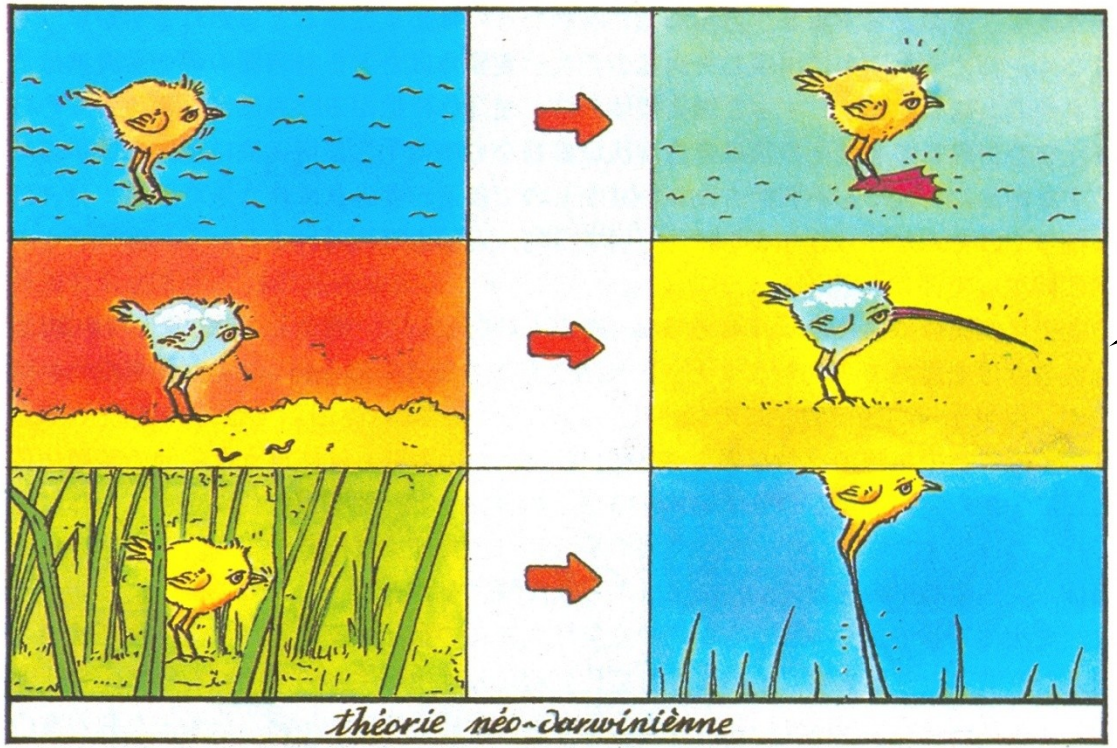


lizards on larger islands have higher variation

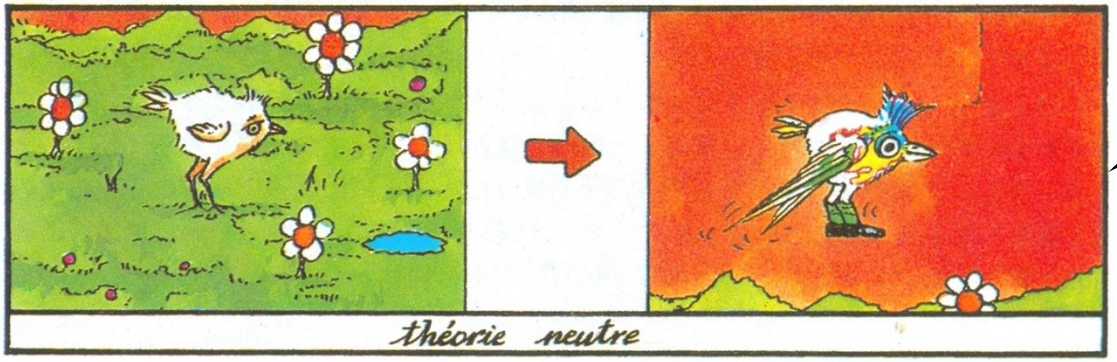


sea level 17 and 12 thousand years ago and nowadays

Evolution of selectively neutral traits is random



Darwinian evolution:
„survival of fittest“



neutral evolution:
„survival of luckiest“

Efective population size

Real populations differ from the WF model (fluctuations of *N*, different reproductive success and mortality, unequal sex ratio, ...)

→ effective population size N_e allows us to measure drift in non-ideal populations

Effective population size = the number of individuals of ideal Wright-Fisher population displaying the same rate of drift as the studied non-ideal population

Like in the inbreeding coefficient there is no single effective population size!!

Some factors decrease N_e relative to N:

overlapping generations

fluctuating population size across generations

different number of breeding males and females

high variation of the number of offspring within populations

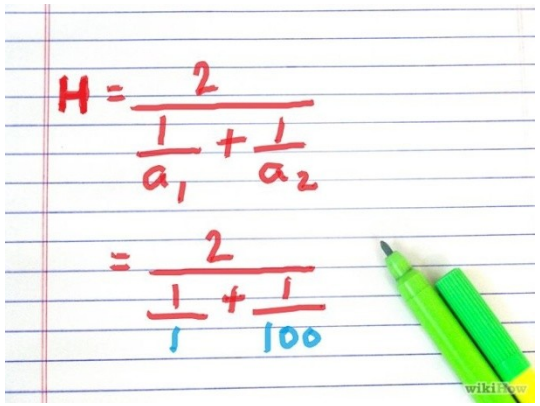
Caution! Under some circumstances
the effective population size can be higher than N!!

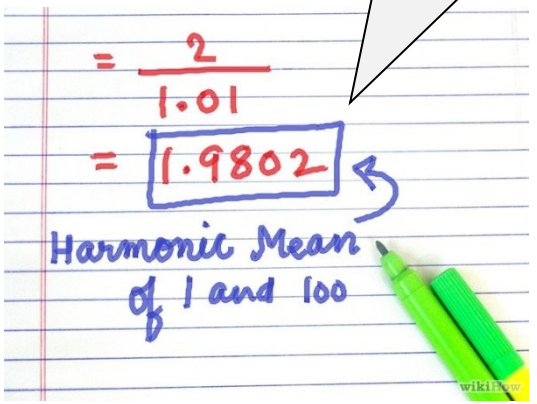
Effect of fluctuating population size:

effective size can be approximated as <u>harmonic mean</u> ⇒ strong influence of small *N*!!

$$N_e = \frac{t}{\frac{1}{N_1} + \frac{1}{N_2} + \cdots + \frac{1}{N_t}}$$

harmonic mean



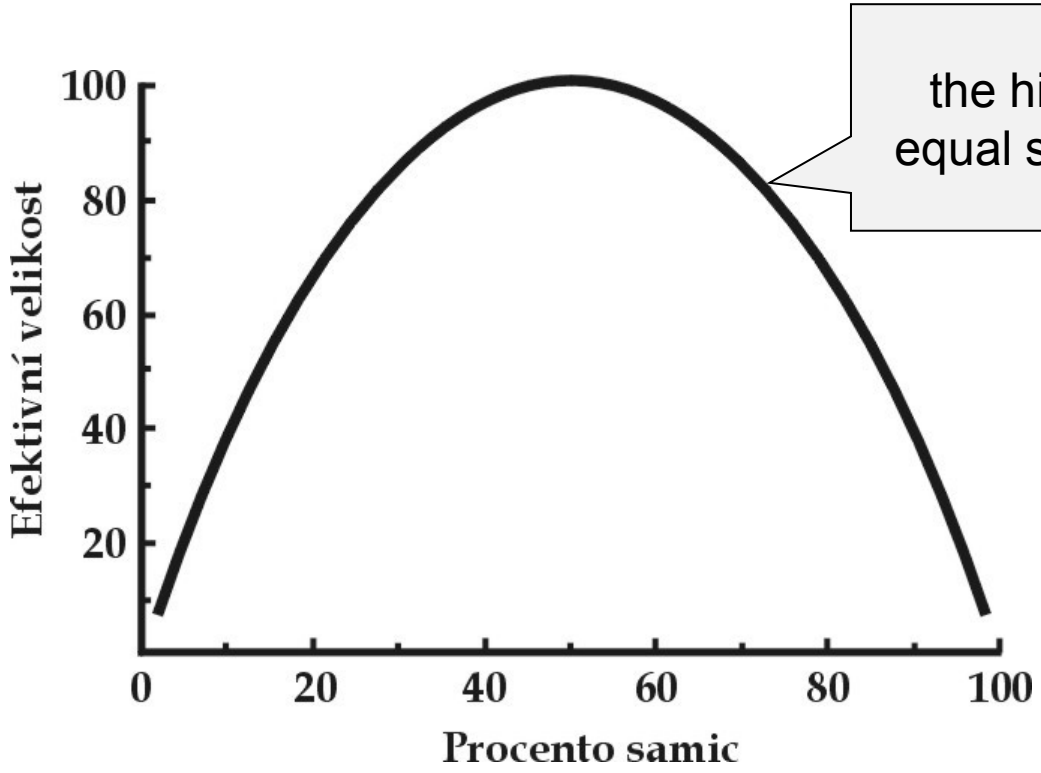


mean much closer to the lower value

Effect of biased sex ratio:

Till now we assumed the same number of breeding males and females

N_m = number of breeding males, N_f = number of breeding females



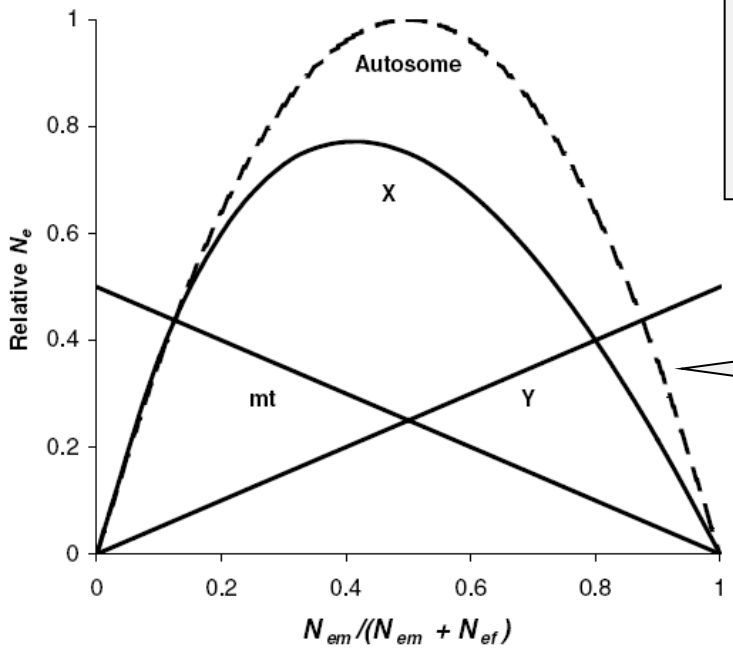
the higher deviation from equal sex ratio, the lower N_e

Effect of biased sex ratio:

$N_m = 1$:

$$N_e = \frac{4N_m N_f}{N_m + N_f} \rightarrow N_e = \frac{4N_f}{N_f + 1} \approx$$

it follows from this formula that if there is only a single breeding male in the population $N_e \approx 4$ regardless of the total number of individuals



effect of sex ratio on N_e different for various genetic traits!

Effect of unequal reproductive success:

southern elephant seal:

sex ratio within a harem 1:40*)

*) effective ratio 1:4-5 due to cuckoldry and short period of male´s dominance (1-2 years)

Reproductive success on the gene level:

If a gene is affected by selection variance of the number of offspring among members of a population is high (individuals with a positive allele have more offspring)

$\Rightarrow$ N_e for this gene is lower than for a neutral gene

Each genetic trait requires its own N_e:

For genes on autosomes, sex chromosomes, and mtDNA there are different effective population sizes:

autosomes:	N_e	$4\ N_e$
X, Z:	$¾\ N_e$	$3\ N_e$
Y, W, mtDNA:	$¼\ N_e$	$1\ N_e$

COALESCENT

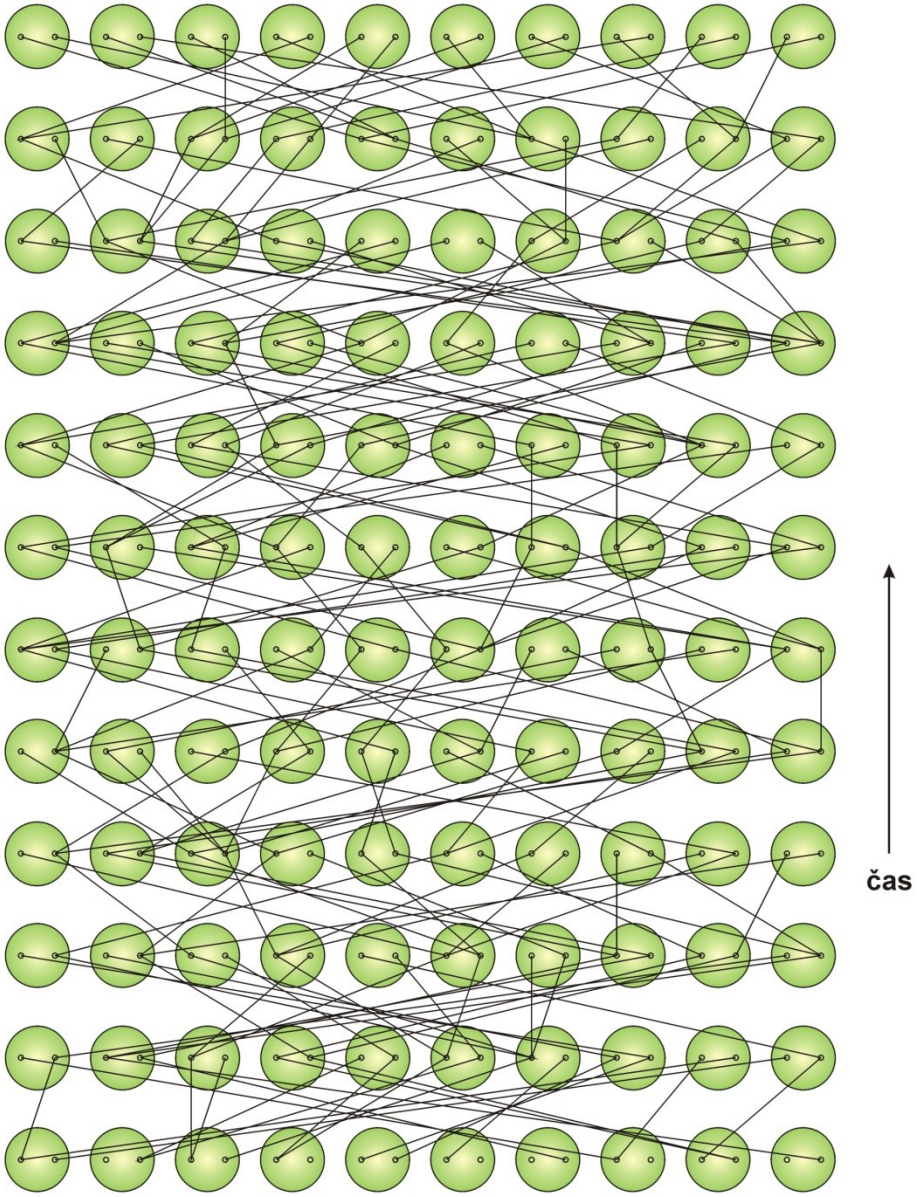
under drift some alleles disappear from a population $\Rightarrow$ when there are no mutations ultimately all gene copies have a common ancestor

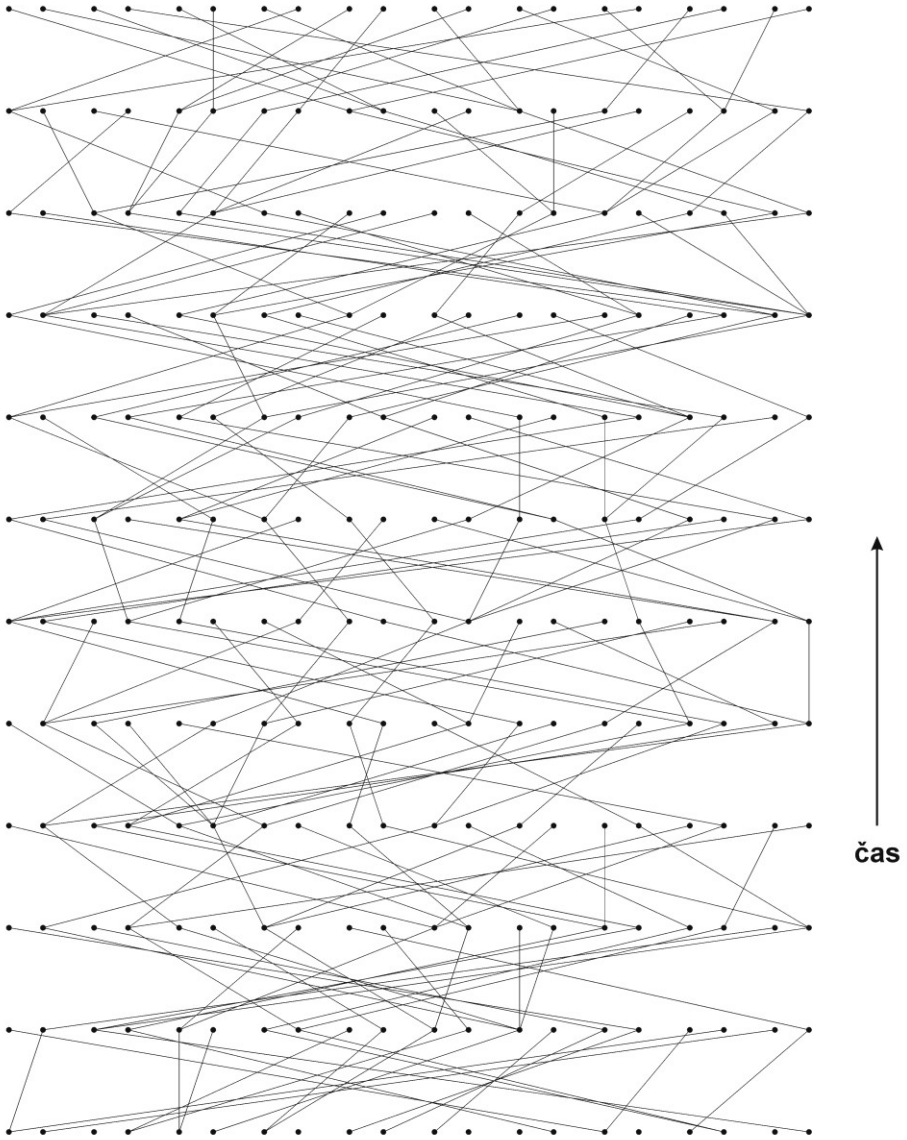
„forward“ approach

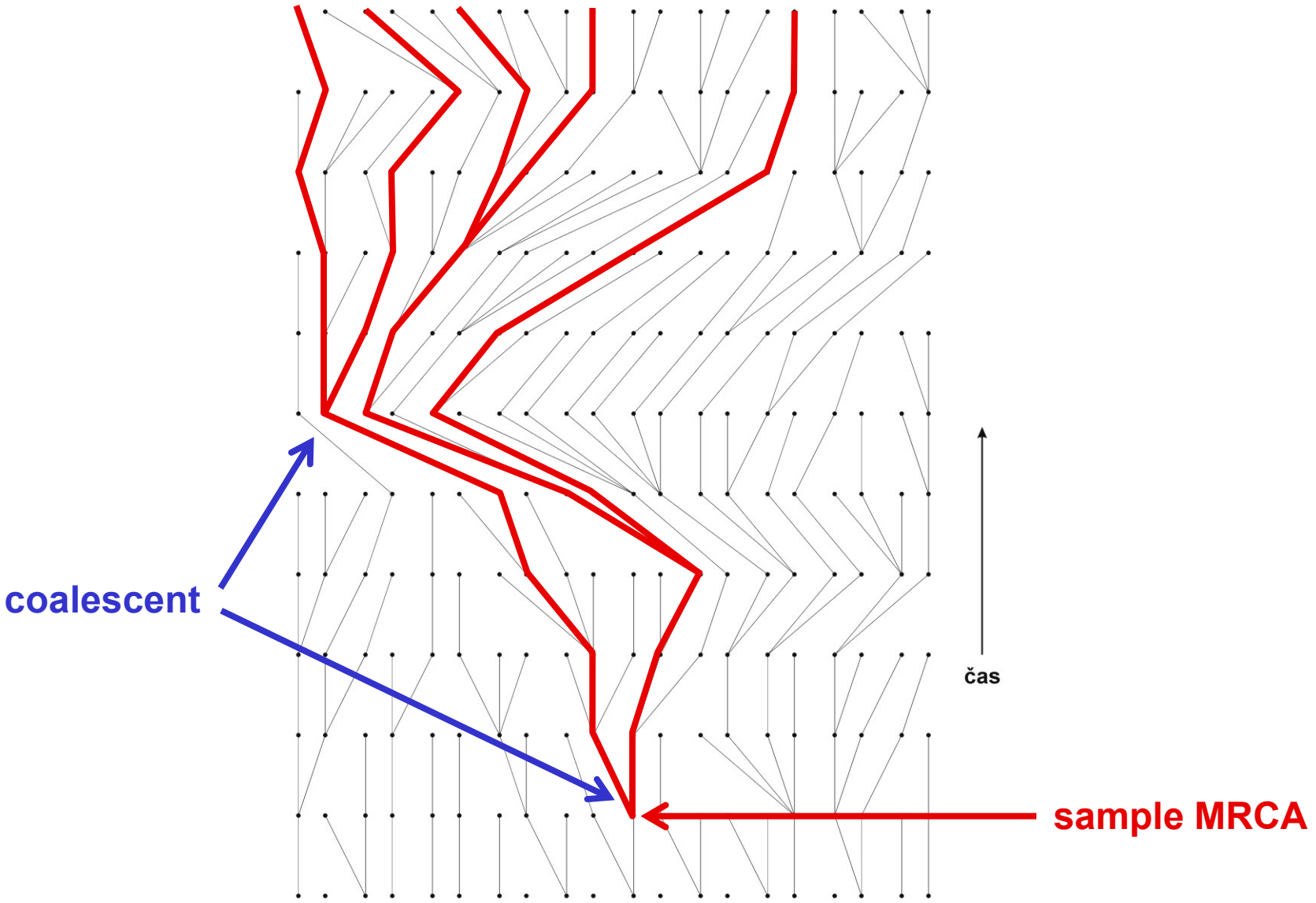
we can proceed also back in time – „backward“ approach $\rightarrow$ moving back in time till two or more gene copies „fuse“ = coalescent event

the most recent common ancestor (MRCA)

Wright-Fisher model:




čas


coalescent
čas
sample MRCA

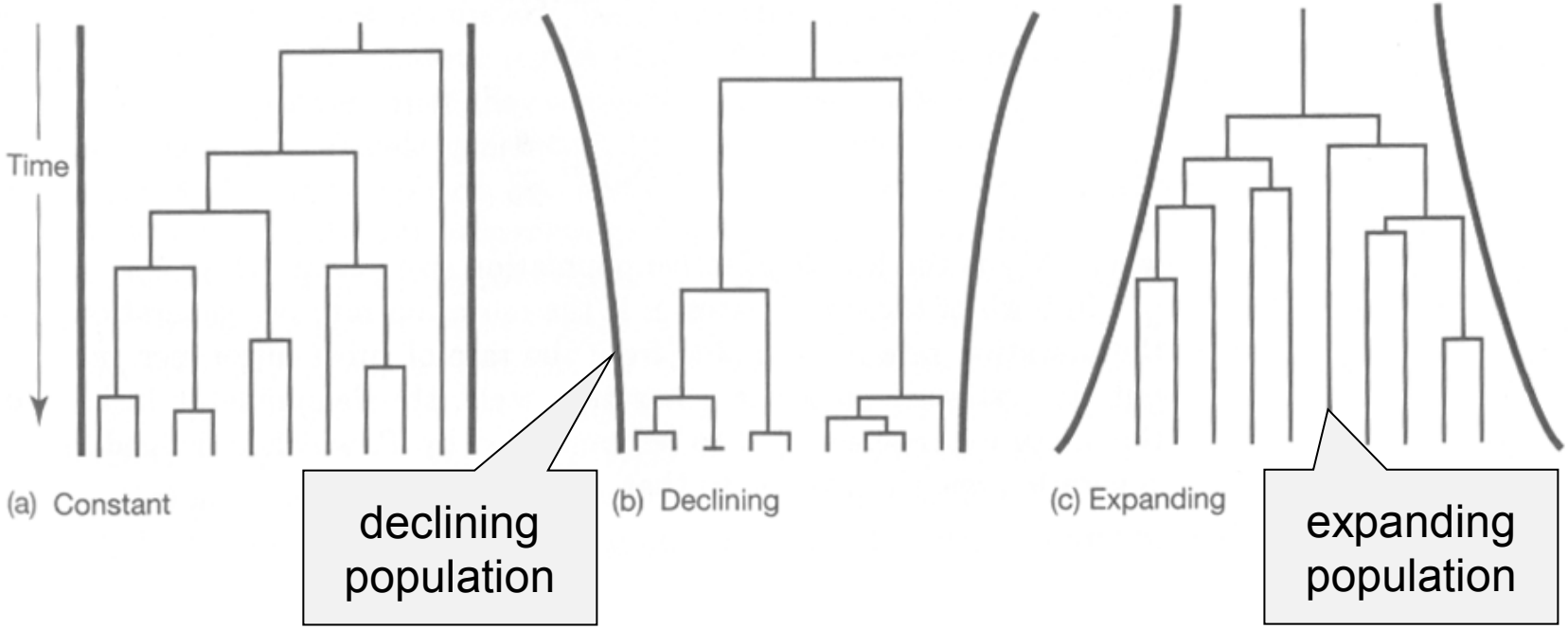
Coalescence and effective population size

from the coalescent theory several interesting consequences follow:

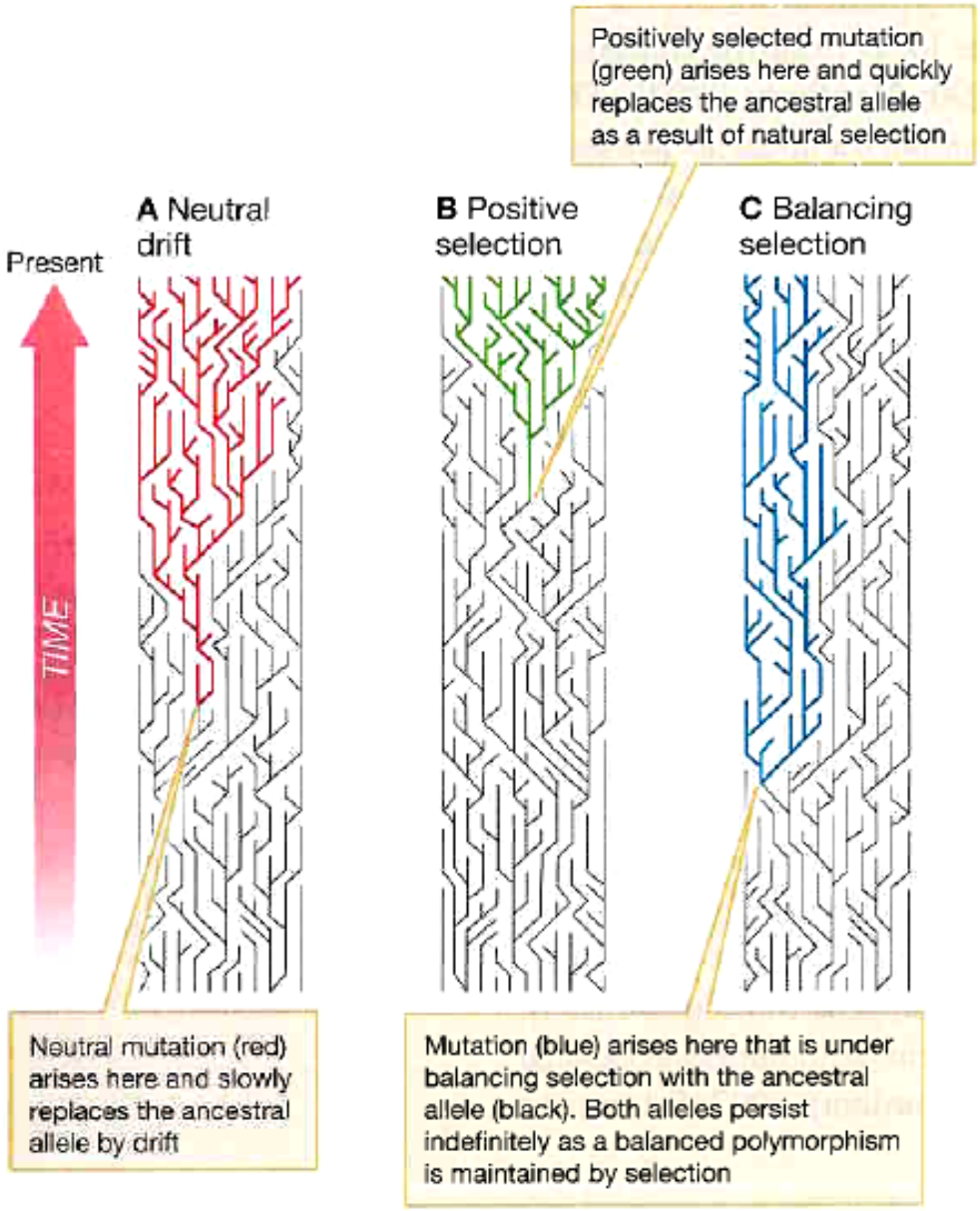
in small populations coalescent rate higher than in large populations

$\Rightarrow$ we can estimate N_e

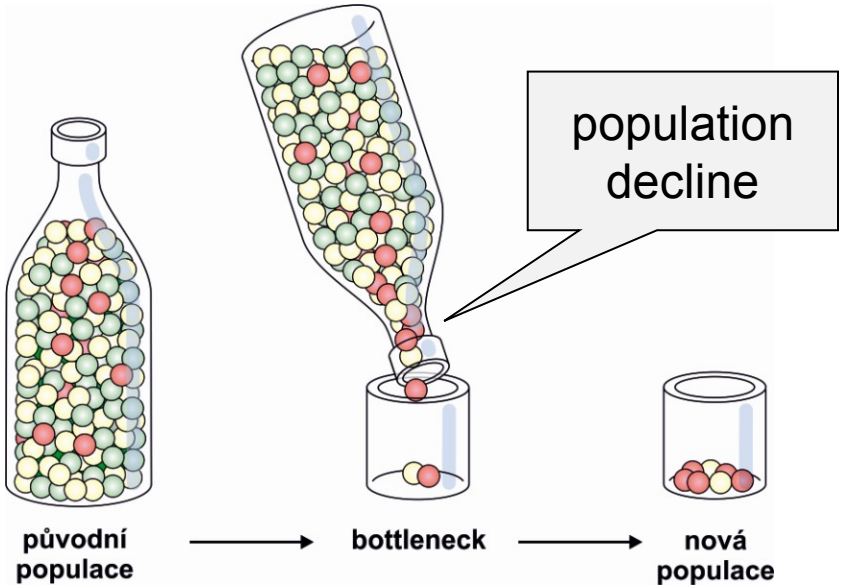
but we can estimate also changes of N_e in time

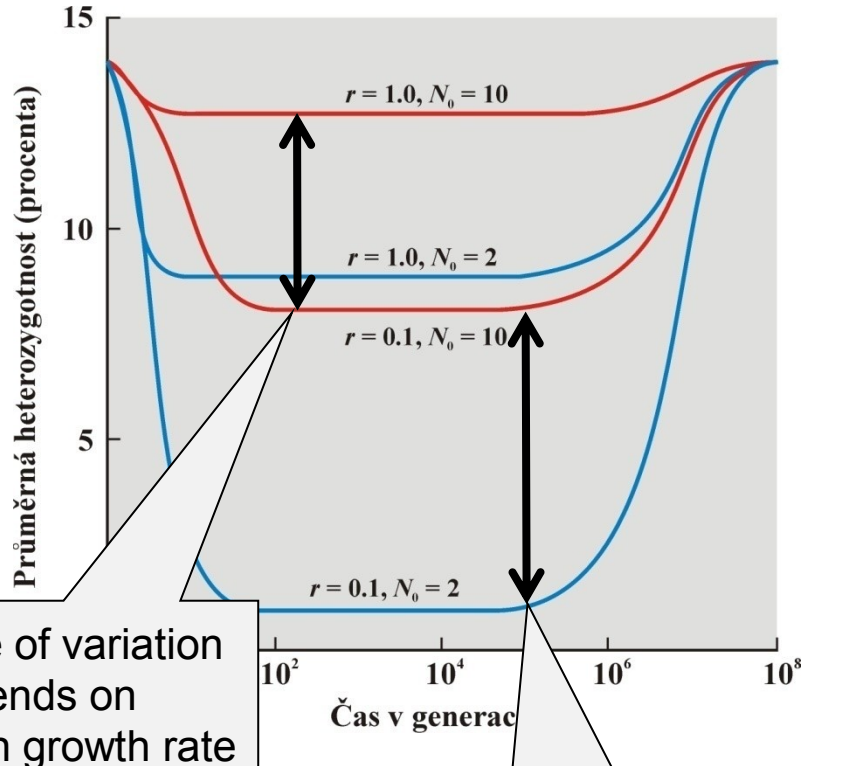


The same effect of selection on the coalescent tree shape:



BOTTLENECK and FOUNDER EFFECT





decrease of variation depends on population growth rate

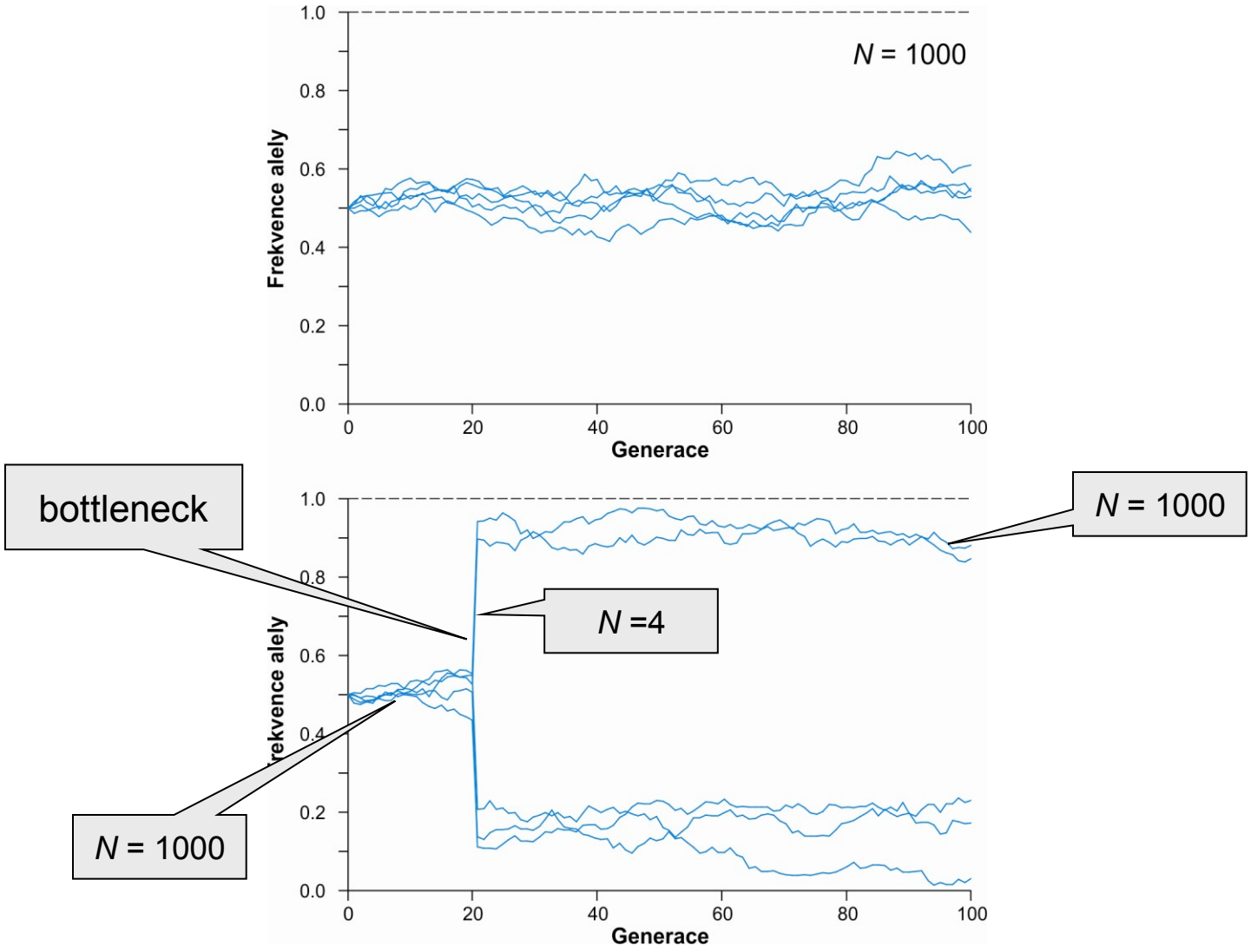
variation more reduced under stronger bottleneck

bottleneck reduces variation

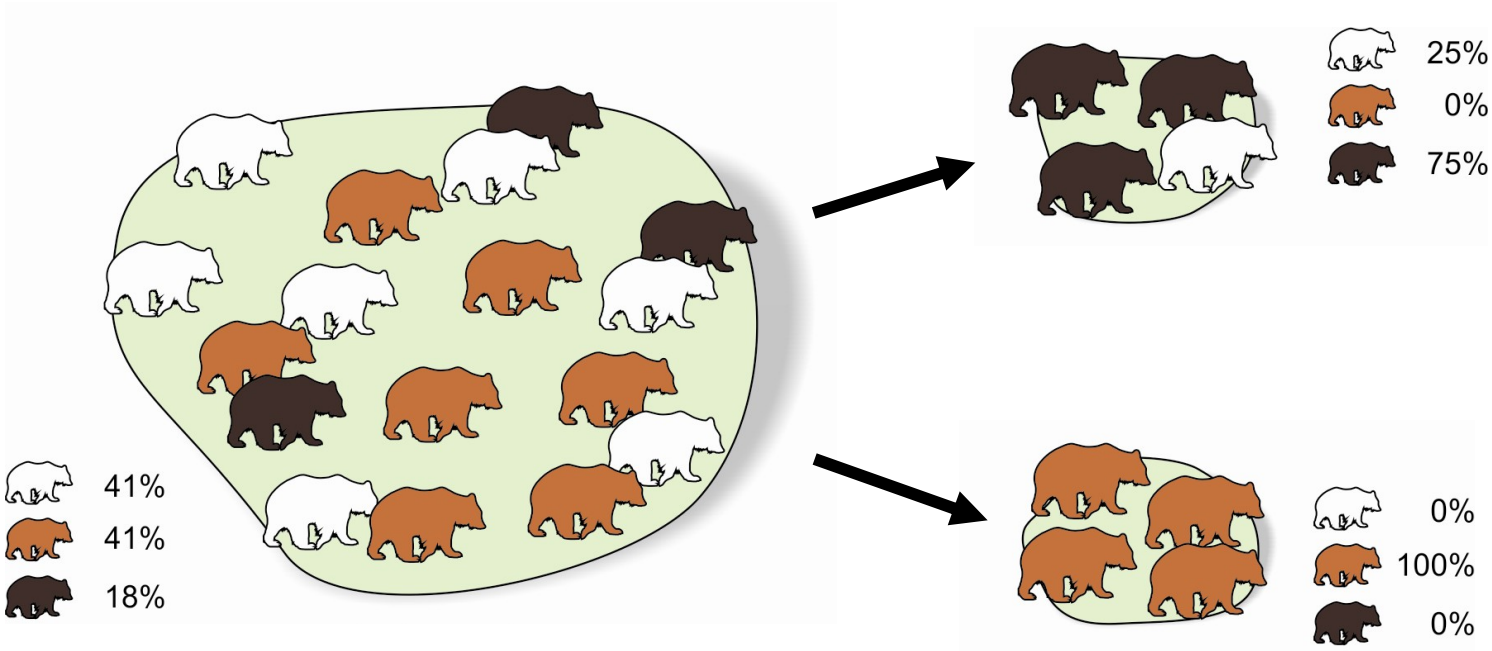
magnitude of this reduction depends on reduction of N_e and duration of bottleneck

rate of decrease of variation different for various genetic traits (autosomes, mtDNA, Y...) – different N_e!

Bottleneck:



Founder effect:



colonization of a novel territory (eg. island)

because of a small numer of founders (even a single pregnant female)
→ random change of allele frequencies
→ reduction of variation

different environmental conditions → speciation

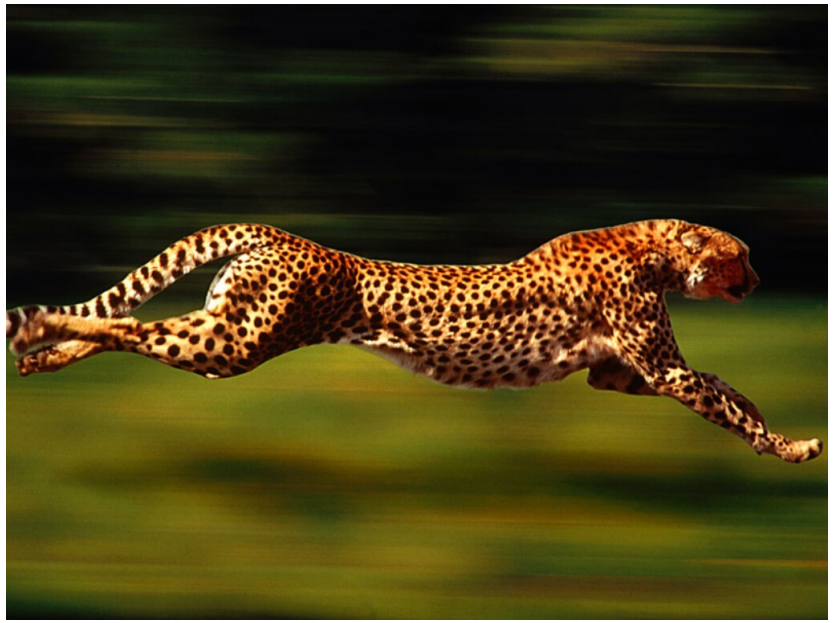
Examples of founder effect and bottleneck
cheetah

30 individuals of *Acinonyx jubatus reineyi* from E Africa, 49 protein loci: only 2 loci polymorphic (P = 0,04), mean heterozygosity H_o = 0,01

98 individuals of *A. j. jubatus* from S Africa: P = 0,02, H_o = 0,0004!

south-African individuals accept skin grafts of the east-African subspecies without problems $\Rightarrow$ monomorphism of MHC genes

assumed strong bottleneck in the past



golden hamster

1930: Israel Aharoni (Hebrew Univ., Jerusalem) – female with offspring

escape of several individuals from captivity

1931: transport of several individuals to Britain 1937: private breeders

Recent genetic analyses including mtDNA → all golden hamsters currently kept in breeding colonies are descendants of a single female, probably that of 1930

mostly presented as an example of bottleneck but it is rather an example of <u>founder effect</u>

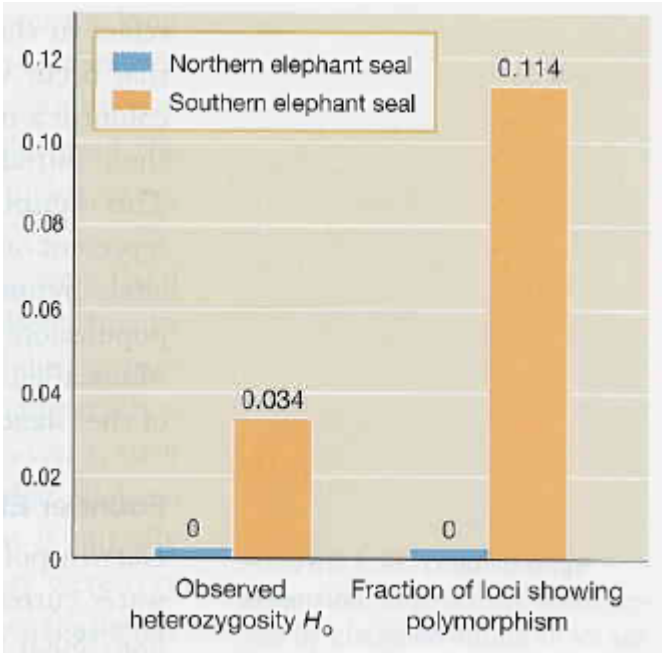
northern elephant seal

Mirounga angustirostris: in 19th century almost eradicated

→ 1892 last 8 individuals on the island of Guadelupe killed for museum collections

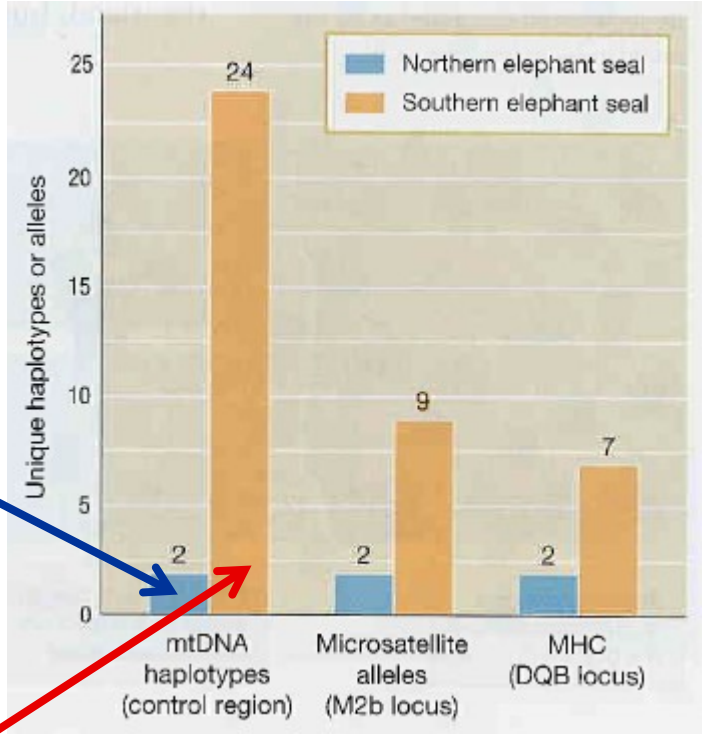
fortunately 10-20 individuals passed unnoticed → today > 100 000 inds.

M. Bonnell a R.K. Selander (1974): blood samples of 159 individuals electrophoresis at 21 loci → no variation
likewise Hoelzel et al. (1993), 62 loci



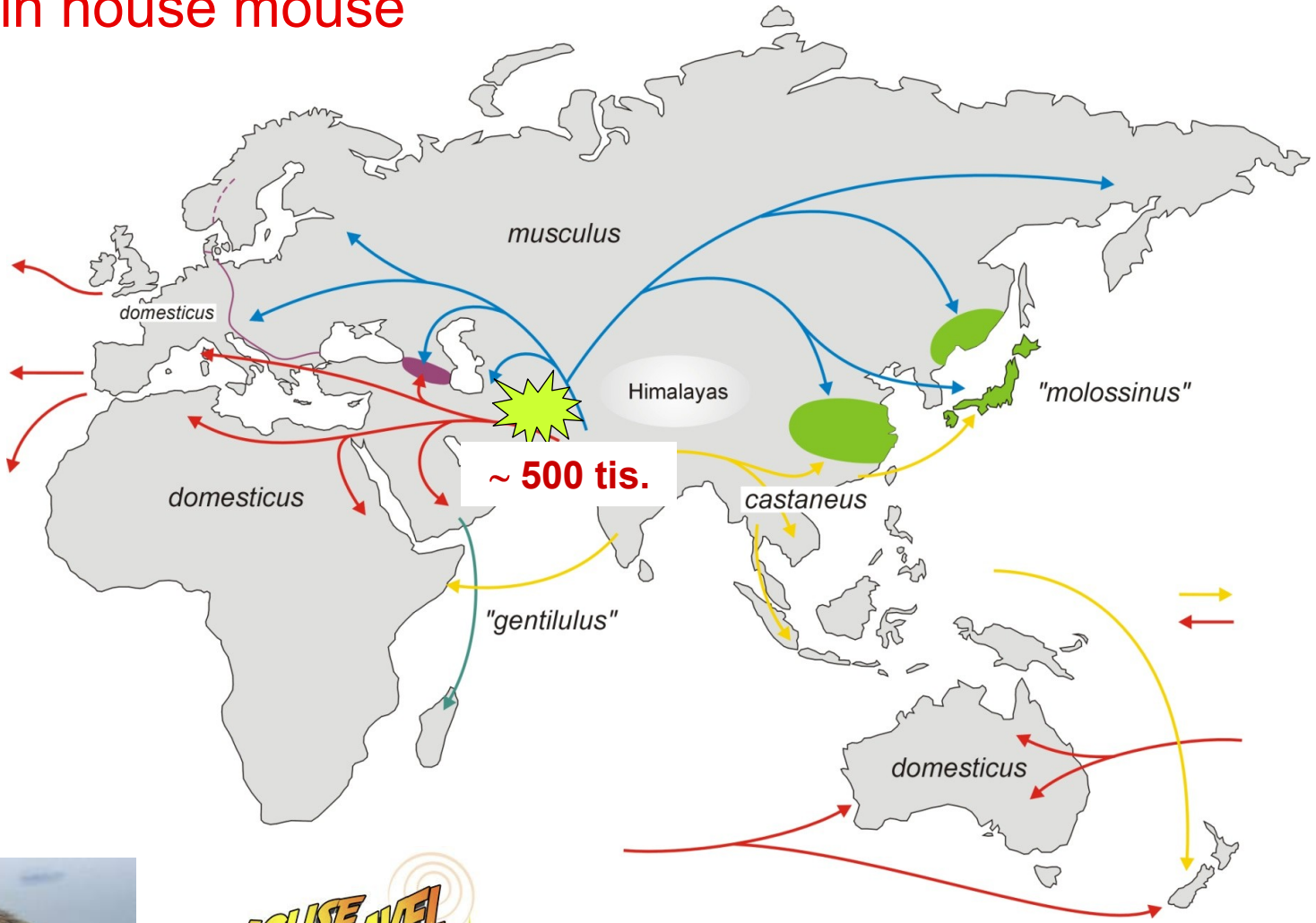
Hoelzel et al. (1999): DNA markers

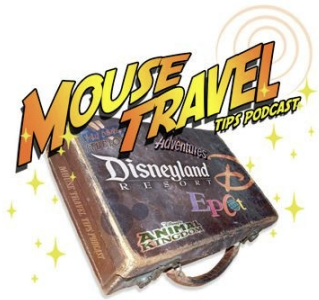
northern elephant seal
(*Mirounga angustirostris*)



southern elephant seal
(*Mirounga leonina*)

FE in house mouse





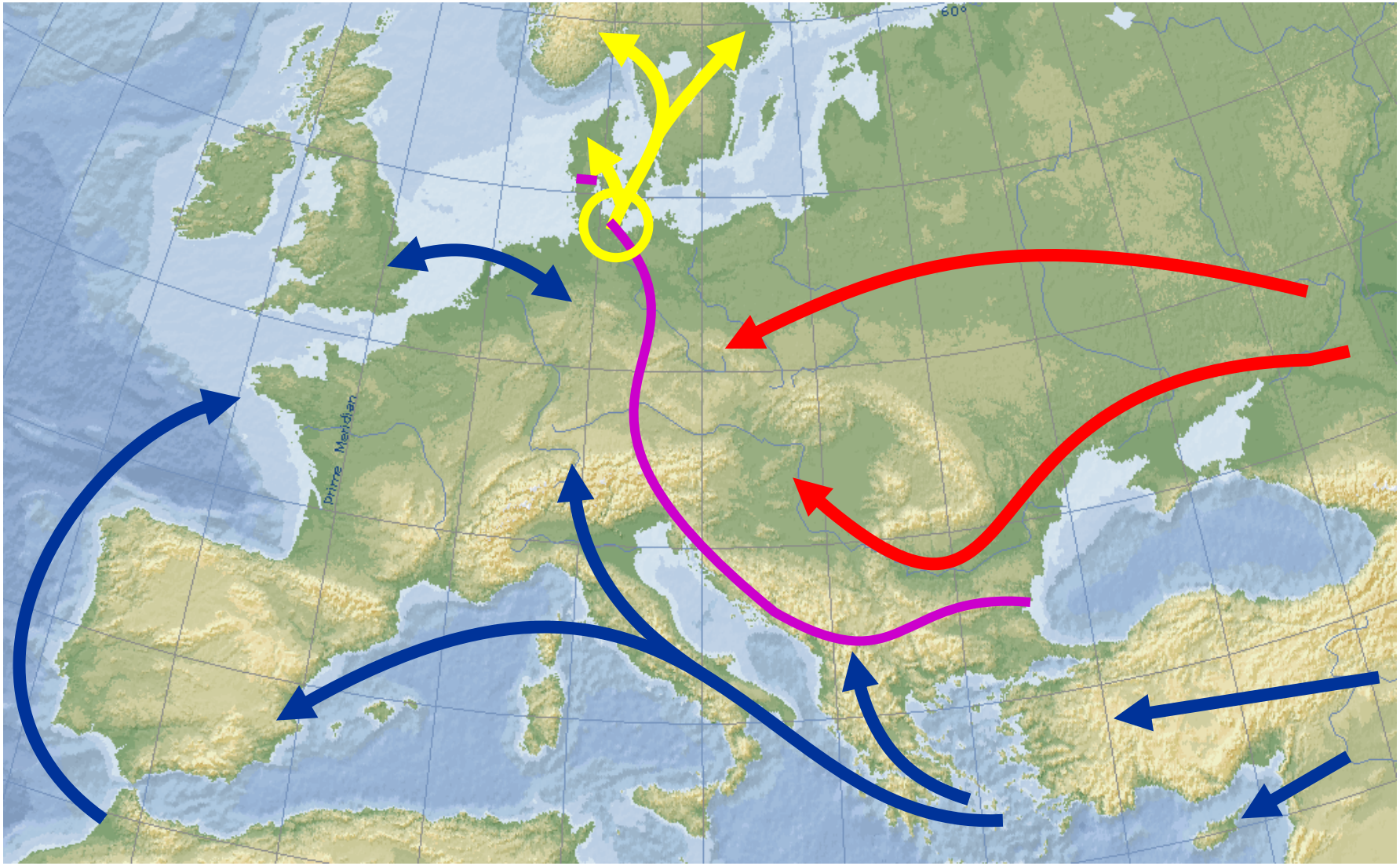
Mus musculus musculus

M. m. domesticus

nuclear DNA

mtDNA

mouse colonization of Europe



humans

a) Las Salinas (Dominican Republic):

Altagracia Carrasco:

several children with at least 4 men

Carrasco heterozygous for substitution T → C in 5th exon of the 5-α-reductase 2 gene ⇒ TGG (Trp) → CGG (Arg) at the 246th position of the protein

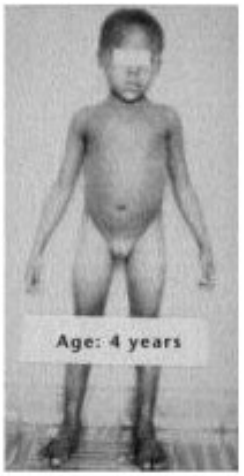
the enzyme catalyzes transformation of testosterone to DHT (dihydrotestosterone)

low activity of the mutant enzyme in homozygotes
⇒ boys have testes but other traits are female

DH

in puberty testosterone production increases
⇒ transformation to men

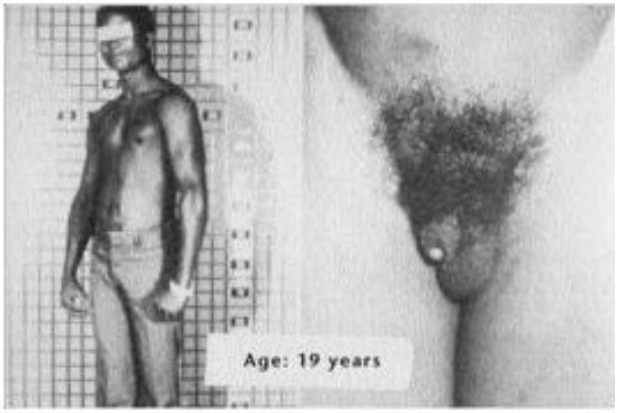
in Salinas high frequency of the mutation ⇒ the word *guevedoces* (= „penis in 12“)


Age: 4 years

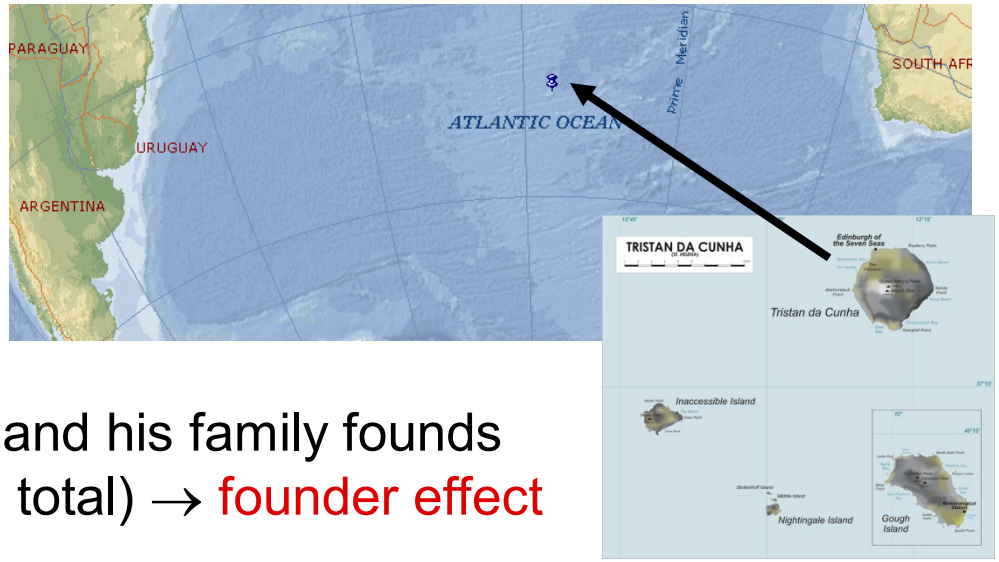
Age: 8 years

Age: 12 years

Age: 18 months


Age: 19 years

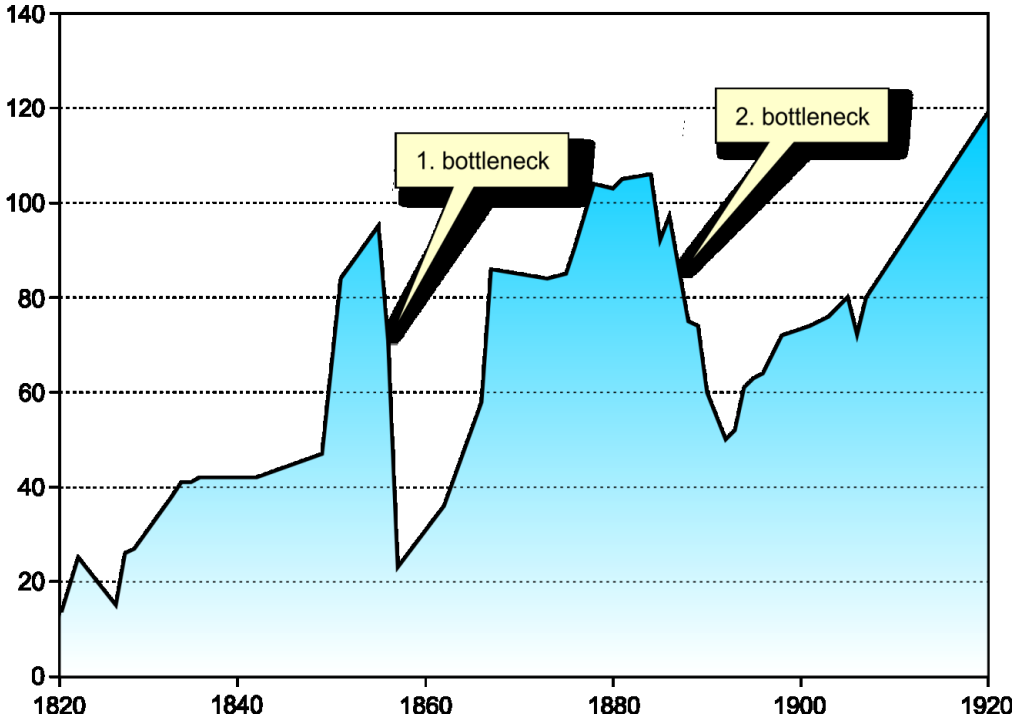
Tristan da Cunha:



1816 military garrison

1817 garrison withdrawn;
Skottish corporal William Glass and his family founds a small colony (20 individuals in total) → founder effect

during 80 years 2 strong bottlenecks

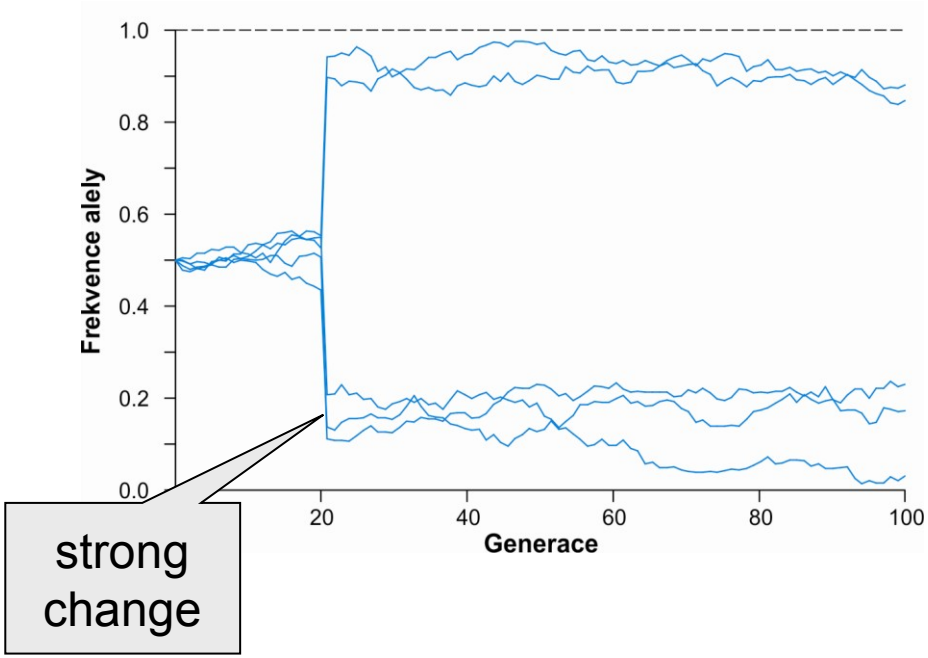


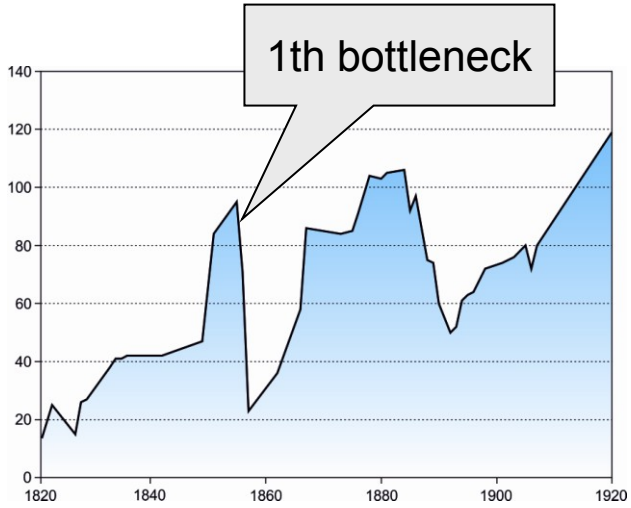
1851: a missionary arrival

1853: death of W. Glass

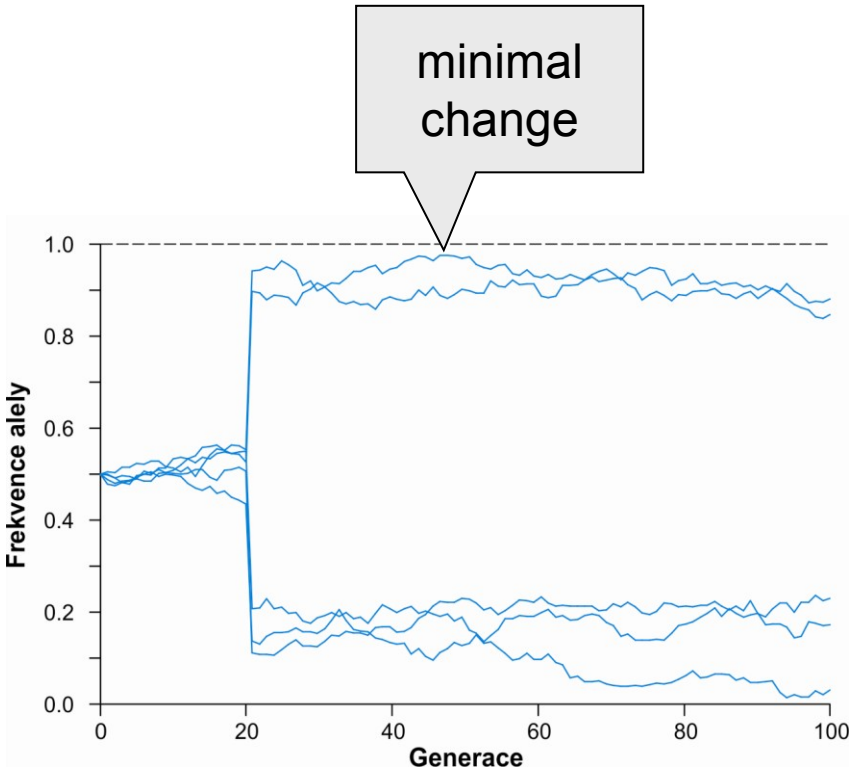
1856: departure of 25 Glass´s descendants to America, departure of other 45 people with the missionary

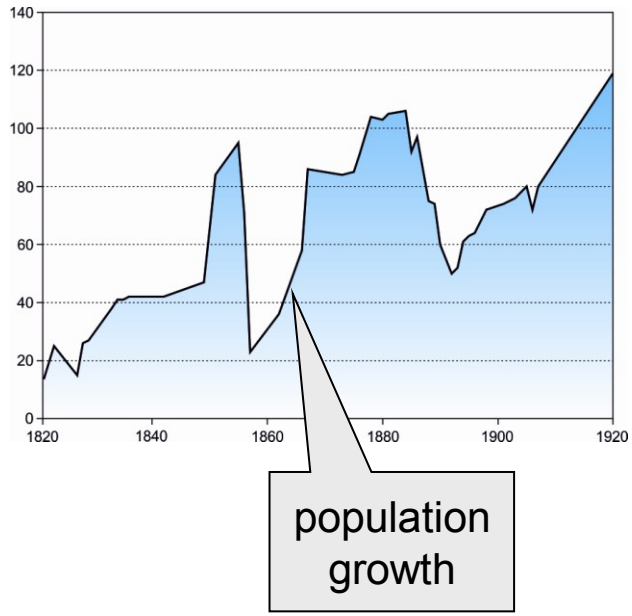
$\Rightarrow$ 103 inds. (1855) $\rightarrow$ 33 (1857) ... 1st bottleneck





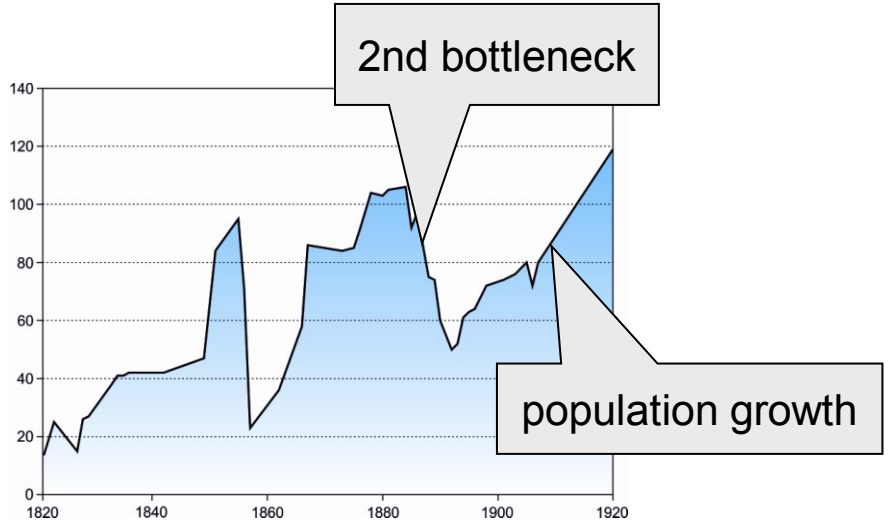
1857–1884: population growth ⇒ conservation of changes caused by previous bottleneck → less changes during 27 years than during 2 years 1855–1857



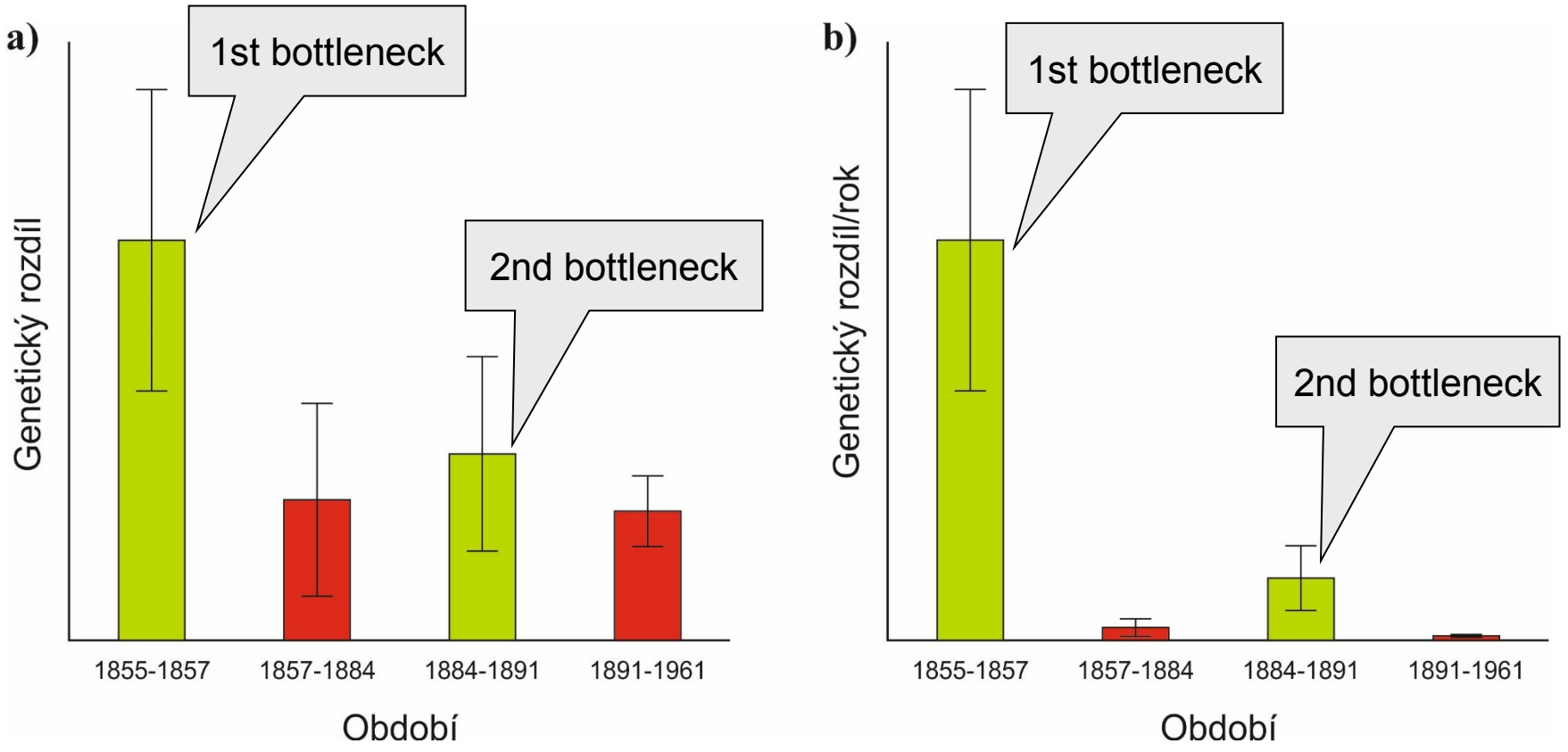


1884–1891: drowning of 15 men, only 4 adult remains, of whic 2 very old („Island of Widows“) → departure of many widows with their children

⇒ 106 inds. (1884) → 59 (1891) ... 2nd bottleneck

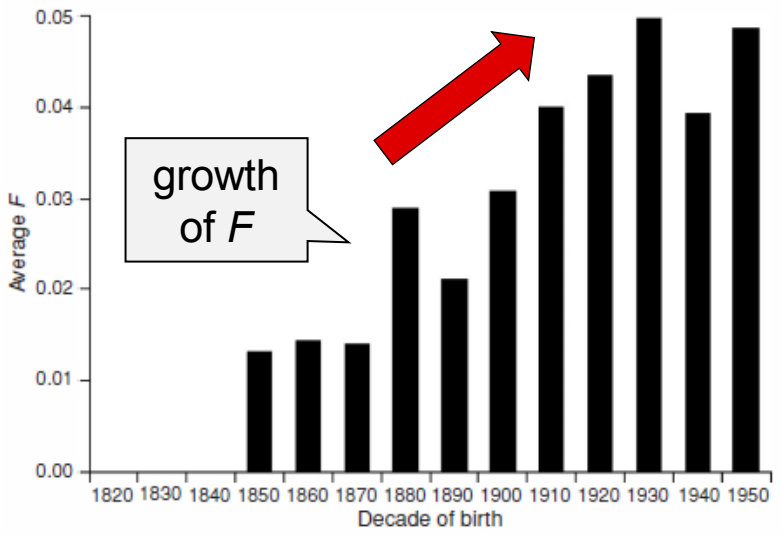


again, the following growth has „frozen“ the changes



Genetic changes during population growth lower than during bottlenecks

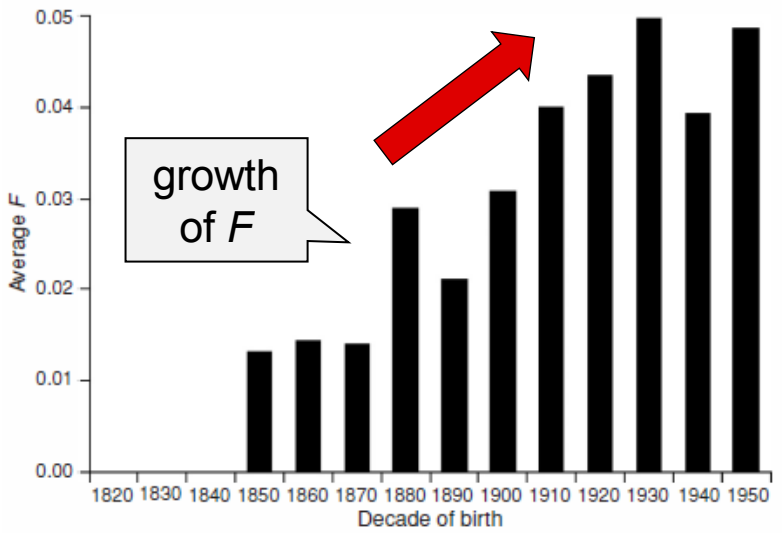
Inbreeding on Tristan da Cunha:



Despite the outbreeding strategy (choice of the least related partner), ie. $F_{IS} < 0$, the level of autozygosity increased

?

Inbreeding on Tristan da Cunha:



Despite the outbreeding strategy (choice of the least related partner), ie. $F_{IS} < 0$, the level of autozygosity increased

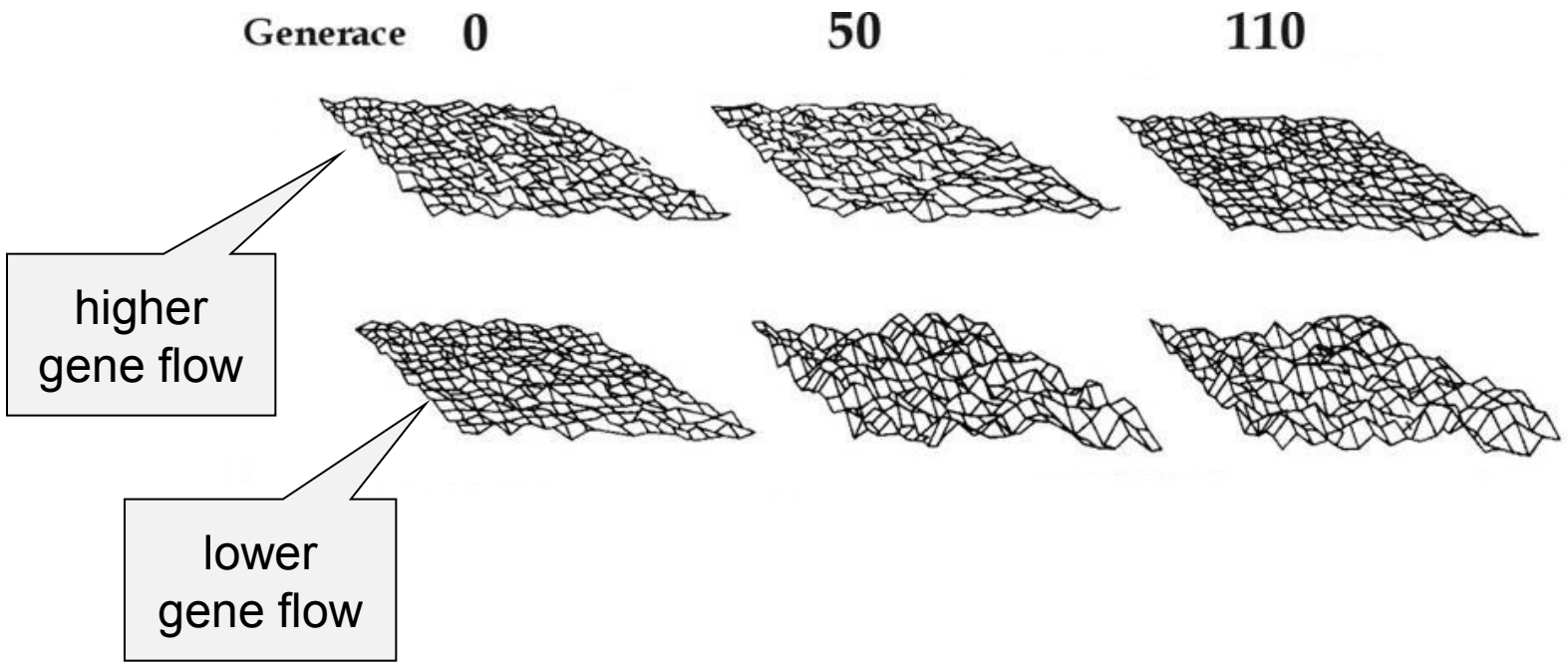
Table 3.3. First Eight Marriages between Biological Relatives on Tristan da Cunha Showing Date of Marriage, Number of Available Women of Marriageable Age,[a] and Number of Available Women Not Related to Groom

Marriage between Relatives	Date of marriage	Number of available women	Number of non relatives
1	1854	7	3
2	1856	9	2
3	1871	1	0
4	1876	1	0
5	1884	7	1
6	1888	8	0
7	1893	3	0
8	1898	1	0

[a] Sixteen years and over, single, and not a sister of the groom.

no unrelated woman available!

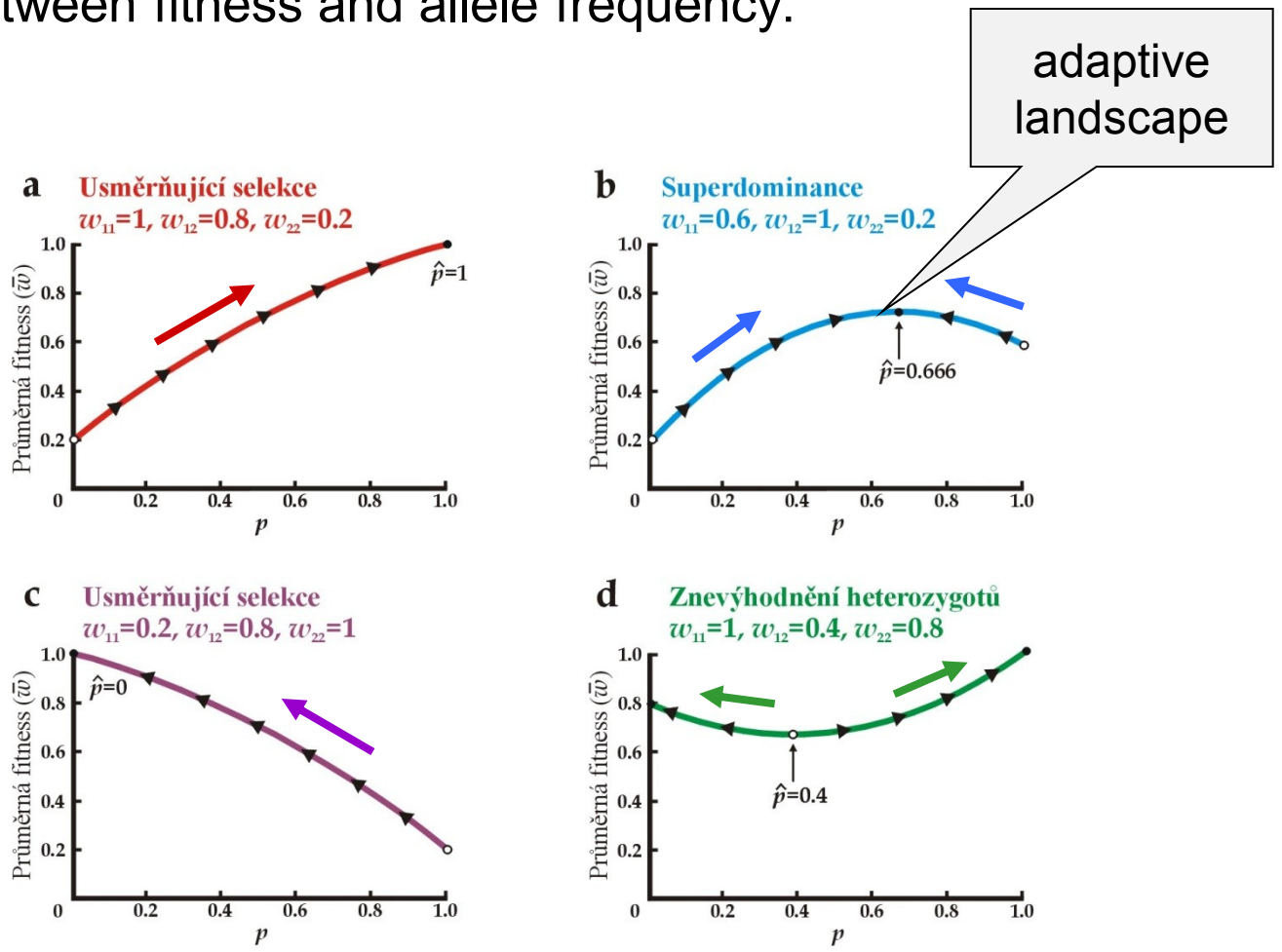
RELATION BETWEEN DRIFT AND GENE FLOW



Gene flow and drift have opposite effects:
drift increases divergence among demes × migration „homogenizes“ demes

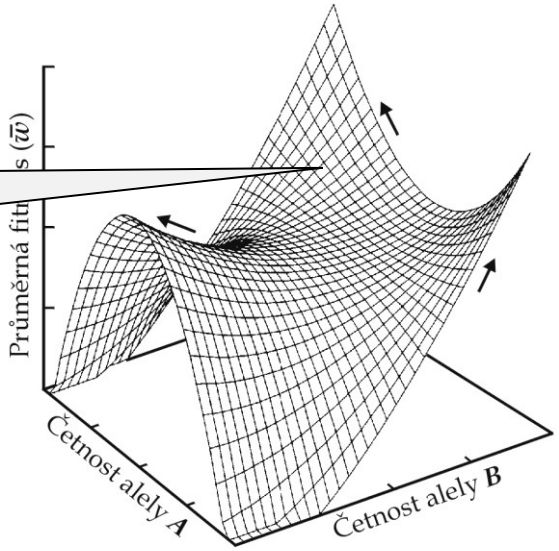
RELATION BETWEEN DRIFT AND SELECTION

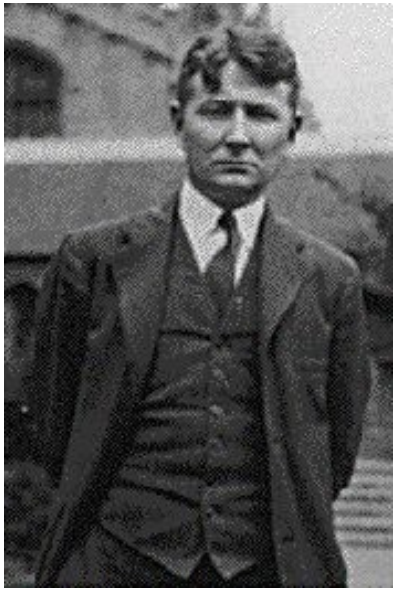
relation between fitness and allele frequency:



Adaptive landscape:

selection „pulls“ the population up





Sewall Wright

The notion of adaptive landscape has 2 mutually incompatible meanings:

1. Allele combinations: fitness values assigned to genotypes
   N genotypes → *N* + 1 dimensions
   discontinuous surface, population = cluster of points

2. Average allele frequencies
   number of dimensions = number of sets of allele frequencies
   continuous surface

Shifting balance theory (SBT)

Assumptions:

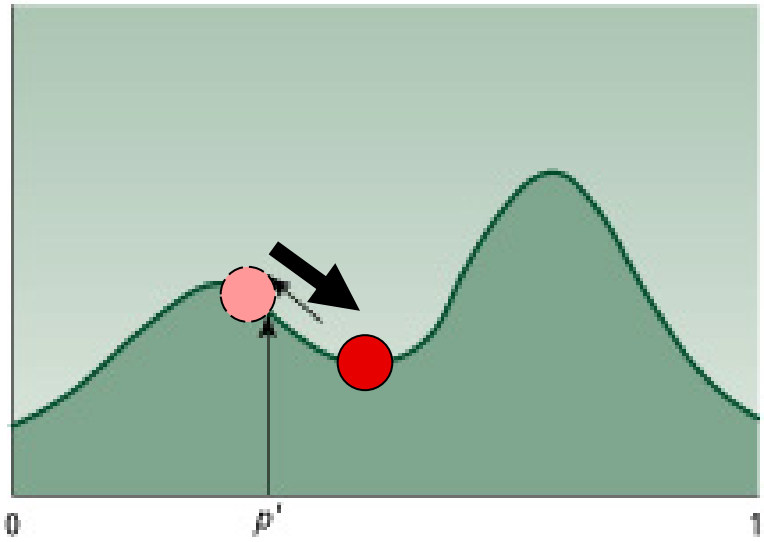
environment changes $\Rightarrow$ populations in constant change

mutations $\Rightarrow$ new dimensions, new ways upwards

small populations (drift) $\Rightarrow$ possibility to move down to adaptive valleys

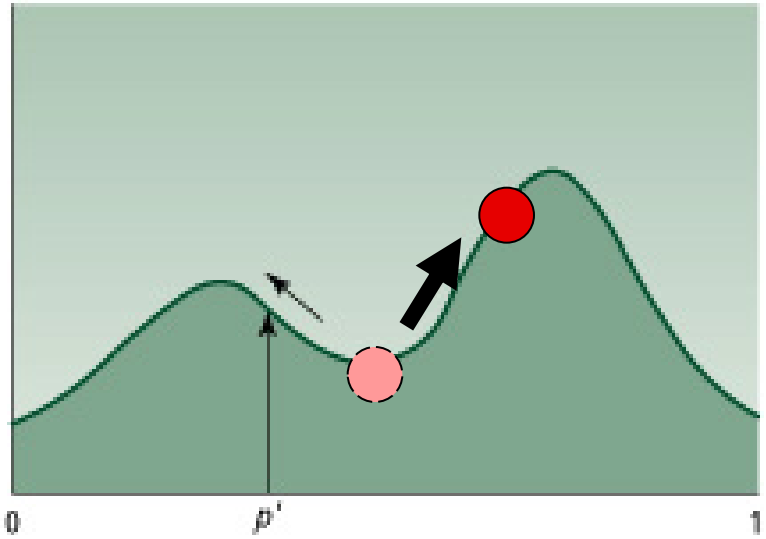
3 phases of SBT:

1. contemporary fitness reduction of a local population due to drift $\rightarrow$ chance to approach the area of attraction of a higher peak



3 phases of SBT:

2. intrademic selection → „pulling“ of the population towards a new peak



3. interdemic selection → spread of the deme´s members at the higher peak to surrounding demes

The whole proces seen as shifting of the balance between drift, intrademic, and interdemic selection

2 views on evolution in populations:

S. Wright

R.A. Fisher

small local populations

combination of selection, drift and migration

epistasis, pleiotropy,
dependence of allele effects on context

speciation as a byproduct of local adaptations in epistatic systems

large panmictic populations

mutation and selection

additive effects of genes,
allele effects independent of context

disruptive or locally divergent selection