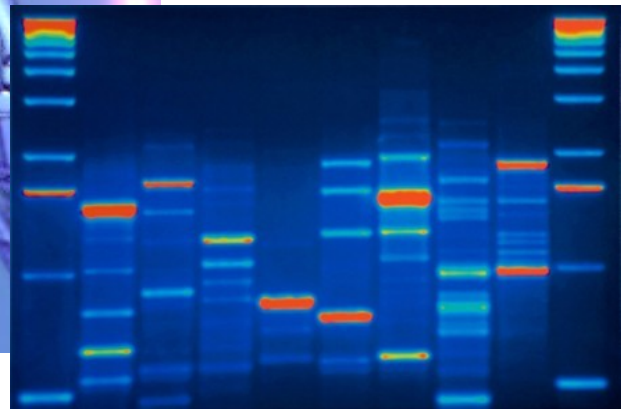
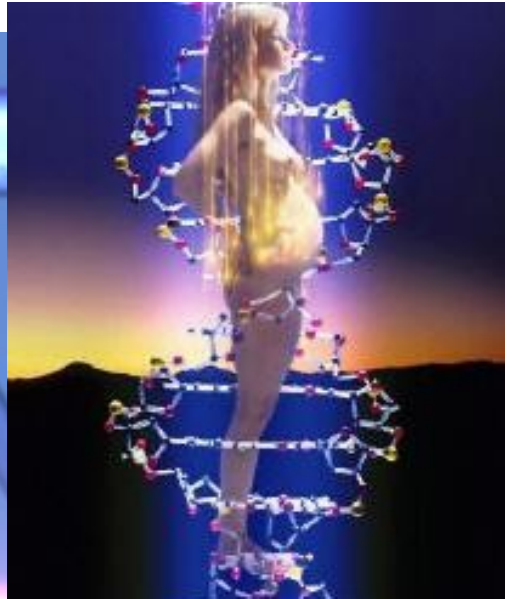
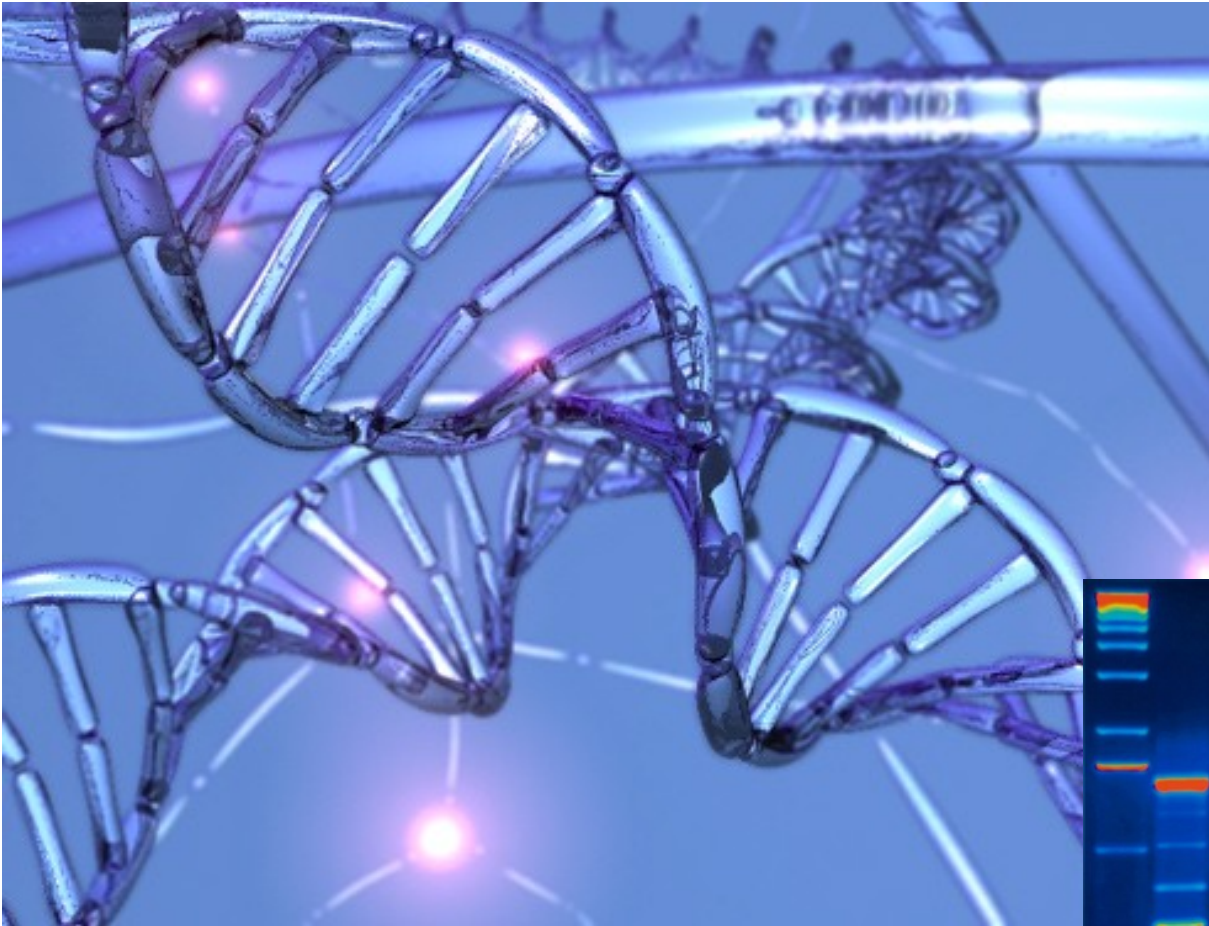


# ORIGIN OF GENETIC VARIATION



## 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

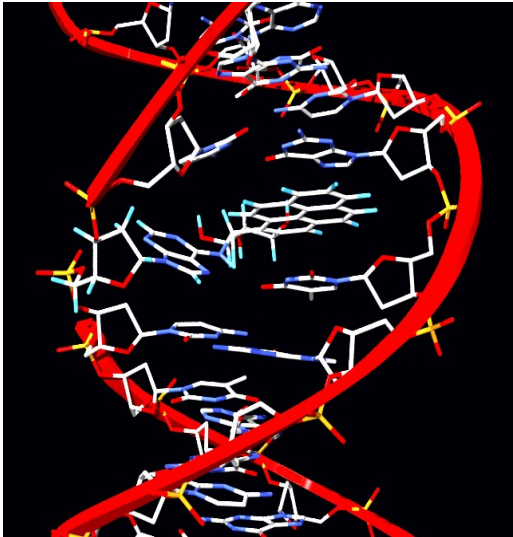
migration (gene flow)

nonrandom mating

natural selection

random genetic drift (incl. bottleneck, founder effect)

(molecular drive)



# MUTATION



spontaneous × induced

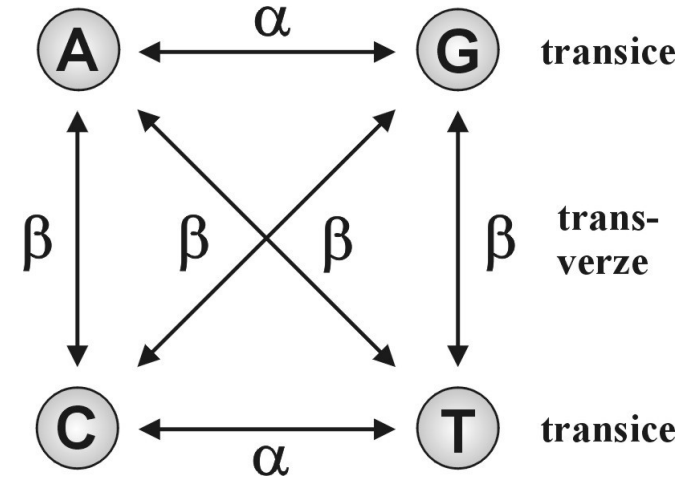
in germ cells × somatic

according to their deleterious/beneficial effect:

beneficial (positive)  
deleterious (lethal, negative)  
neutral

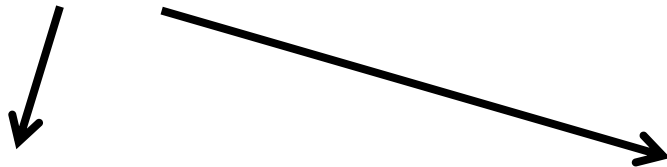
## According to effect

point (gene)  
chromosomal  
genome



## Point mutations:

**substitutions** (transitions, transversions)



**synonymous**

GTC → GTA  
Val → Val

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

**nonsynonymous**

missense  
nonsense

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

back mutations: generally 10-times lower frequencies

recurrent mutations → mutation pressure:

eg. when allele frequency  $A = 0,5$ ;  $2N = 2000$ :

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

after 100 generations → 0,55 ...

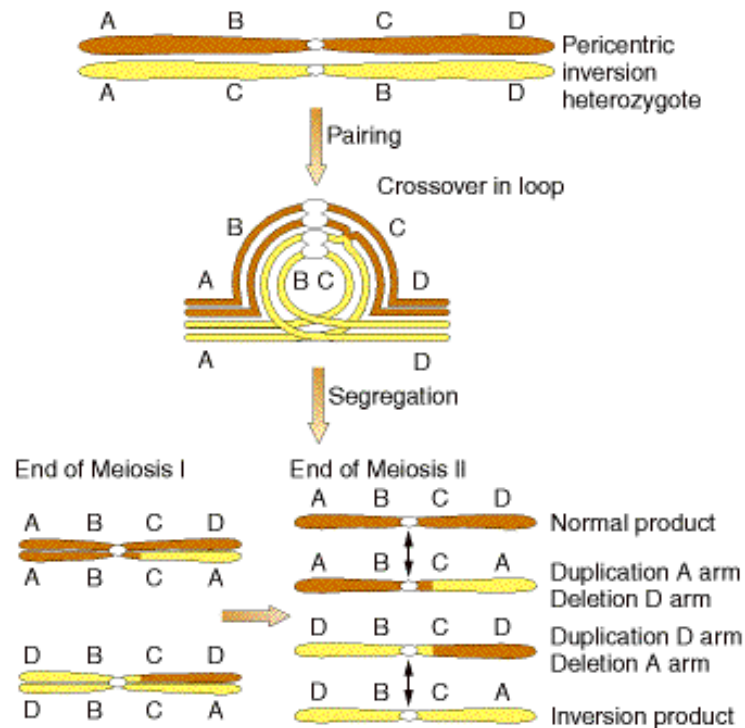
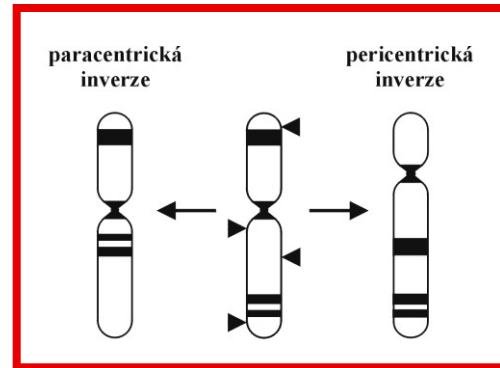
⇒ change of allele frequencies by mutations very slow

# Chromosomal mutations (chr. rearrangements)

## inversions

pericentric

paracentric



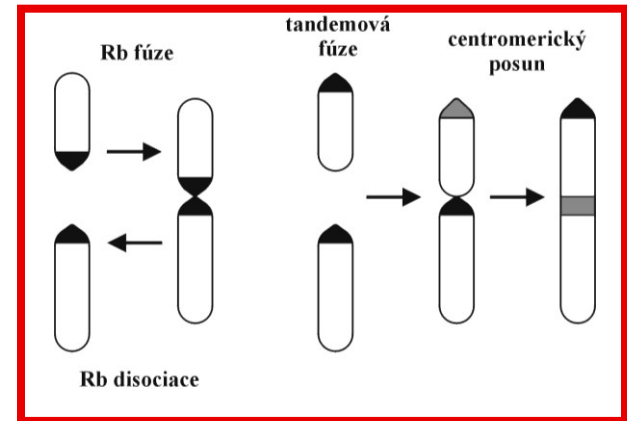
translocations

fusions and dissociations

(Robertsonian translocations)



house mouse



XX XX XX XX XX XX XX XX XX XX

XX XX XX XX XX XX XX XX XX XX

XX XX XX XX XX XX XX XX XX XX XX XX



# 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

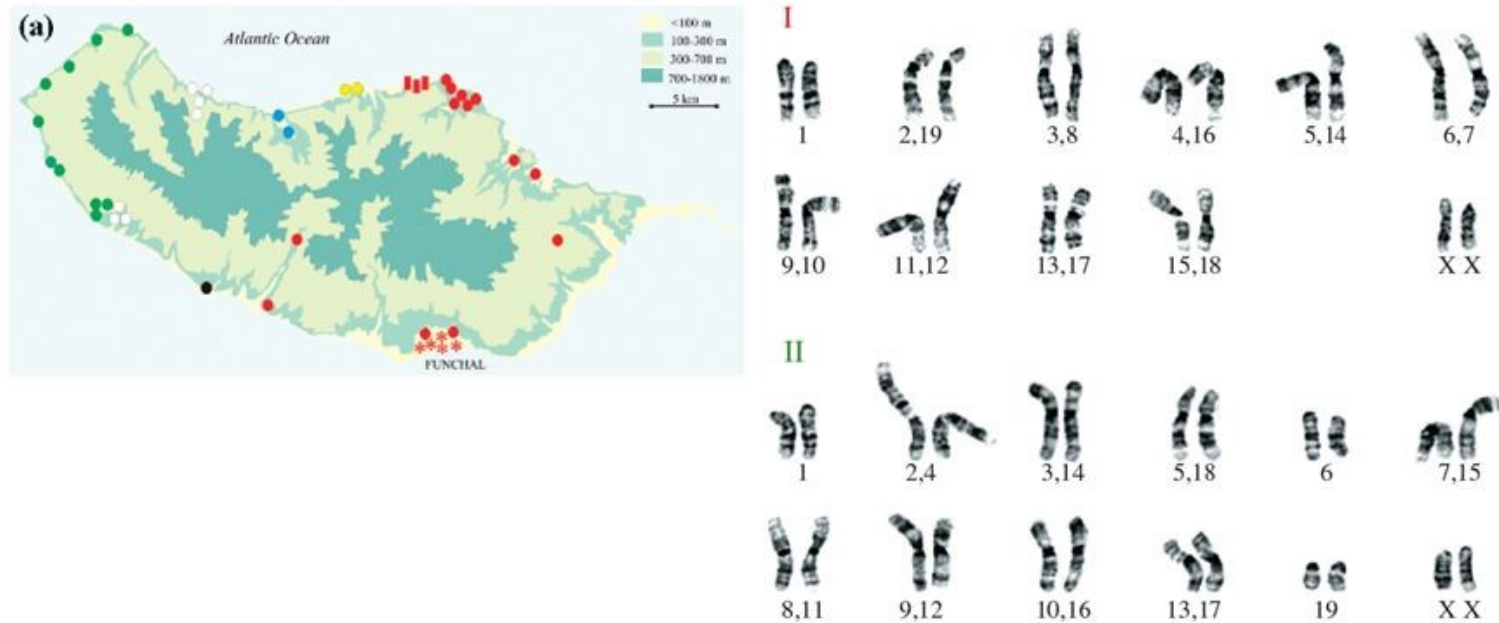


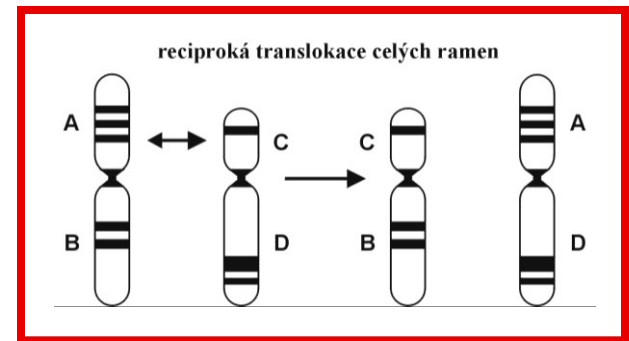
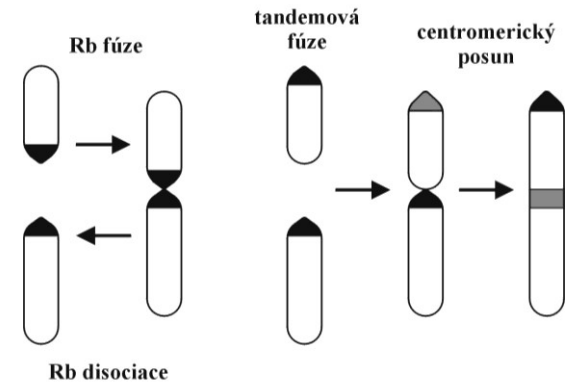
Fig. 13.30

# translocations

## fusions and dissociations

### whole-arm reciprocal translocations (WART)

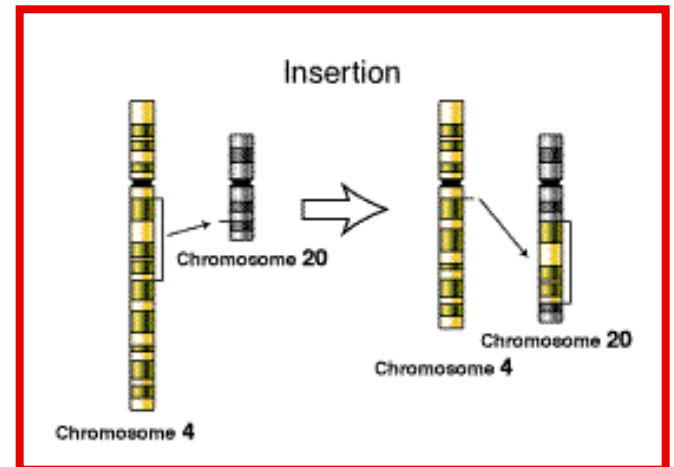
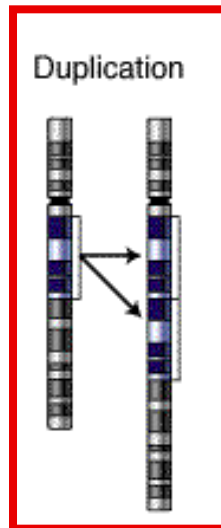
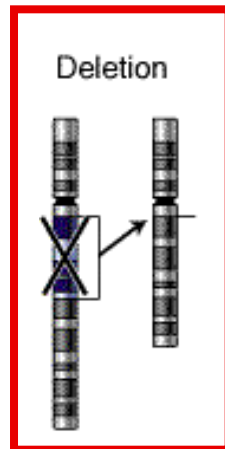
house mouse



deletions

duplications

insertions



# Genome mutations

## -somes (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 ( $\mu$ )

## mutation effects random, position and rate nonrandom

transitions > transversions

mutation „hotspots“: CpG in animals (methylated C  $\rightarrow$  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  $\Rightarrow$  >  $\mu$

RNA viruses (HIV)

parasites

antigens, immunoglobulins

>  $\mu$  of somatic mutations

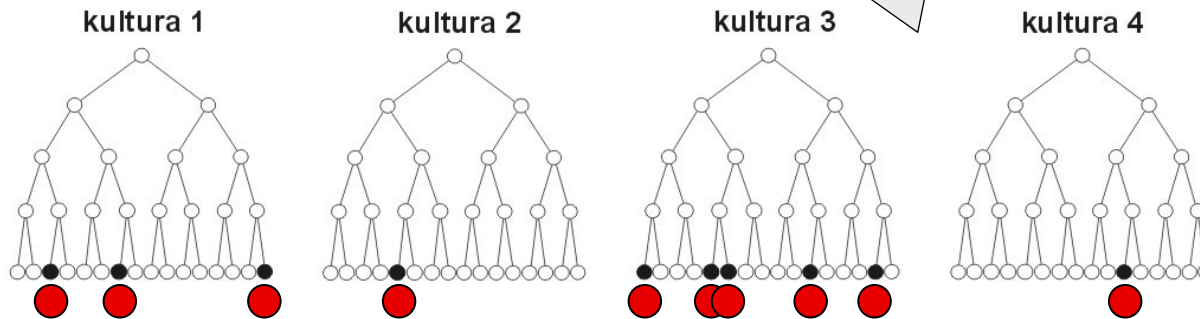
males > females: humans 6x, rodents, fox: 2x ... more cell divisions in germ cells

# Adaptive (directed) mutations?

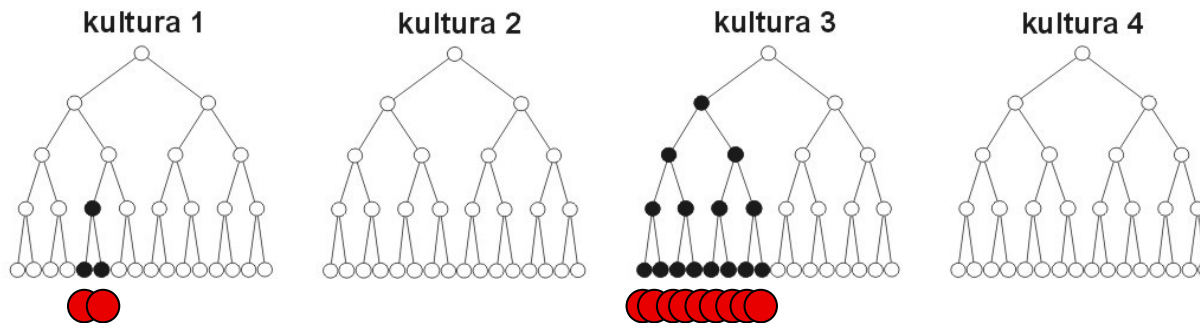
Max Delbrück, Salvador Luria (1943):  
fluctuation test



(a) mutace vyvolané prostředím



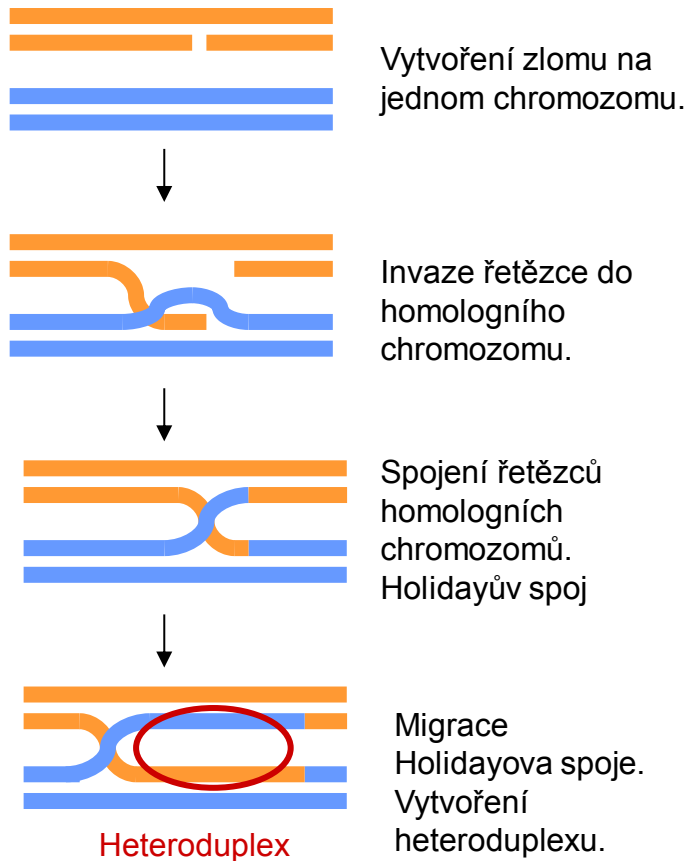
(b) náhodné mutace



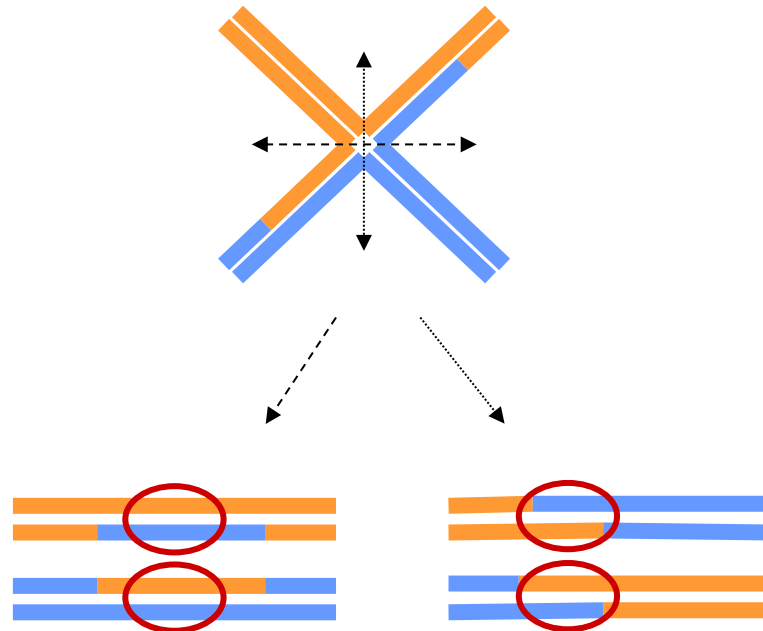
# RECOMBINATIONS

mutations → new alleles

recombinations → new genotypes (exception = intragenomic recombination)



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

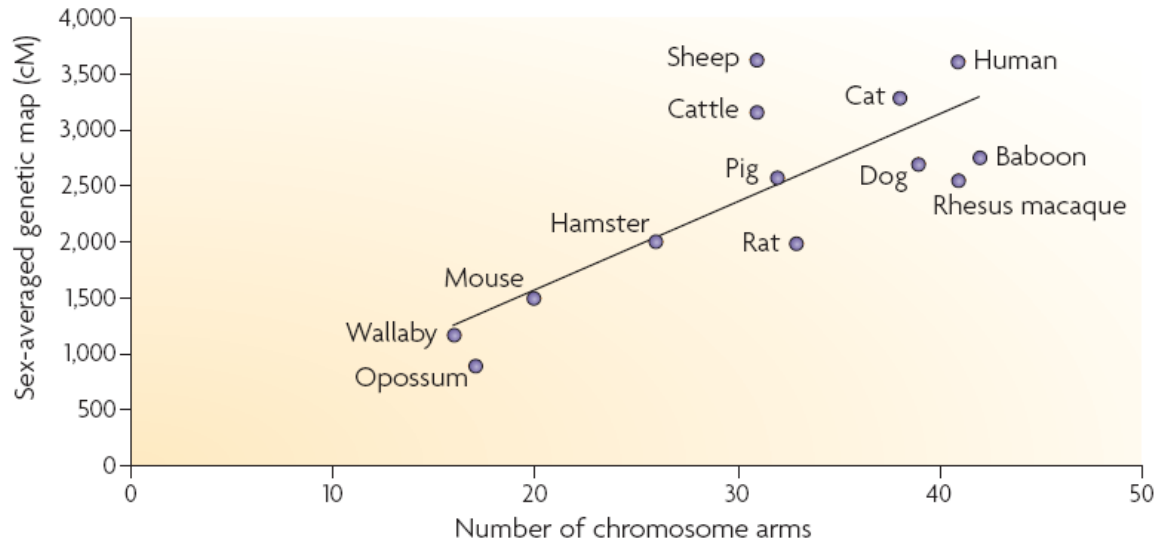
demise of 1 point often compensated by increased activity of a neighbour point

## 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:

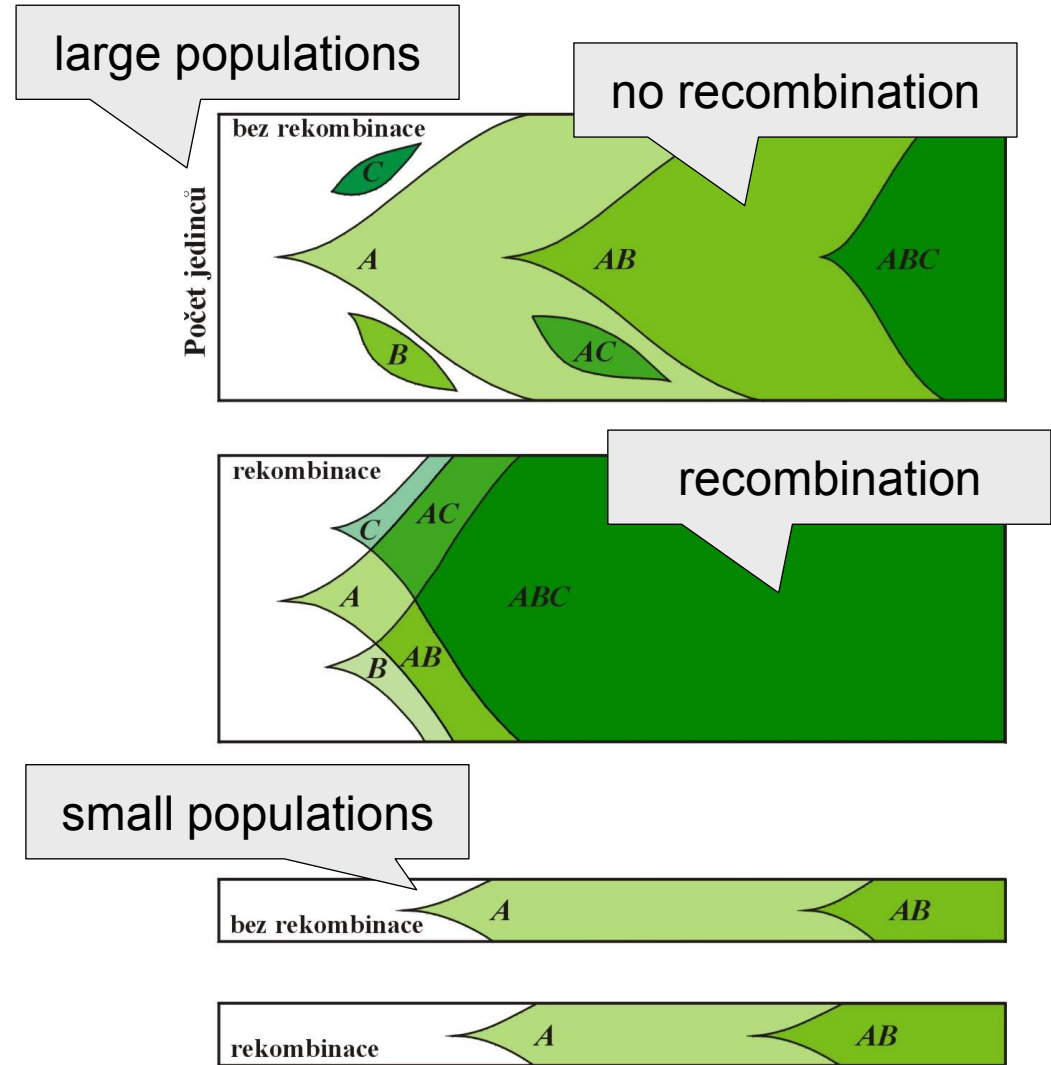
- species with more small chromosomes → more recombinations than species with less large chromosomes
- correlation with the number of arms: more recombinations in karyotypes with large numbers of chrom. arms (at least 1 c-o/arm to avoid aneuploidies?)



# EVOLUTIONARY CONSEQUENCES OF RECOMBINATION:

Recombination  
and polymorphism:

absence of recombination  
⇒ linkage disequilibrium



# EVOLUTIONARY CONSEQUENCES OF RECOMBINATION:

## Recombination and polymorphism:

positive selection: **selective sweep**

hitchhiking (draft)

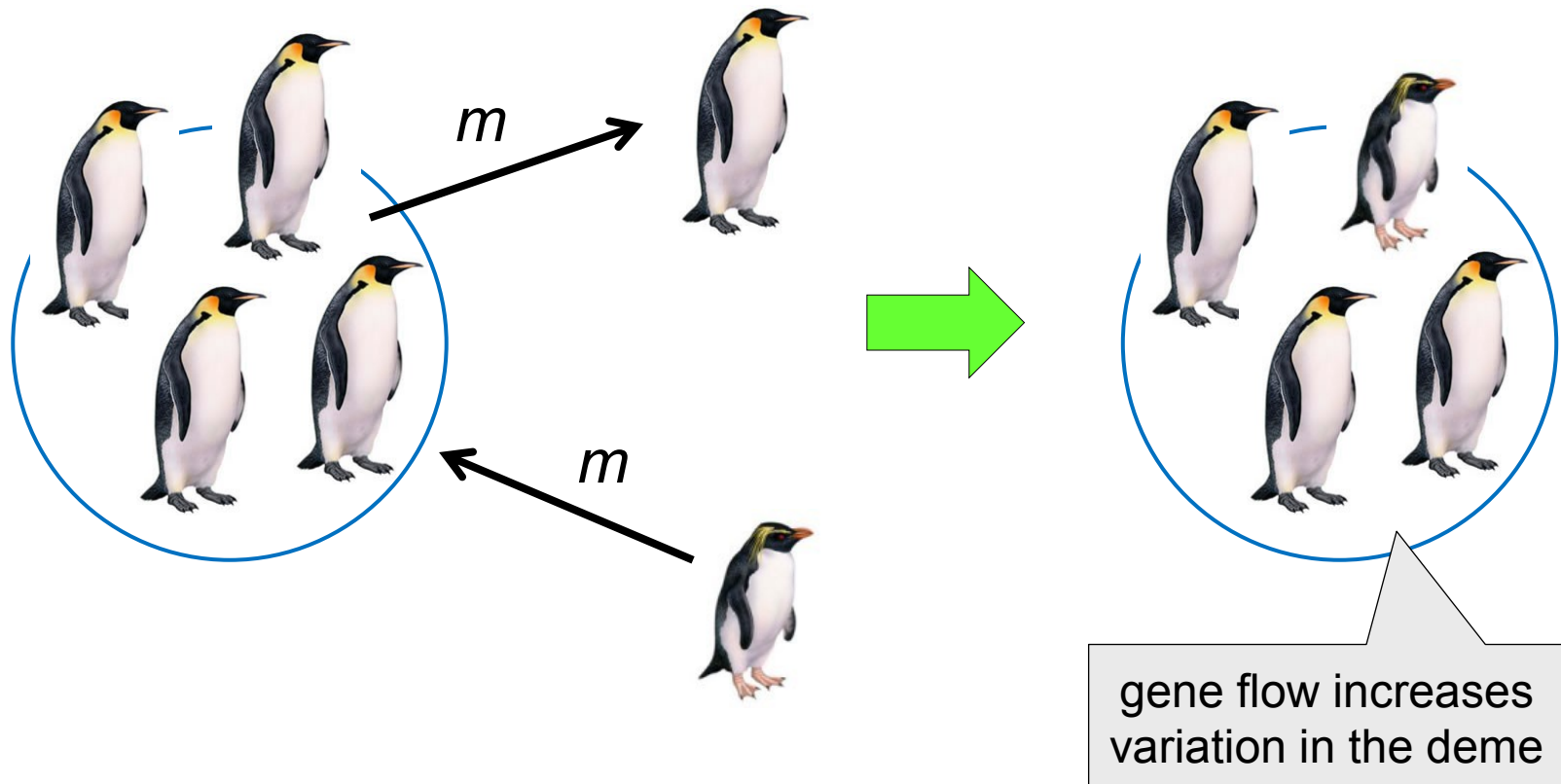
more frequent appearance of rare alleles

negative selection: **background selection**

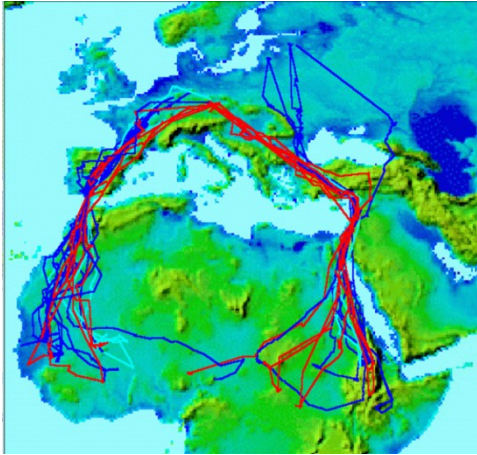
→ **loss of polymorphism**

# MIGRATION (GENE FLOW)

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



# MIGRATION (GENE FLOW)



long-distance  
migration but no  
gene flow



gene flow but no  
migration

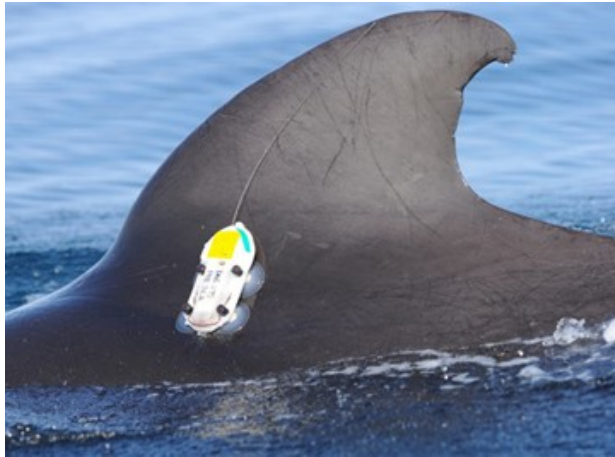


# METHODS OF GENE FLOW ESTIMATION:

## 1. direct

### capture-mark-recapture (CMR)

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

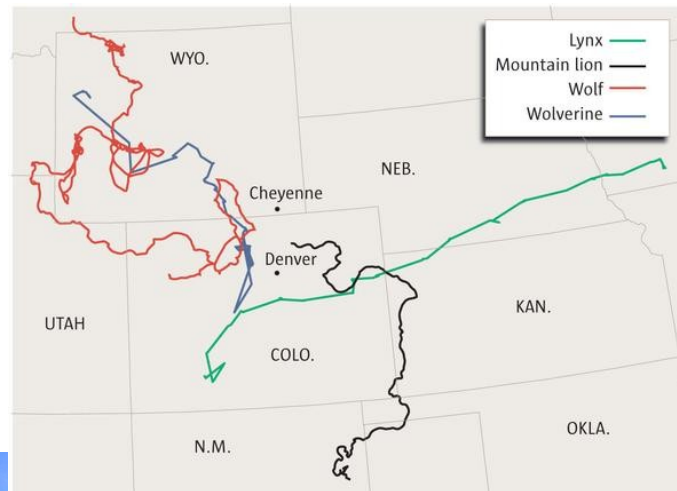


# 1. Direct methods

remote tracking – telemetry

transmitters, antennas; GPS systems

... more expensive, time consuming



**Risk of underestimation of gene flow!!**



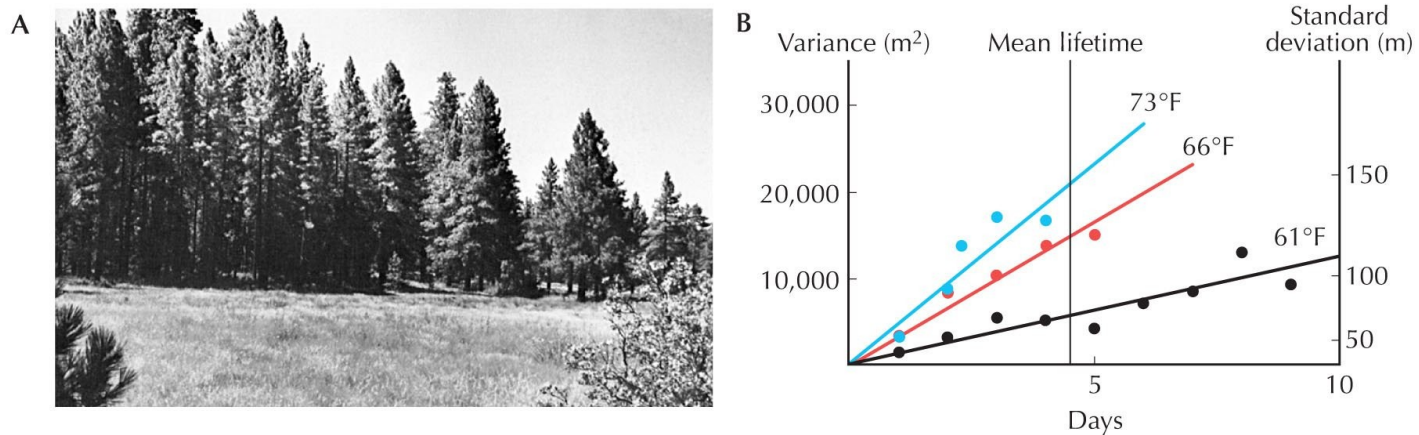
## 2. Indirect methods

### molecular markers

gene flow models

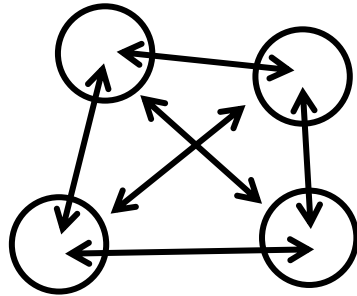
maximum likelihood and Bayesian programs

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).

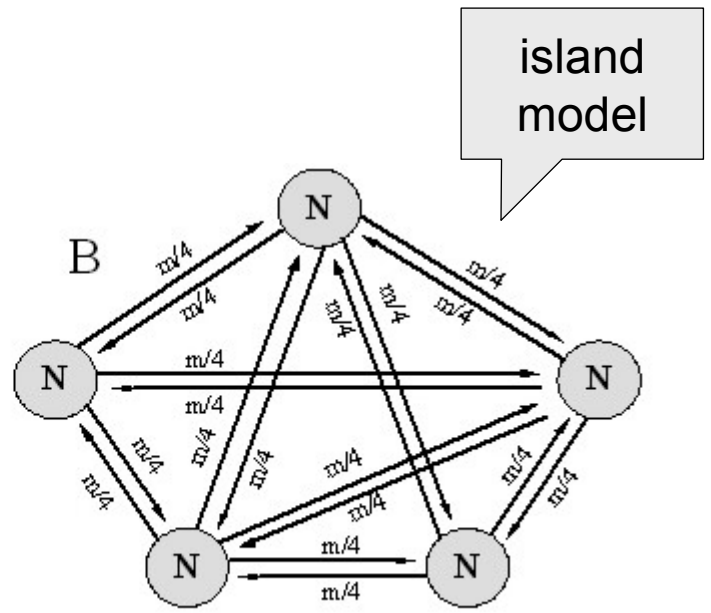
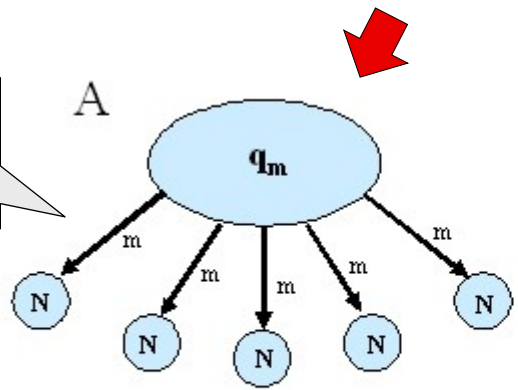
# A) Island model



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

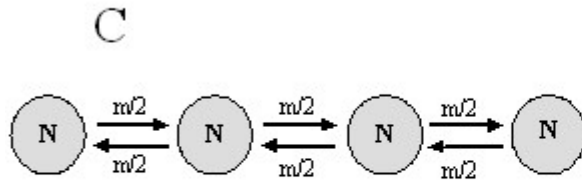
Island model can be also asymmetric:

continent-island model

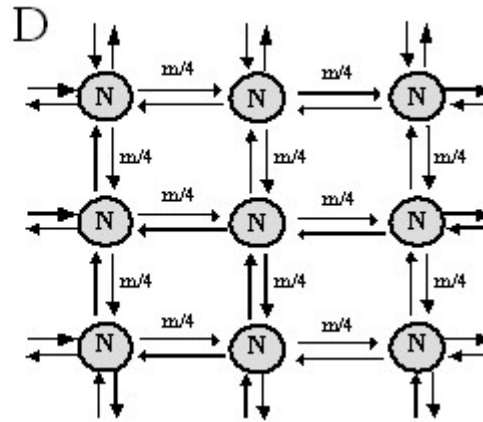


island model

## 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



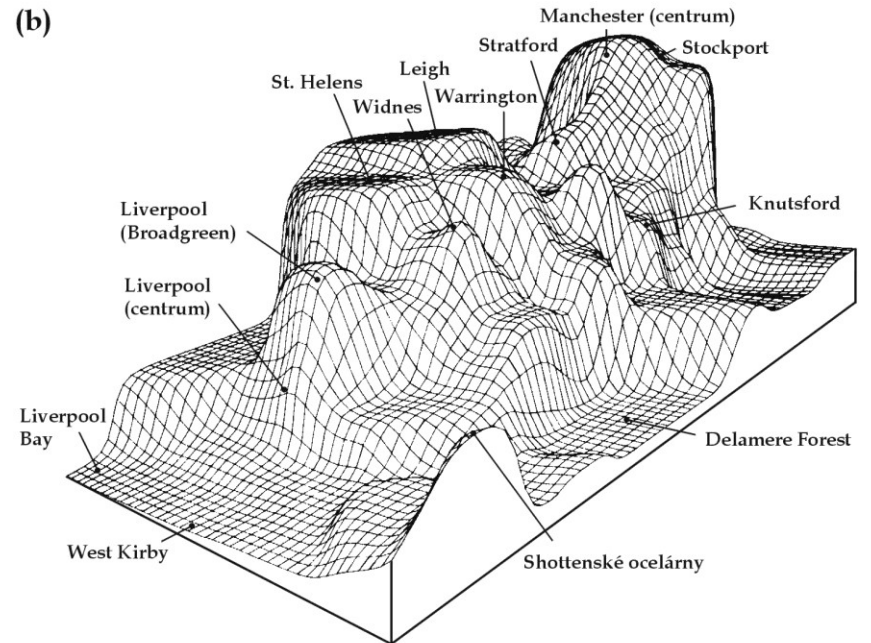
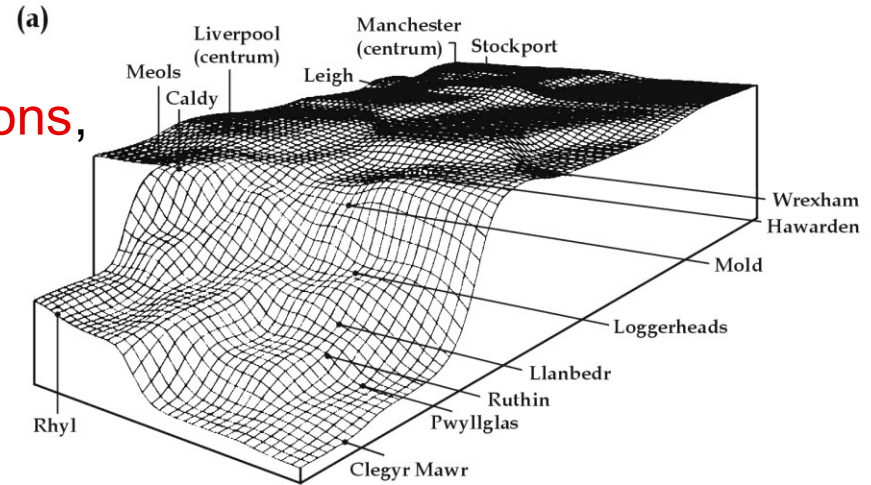
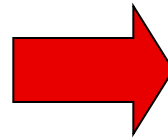
*L. parryae*

# Gene flow consequences:

genetic homogenization of subpopulations,  
preventing their genetic divergence

in many species migration severely  
reduced

Eg.: melanic forms of moths  
in England

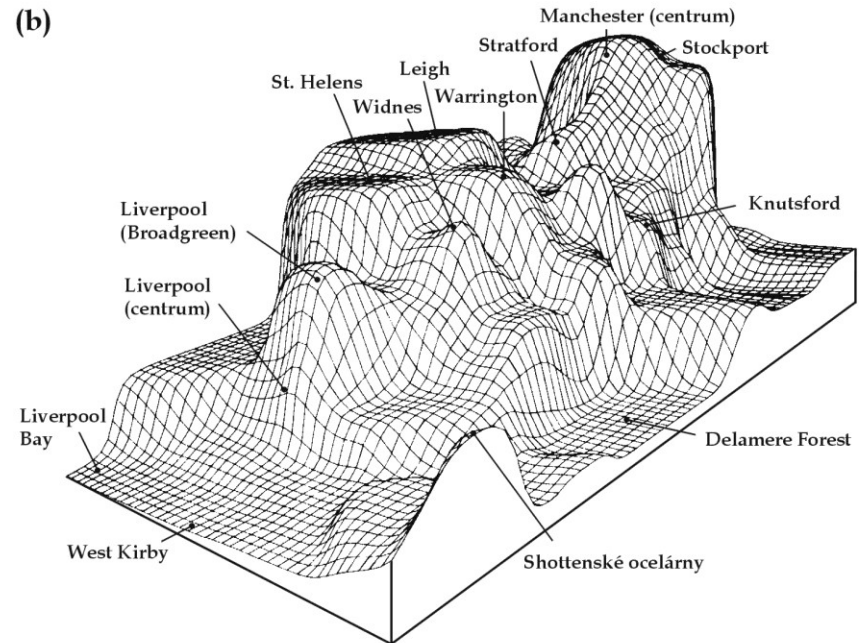
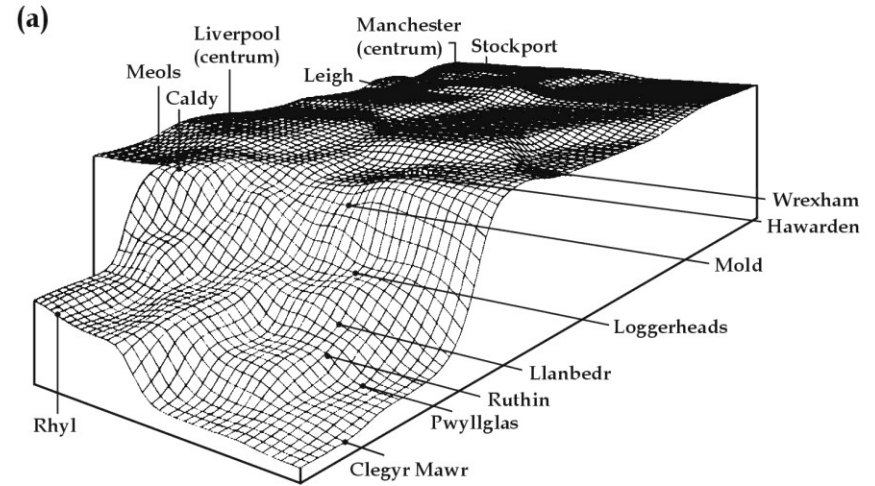




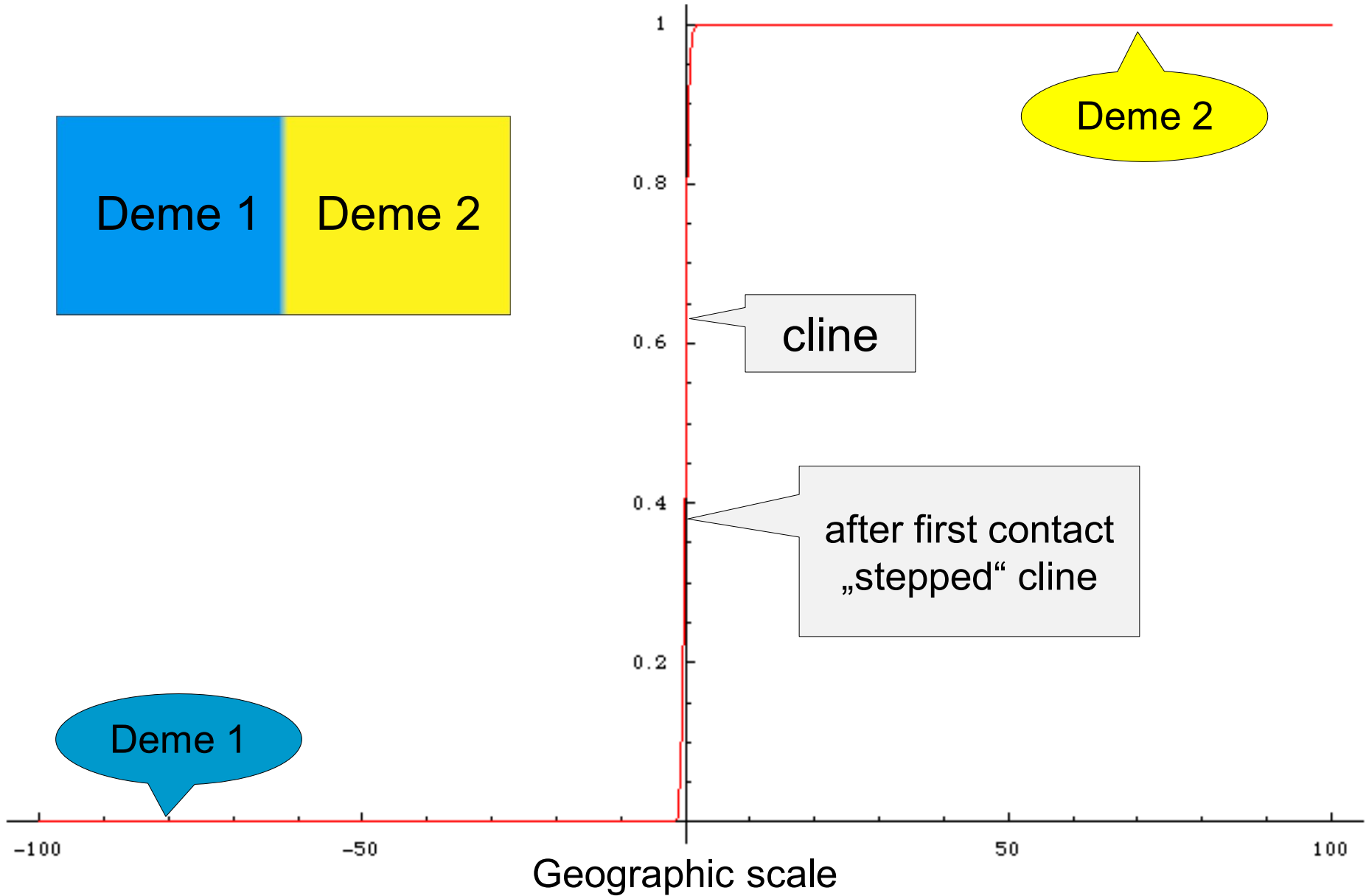
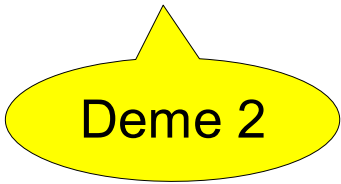
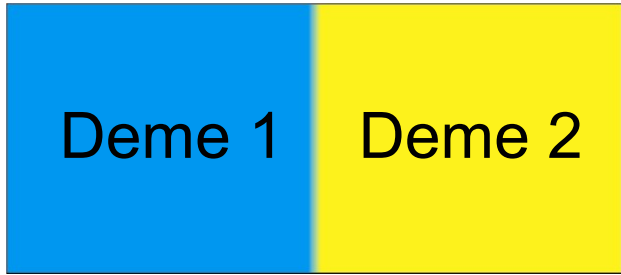
peppered moth (*Biston betularia*)

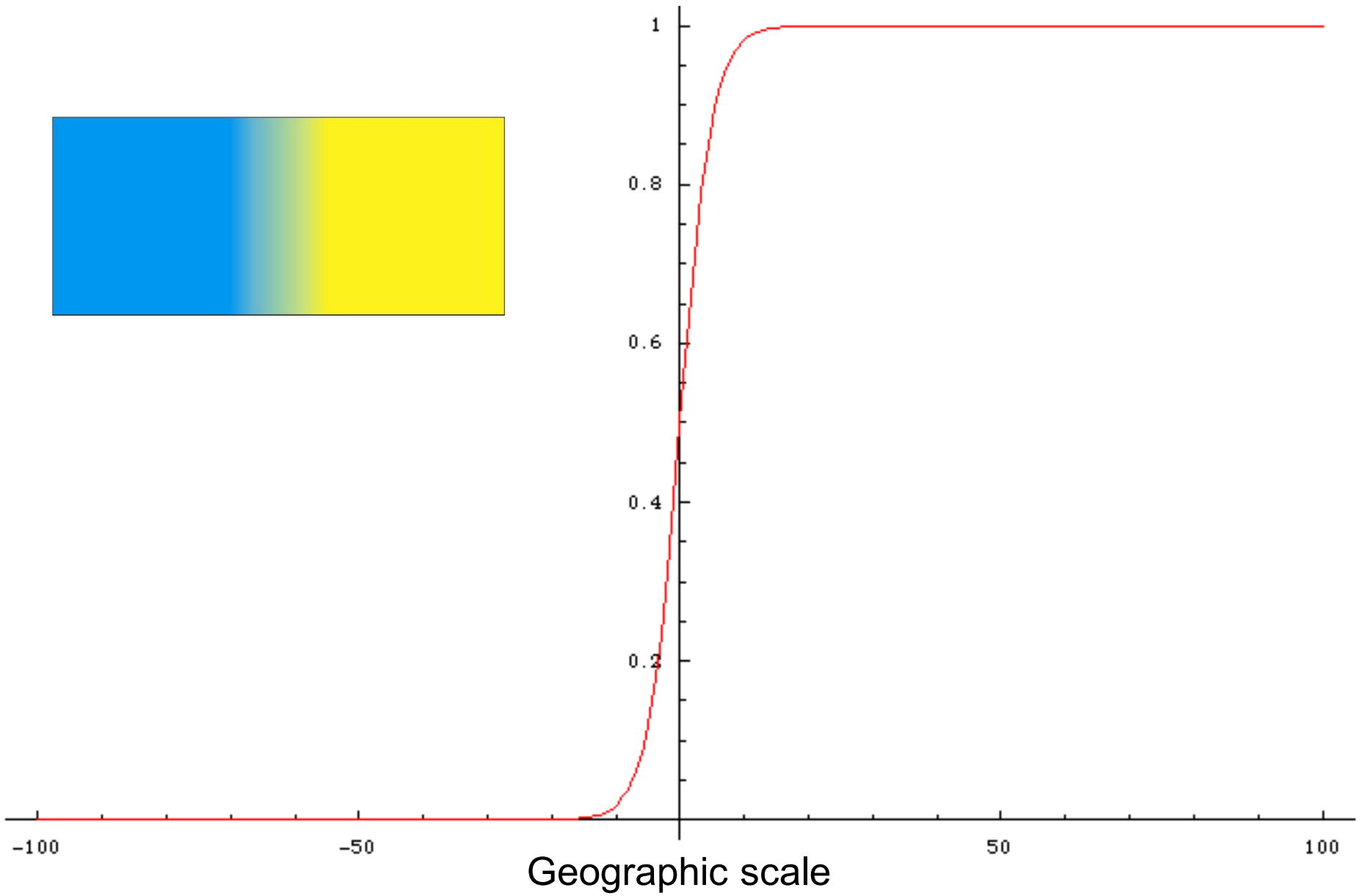
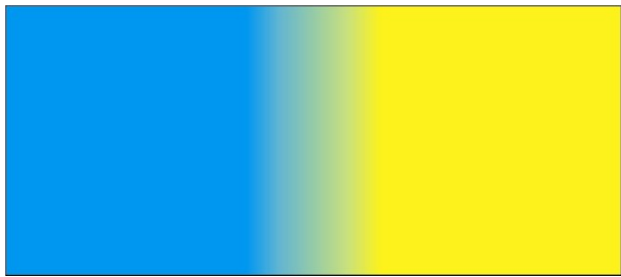


scalloped hazel  
(*Odontoptera [Gonodontis] bidentata*)

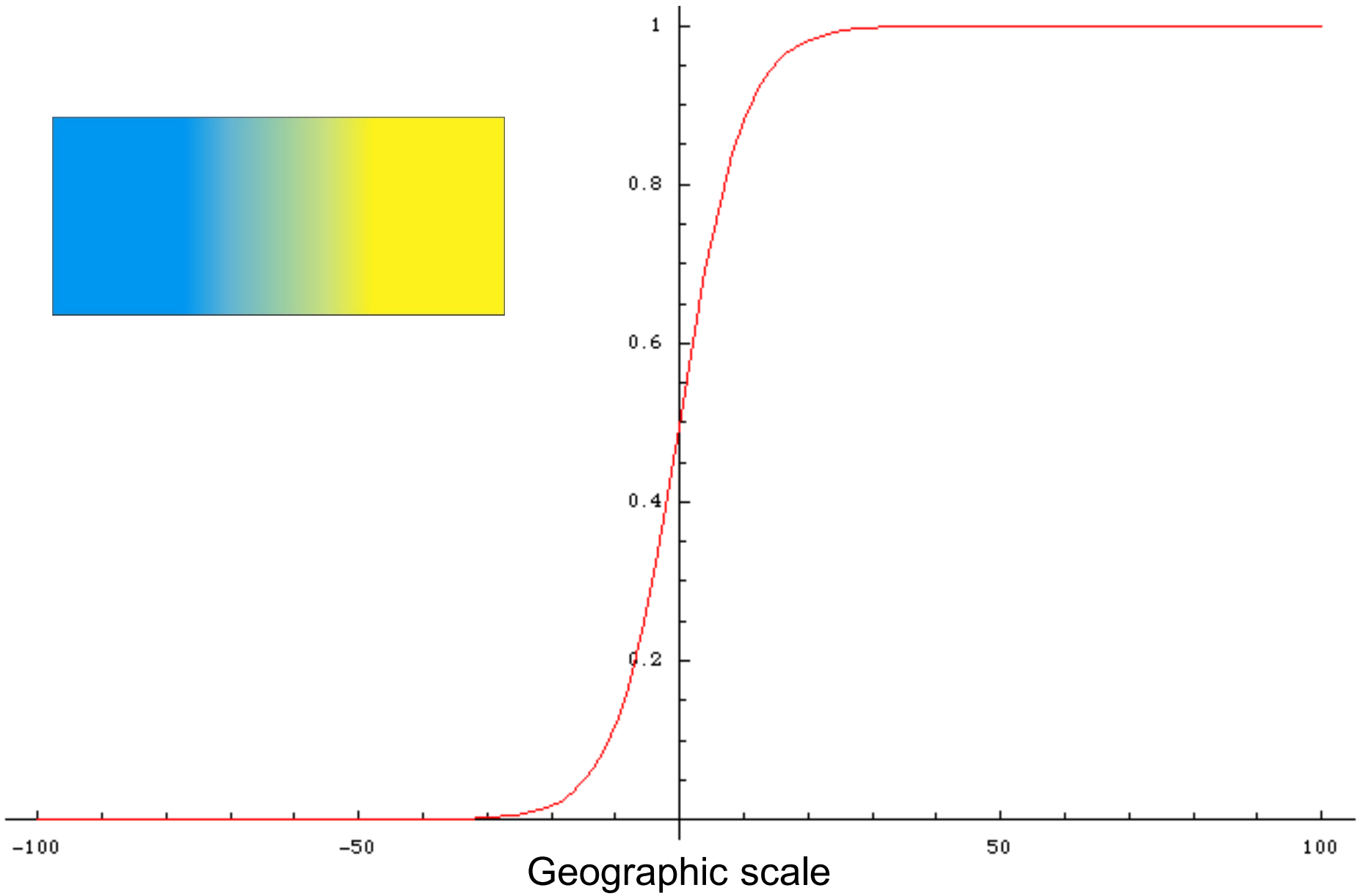
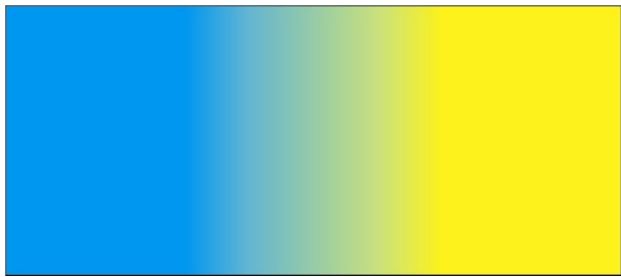


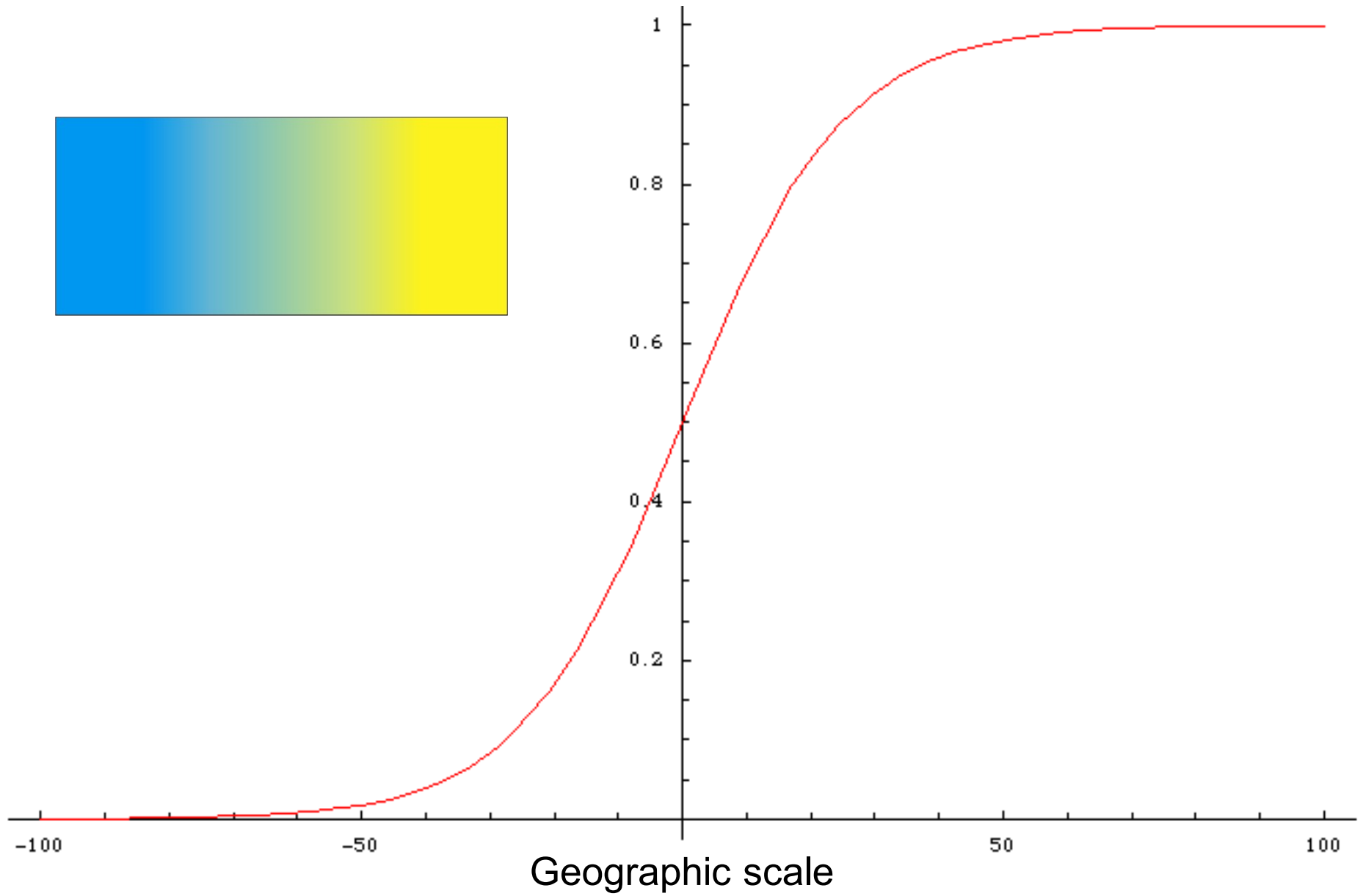
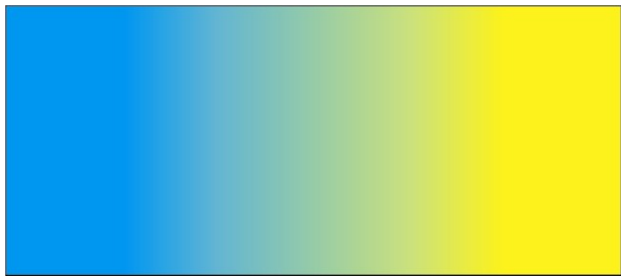
# Diffusion of neutral alleles due to gene flow between demes

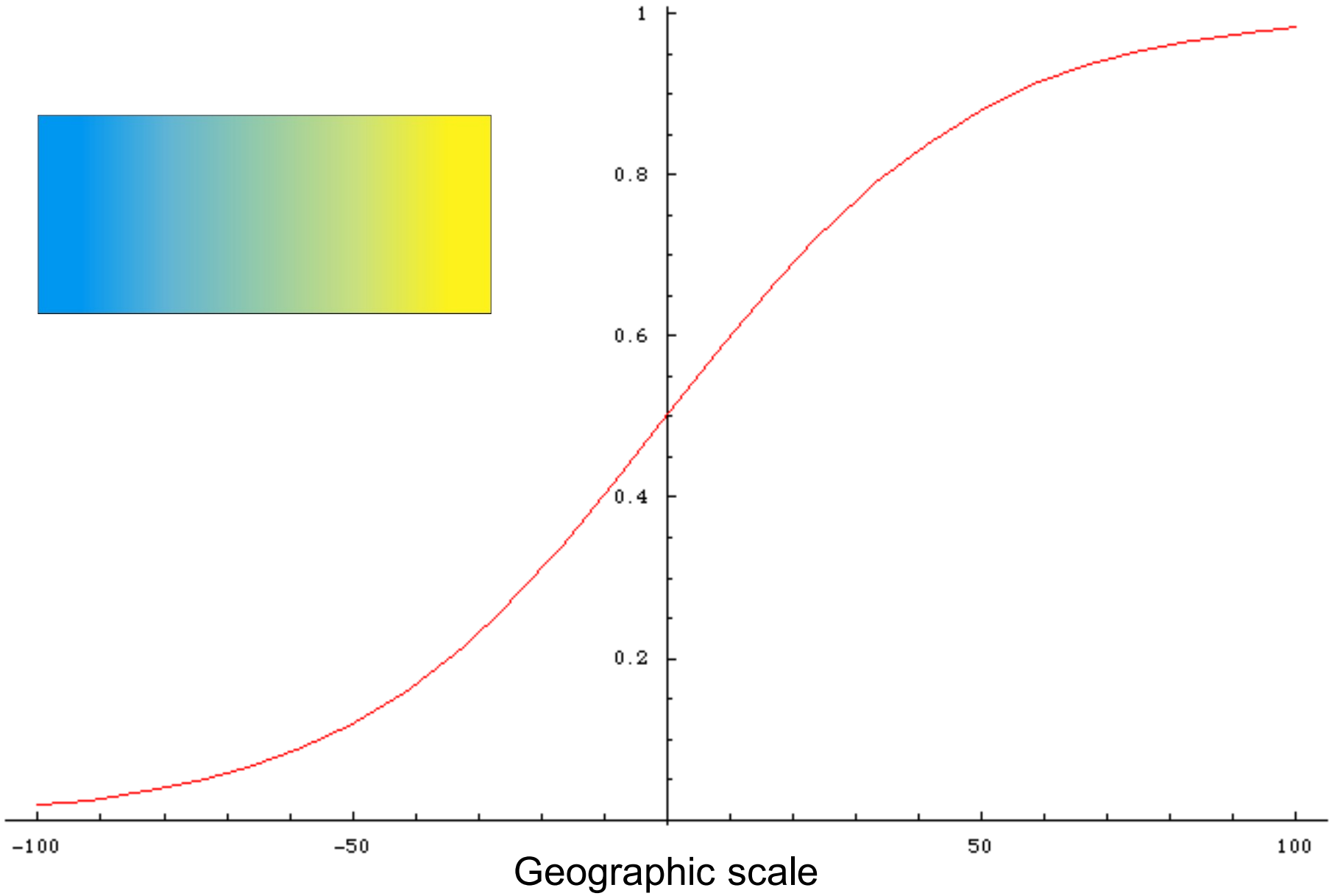
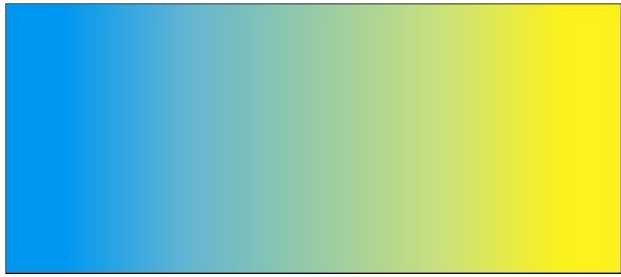




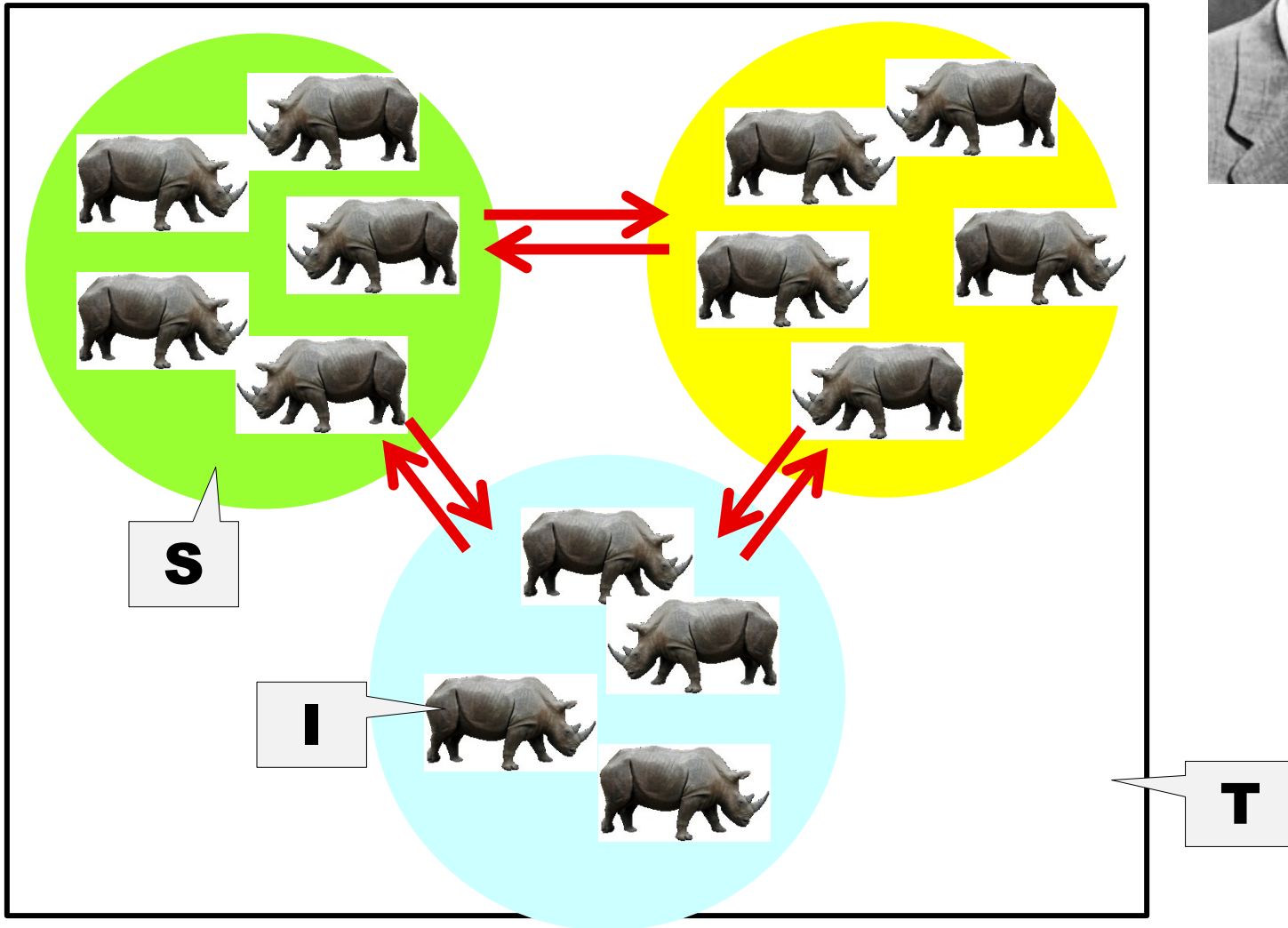








# Sewall Wright - F-statistics:



$F_{IS}$  (= 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

$$F_{IT} = (H_T - H_I)/H_T \quad (1 - F_{IS})(1 - F_{ST}) = 1 - F_{IT}$$