ORIGIN OF GENETIC VARIATION

Consequence of the H-W principle:

if the assumptions of the H-W population hold true, polymorphism can be maintained solely by random mating and Mendelian inheritance

BUT!

real populations usually differ from the model:

population size finite

mating may be nonrandom

migration

selection

emergence of new alleles by mutation

MAIN MICROEVOLUTIONARY MECHANISMS:

mutation (incl. transposition)

recombination

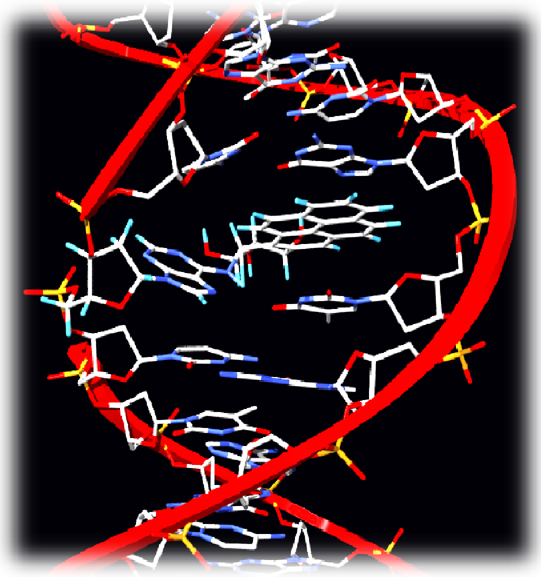
migration (gene flow)

nonrandom mating

natural selection

random genetic drift (incl. bottleneck, founder effect)

(molecular drive)



MUTATION

spontaneous × induced

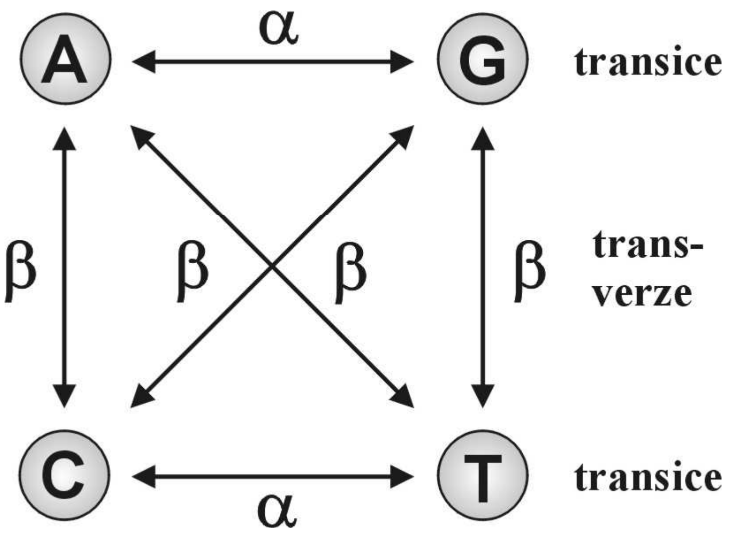
in germ cells × somatic

according to their deleterious/beneficial effect:

beneficial (positive)
deleterious (lethal, negative)
neutral

According to effect

point (gene)
chromosomal
genome



Point mutations:

substitutions (transitions, transversions)

synonymous

nonsynonymous

GTC → GTA
Val → Val

GTC → TTC
Val → Phe
AAG → TAG
Lys → *ochre* (stop)

missense
nonsense

insertions ACGGT → ACAGGT
delections ACGGT → AGGT
} indels → shift of reading frame

back mutations: generally 10-times lower frequencies

recurrent mutations → mutation pressure:

eg. when allele frequency $A = 0{,}5$; $2N = 2000$, i.e. : $N_A = 1000$

after 1st generation → $N = 1001$ ⇒ increase to 0,5005

after 100 generations → 0,55 ...

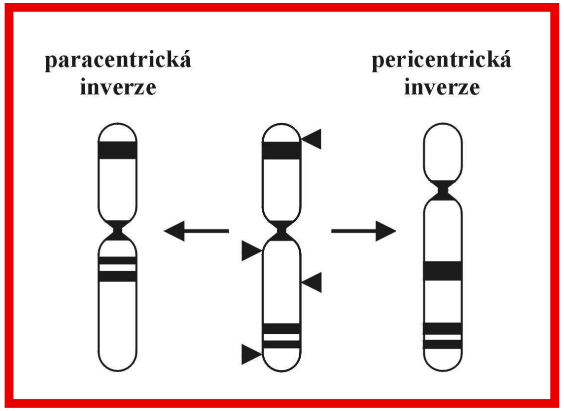
⇒ change of allele frequencies by mutations very slow

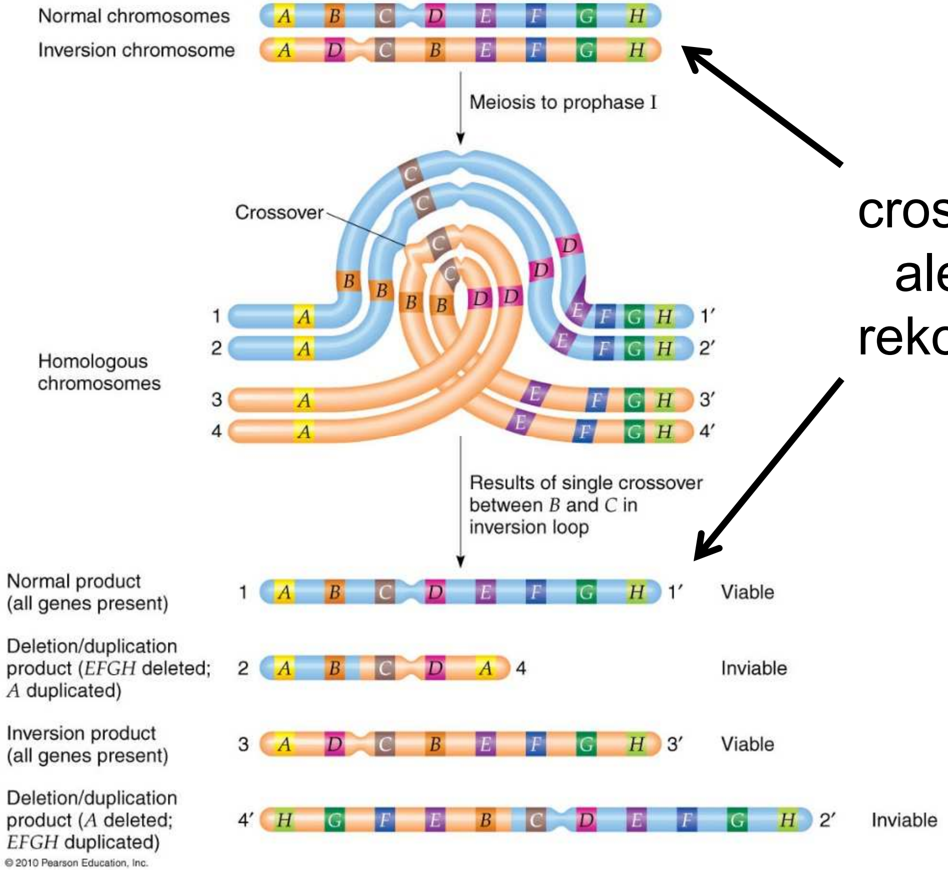
Chromosomal mutations (chr. rearrangements)

inversions

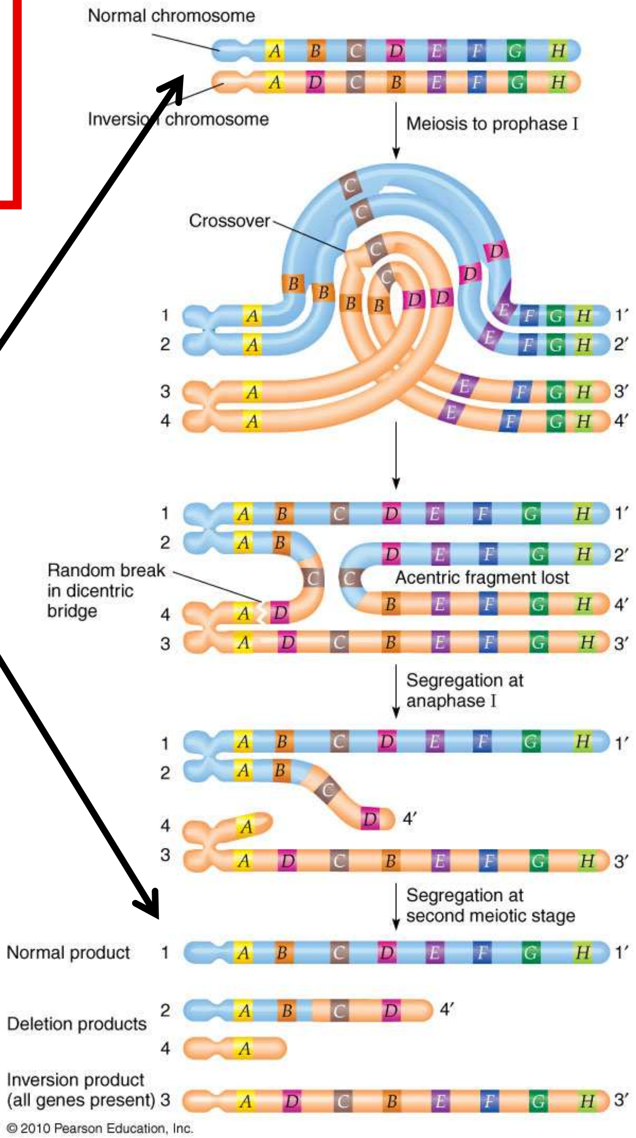
pericentric

paracentric





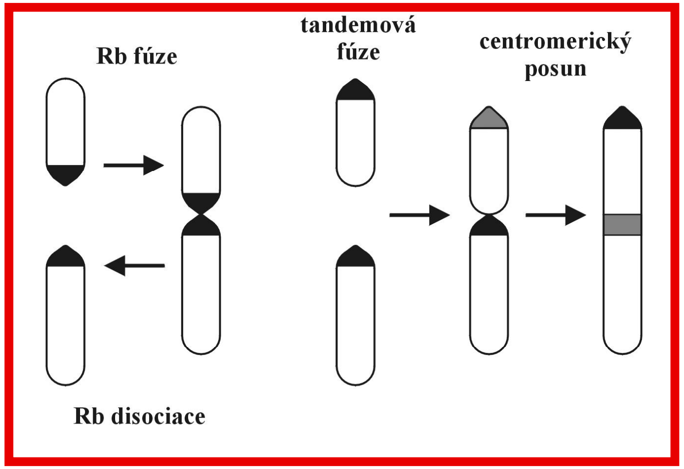
crossing-over
ale žádná
rekombinace!

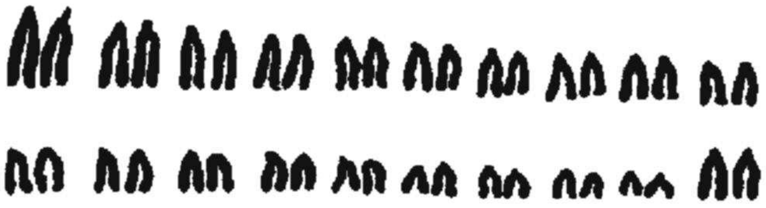


translocations

fusions and dissociations
(Robertsonian translocations)

house mouse





Rapid chromosomal evolution in house mice on the island of Madeira

One population of mice introduced to island in 1400s

Two populations evolved different sets of Robertsonian translocations, hybrid offspring are sterile

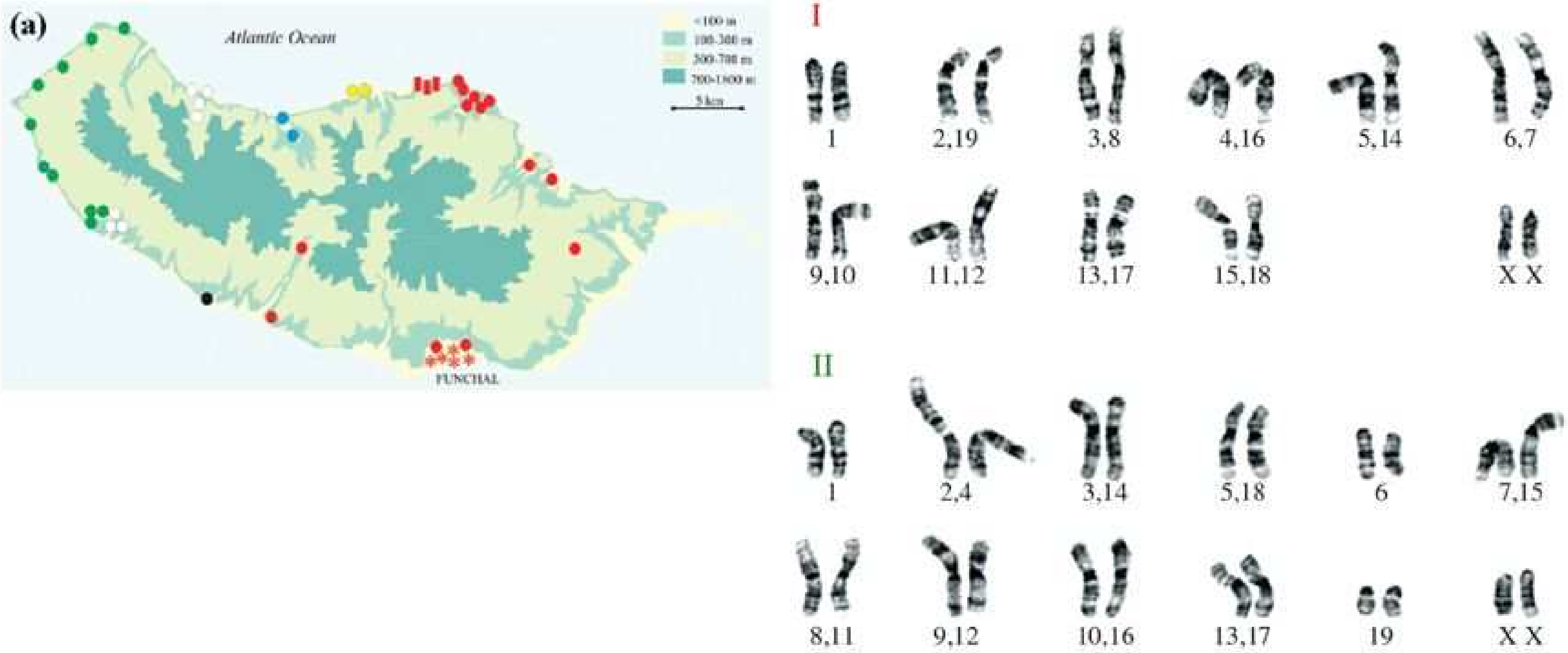


Fig. 13.30

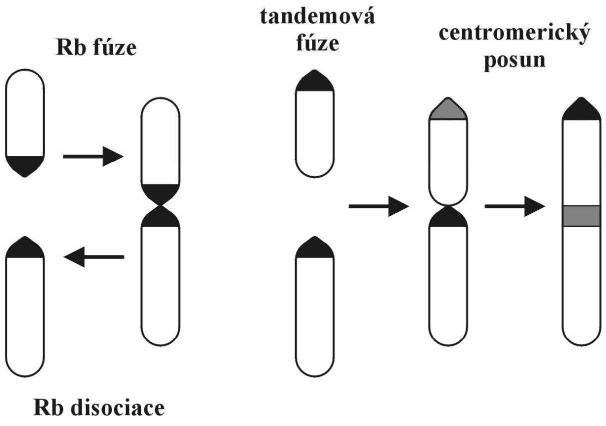
Hartwell et al., 4th edition, Chapter 13

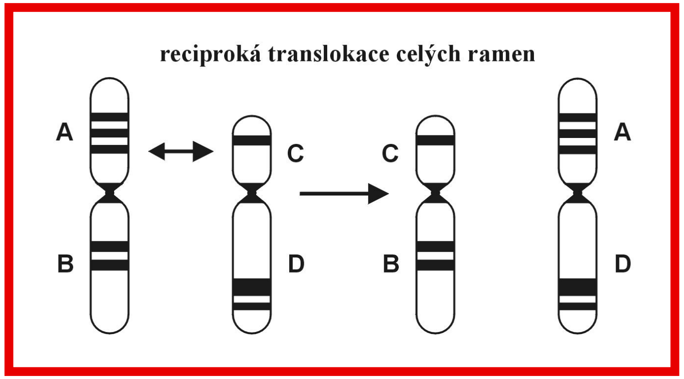
translocations

fusions and dissociations

whole-arm reciprocal translocations (WART)

house mouse

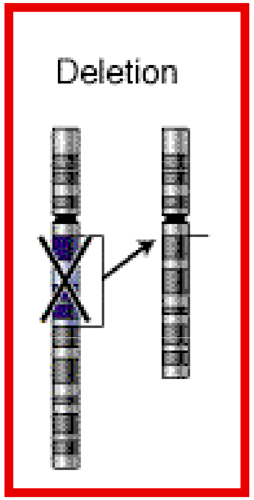


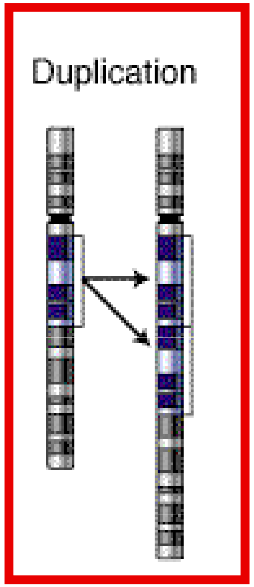


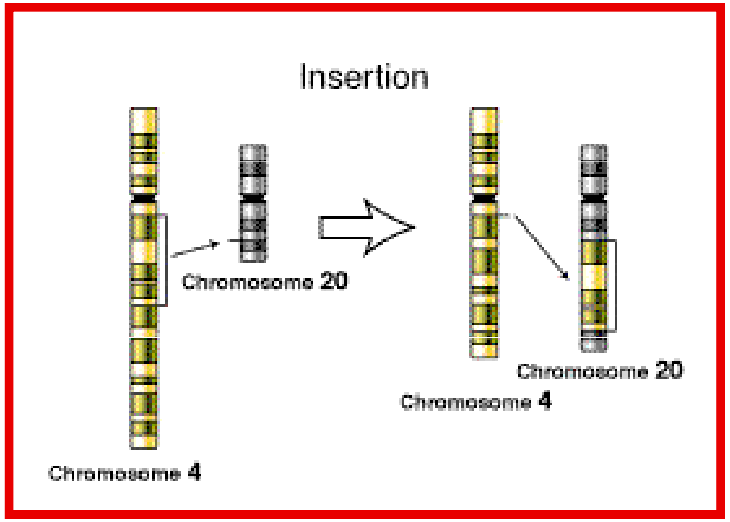
deletions

duplications

insertions







Genome mutations

-somies (monosomies, trisomies)

mostly incompatible with life

monosomies: the only viable = X0 (Turner syndrom)

trisomies: imbalance in gene dosage (increased expression of the trisomic pair)

viable trisomies : XXY, XXX, XYY, Patau syndrom (chr. 13), Edwards s. (chr. 18), Down s. (chr. 21)

-ploidies (polyploidy)

especially plants

in animals less frequent (invertebrates, fishes, amphibians)

during the vertebrate evolution 2 rounds of whole genome duplications (2R-hypothesis)

polyploid individuals usually bigger (increased cell volume)

odd multiples of the genome → problems in meiosis ⇒ reproductive barrier (not always – eg. triploid frogs)

autopolyploidy: combination of two identical genomes

fusion of cells
endoreplication
abortive cell cycle

allopolyploidy: combination of two different genomes

fusion of diploid gametes
polyspermy

Randomness and mutation rate (μ)

mutation effects random, position and rate nonrandom

transitions > transversions

mutation „hotspots“: CpG in animals (methylated C → T); TpT in Procaryota „SOS reactions“ in Bacteria, minisatellites (VNTR), microsatellites (STR)

mtDNA > nuclear DNA

sex chromosomes > autosomes

influence of proximity of the replication start, centromeres, telomeres, repetitive sequences, intensity of transcriptions

cold-blooded animals: > temperature $\Rightarrow$ > μ

RNA viruses (HIV)

parasites

antigens, immunoglobulins

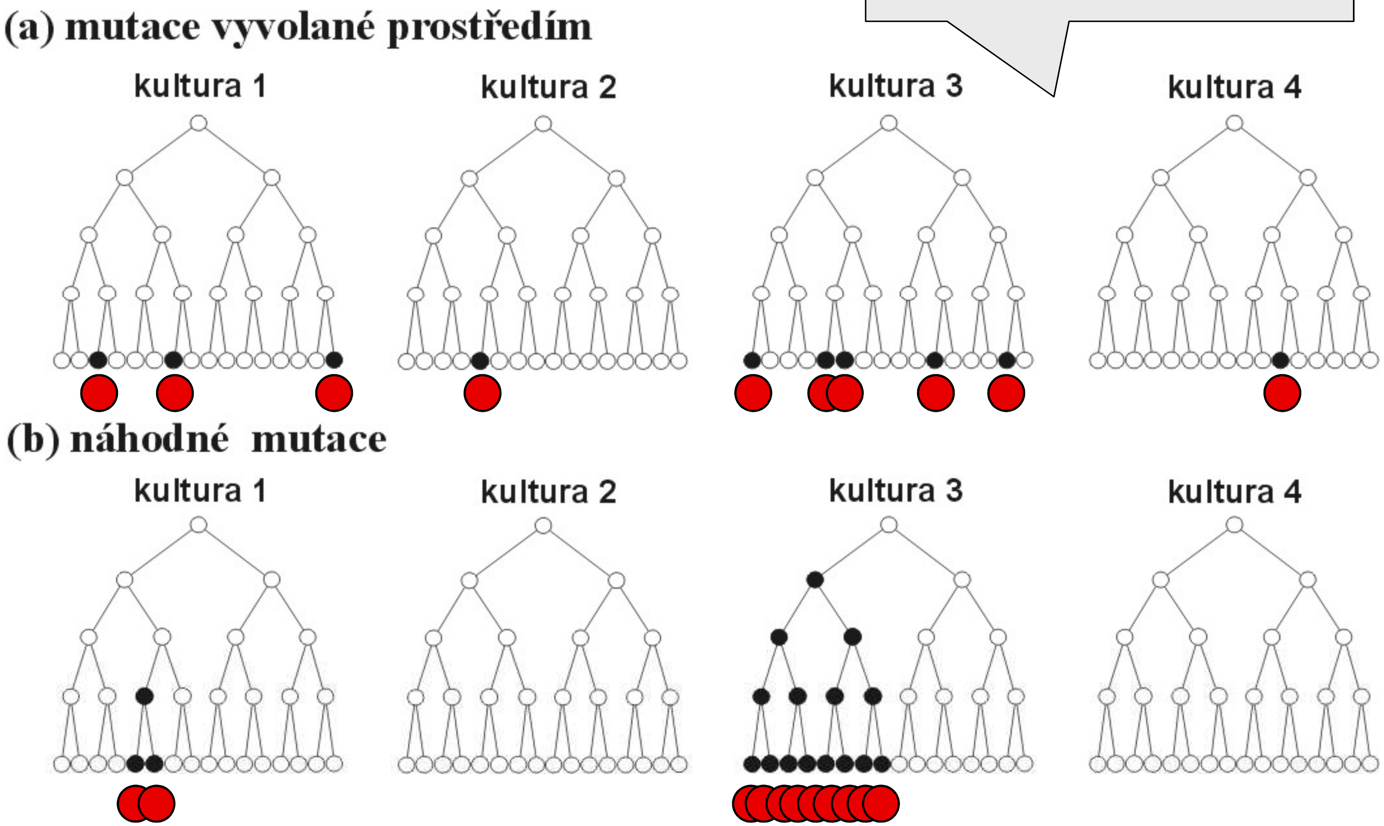
> μ of somatic mutations

males > females: humans 6x, rodents, fox: 2x … more cell divisions in germ cells

Adaptive (directed) mutations?

Max Delbrück, Salvador Luria (1943):
fluctuation test

directed mutations

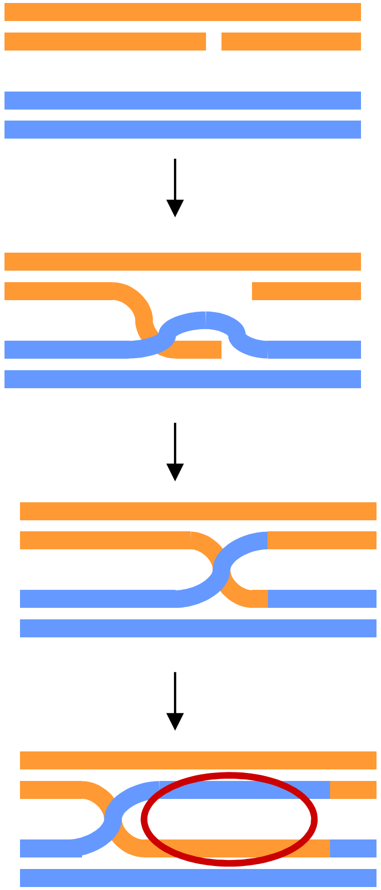


random mutations

RECOMBINATIONS

mutations → new alleles

recombinations → new genotypes (exception = intragenomic recombination)



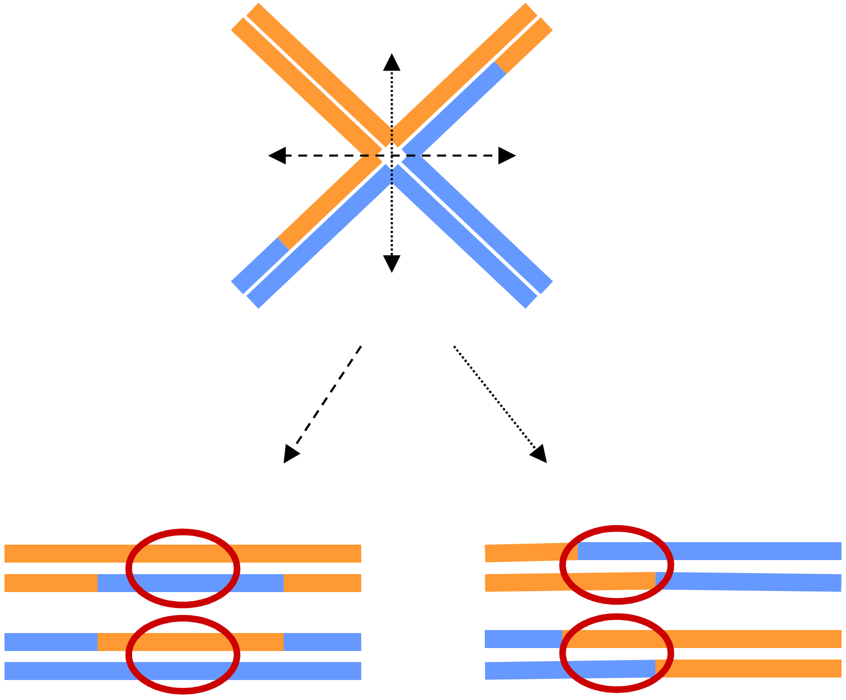
Vytvoření zlomu na jednom chromozomu.

Invaze řetězce do homologního chromozomu.

Spojení řetězců homologních chromozomů. Holidayův spoj

Migrace Holidayova spoje. Vytvoření heteroduplexu.

Štěpení Holidayova spoje



in many organisms crossing-over important for right meiosis
(at least 1 c-o per chromosome, otherwise aneuploidies)

women with > c-o → > children

children of older women → > recombinations

differences in various parts of chromosome (near centromeres and telomeres etc., differences among organisms)

small chromosomes > recombination frequencies

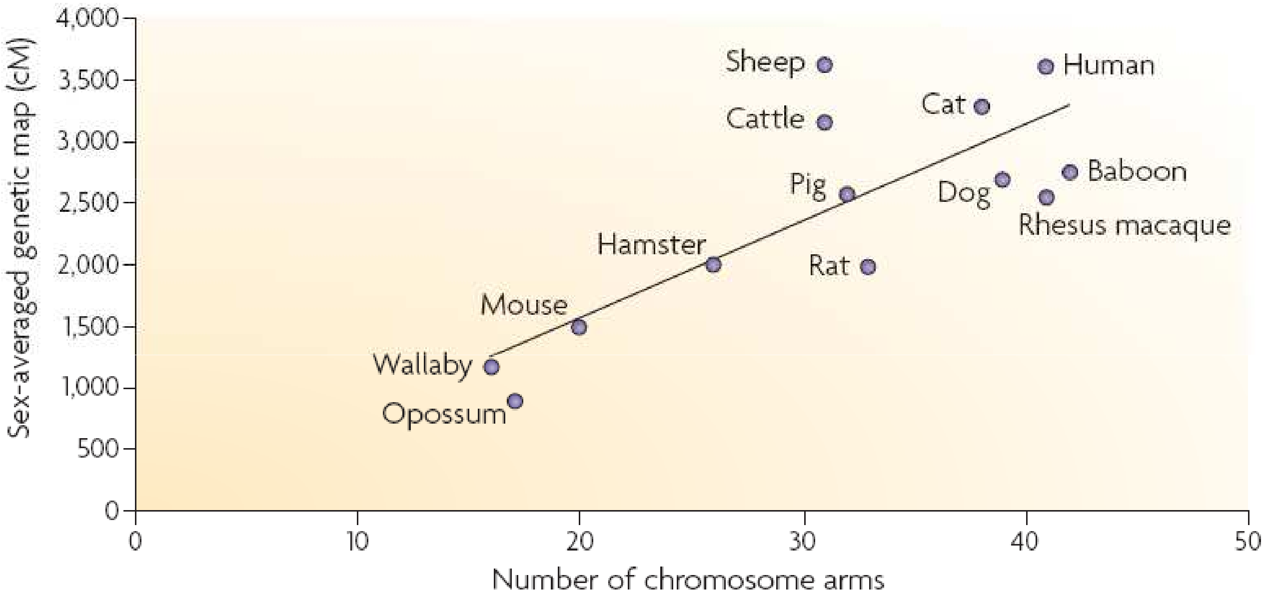
recombination „hotspots“:
humans ~25 000
absent in *Drosophila* and *Caenorhabditis elegans*
frequent appearance and disappearance
demise of 1 point often compensated by increased aktivity of a neighbour point

differences in recombination rate between sexes:

- Haldane-Huxley rule: if one sex doesn´t recombine, it is the heterogametic sex
- if both sexes recombine, mostly in females > recombinations (man 1,7x, mouse 1,3x)

differences between species:

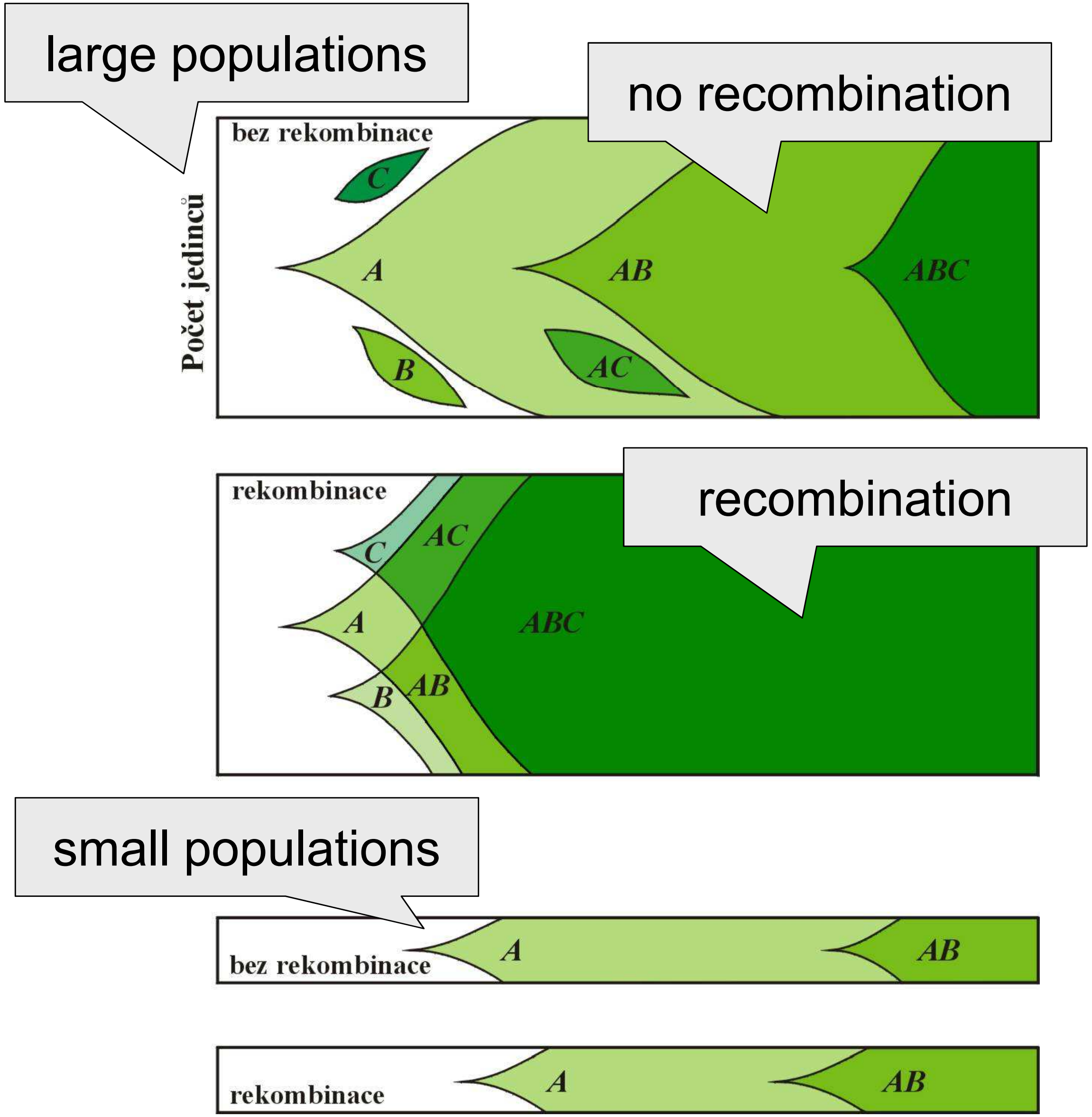
- species with more small chromosomes → more recombinations than species with less large chromosomes
- correlation with the number of arms: more recombinations in karyotypes with large numbers of chrom. arms (at least 1 c-o/arm to avoid aneuploidies?)



EVOLUTIONARY CONSEQUENCES OF RECOMBINATION:

Recombination and polymorphism:

absence of recombination
⇒ linkage disequilibrium


large populations

no recombination

recombination

small populations

EVOLUTIONARY CONSEQUENCES OF RECOMBINATION:

Recombination and polymorphism:

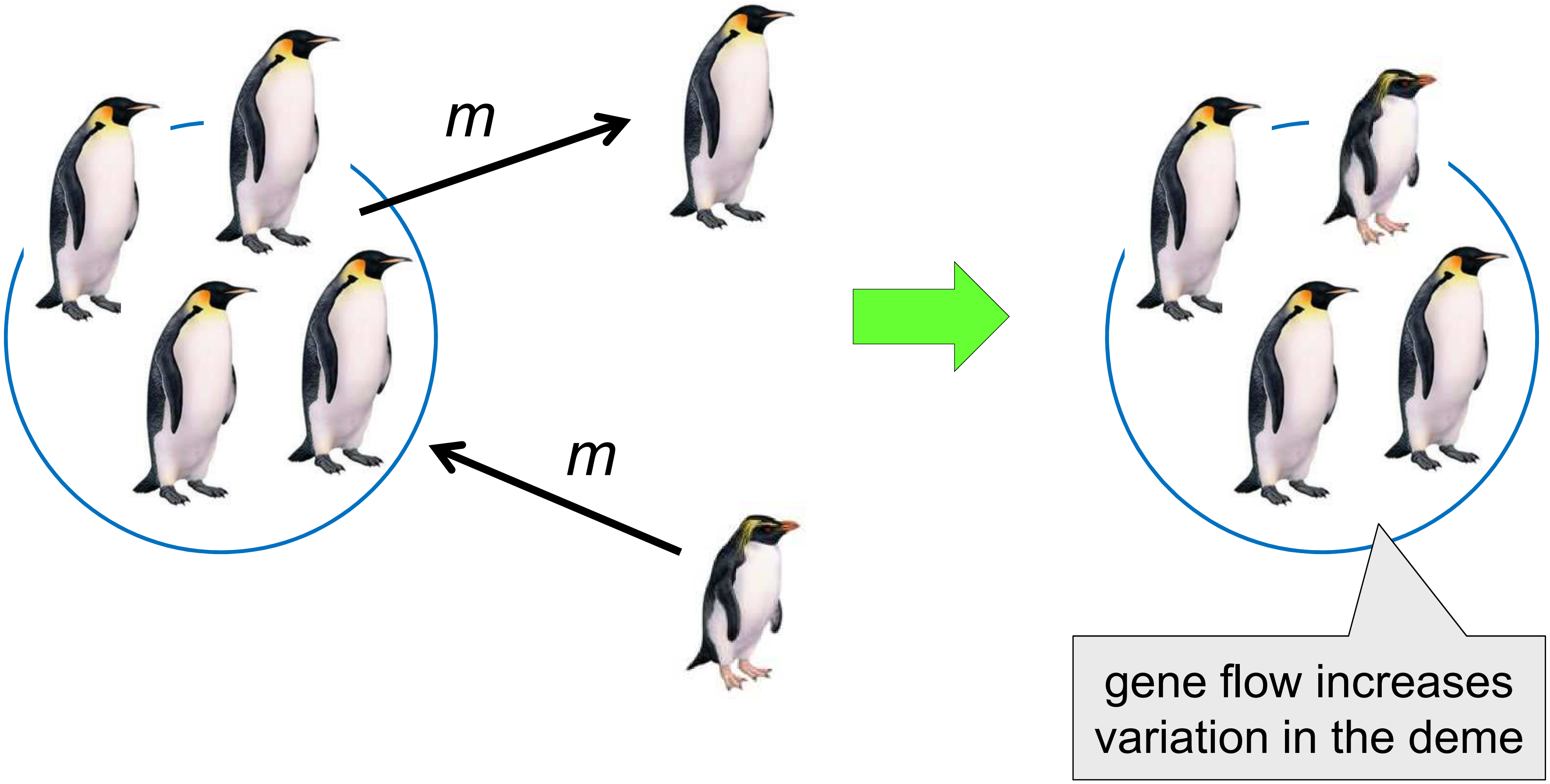
pozitivní selekce: *hitchhiking* (*draft*) → *selective sweep* (selekční smetení)

negativní selekce: *background selection*

→ loss of polymorphism

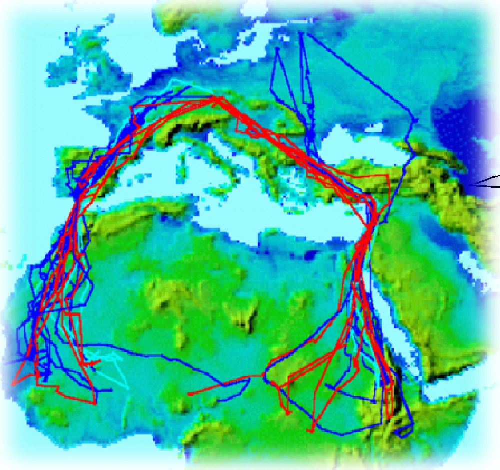
MIGRATION (GENE FLOW)

Migration rate, *m* = proportion of gene copies appearing in the population by immigration from other populations in the given generation



gene flow increases variation in the deme

~~MIGRATION (~~GENE FLOW~~)~~



long-distance migration but no gene flow

gene flow but no migration

METHODS OF GENE FLOW ESTIMATION:

1. direct

capture-mark-recapture (CMR)

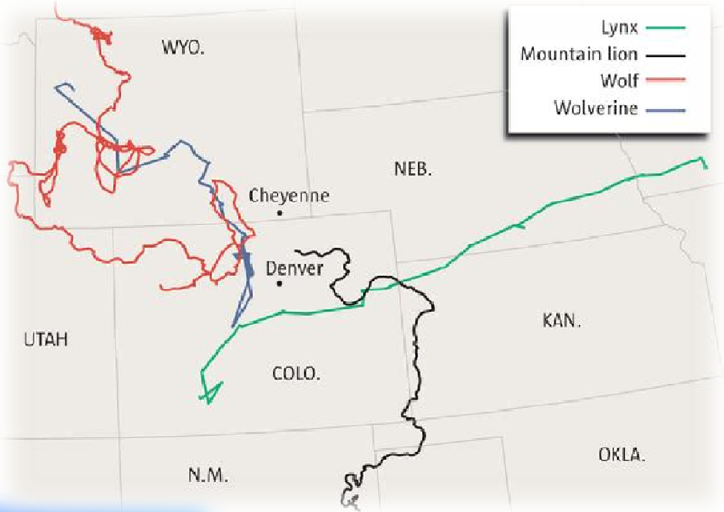
finger clipping, special dyes, tattooing, tags, rings, collars, genetic marking

1. Direct methods

remote tracking – telemetry

transmitters, anntenas; GPS systems

... more expensive, time consuming



Risk of underestimation of gene flow!!

2. Indirect methods

molecular markers

gene flow models

maximum likelihood and Bayesian programs

dispersal: distance between parents and offspring

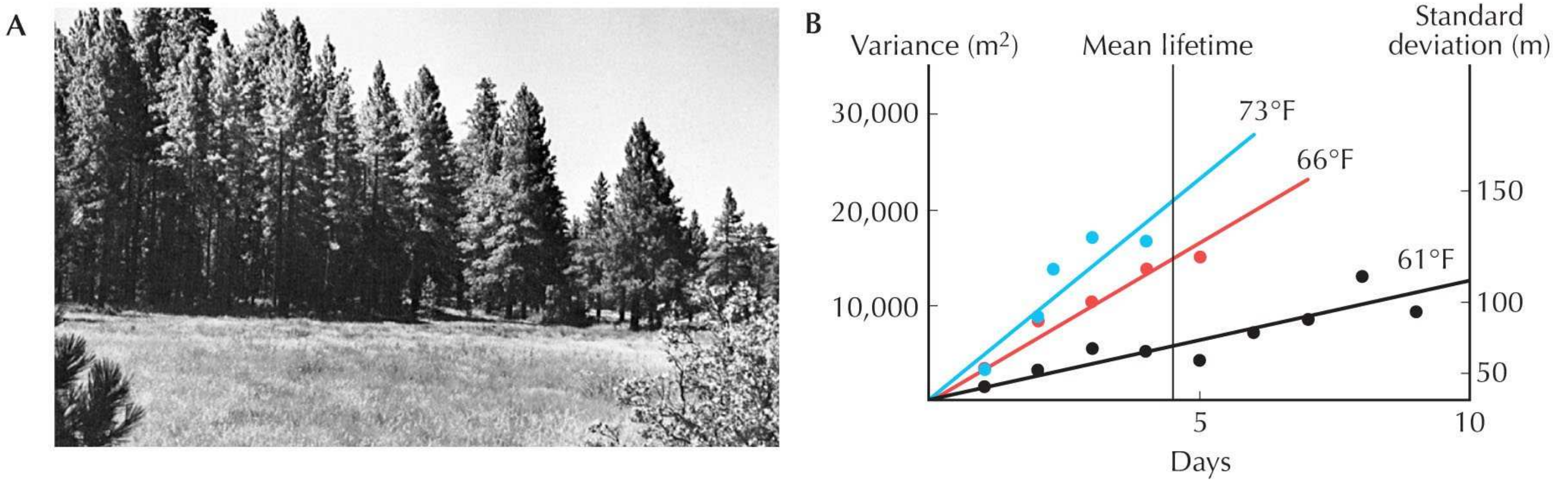
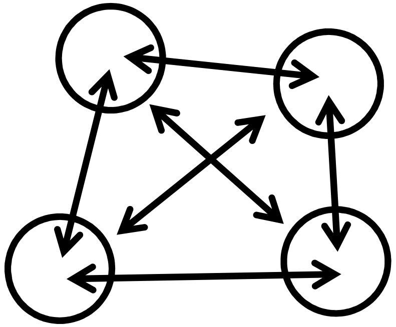


FIGURE 16.4. Dobzhansky and Wright (1943) measured the rate of dispersal of *Drosophila pseudoobscura* by releasing marked flies at sites in the Sierra Nevada, California (*A*). Over the following days, flies were caught in a series of traps. The graphs (*B*) show how the variance of the distribution of marked flies increased over time. The three sets of points show results from experiments at different times during the summer: Rates of movement increase strongly with temperature. The rate of diffusion of genes is estimated by assuming a mean lifetime of 4.5 days (*vertical line*).

A) Island model

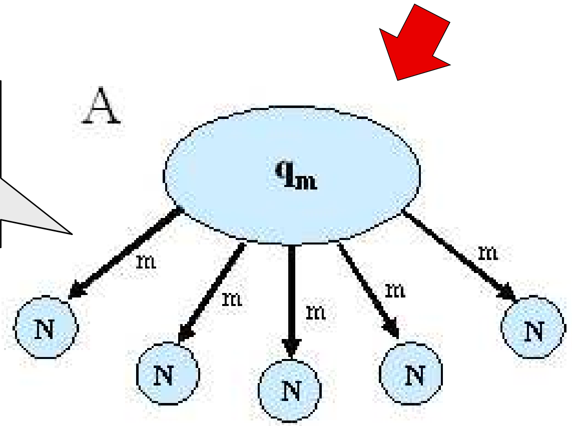


S. Wright (F-statistics): $F_{ST} = 1/(4Nm + 1) \Rightarrow Nm = (1/F_{ST} - 1)/4$

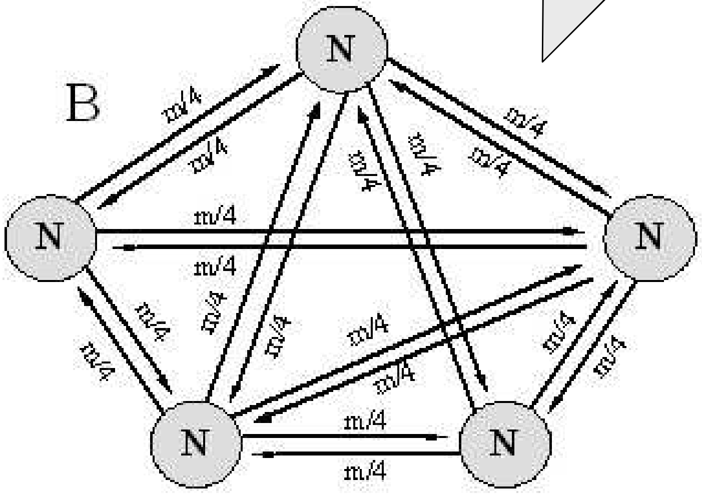
… Nm = number of migrants per generation

Island model can be also asymmetric:

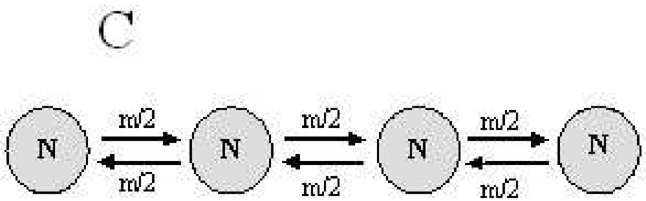
continent-island model



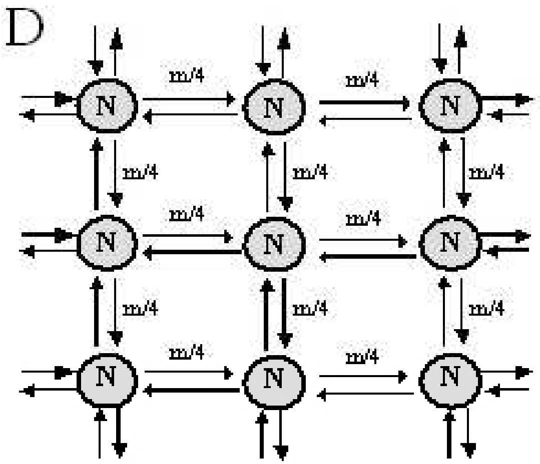
island model



B) Isolation by distance models
discontinuous = stepping stone model



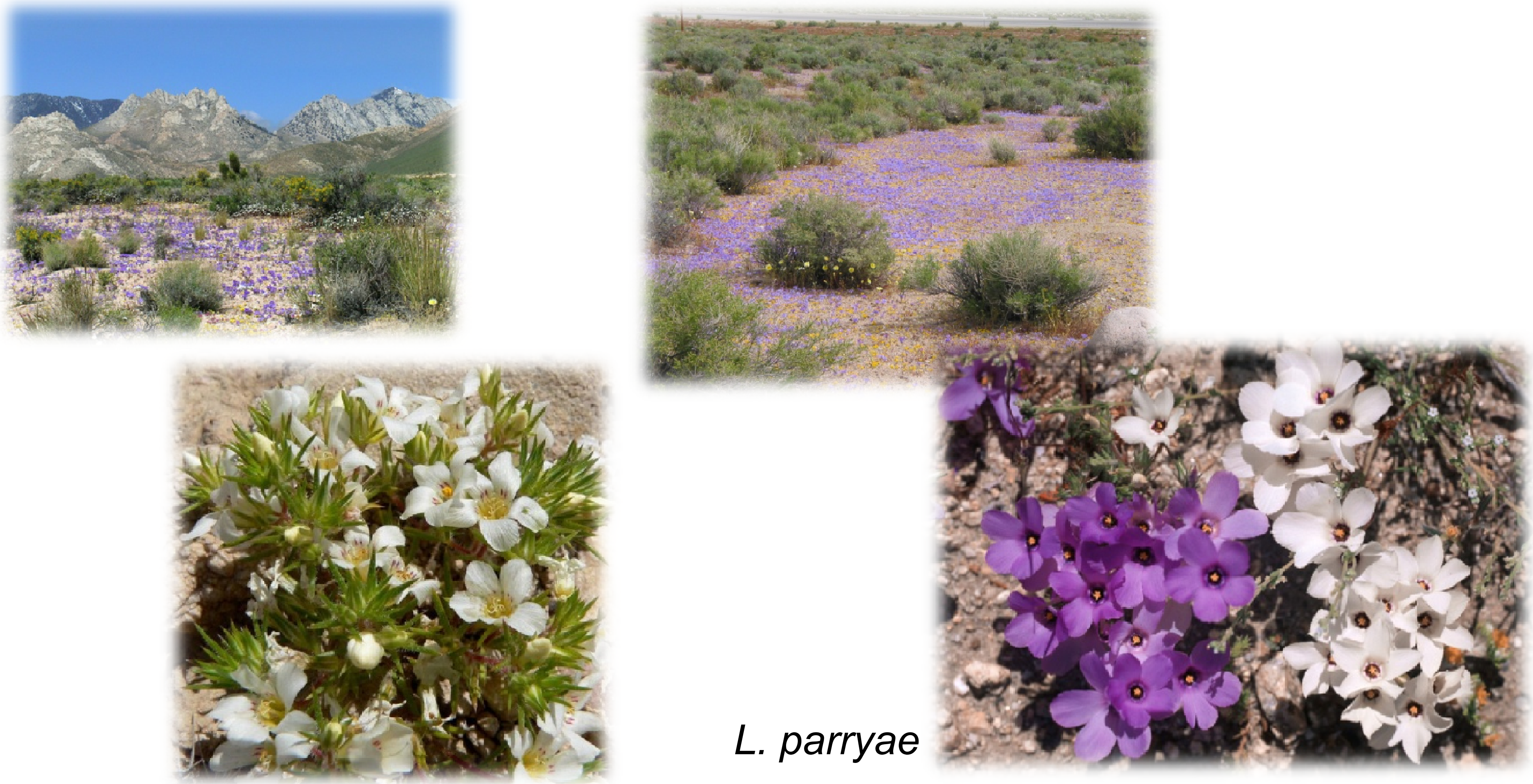
1D stepping-stone model



2D stepping-stone model

B) Isolation by distance models continuous

Linanthus parryae (Polemoniaceae), Mojave Desert (California)
T. Dobzhansky, Sewall Wright



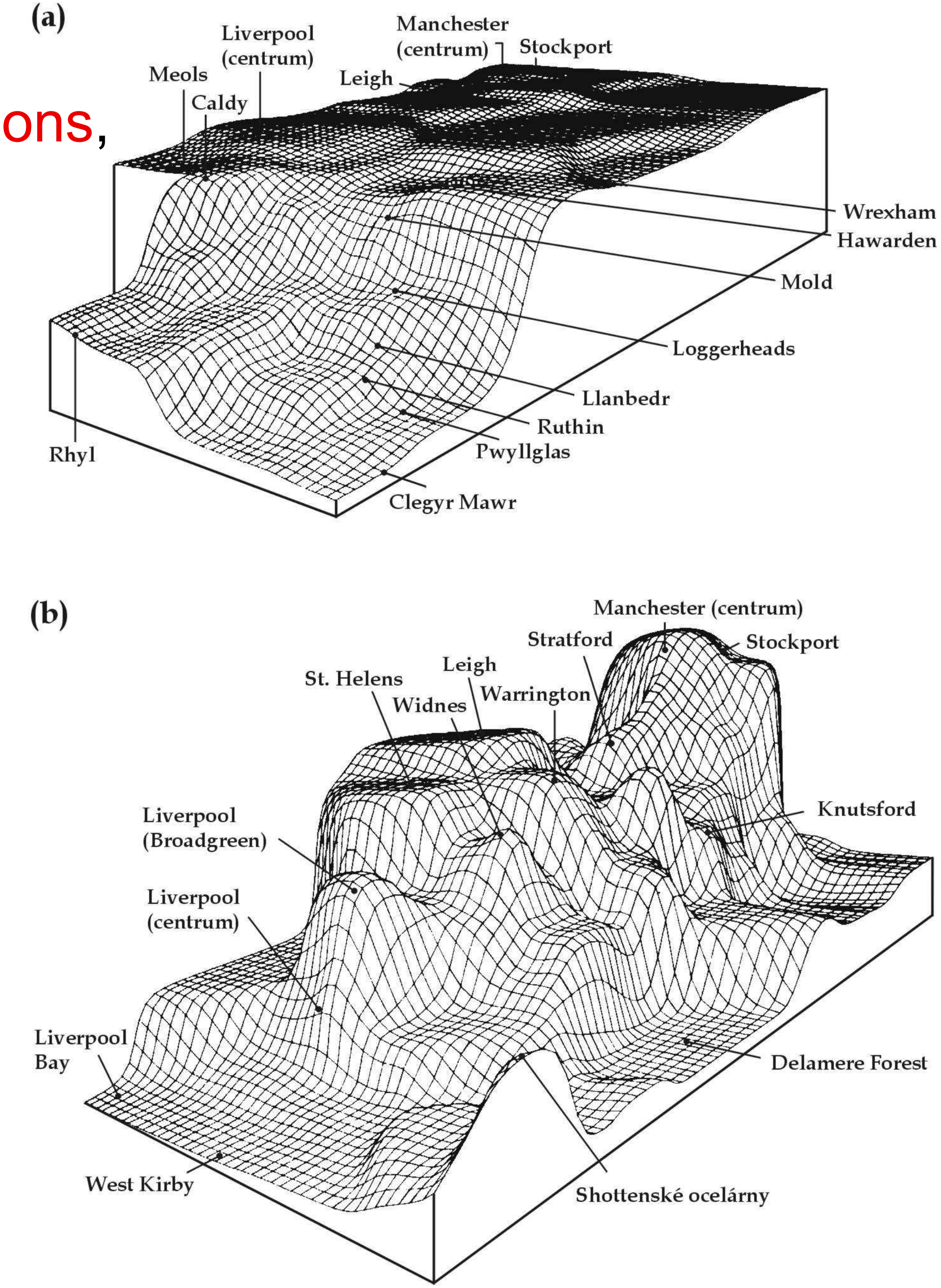
L. parryae

Gene flow consequences:

genetic homogenization of subpopulations, preventing their genetic divergence

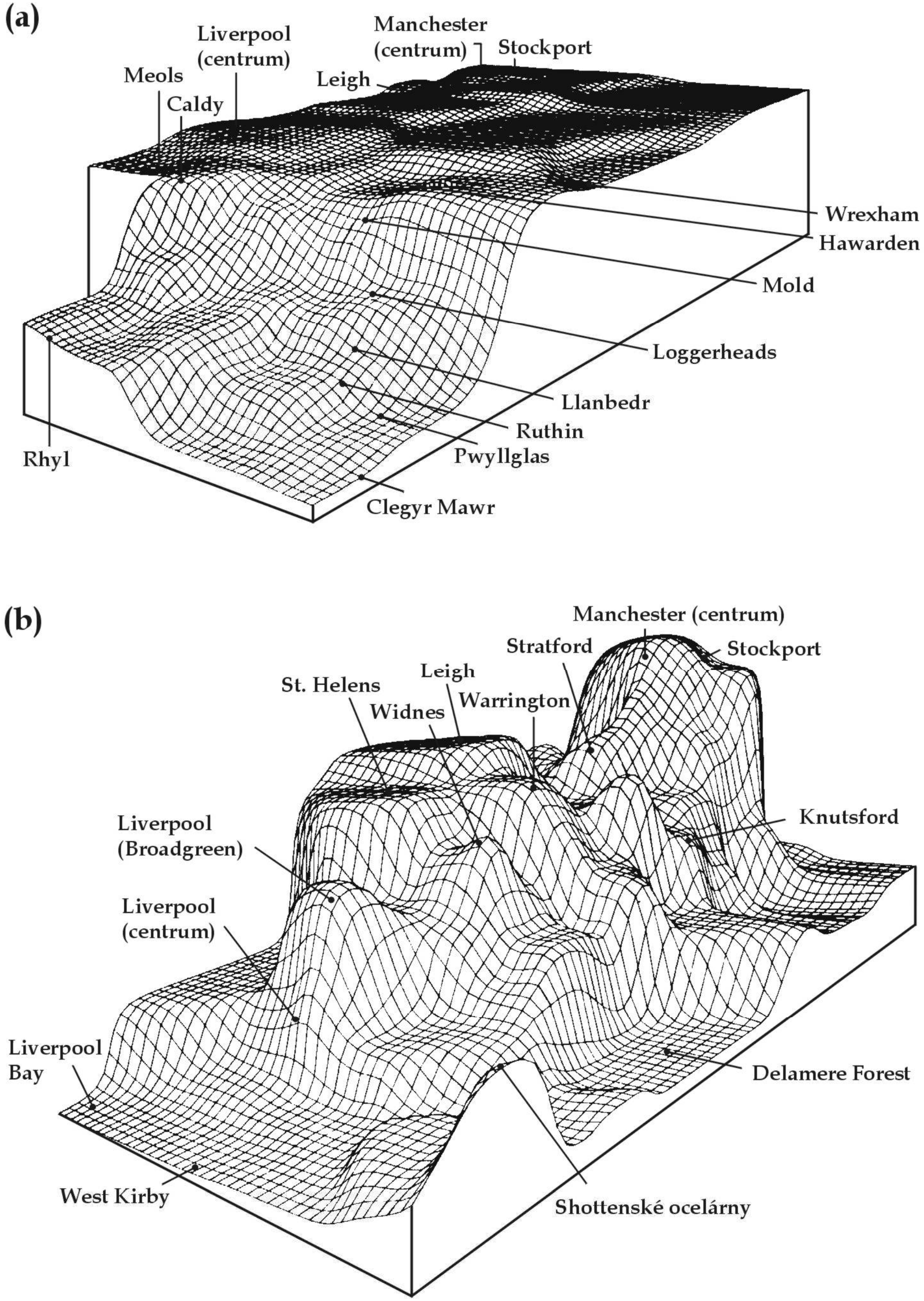
in many species migration severely reduced

Eg.: melanic forms of moths in England

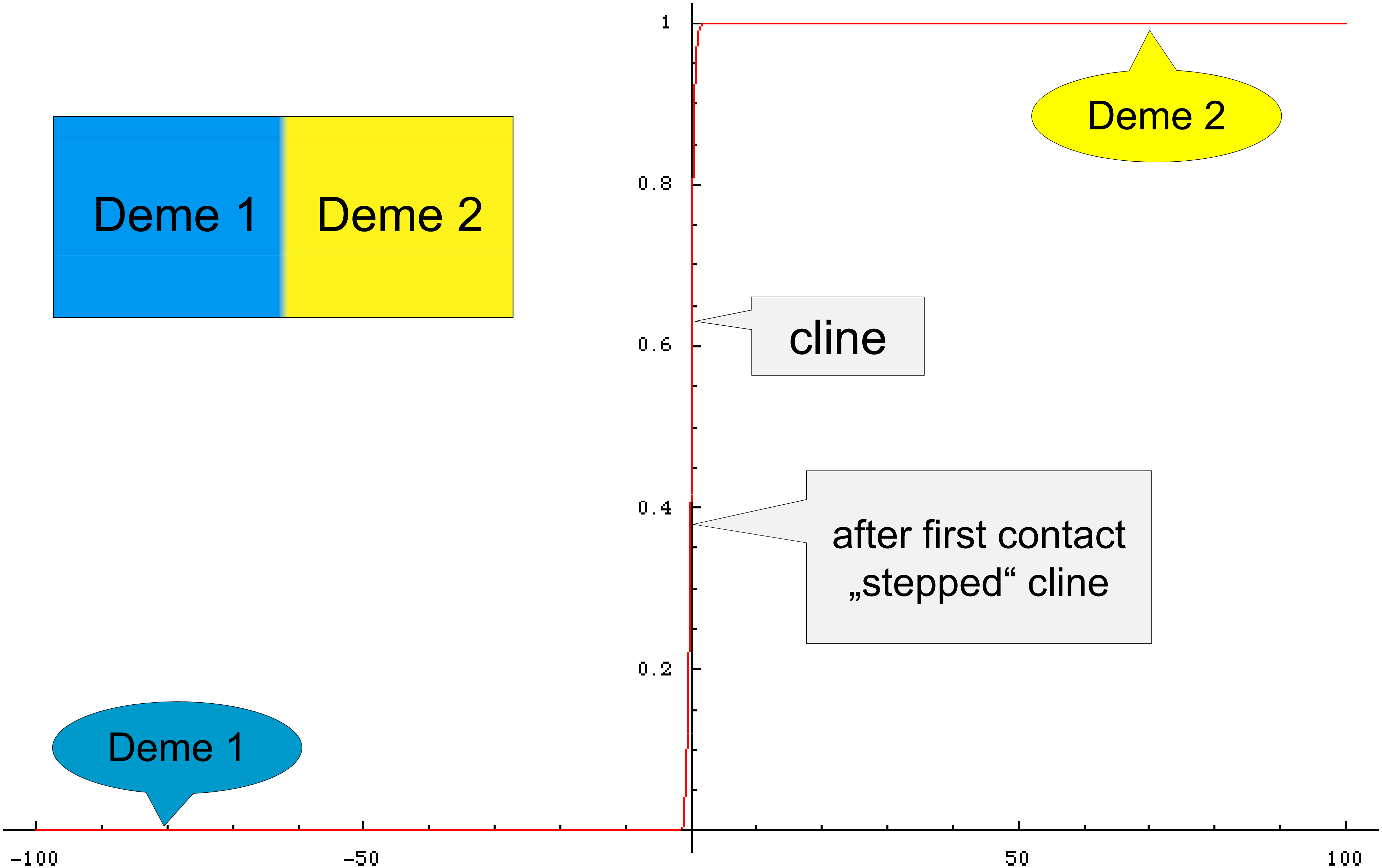


peppered moth (*Biston betularia*)

scalloped hazel
(*Odontoptera* [*Gonodontis*] *bidentata*)

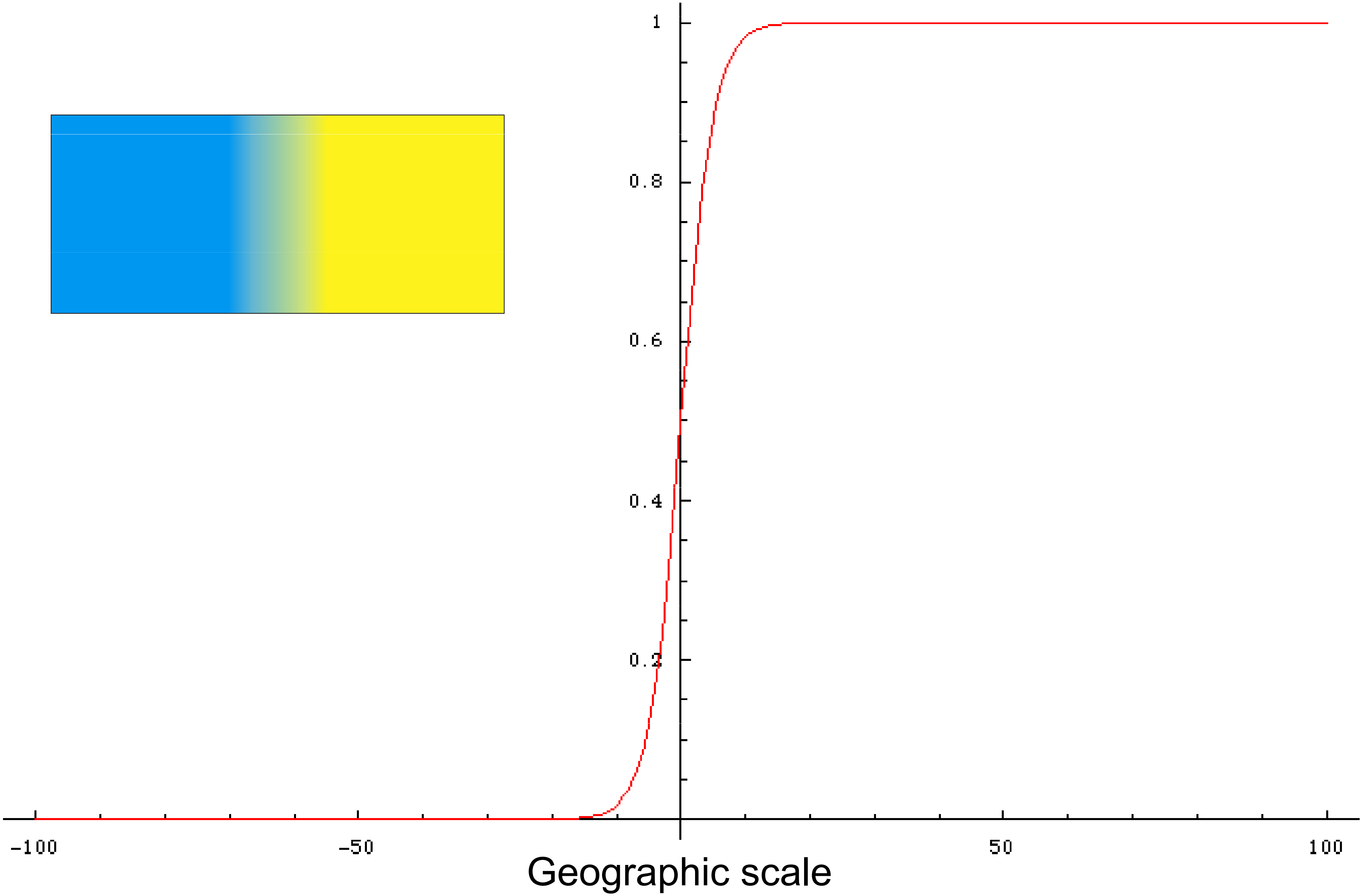


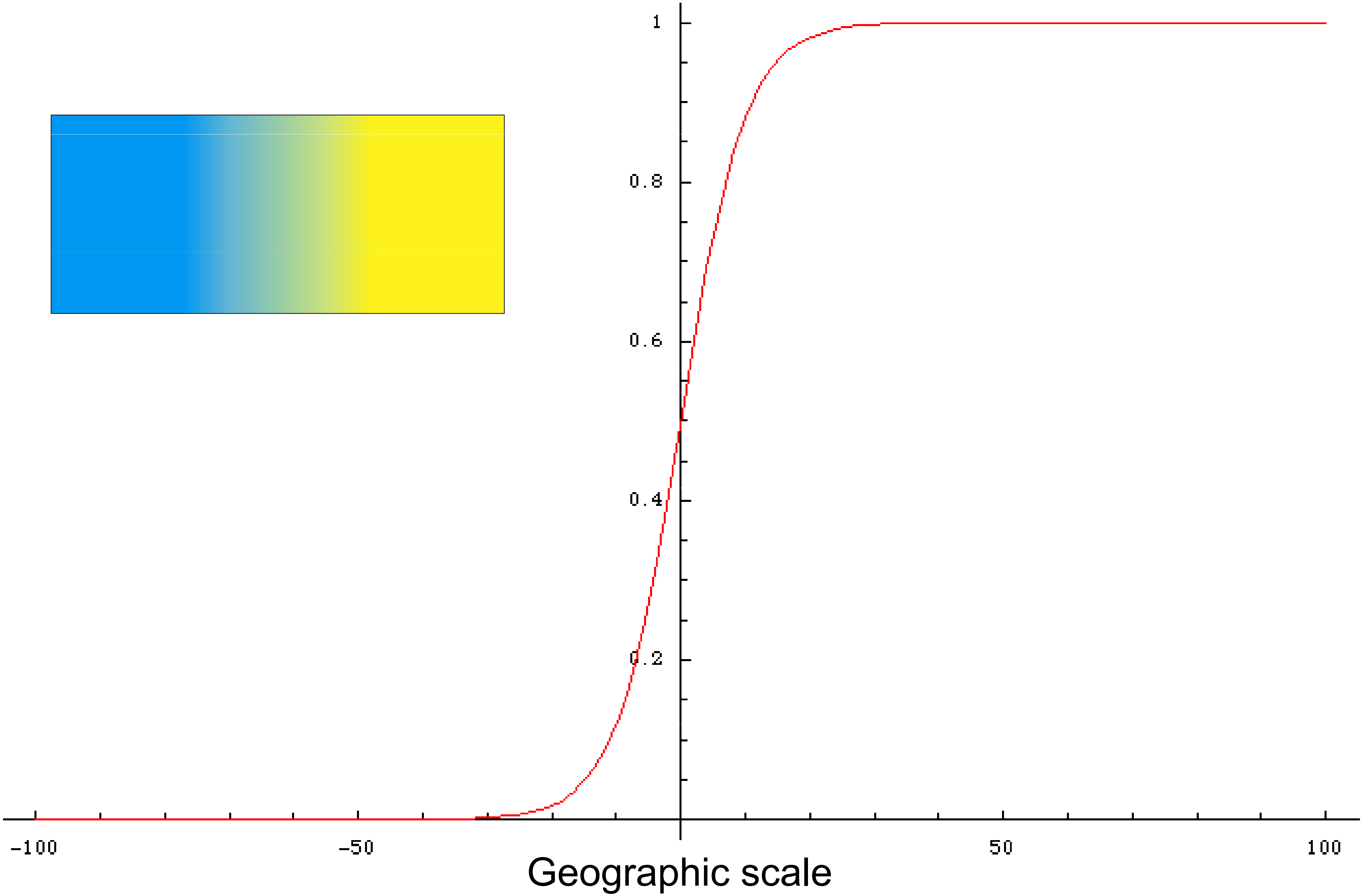
Diffusion of neutral alleles due to gene flow between demes



cline

after first contact
„stepped“ cline


1
0.8
0.6
0.4
0.2
-100
-50
50
100
Geographic scale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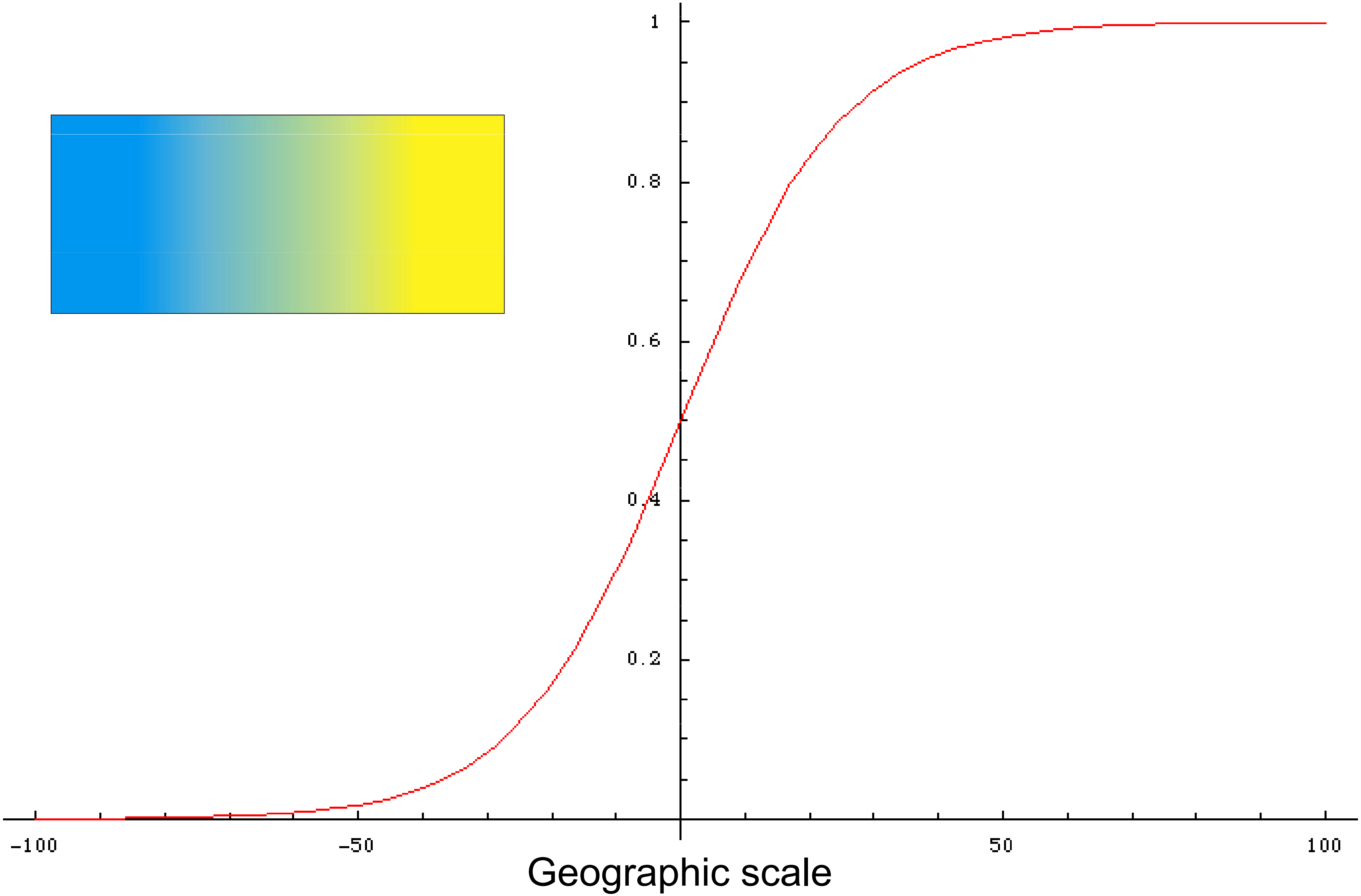
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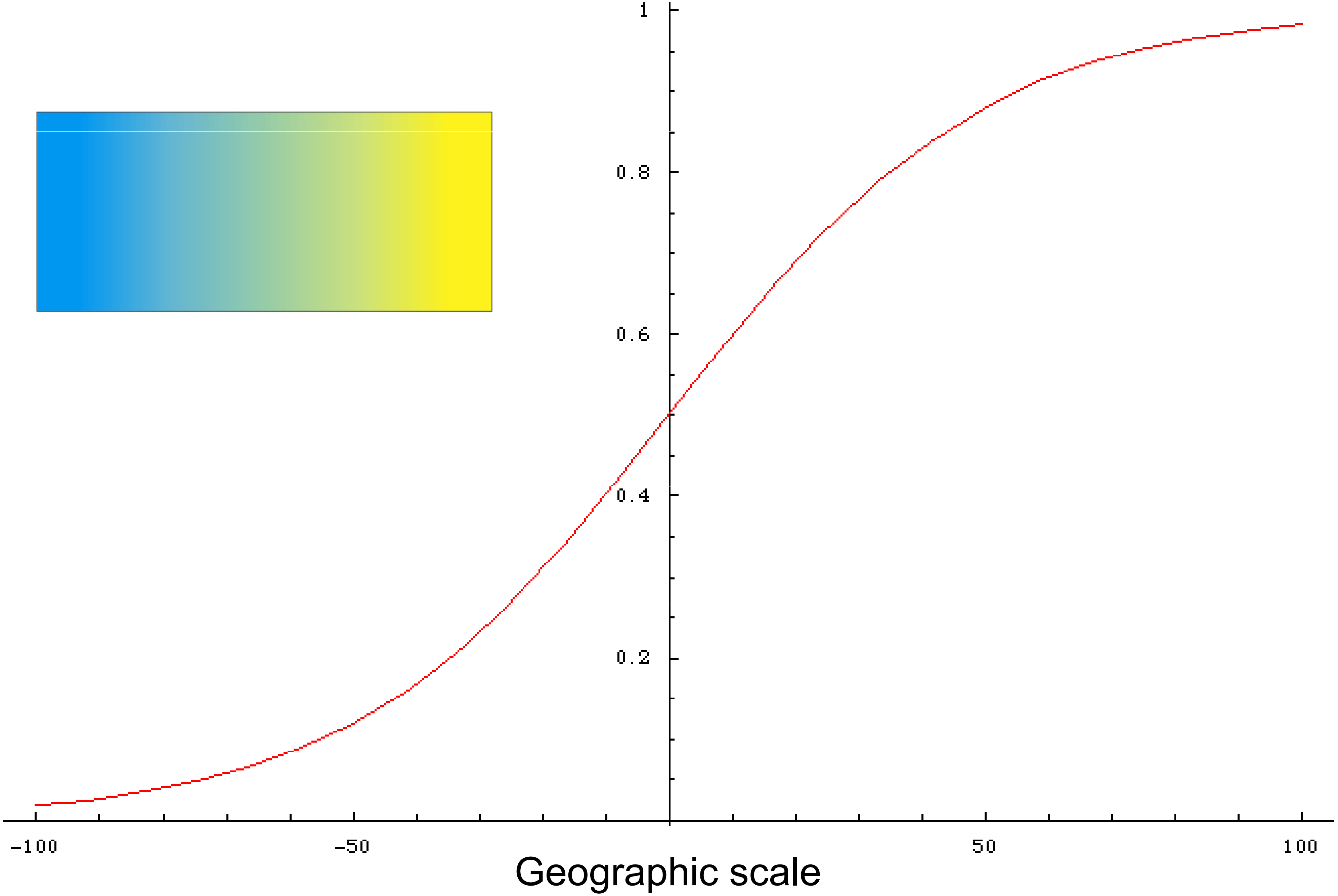
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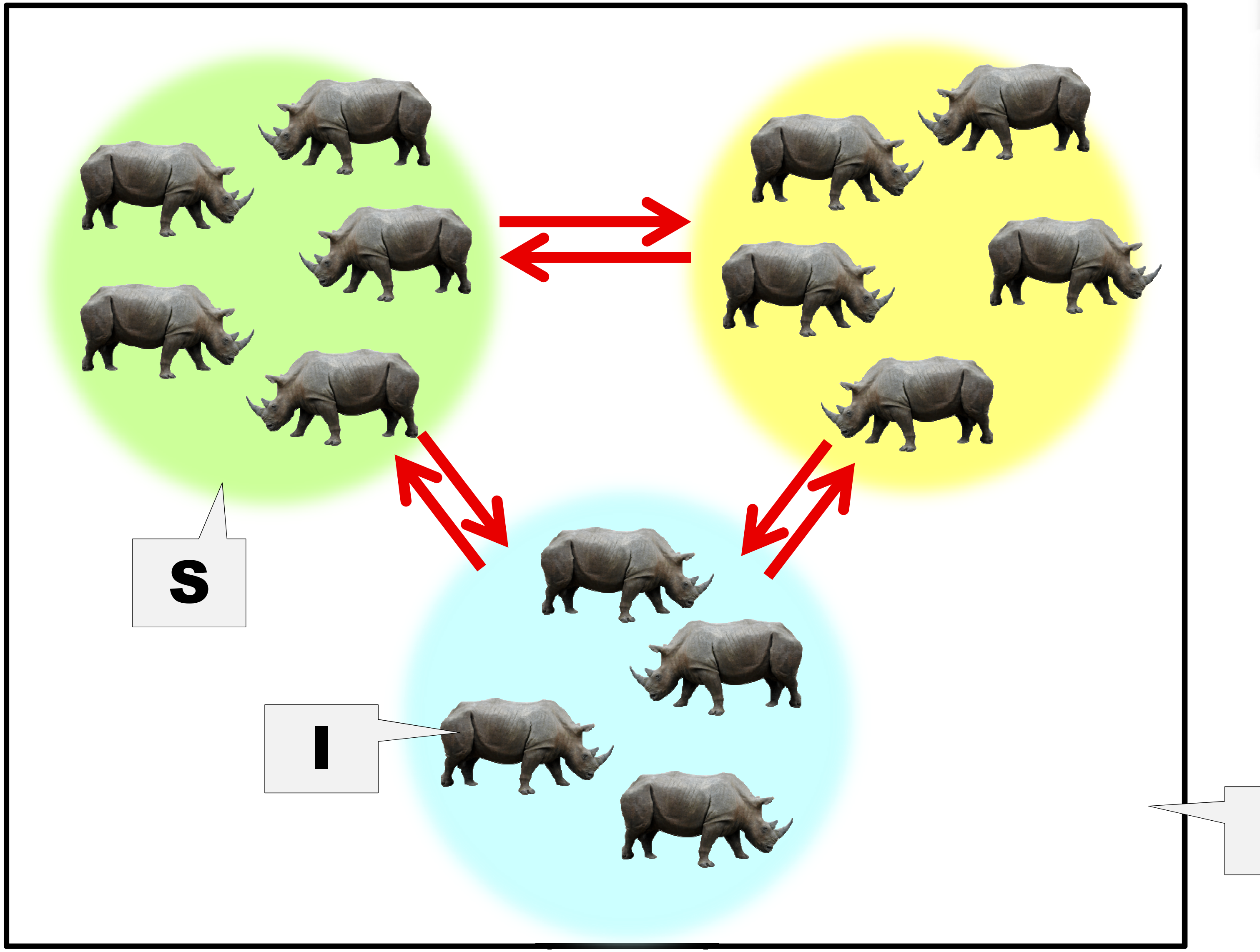
Geographic scale

Sewall Wright - F-statistics:



F_{IS} (= inbreeding coefficient)

→ reduction of HZ in a subpopulation due to inbreeding

$F_{IS} = (H_S - H_I)/H_S$ $\quad -1 \leq F_{IS} \leq +1$

F_{ST} (= fixation index) → reduction of HZ due to population substructuring

$F_{ST} = (H_T - H_S)/H_T$ $\quad 0 \leq F_{ST} \leq +1$

F_{IT} → reduction of HZ both due to population substructuring and inbreeding

$F_{IT} = (H_T - H_I)/H_T$

$(1 - F_{IS})\,(1 - F_{ST}) = 1 - F_{IT}$