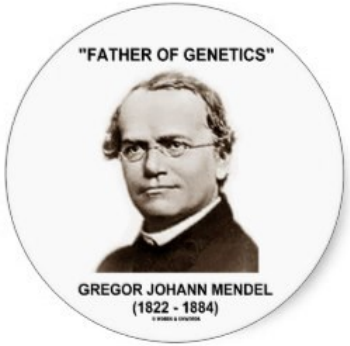
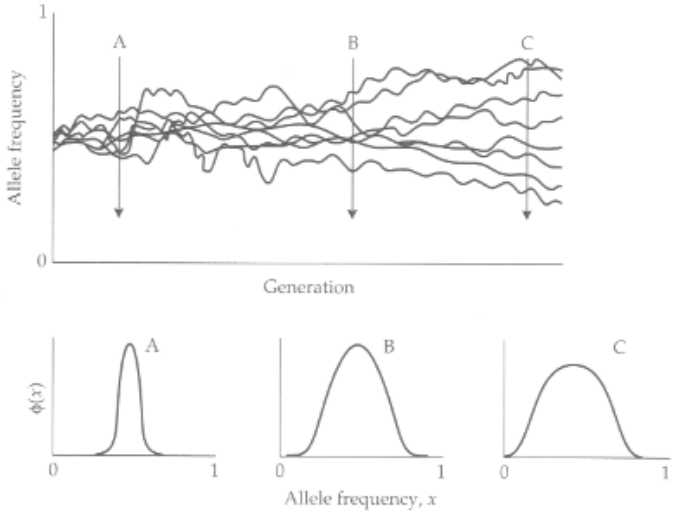
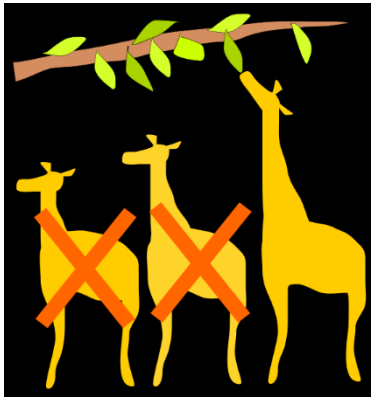
# Opakování z minula




- Ideální populace (Fischer Wright model)
- Hardy-Weinberg principle
  odchylky: inbreeding, asortativní křížení, pop. struktura
- Drift (náhoda)
- Effective population size



# Selection

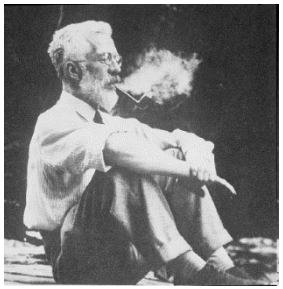


## Mutace přeci nemusí být jen neutrální

- Mohou ovlivnit fitness

- Alela → fenotyp → fitness (*W*)
  - Fitness: schopnost přežít a dožít se reprodukčního věku, najít si partnera, plodnost a počet potomků
  - Components of fitness: viability, mating success, fertility and fecundity

- Mutace neutrální, výhodné, nevýhodné



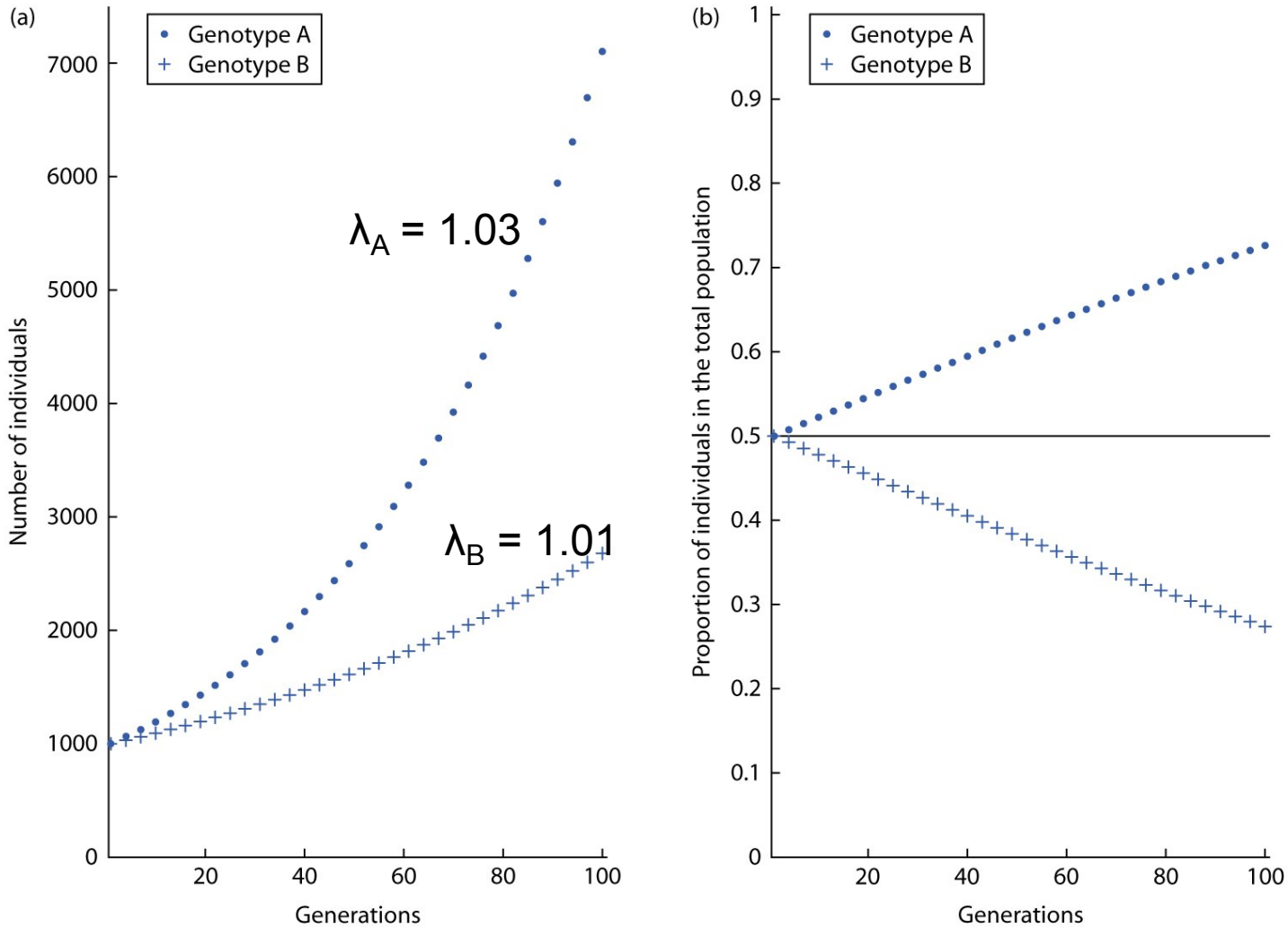
- Fundamental theorem of natural selection aneb bez rozdílů nečekejme vylepšení

  *The rate of increase in fitness of any organism at any time is equal to its genetic variance in fitness at that time* (Fisher 1930)

# Clonal reproduction, no upper limit of pop size

$$N_{t+1} = \lambda N_t$$

relative fitness $w = \lambda_A / \lambda_B$



# Diploidní model

- Fitness vztažena **ke genotypu**.

  (Jaká je fitness jedinců s konkrétním genotypem VZHLEDEM k jiným genotypům?)

- Začleníme fitness mutací (genotypů) do jednoduchých modelů klasické populační genetiky

- Genotyp, který zanechá nejvíce potomků $w = 1$, letální genotyp $w = 0$
- Ostatní genotypy: $w$ mezi 0 do 1, $w = 1 - s$, $s$ je selekční koeficient

- Diploidní organismy:
- $s$ míra selekce proti homozygotům
- $h$ míra dominance
- $hs$ míra selekce proti heterozygotům
- Alternativně $w_{11}$, $w_{12}$, $w_{22}$

**TABLE 6.2 DIPLOID SELECTION FOR SURVIVORSHIP (VIABILITY)**

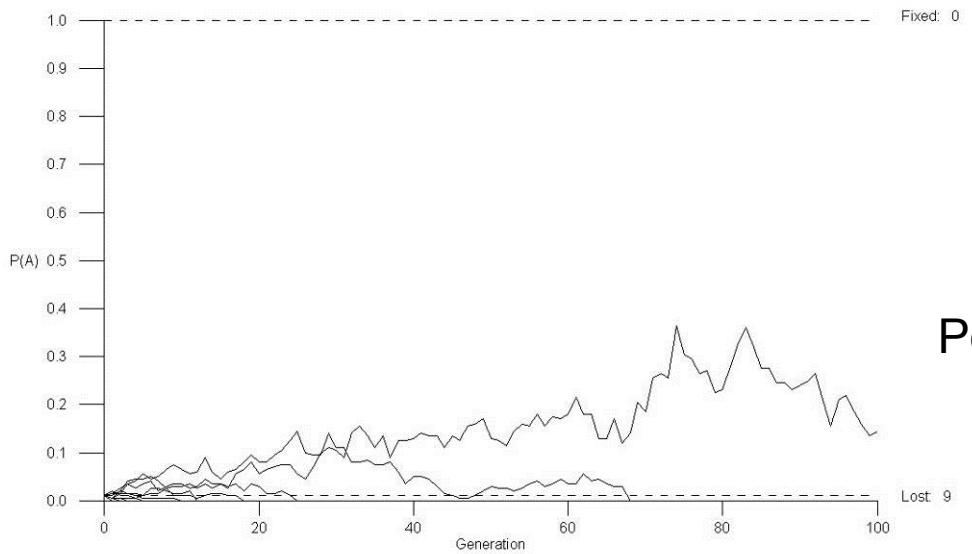
| | Genotype | | | Total |
|---|---|---|---|---|
| Generation $t-1$ | $AA$ | $Aa$ | $aa$ | |
| Frequency before selection | $p^2$ | $2pq$ | $q^2$ | $1 = p^2 + 2pq + q^2$ |
| Relative fitness (viability) | $w_{11}$ | $w_{12}$ | $w_{22}$ | |
| After selection | $p^2w_{11}$ | $2pqw_{12}$ | $q^2w_{22}$ | $\bar{w} = p^2w_{11} + 2pqw_{12} + q^2w_{22}$ |
| Normalized | $\frac{p^2w_{11}}{\bar{w}}$ | $\frac{2pqw_{12}}{\bar{w}}$ | $\frac{q^2w_{22}}{\bar{w}}$ | |
| Generation $t$ | $p' = \frac{p^2w_{11} + pqw_{12}}{\bar{w}}$ | | | |
| | $q' = \frac{pqw_{12} + q^2w_{22}}{\bar{w}}$ | | | |

- Dosadíme do HW rovnováhy
- Známe-li frekvence alel a fitness genotypů (*s* a *h*), můžeme predikovat změny frekvencí v dalších generacích.
- Pozor! Současně má vliv i drift!

# Modes of selection

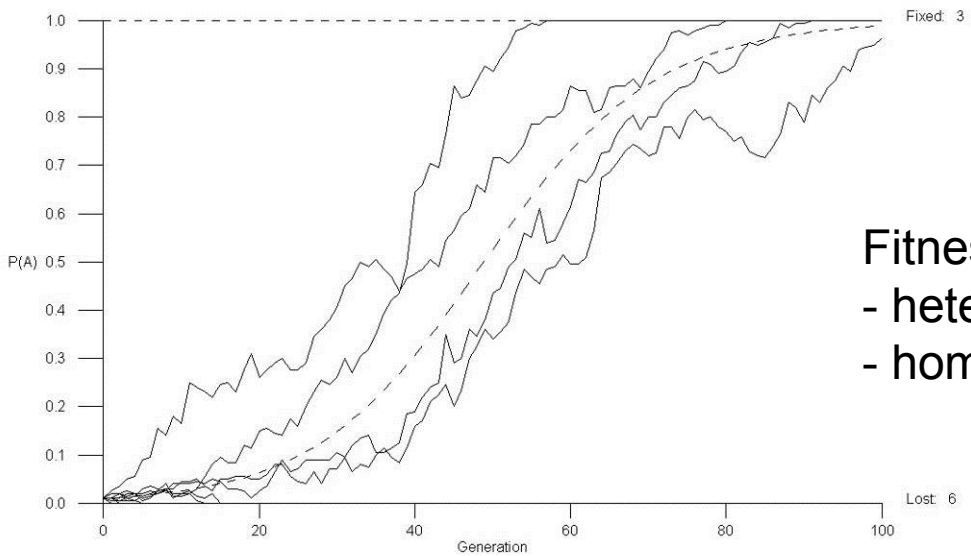
- Positive (adaptive Darwinian) selection
- Negative (purifying) selection

# Positivní selekce




Simulováno 10 populací
Nová mutace
N je 100

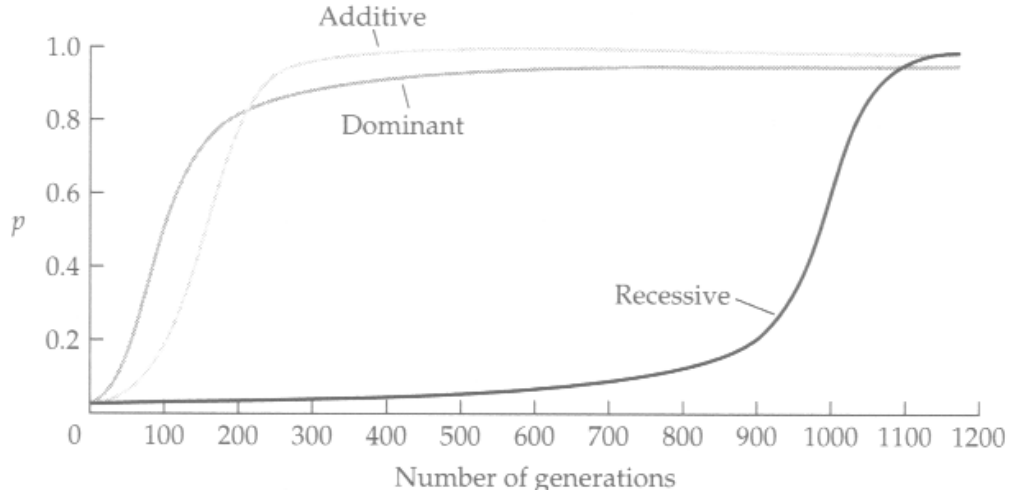
Pouze drift

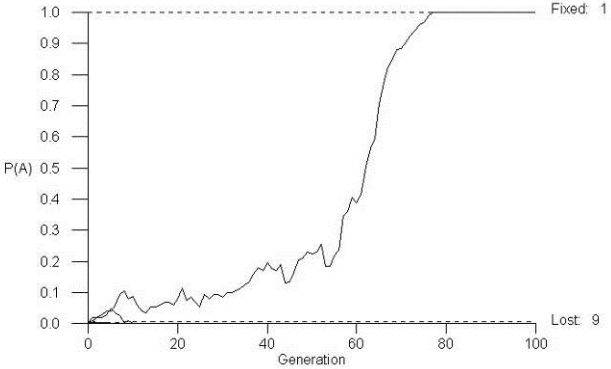



Fitness
- heterozygota pro novou alelu o 10 % vyšší
- homozygota o 20 % vyšší

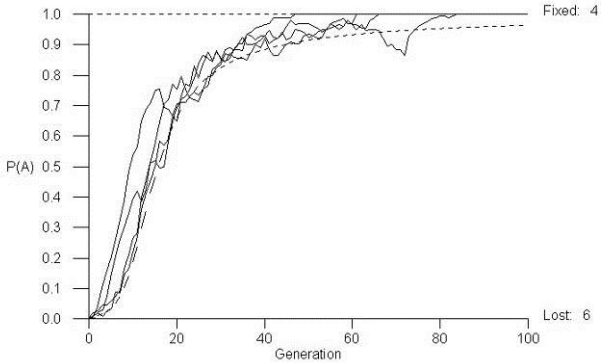
# Fixation of advantageous allele

- Initial allele qrequency
- Selection coefficient
- Degree of dominance

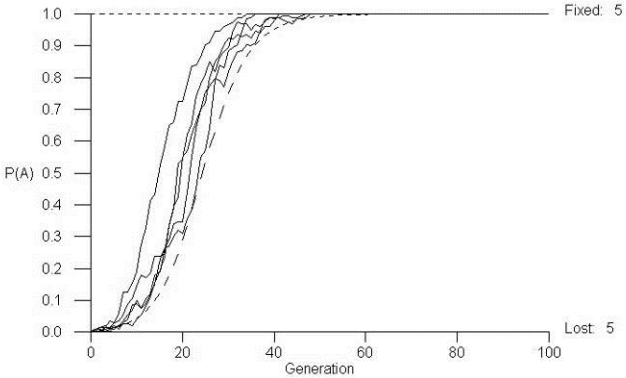



recesivní



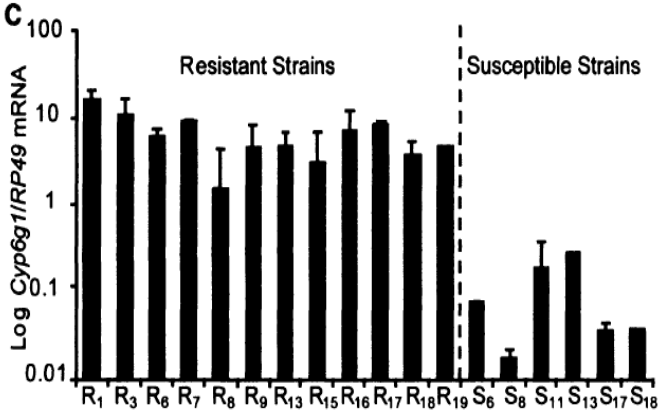

dominantní



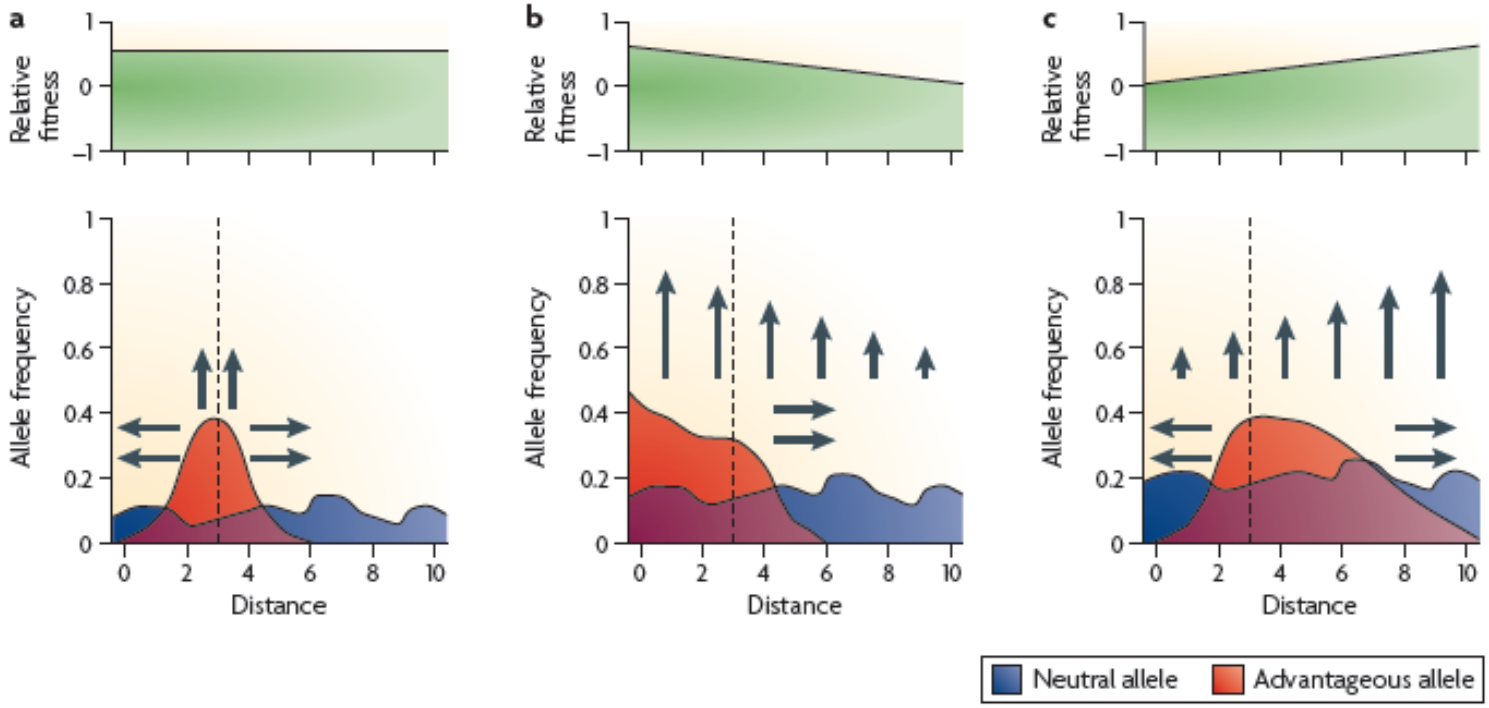

aditivní

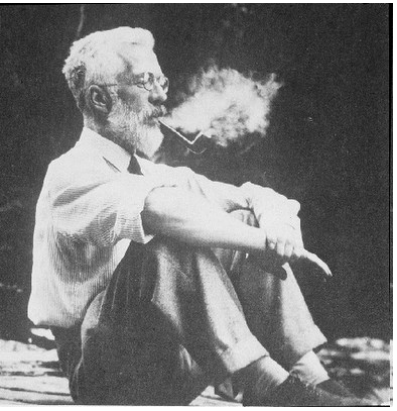
# *D. melanogaster* a rezistence vůči DDT

- Stejný gen u různých populací
- Stejná alela s globálním rozšířením
- Zvýšená transkripce
- Transposon v 5’ oblasti



# ‘wave of advance’ model



# Industriální melanismus a *Biston betularia*

drsnokřídlec březový

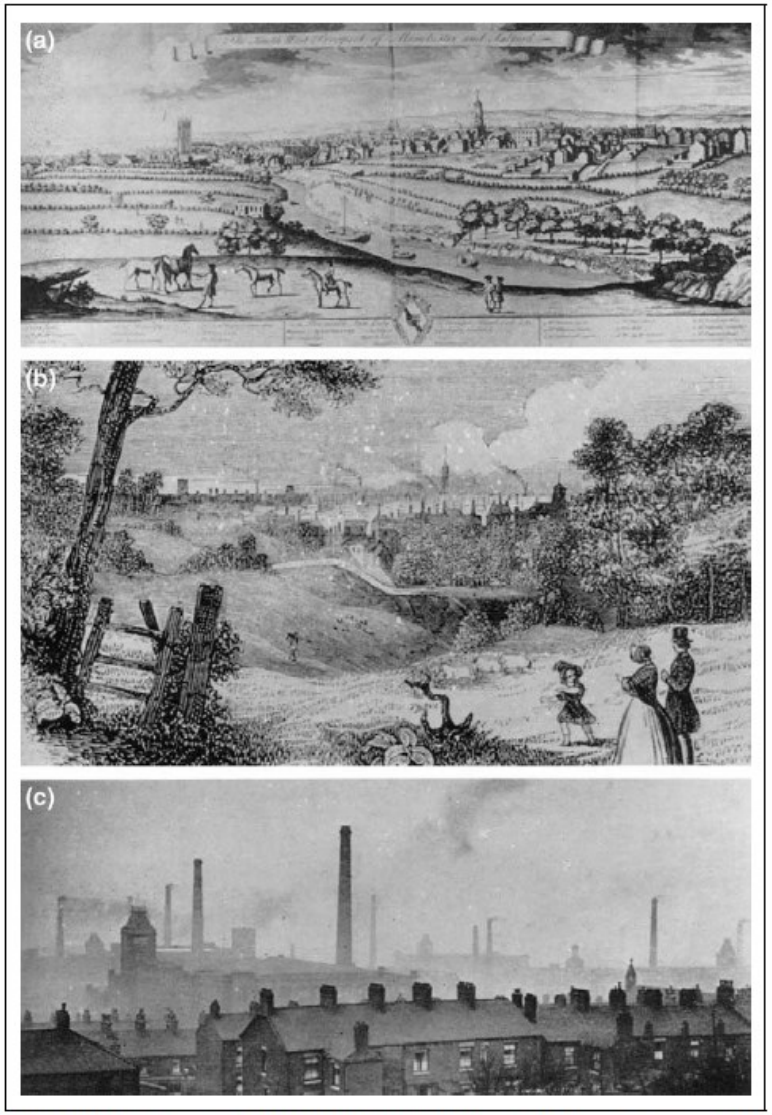
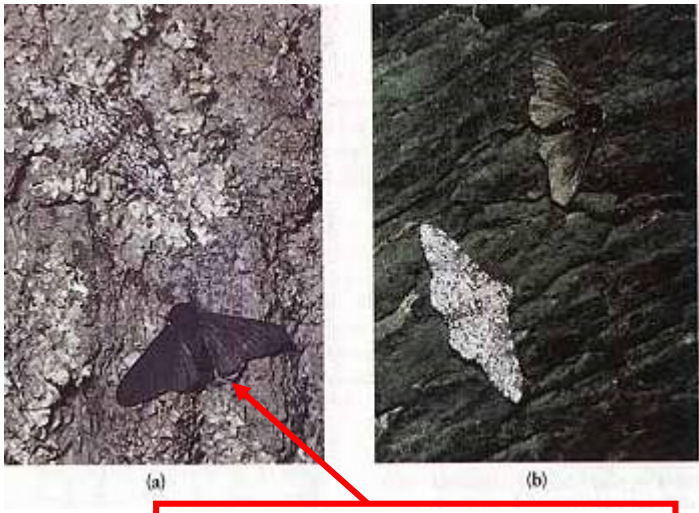
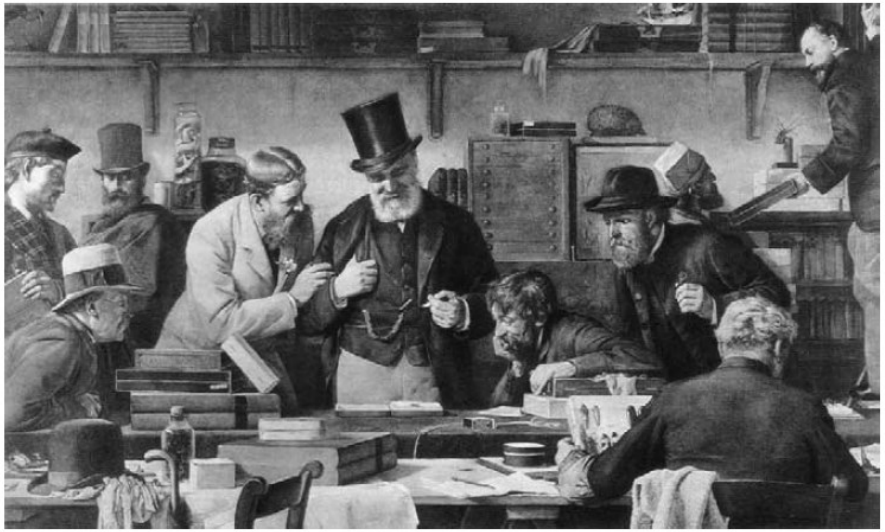



**Figure 1.** The development of industry. A view of Manchester from the surrounding countryside in (a) 1730, (b) 1860 and (c) 1954. (a) and (b) were made from the same spot, and (c) is a photo of the same district.




Carbonaria phenotype



*Henry Bernard Davis Kettlewell*

Carbonaria – poprvé se objevila asi začátkem 19. stol.
↑ frekvence i na > 90 %
↑ šíření na značné území

20. stol.
↓ frekvence
Nápadný ústup až vymizení
Souvisí se znečištěním ovzduší a následným zlepšením podmínek

*s* až 0.2

Různá predace ptáky

Zřejmě jedinečná událost
Mutace na Chr 17

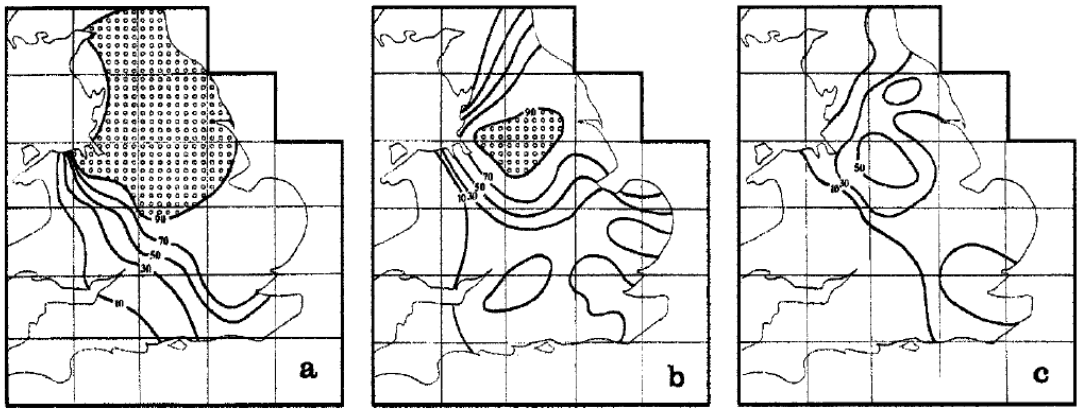



FIGURE 1. FREQUENCY OF *CARBONARIA* IN ENGLAND AND WALES AT AT THREE SUCCESSIVE PERIODS
(**a**) Mid-20th century, derived from data of Kettlewell 1973, Lees and Creed 1975, Steward 1977a, Bishop et al. 1978a, and Lees 1981; (**b**) 1983–1984, from the Open University survey (Cook et al. 1986); (**c**) 1987–1999, from Rothamsted Insect Survey (Cook et al. 2002) and other recent records. The grid shows Ordnance Survey 100 km squares.

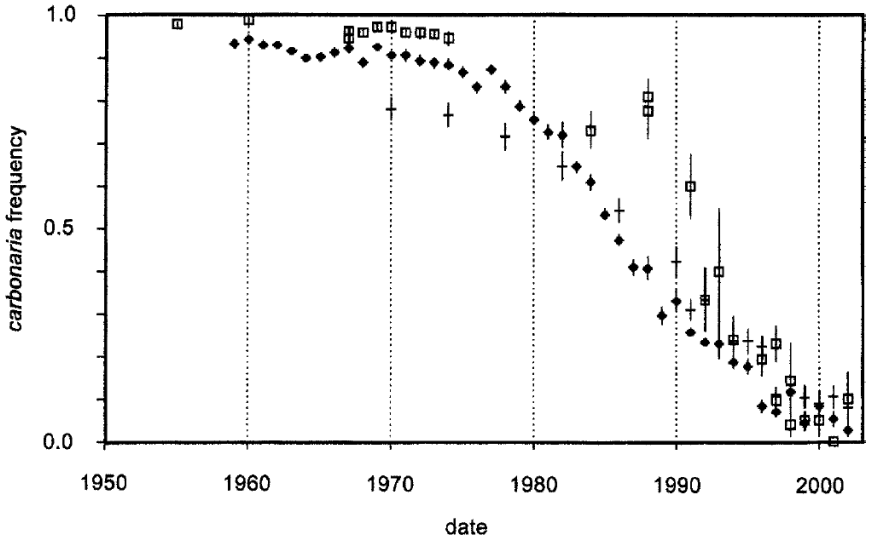
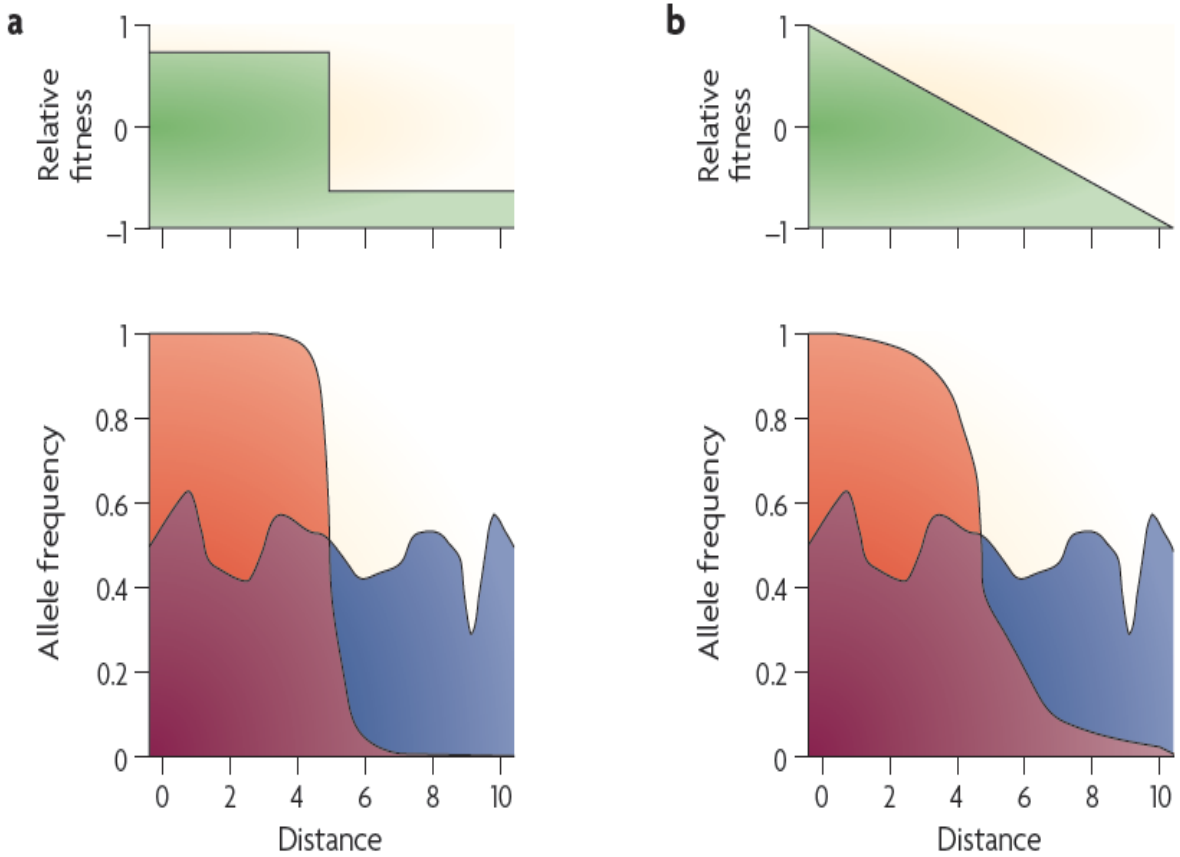


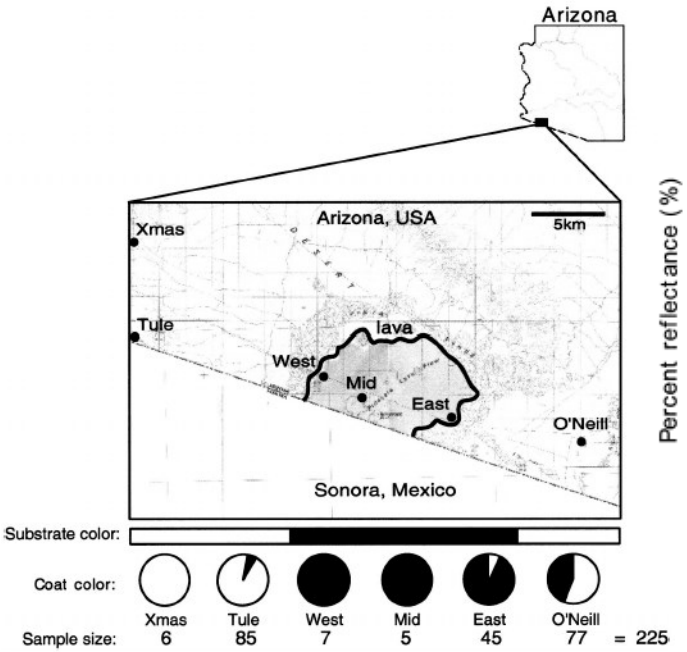

FIGURE 2. DECLINE IN *CARBONARIA* FREQUENCY IN THE 20TH CENTURY
Solid circles: Caldy, Wirral, northwestern England near the border with northern Wales (Clarke et al. 1985; Clarke et al. 1990; Clarke et al. 1994; Grant et al. 1996); squares: Manchester area, northwestern England (Bishop et al. 1978a; Cook et al. 1999); crosses: Kent, east of London (West 1994, 2003). Vertical lines are standard errors.

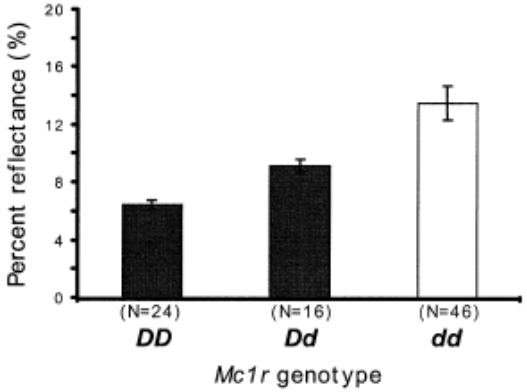
# spatially varying selection
# Isolation by adaptation IBA



pytlouš

# *Chaetodipus intermedius*



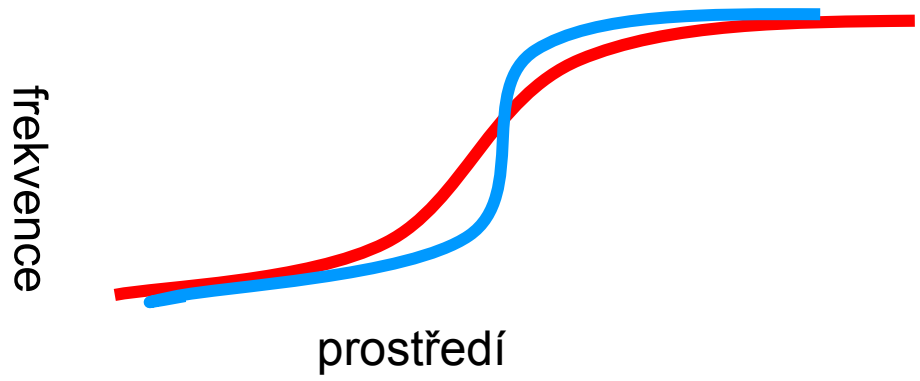



Korelace prostředí – fenotyp – genotyp

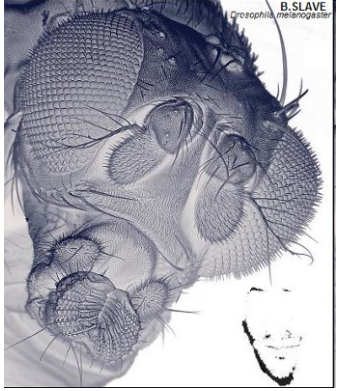
Spíše výjimka

# Klina

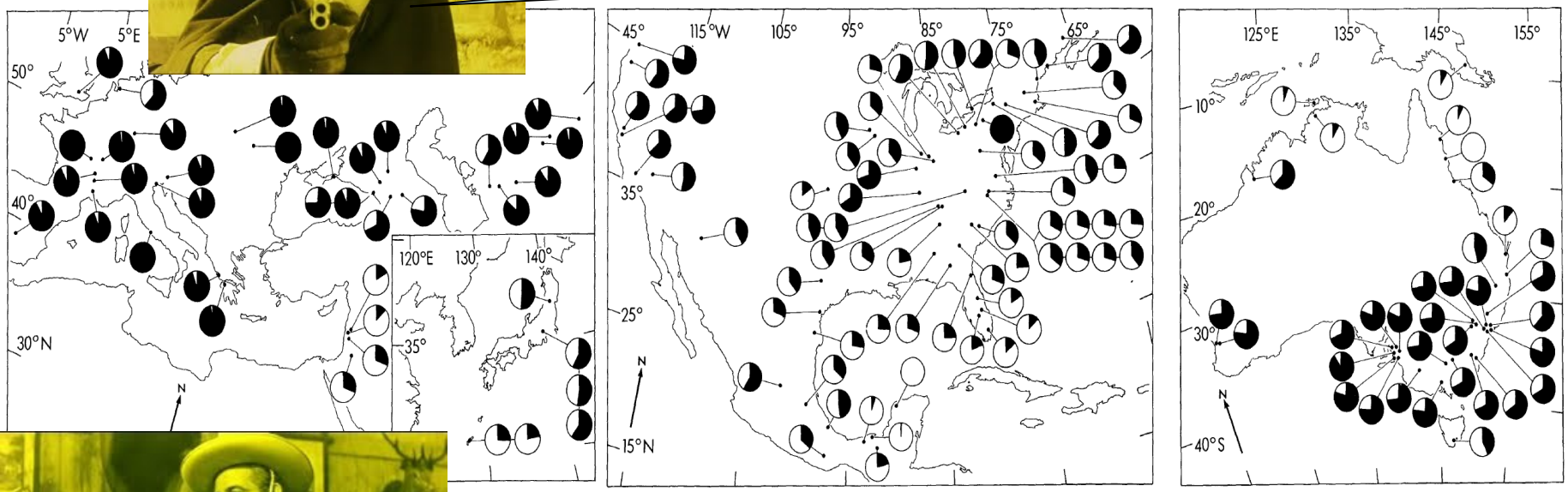
- Gradient frekvencí
- Korelace s prostředím



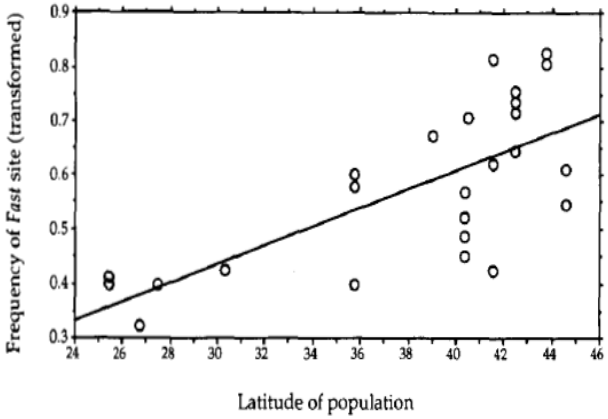
# *Drosophila melanogaster*, rezistence k alkoholu a *Adh*



Alkohol podávaný v malých dávkách neškodí v jakémkoli množství



...mým sedmým nebem zůstane Kolaloka jen




- Rezistence stejně klinální na různých kontinentech
- Alely na *Adh* obdobná klina
- Selekce?
- Více než 350 článků
- Vztah stále nejasný
- Další hráči (aldehyd dehydrogenáza)

# Vznik kliny jinými mechanismy

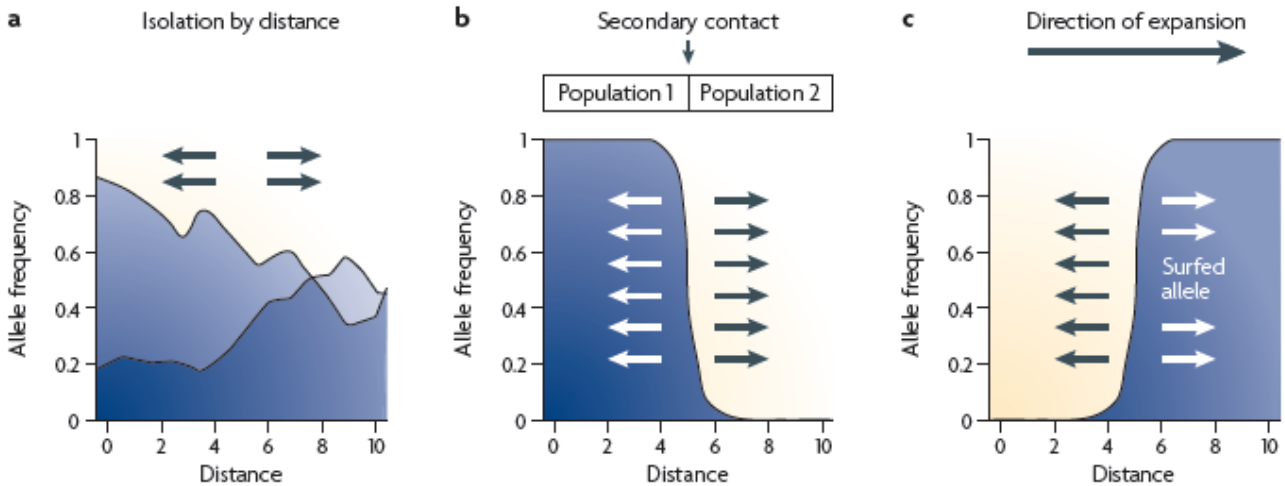
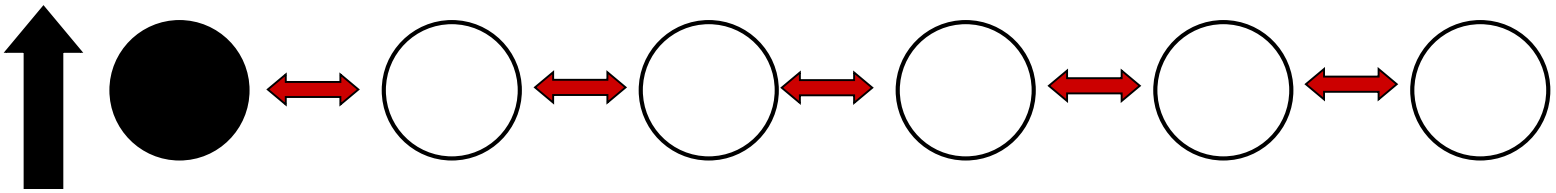
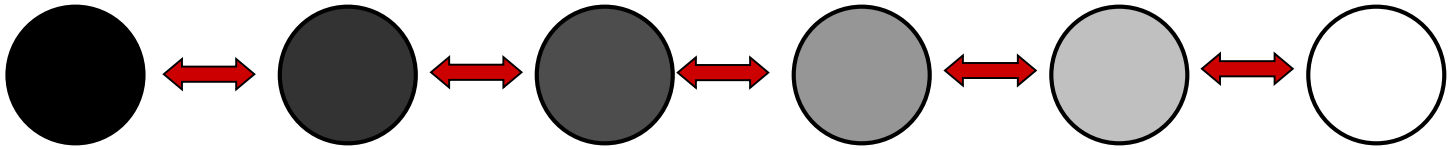


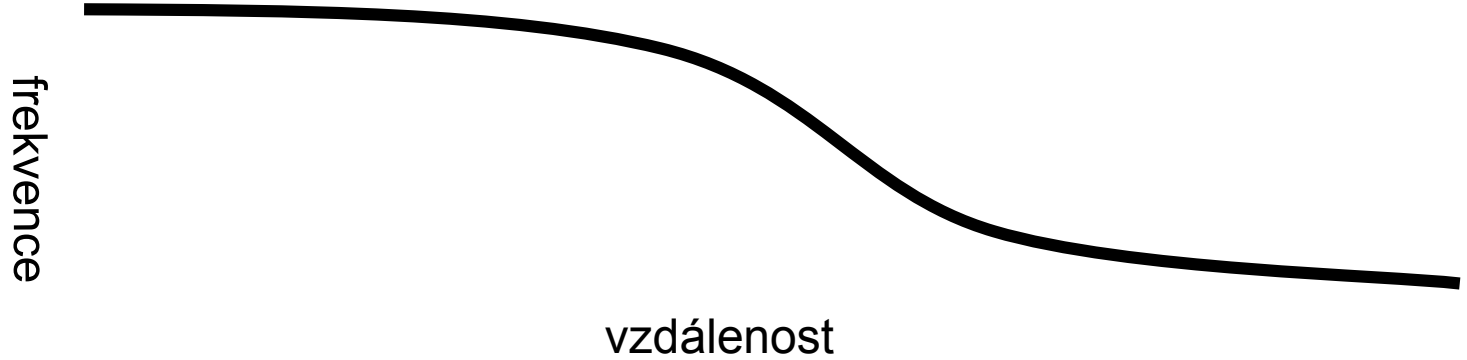

Figure 3 | **Neutral scenarios that produce clines in allele frequencies. a** | Isolation by distance. Under models of isolation by distance, many neutral alleles will show cline patterns, especially along geographic axes with the least gene flow. **b** | Secondary contact between two populations. With secondary contact, neutral alleles will transiently show a cline pattern at the contact zone between the two populations. Note that the allele frequency in the two source populations need not be 0 and 1, as shown here; clines along the secondary contact zone will form even if the allele frequency difference between the two populations is more modest. **c** | Expansion into new territory giving rise to serial founder effects and 'allele surfing'. In this extreme allele surfing scenario, the allele frequency has increased to fixation immediately after expansion. In each case, arrows indicate that none of these patterns is stable — dispersal and drift will erode the clines over time. In all three models, cline patterns that arise can potentially be confused with cline patterns that are expected as a result of selection.

# Isolation by distance

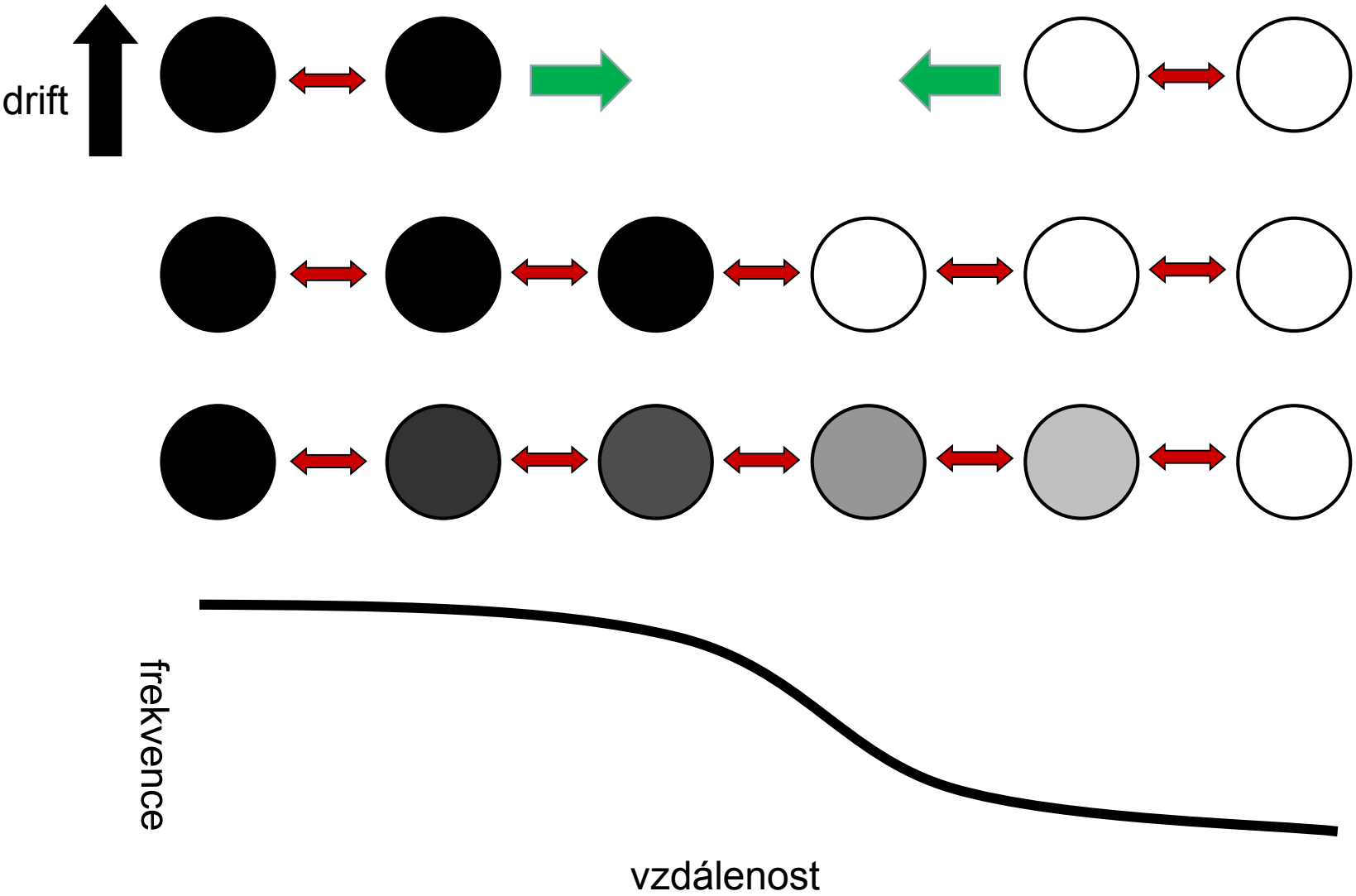


Zvýšení frekvence
nové alely driftem

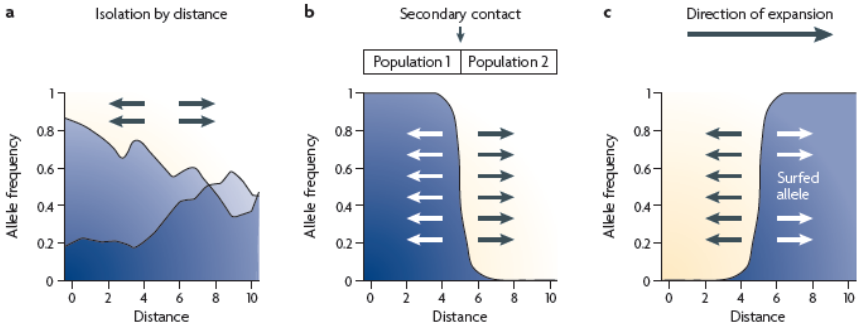




# Secondary contact



# Vznik kliny jinými mechanismy

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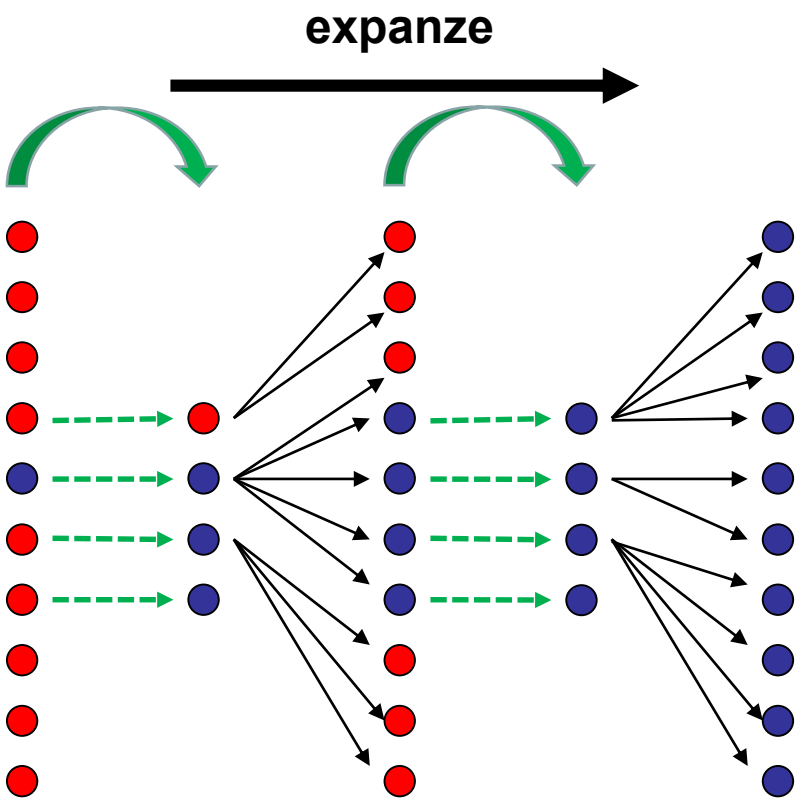



Zvýšení frekvence vzácné alely

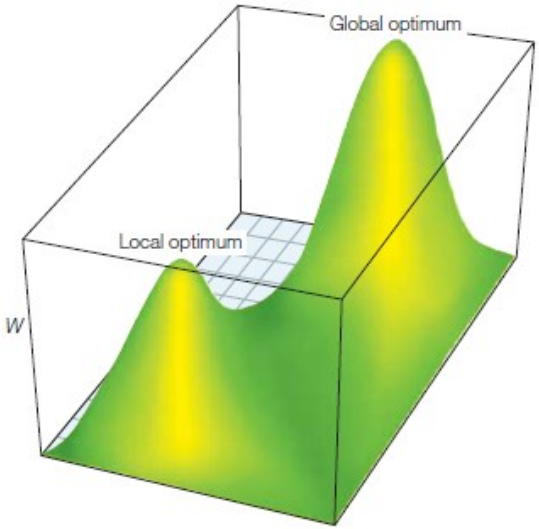
# Selekce a drift

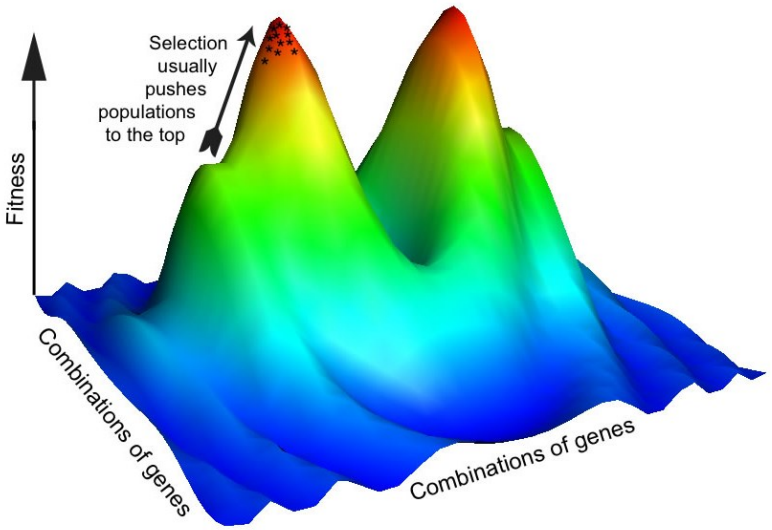
## Shifting balace theory (Sewall Wright)

- Malé populace
  → teoreticky možnost fixace i selekčně nevýhodných mutací

- Překonání nevýhodných mezistavů
  → crossing walleys in adaptive landscape

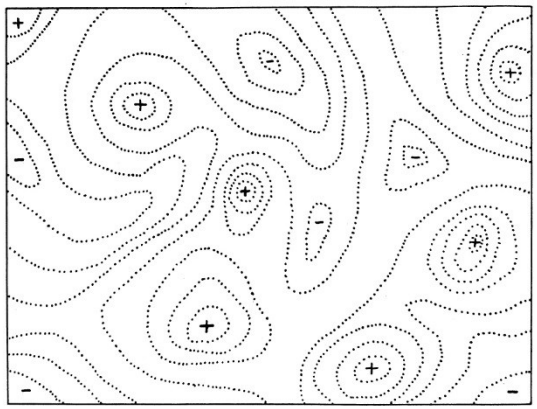
- Dále šíření pomocí **Interdemic selection**

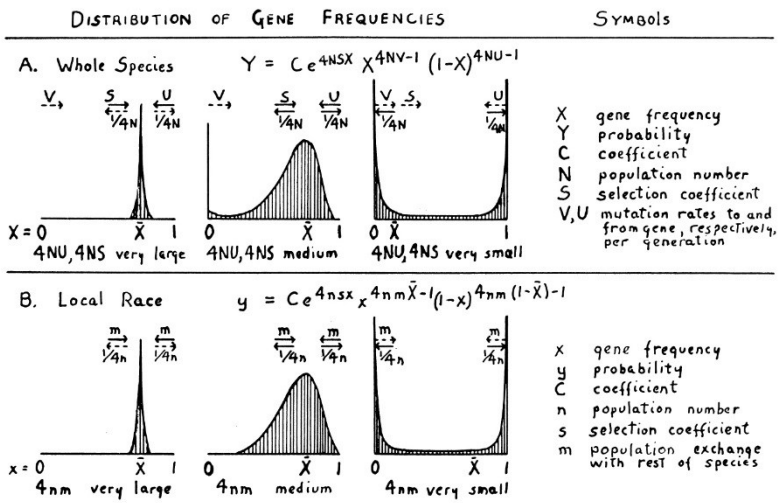


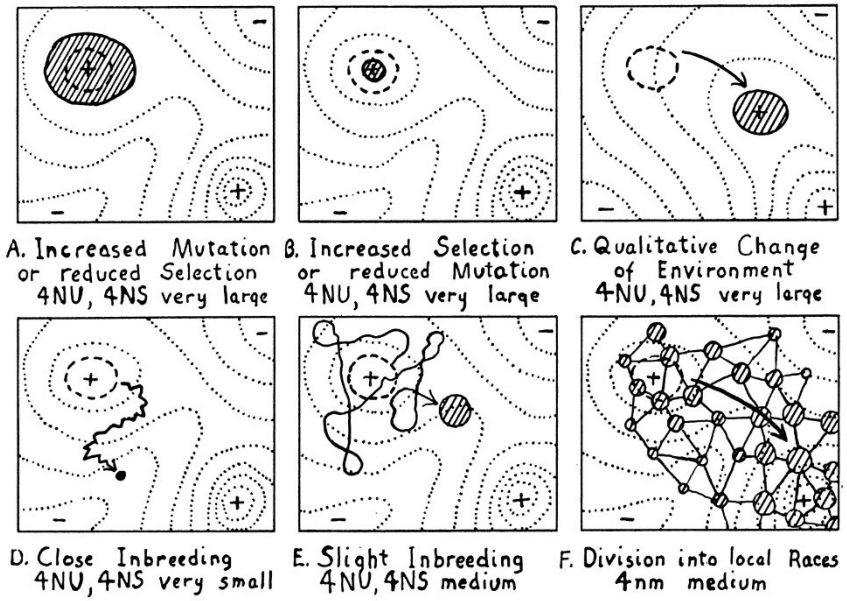



# Shifting balace theory (Sewall Wright)

Controversy, little support from nature

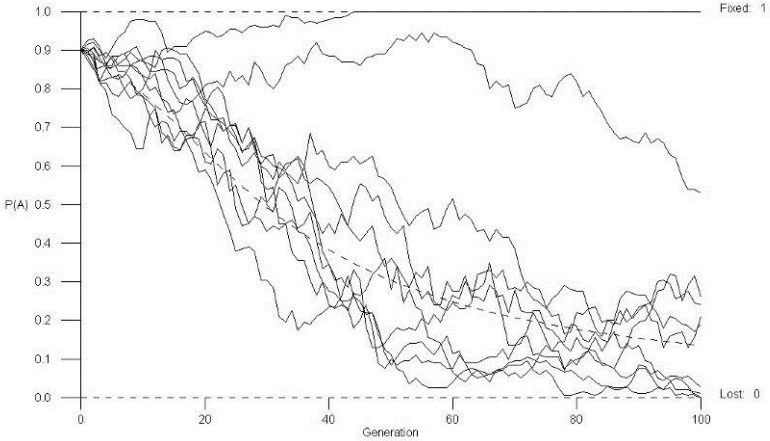




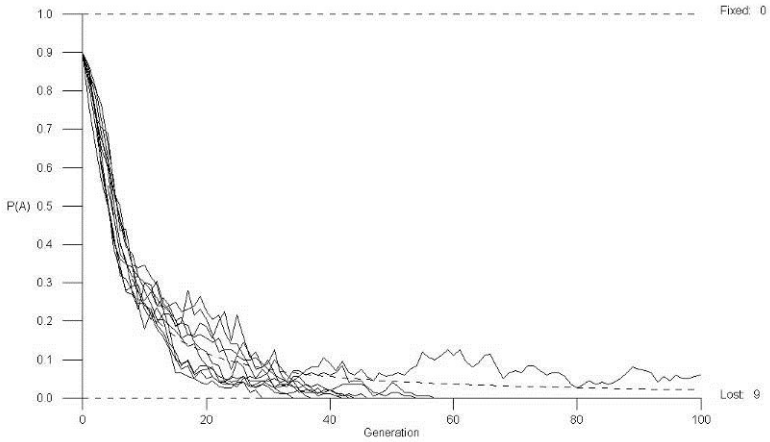
# Negativní (purifikující) selekce

Selekce jen proti recesivním homozygotům
pokles frekvence alely
pro různá *s*



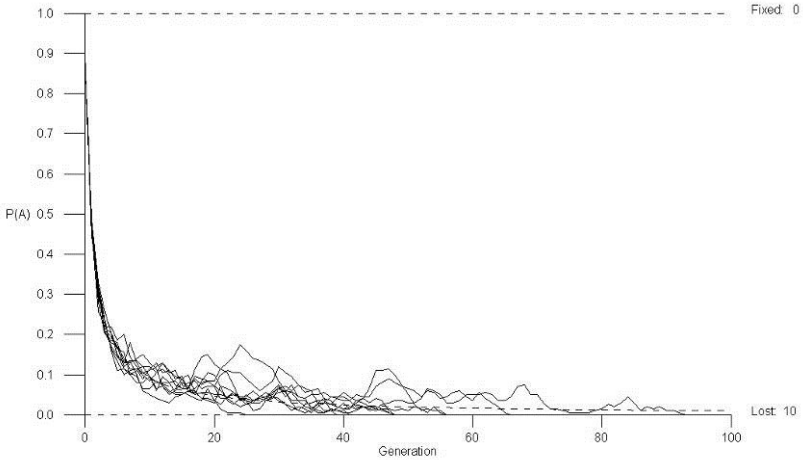

*s* = 0.1

*W fitness*
*s selekční koeficient*
*W = 1 – s*

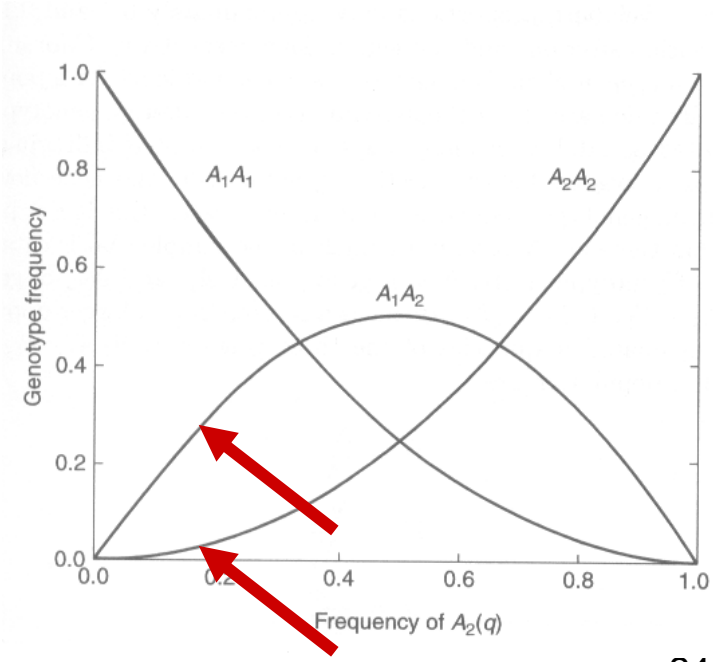



*s* = 0.5

Asymptotický tvar
→ eliminace alely může trvat velmi dlouho
alela schována v heterozygotech




*s* = 1



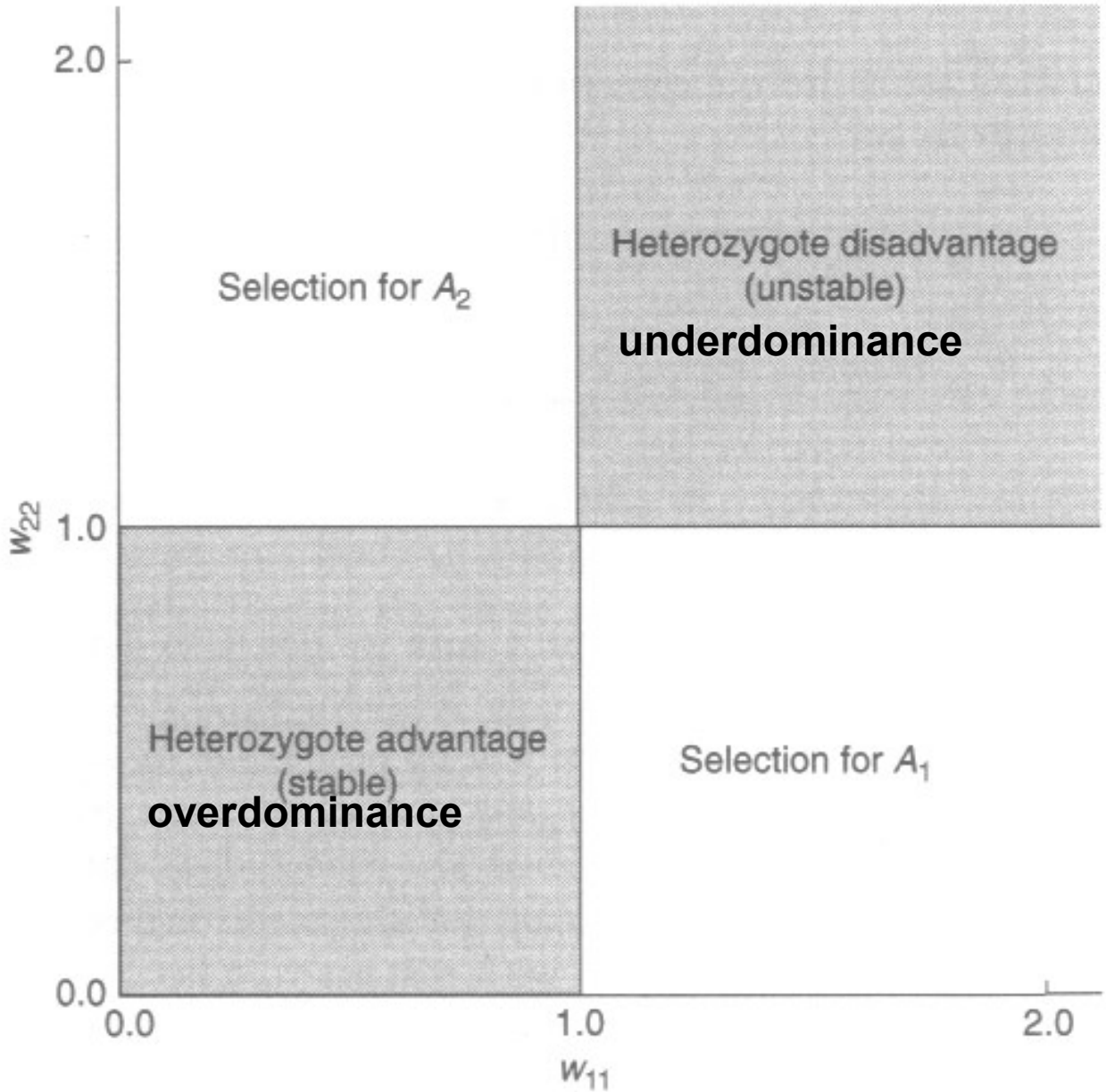
# Vliv rekurentních mutací

- **A** opakovaně mutuje do **a**
- Zpětné mutace málo pravděpodobné
- → rovnováha mezi selekcí a mutacemi
- → stálá frekvence recesivní škodlivé (i letální) alely v populaci

$$\sqrt{\frac{\mu}{s}}$$

- Příklad: aa letální, s = 1, $\mu = 10^{-6}$ → rovnovážná frekvence a = 0.001
- → každý si zřejmě neseme několik recesivních hodně škodlivých alel (odhad 3-8)

# Role heterozygotů




Fitnesses

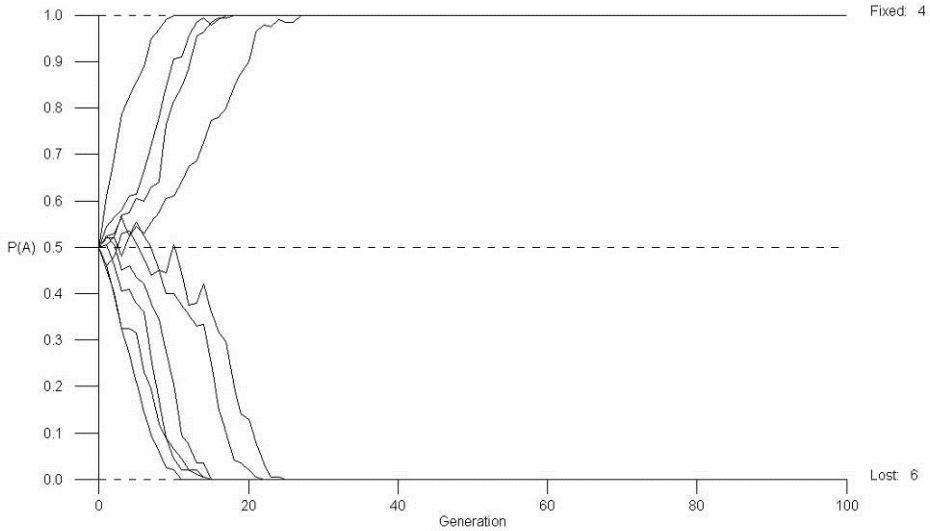
$A_1A_1$ $w_{11}$

$A_2A_2$ $w_{22}$

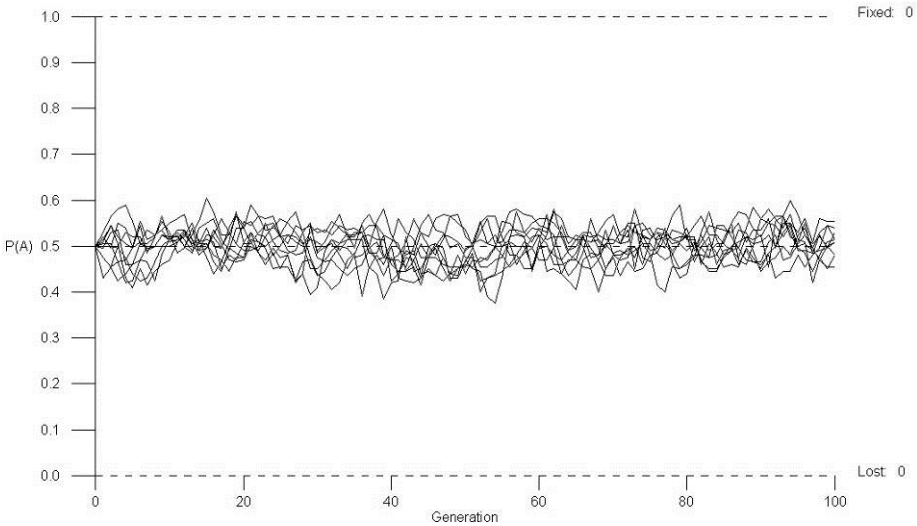
$A_1A_2$ 1

# Role heterozygotů

- Against
  - heterozygotes (heterozygote disadvantage, underdominance), nestabilní
  - homozygotes (heterozygote advantage, overdominance), stabilní
    → frekvence alel odpovídá *w* homozygotů



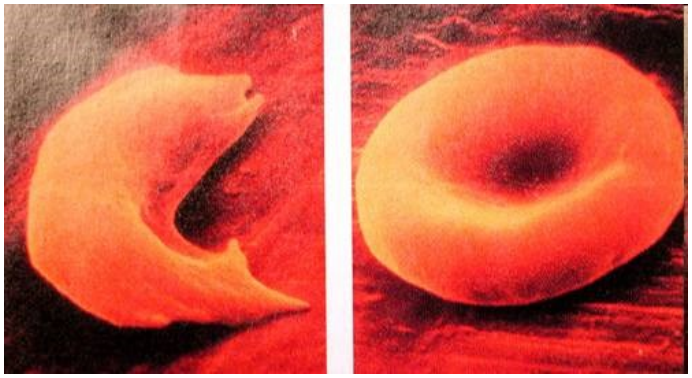

underdominance

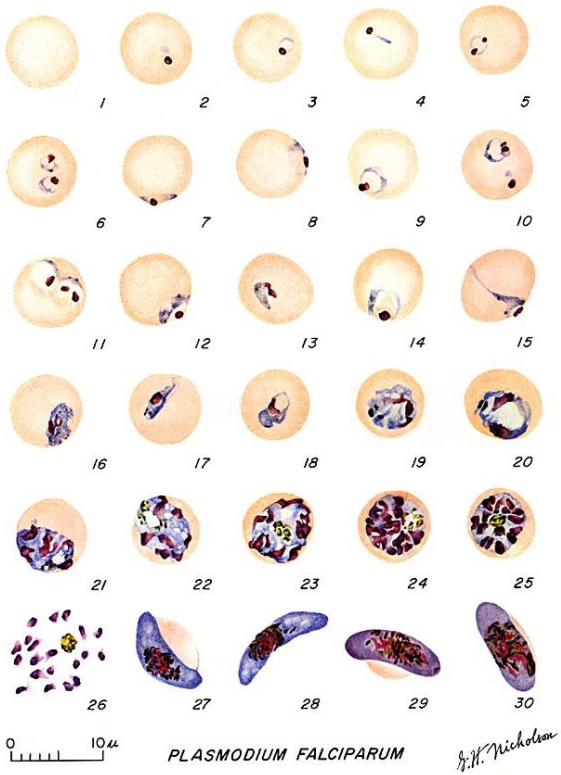


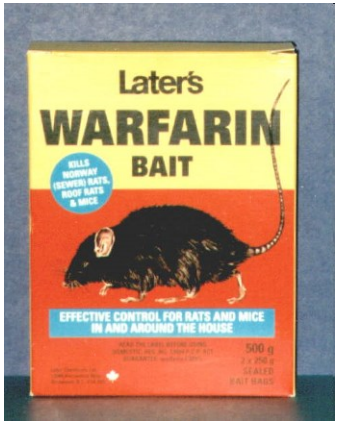

overdominance

# overdominance

- Příklady (učebnicové):
- srpková anémie a výskyt malárie
  - hemoglobin *β* locus
  - normal (*A*), sickle-cell (*S*) allele
  - (je i *C* alela, rezistence a *CC* bez problémů)
- Warfarin a potkani:
  - Warfarin – jed, antikoagulant
  - Dominantní alela rezistence
  - Rychlé šíření
  - Homozygoti potřebují 20x více vitamínu K
  - (Někdy asi nepotřebují)







# Balancing selection

- Overdominance  je jen jedním z příkladů

  **Balancing selection** = selekce udržující polymorfismus

- Jsou i jiné příčiny

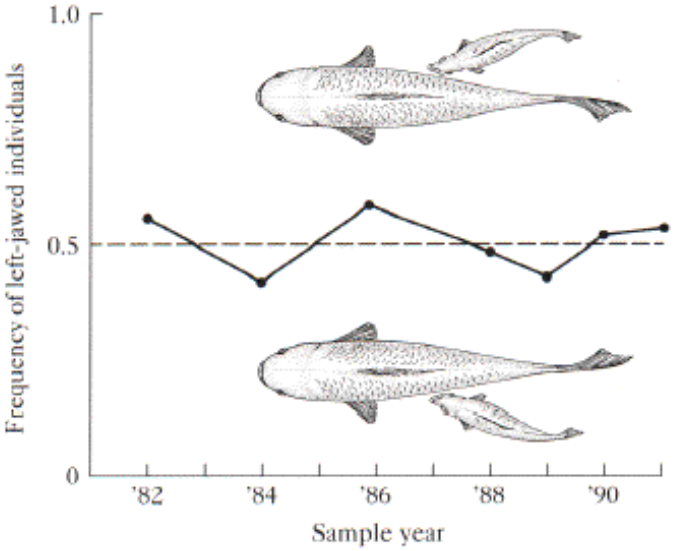
- Často – **Frequency-dependent selection**

  *The fitness of a genotype depends on the genotype frequencies in the population.*

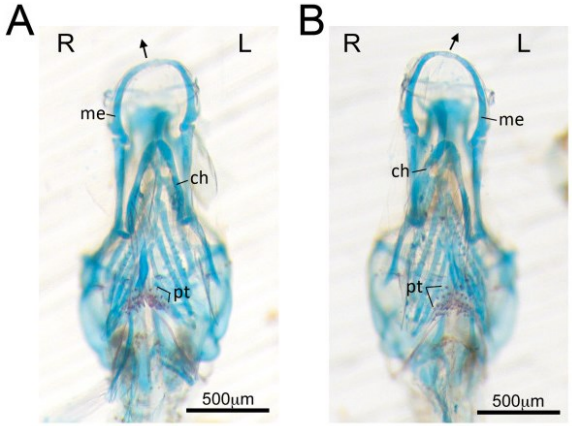
- Inverse frequency - dependent selection

  the rarer a phenotype → the greater its fitness

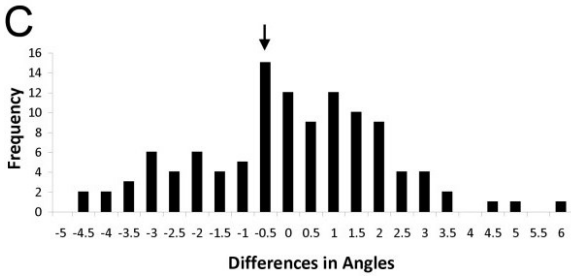
# Frequency dependent selection




*Loxia curvirostra*
křivka obecná (i jiné druhy)
dva typy překřížení zobáku
(není to záležitost jednoho genu, nízká heritabilita)




*Perissodus microlepis*
cichlida z jezera Tanganyika
žere šupiny jiných ryb
dva typy natočení tlamy
stabilní rovnovážná frekvence
? dominant (lefty) allele - homozygous lethal
versus Van Dooren et al.
– asymetrie je plastická, víc genů

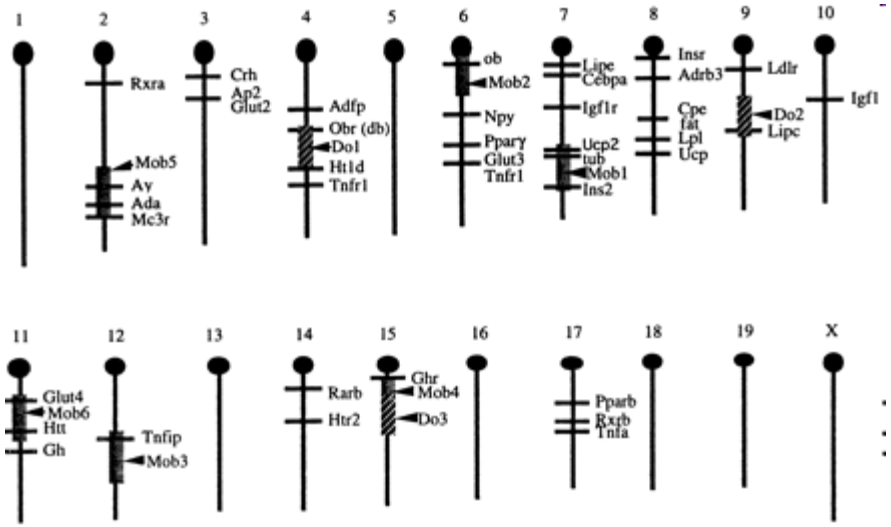


## The evolution of the sex ratio by frequency-dependent selection

- Sex ratio 0.25 (1 male to 3 females)
- Each female – 4 offspring
- → male – 12 offspring
- Grandchildern
  (3*4) through daughters + (1*12) through sons = 24
- Mutation → sex ratio 0.5
- → Grandchildern
  (2*4) through daughters + (2*12) through sons = 32
- → allele will increase in frequency
- → sex ratio 0.5 = **evolutionary stable strategy** (ESS)

# Geny nejsou osamělí hráči

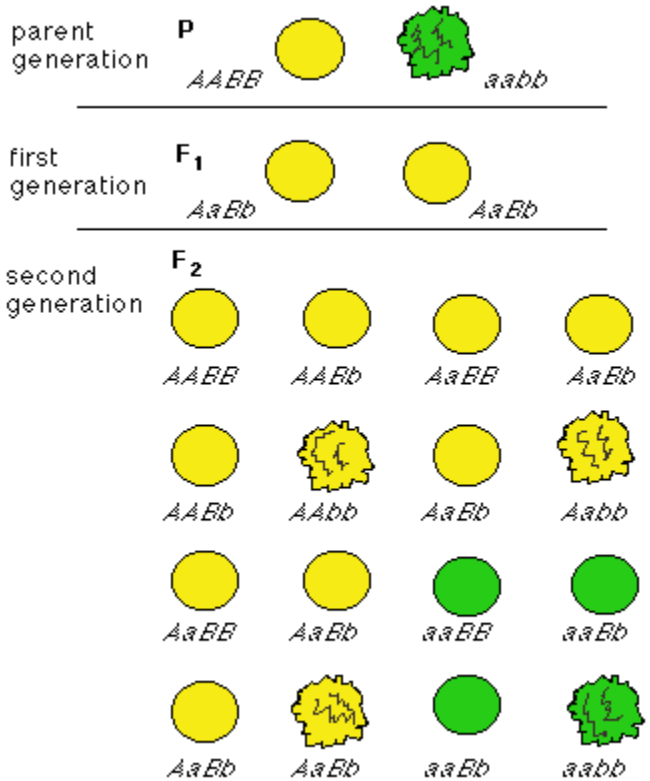
- Pleiotropie
- Epistáze
- Linkage

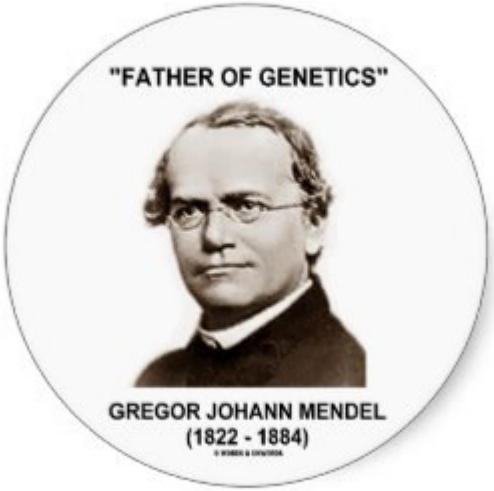



- Doposud modely s jedním genem (lokusem)
- Selekce na více (mnoho) genů současně

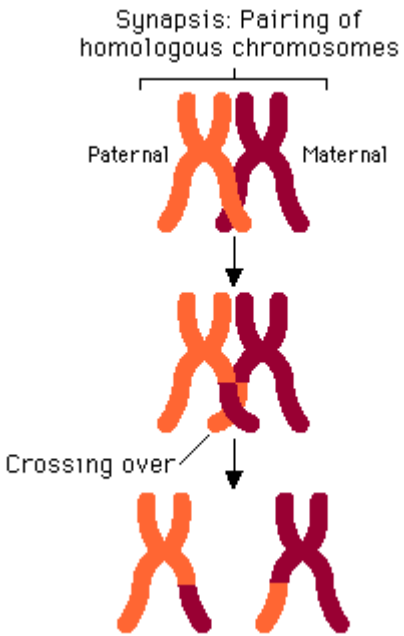
# Mendel's second "law"

Predicts independent assortment of multiple loci:
during gamete formation, the segregation of alleles of one gene is independent of the segregation of alleles of another gene.



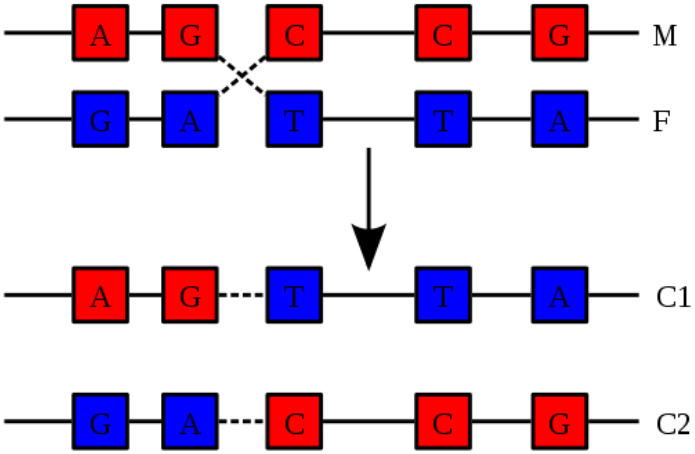



# Linkage disequilibrium




- = gamete disequilibrium
  = gametic phase disequilibrium

- Nonrandom association of alleles at different loci into gametes

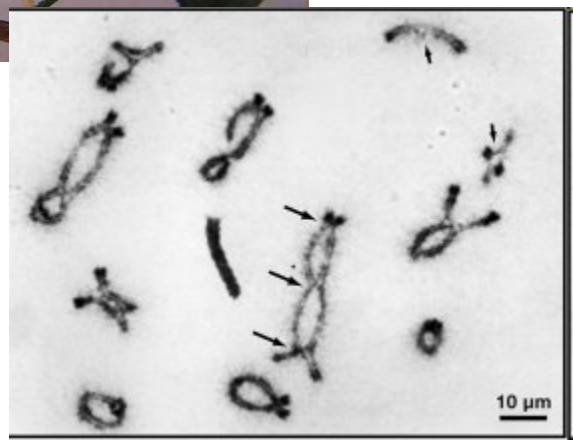



- Linkage versus epistasis

- Linkage
  = co-inheritance caused by physical location

# Rekombinace

Meiotic crossovers → chiasmata




- Nové kombinace alel
- Odstranění škodlivých mutací
  (z blízkosti výhodných alel)
- Správná disjunkce

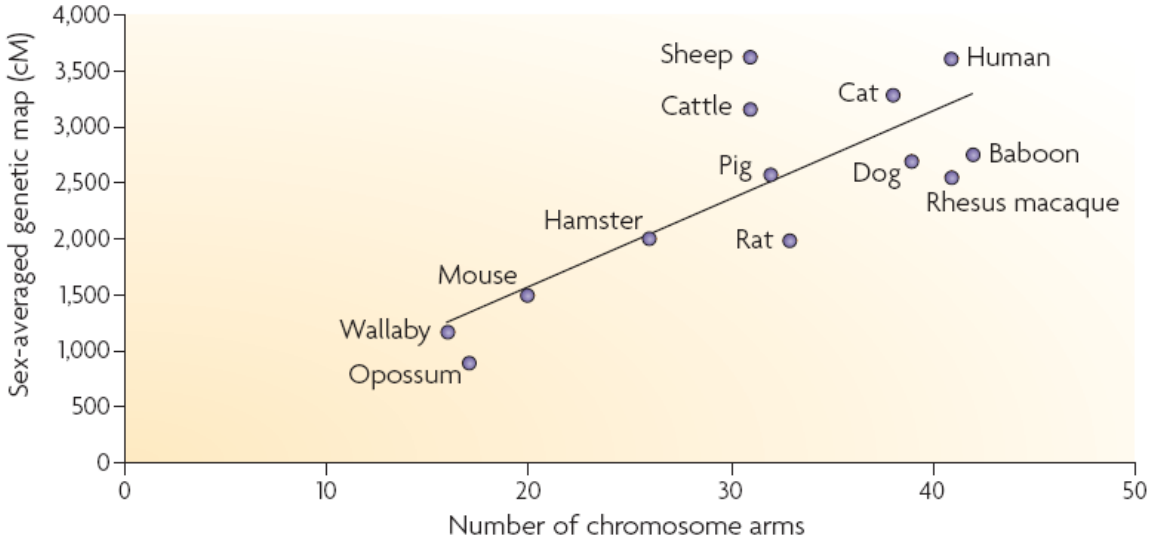
- obligate crossover
- crossover interference
  = nebývají blízko sebe

  *CO decreases the likelihood*
  *that another forms nearby on the same chromosome*

- → malé Chr – jeden CO
  → velké víc ale daleko od sebe

# Míra rekombinace

## Liší se mezi druhy



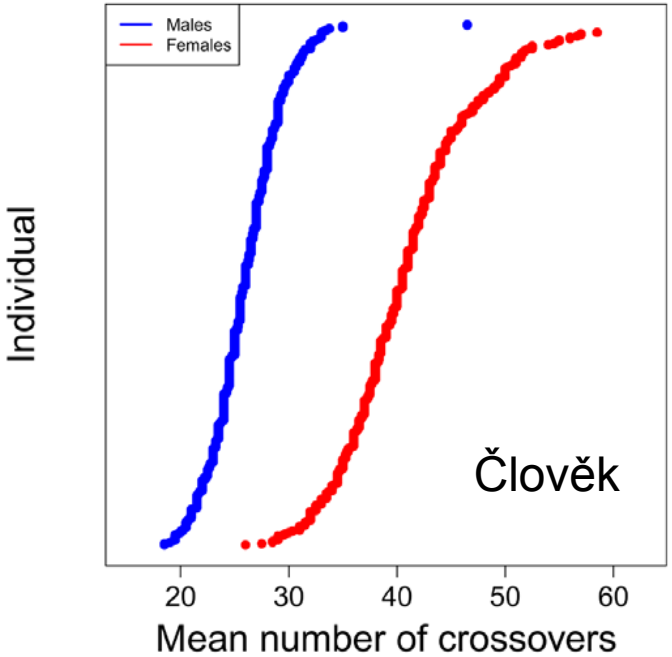

- Druhy s více malými chromosomy rekombinují více než druhy s málo velkými chromosomy.
- Korelace míry rekombinace s celkovým počtem ramen chromosomů (dokonce lépe než s celkovým počtem chromosomů)
- Vyjadřuje nutnost jednoho crossing-overu na chromosomální rameno, aby bylo zabráněno aneuploidiím?

- **Rozdíly mezi pohlavími**

- extrémní případ:
  *Haldane-Huxley rule*
  Pokud jedno pohlaví nerekombinuje vůbec, je to pohlaví heterogametické

- Vedlejší produkt omezení rekombinace pohlavních chromosomů?

- male-female difference in gametic selection = větší selekce → větší snaha zachovat úspěšné haplotypy

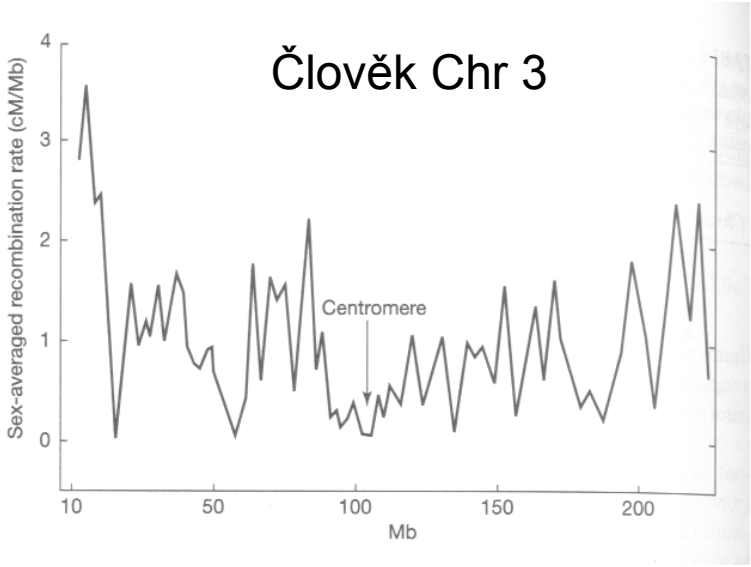


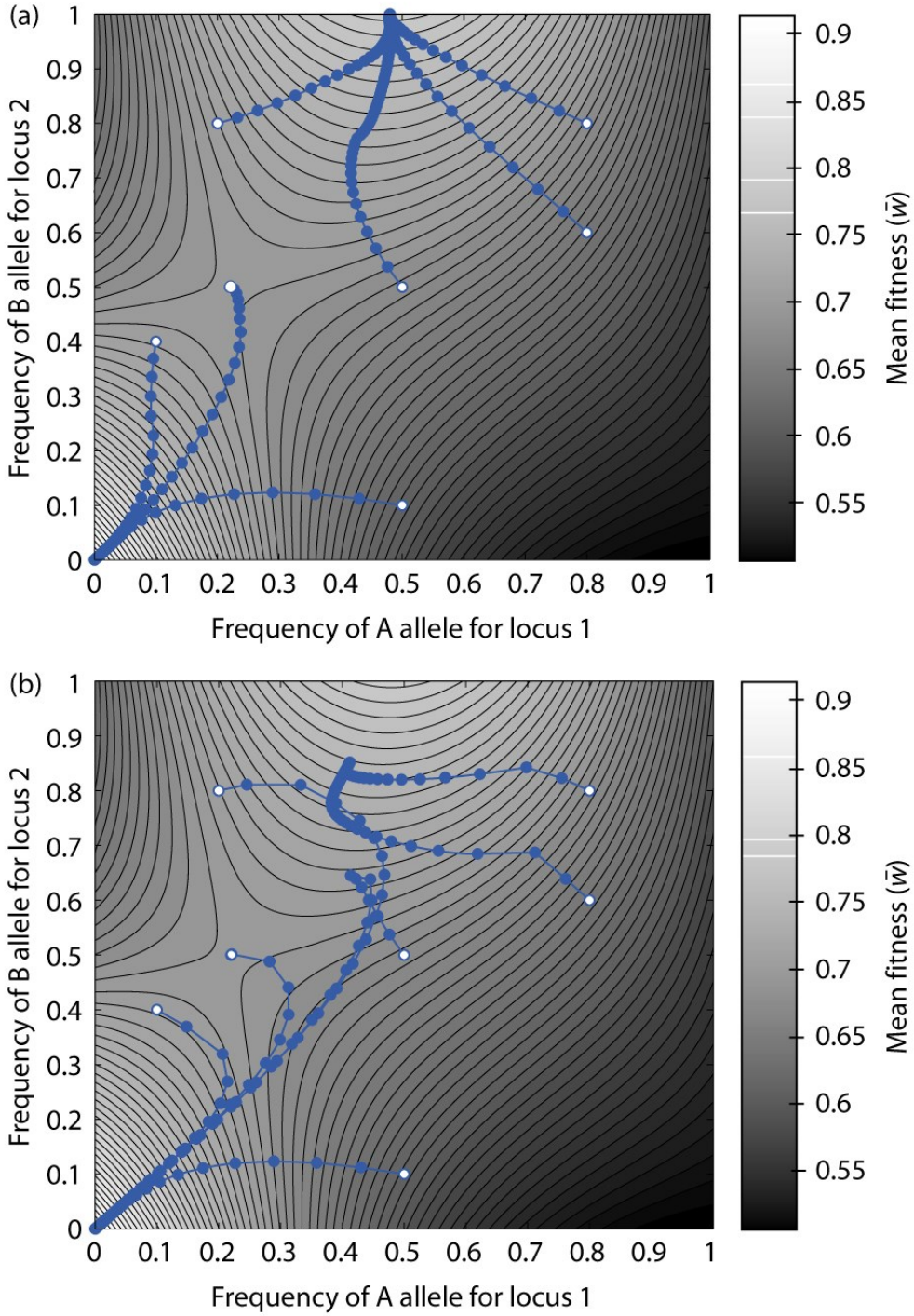

| Species | $r_m$ [a] | $r_f$ [b] | Heterogametic Sex |
|---|---|---|---|
| *Drosophila* | 0 | + [c] | Male |
| *Bombyx* | + | 0 | Female |
| *Apotettix* | − | + | Male |
| *Paratettix* | − | + | Male |
| *Mus/Rattus* | − | + | Male |

Listed are the data available to Haldane [4] when he proposed the Haldane-Huxley rule.
[a] $r_m$ represents recombination in males.
[b] $r_f$ represents recombination in females.
[c] Plus and minus symbols indicate the direction of heterochiasmy, and zero indicates achiasmy.

# Rozdíly

- Mezi jedinci
  - pod selekcí
  - málo rekombinací → snížení fertility
- Mezi chromosomy
- Uvnitř chromosomů
- Hotspots
  - 1-2 kb
  - Myers motif: 13-mer CCNCCNTNNCCNC
  - zinc finger array of PRDM9
  - Jiné hotspots u člověka a šimpanze
  - Hotspoty u lidí jsou polymorfní




(a)
Frequency of B allele for locus 2
Frequency of A allele for locus 1
Mean fitness ($\bar{w}$)
0
0.1
0.2
0.3
0.4
0.5
0.6
0.7
0.8
0.9
1
0.55
0.6
0.65
0.7
0.75
0.8
0.85
0.9
(b)
Frequency of B allele for locus 2
Frequency of A allele for locus 1
Mean fitness ($\bar{w}$)

# Linkage disequilibrium

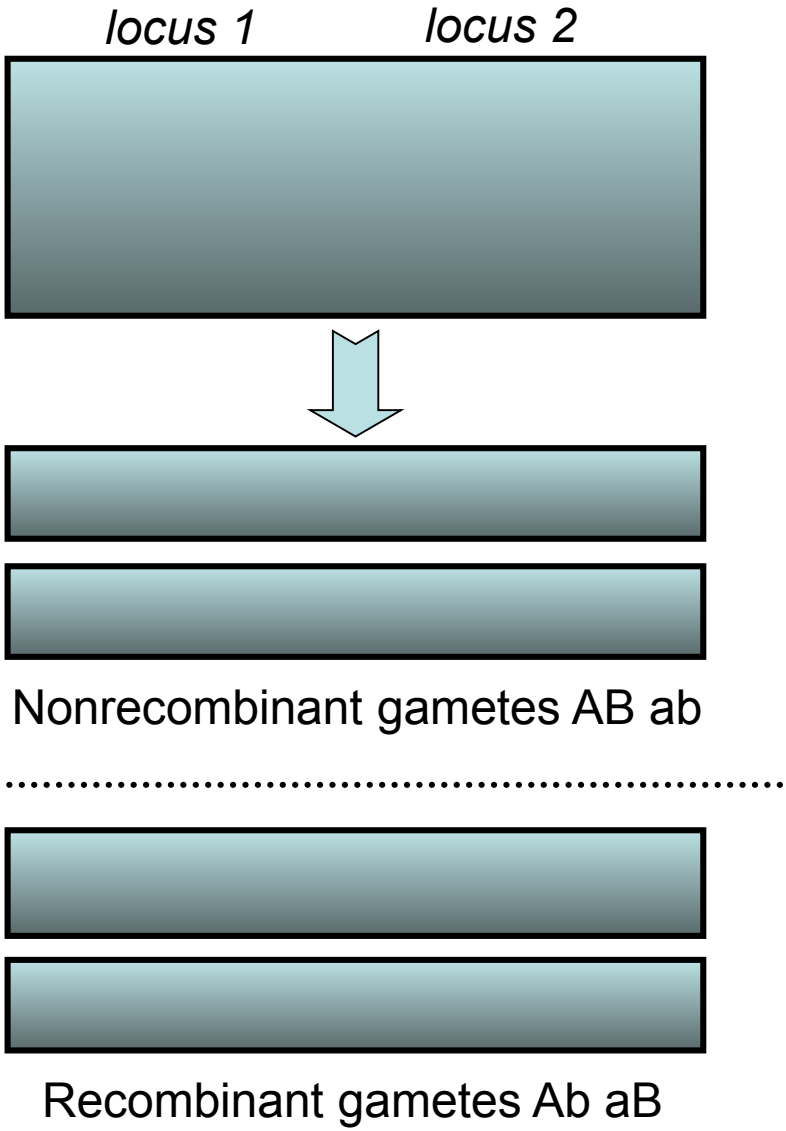
- = gamete disequilibrium
  = gametic phase disequilibrium

- Nonrandom association of alleles at different loci into gametes

- Linkage versus epistasis

- AB/ab
- Nonrecombinant gametes AB ab
- Recombinant gametes Ab aB
- Recombination fraction
  ***r (c)*** = rate of recombination
- Geny daleko (nebo na jiných Chr) ***r*** = 0.5
- Geny velice blízko ***r*** = 0
- ***r*** < 0.5 → linked genes

Two loci genotypes Aa Bb



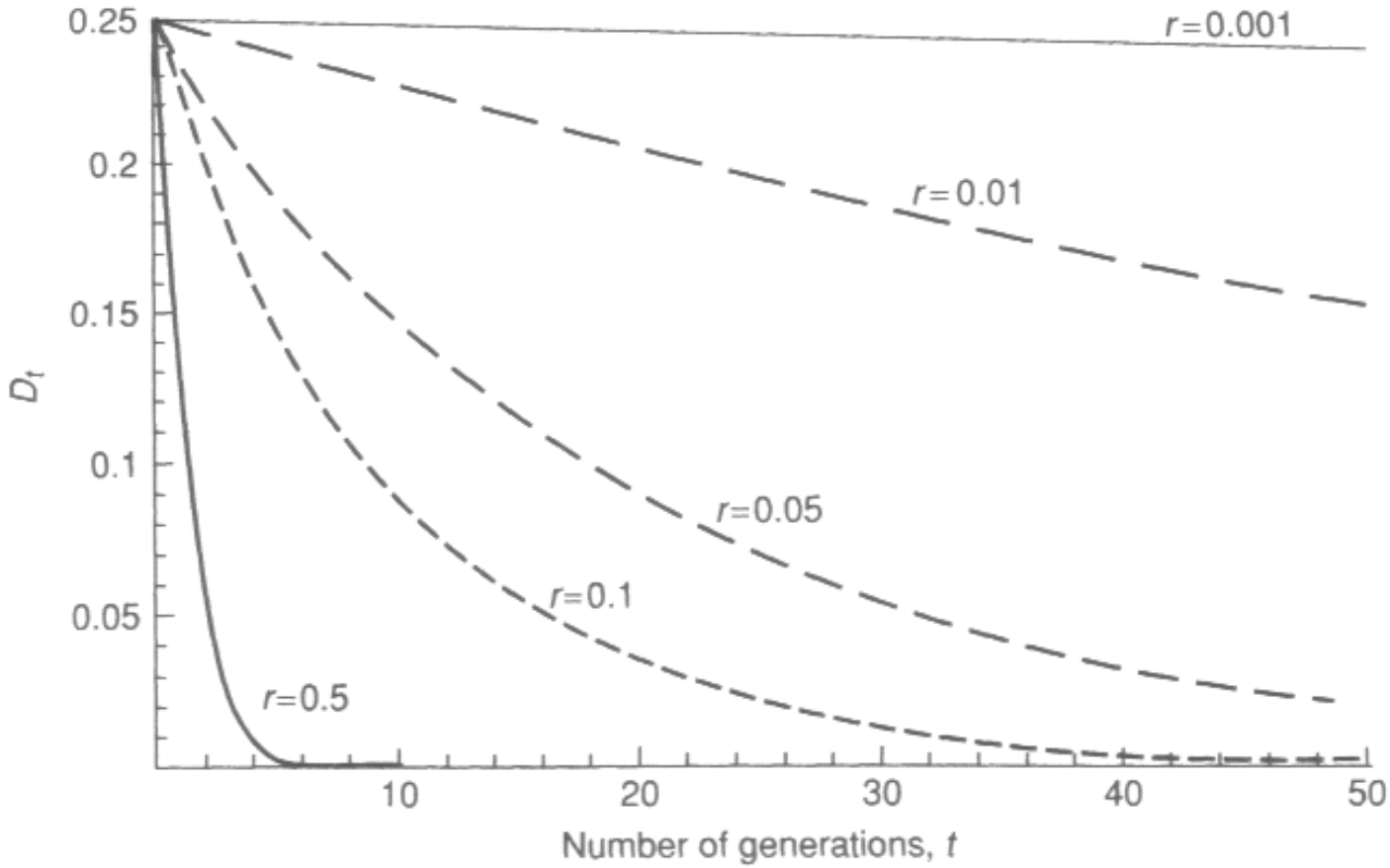
| A | B |
|---|---|
| a | b |

# Linkage (dis)equilibrium

*g* frekvence gamet, $\boldsymbol{D} = g_{AB}g_{ab} - g_{Ab}g_{aB}$

| **Frekvence gamet** | | | Alely genu 1 | |
|---|---|---|---|---|
| | | | **A** | **a** |
| | | | $p1$ | $p2$ |
| Alely genu 2 | **B** | $q1$ | **AB** $g_{AB} = p1q1$ | **aB** $g_{aB} = p2q1$ |
| | **b** | $q2$ | **Ab** $g_{Ab} = p1q2$ | **ab** $g_{ab} = p2q2$ |

K ustavení linkage equilibrium (vymizení LD) je třeba několika (mnoha) generací
Srovnej s jednou generací pro HW equilibrium




***r (c)*** = rate of recombination, ***D*** = *g*AB*g*ab - *g*Ab*g*aB = LD parameter

- V praxi výpočet z genotypů

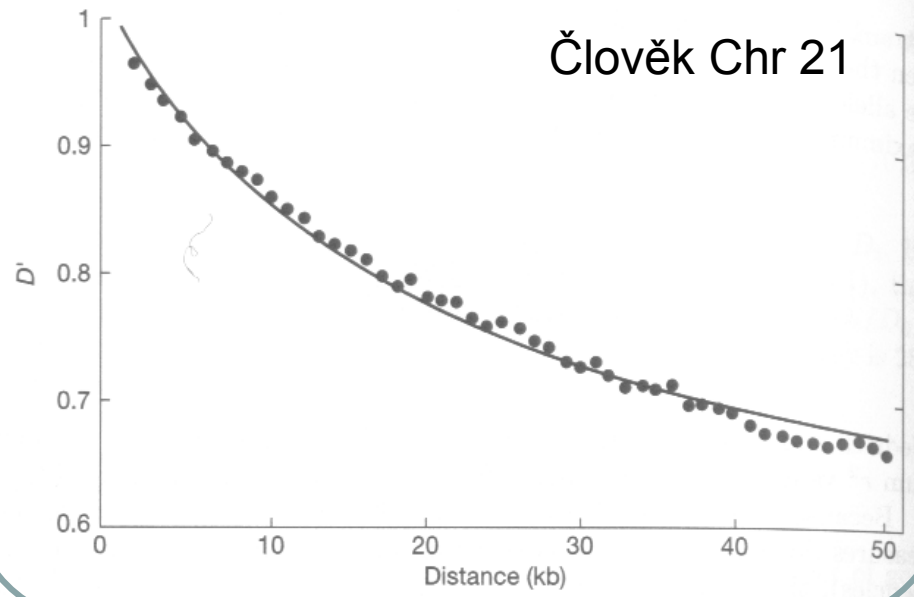
- Normalized linkage disequilibrium
- *D*', Děleno maximální teoretickou hodnotou (od 0 do 1)

- ***R***$^2$ (od 0 do 1), odchylky se dobře testují

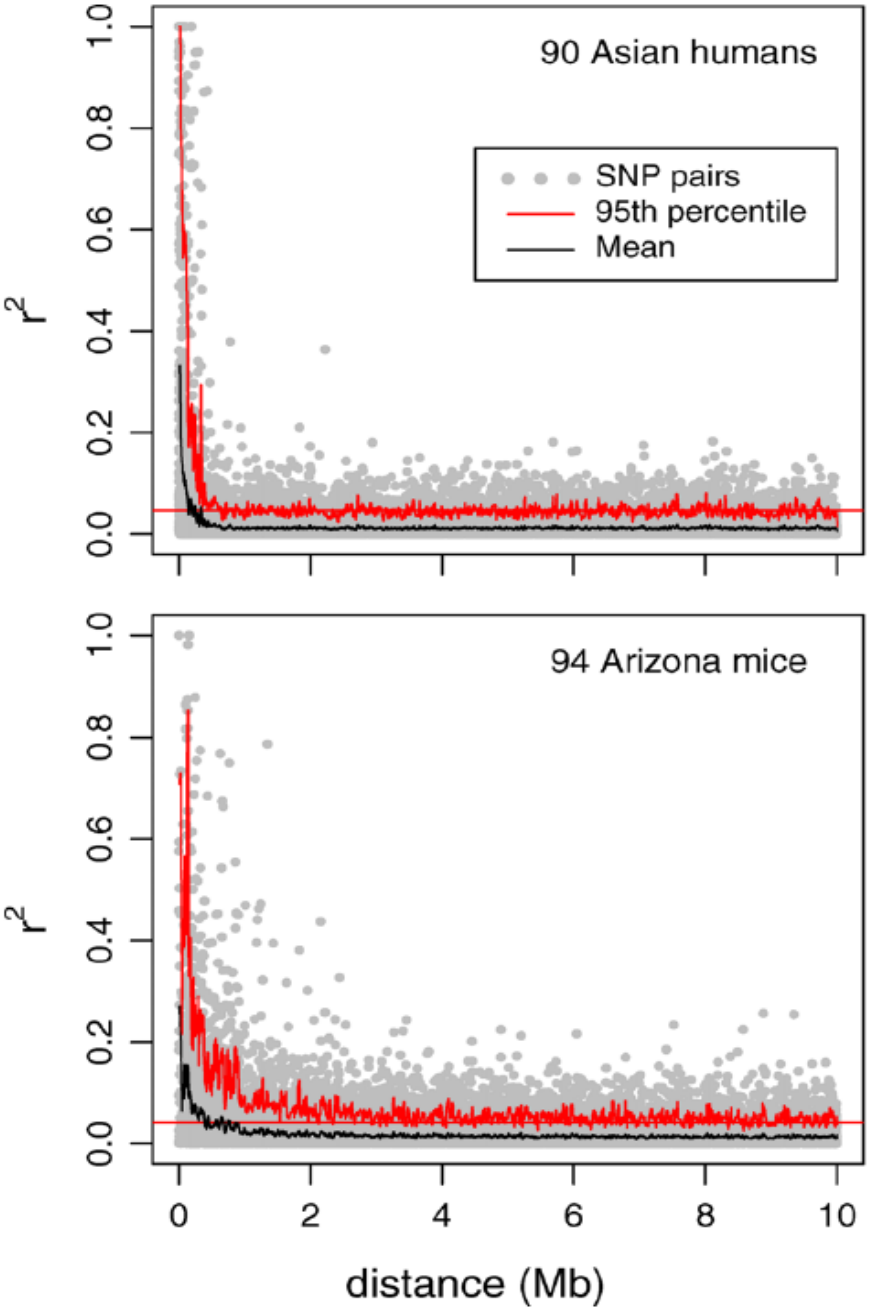
$$D' = \frac{D}{\min(f_A f_b, f_a f_B)} \text{ if } D > 0$$
$$= \frac{-D}{\min(f_A f_B, f_a f_b)} \text{ if } D < 0$$

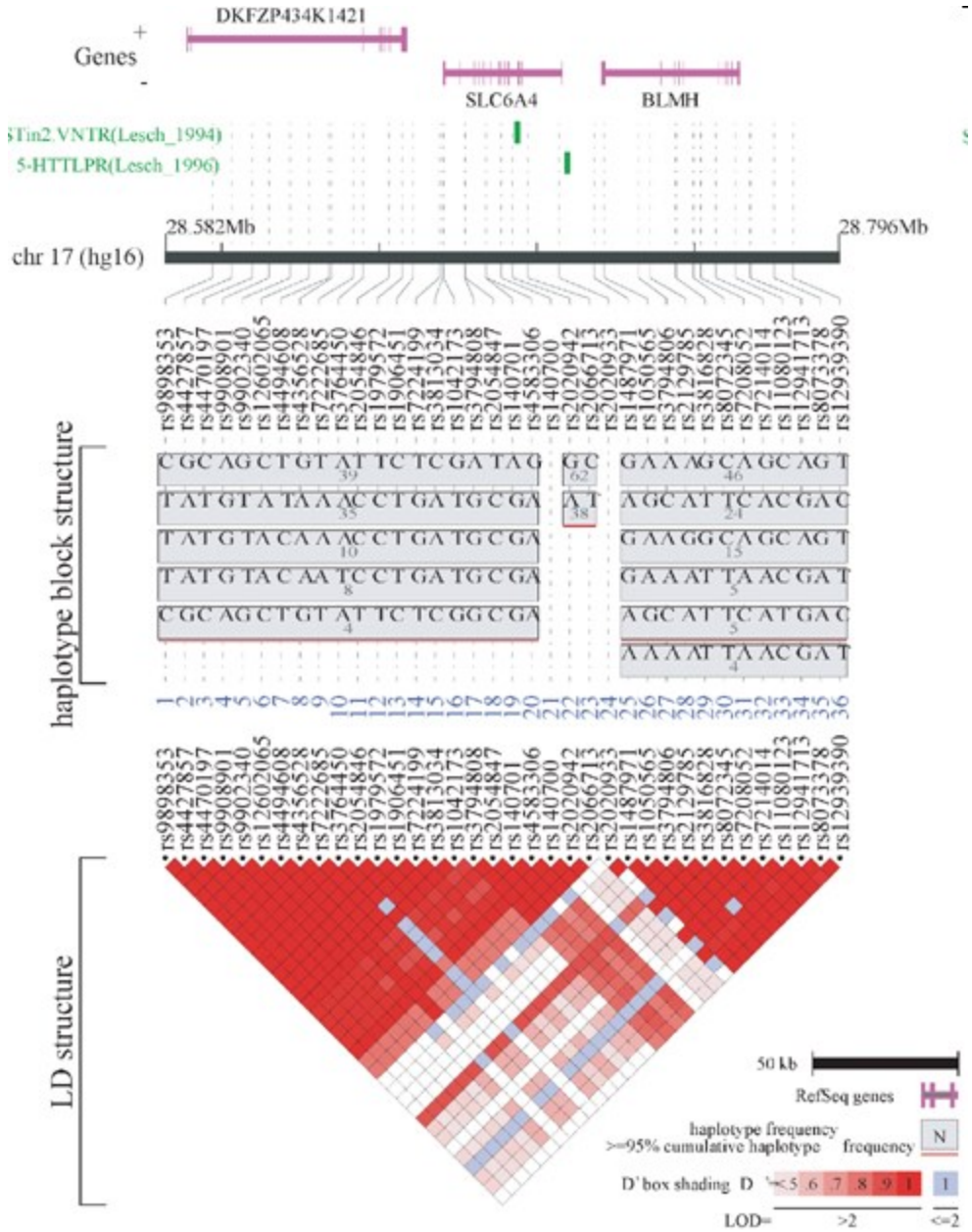
$$r^2 = \frac{D^2}{f_A f_a f_B f_b}$$

LD klesá se vzdáleností



*Laurie et al. 2007*



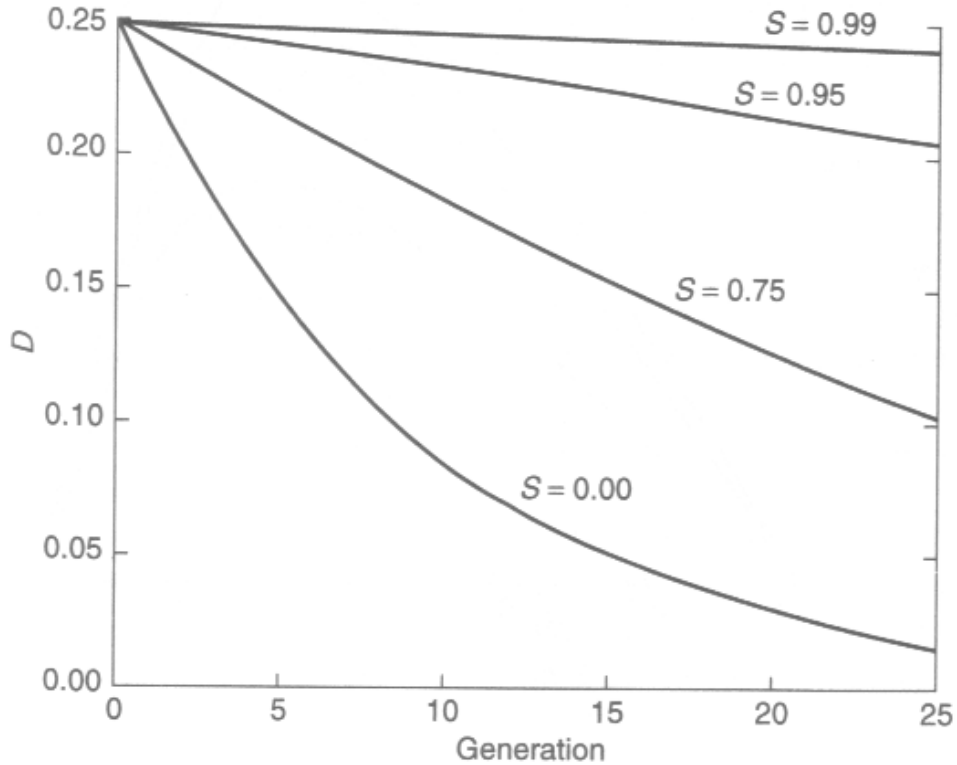

DKFZP434K1421
Genes
SLC6A4
BLMH
5Tin2.VNTR(Lesch_1994)
5-HTTLPR(Lesch_1996)
28.582Mb
28.796Mb
chr 17 (hg16)
haplotype block structure
LD structure
50 kb
RefSeq genes
haplotype frequency
>=95% cumulative haplotype frequency
N
D' box shading D'
<.5 .6 .7 .8 .9 1
1
LOD=
>2
<=2

# Sources of linkage disequilibrium

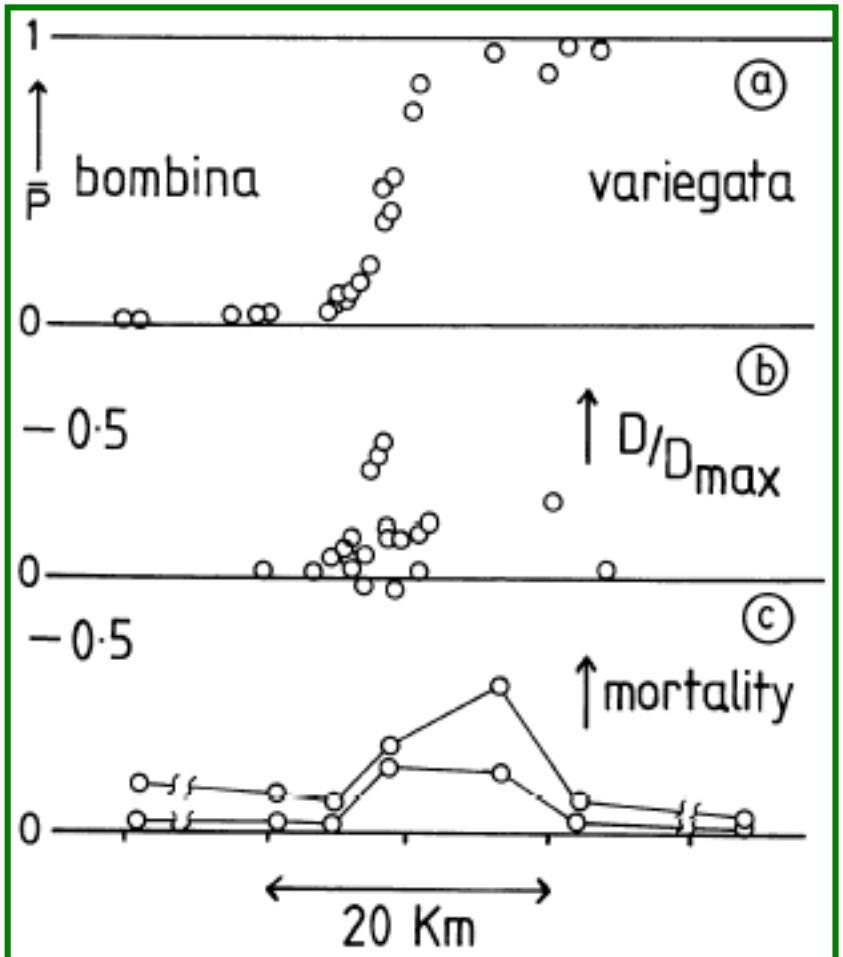
- Mutation (nové alely)
- Finite population size (stochatický proces, drift)
- Nonrandom mating (inbreeding)
- Gene flow (admixture, hybridization)
- Selection - Multilocus selection (epistasis, supergenes, coadapted gene complex)

- Nonrandom mating (inbreeding)

- Selfing



# Gene flow (hybridization)

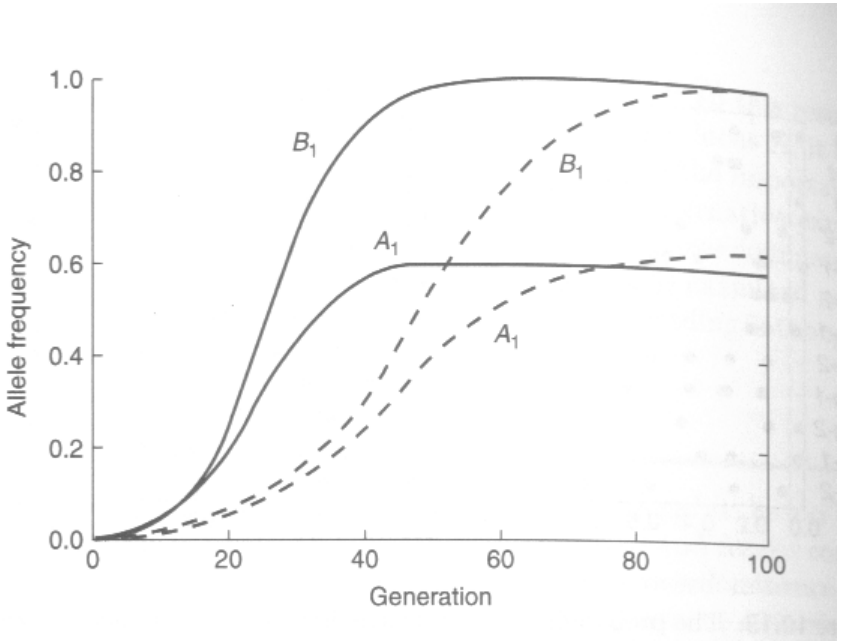


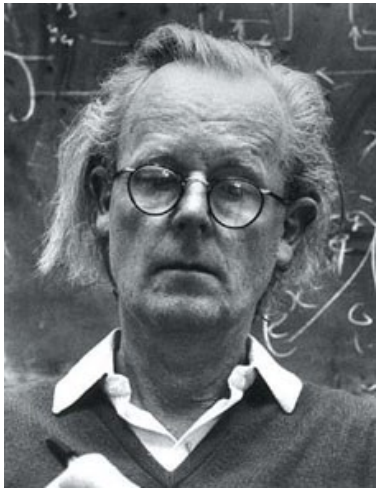
# linkage disequilibrium

- **Lokus ovlivněn selekcí na jiné lokusy**
- background selection
- genetic hitchhiking
- selective sweep

# genetic hitchhiking

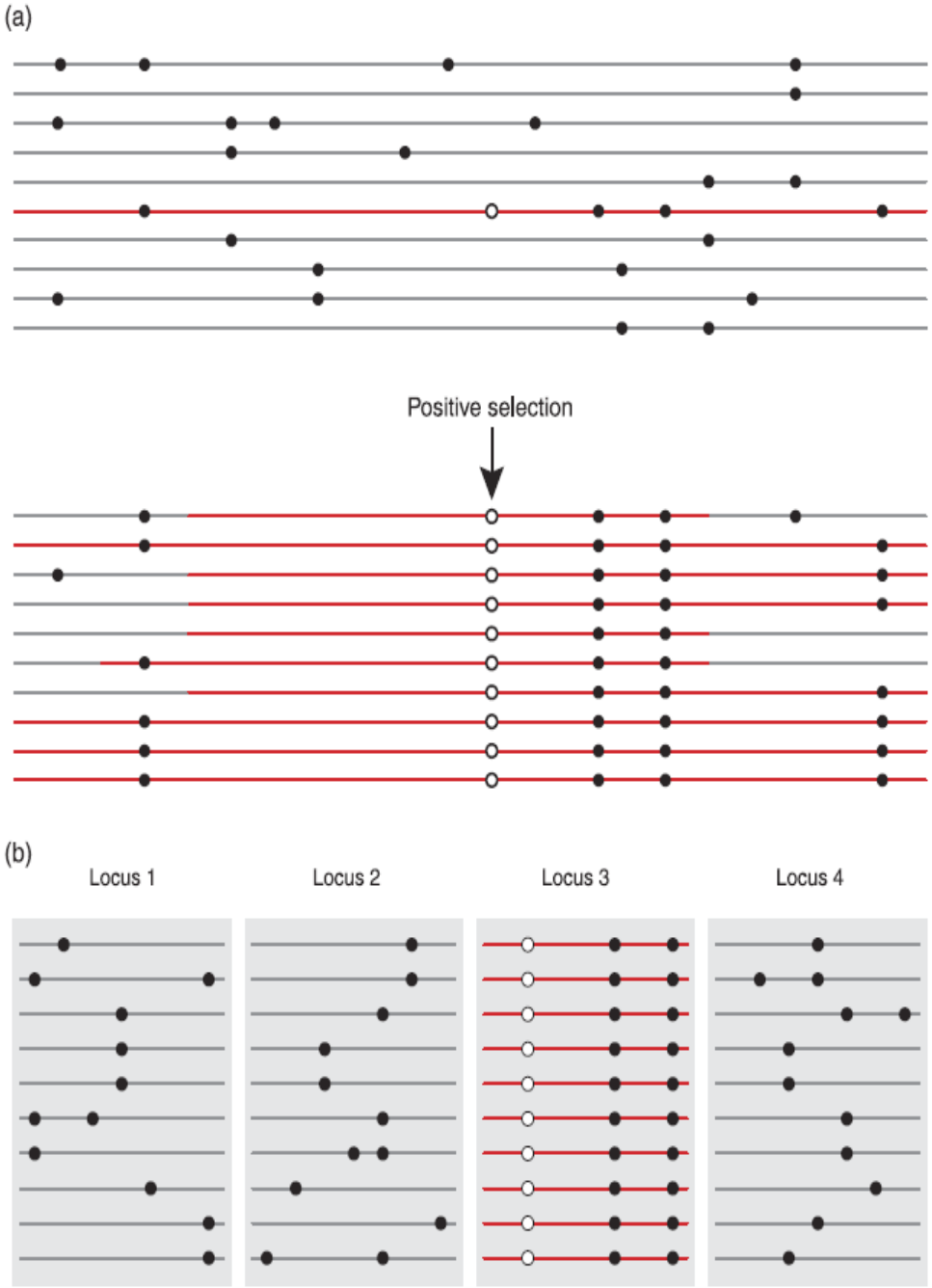
- Změna (vzrůst) frekvence neutrální alely díky asociaci s alelou jiného lokusu pod selekcí



*J. M. Smith*

# Selective sweep, hitchhiking



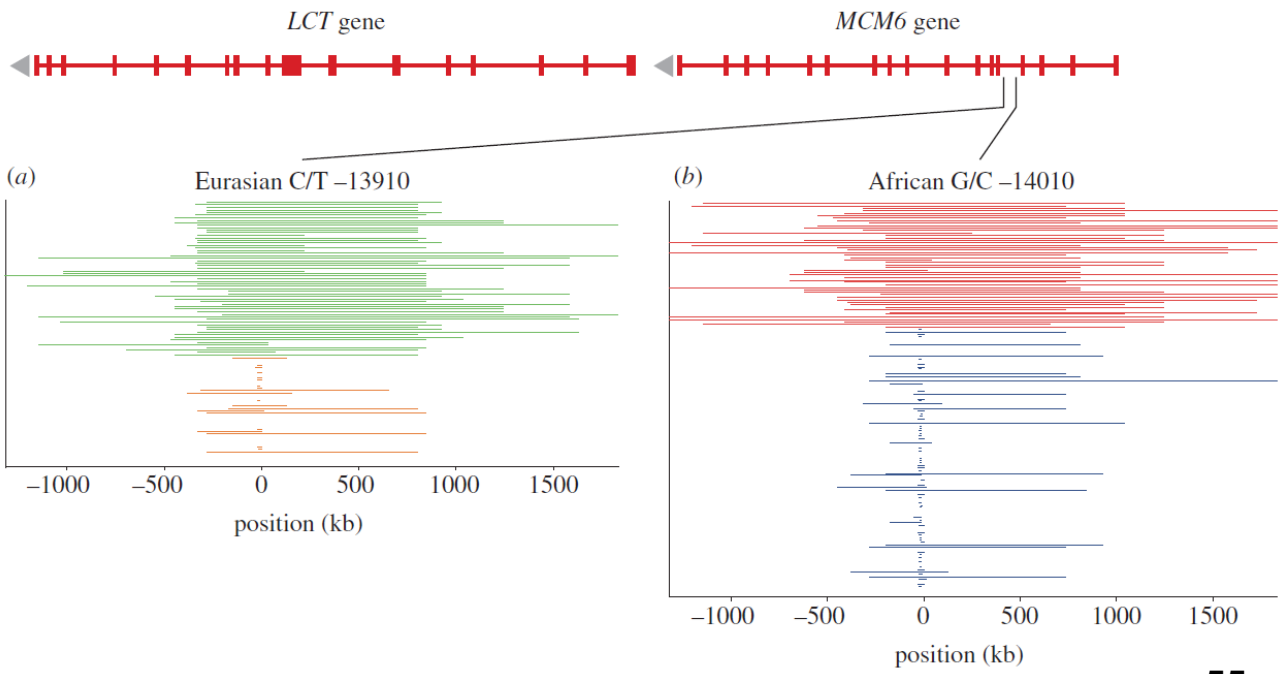
# *Drosophila*, Gonzáles et al. 2008

**FBti0019065**

| SS number | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 | 11 | 12 | 13 | 14 | 15 | 16 | 17 | 18 | 19 | 20 | 21 | 22 | 23 | 24 | 25 | 26 | 27 | 28 | 29 | 30 | 31 | 32 | 33 | 34 | 35 | 36 | 37 | 38 | 39 | 40 | 41 | 42 | 43 | 44 | 45 | 46 | 47 | 48 | 49 | 50 | 51 | 52 | 53 | 54 | 55 | 56 | 57 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Gene | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Length | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Position | -1323 | -1248 | -1227 | -1108 | -919 | -724 | -672 | -515 | -513 | -373 | -357 | -311 | -275 | -226 | -225 | -223 | -217 | -208 | -185 | -155 | -132 | -108 | -65 | -63 | -18 | 3 | 21 | 28 | 31 | 36 | 38 | 178 | 230 | 250 | 451 | 459 | 501 | 502 | 504 | 512 | 569 | 577 | 609 | 734 | 756 | 818 | 922 | 929 | 964 | 1376 | 1420 | 1427 | 1448 | 1464 | 1562 | 1604 | 1680 |
| Wi3 | C | G | C | G | C | A | G | A | A | G | C | C | G | G | G | G | A | G | T | A | C | G | T | A | T | T | A | T | G | G | A | T | G | G | C | A | G | A | | | | | | | | | | | | | | C | C | G | C | A | A |
| Wi41.5 | . | . | . | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| Wi77 | . | . | . | C | . | . | . | . | . | . | . | . | . | T | . | . | G | . | . | . | . | . | . | . | . | . | T | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| Wi98 | . | . | . | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| We4 | . | . | A | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| We10 | . | . | . | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| We25 | . | . | . | C | T | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| We44 | . | . | . | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| We47 | T | . | A | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | T | . | . | . | G | . |
| We67 | . | . | . | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| We80 | . | . | . | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | | | | | | | | | | | | | | . | . | A | . | G | C |
| AE003499 | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | A | . | . | . | . | . | . | . | . | . | . | . | A | . | G | C |
| Wi1 | . | . | . | C | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | G | T | C | G | T | . | A | . | . | C | A | T | . | A | A | - | . | A | G | A | A | G | . | . | A | . | . | . | . | . | . | . | . | A | . | G | C |
| Wi15 | . | C | . | C | . | C | T | T | . | . | . | A | . | . | . | T | . | . | . | . | T | C | . | T | C | A | . | . | . | . | . | . | . | . | . | . | A | G | A | . | G | - | . | . | . | A | . | . | C | . | . | . | T | A | . | G | . |
| Wi18 | . | . | . | C | . | . | . | . | T | A | . | . | C | . | A | . | C | . | C | . | T | C | G | T | . | A | . | . | . | . | . | . | . | . | A | T | A | G | A | . | G | . | T | . | . | . | . | . | . | . | . | . | . | A | . | G | C |
| Wi45 | . | . | . | C | . | . | . | . | . | . | . | . | . | . | . | . | . | A | C | G | T | C | . | T | C | A | . | C | . | A | T | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | A | . | G | C |
| Wi68 | . | . | . | C | . | . | . | T | . | . | . | . | C | . | A | . | . | A | C | G | T | C | . | T | C | A | . | C | . | A | T | . | . | . | . | . | . | . | . | . | G | . | . | . | . | . | . | . | . | . | . | . | . | A | . | G | C |
| Wi83 | . | . | . | C | . | . | . | T | . | . | . | A | . | . | . | T | . | . | C | G | T | C | . | T | C | A | . | C | . | A | T | G | . | . | . | . | . | . | . | . | G | . | . | . | . | . | . | . | . | . | . | . | . | A | . | G | . |
| We7 | . | . | . | C | . | . | . | T | . | . | T | . | C | - | - | . | . | . | C | G | . | C | . | T | . | A | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | T | . | . | . | . | . | . | . | . | . | . | A | A | G | C |
| We28 | . | . | . | C | . | . | . | T | . | . | T | . | C | - | - | . | . | . | C | G | . | C | . | T | . | A | . | . | . | . | . | . | . | . | . | A | T | . | . | . | . | . | . | . | . | . | A | . | . | . | A | . | . | . | A | . | G | C |
| We33 | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | A | T | . | . | . | . | . | . | . | . | . | . | . | . | . | . | A | . | . | . | . | . | . | . | . | A | . | G | C |
| We63 | . | . | . | C | . | . | . | T | . | . | T | . | C | - | - | . | . | . | C | G | . | C | . | T | . | A | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | T | . | . | . | . | . | . | . | . | . | . | A | A | G | C |
| We70 | . | C | . | C | . | C | T | T | . | . | . | A | . | . | . | T | . | . | . | . | T | C | . | T | C | A | . | . | . | . | . | . | . | . | . | . | A | G | A | . | G | - | . | . | . | A | . | . | C | . | . | . | T | A | . | G | . |
| We91 | . | C | . | C | . | C | T | T | . | . | . | A | . | . | . | T | . | . | . | . | T | C | . | T | C | A | . | . | . | . | . | . | . | . | . | . | A | G | A | . | G | - | . | . | . | A | . | . | C | . | . | . | T | A | . | G | . |

# Člověk a „selective sweeps“

- genes in pigmentation pathways
- components of the dystrophin protein complex
- clusters of olfactory receptors
- genes involved in nervous system development and function
- immune system genes
- heat shock genes
- centromeric regions

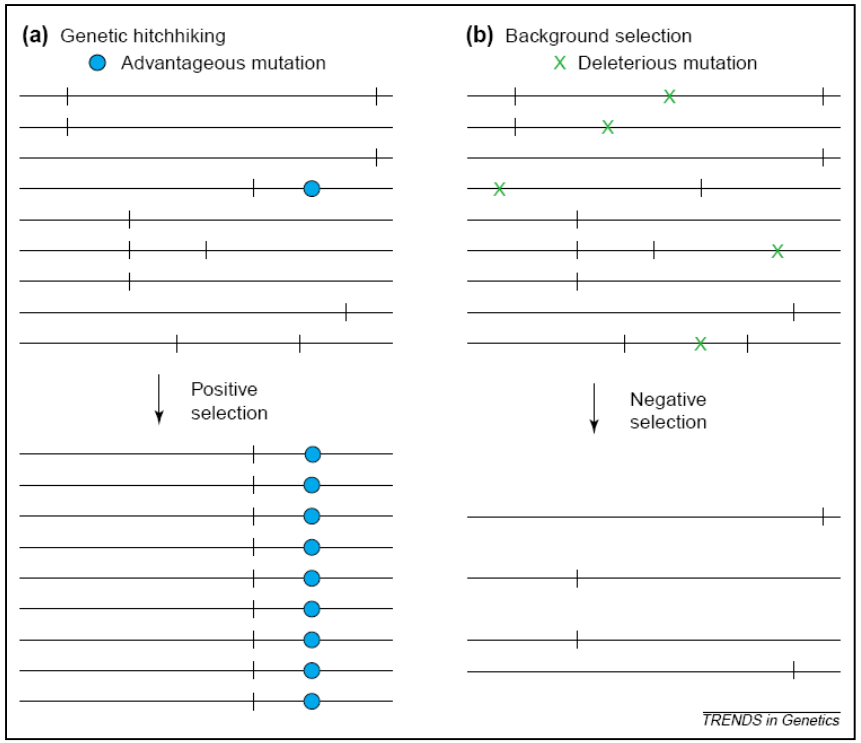


# Background selection

- Asociace s alelami jiných lokusů, na které působí selekce
- Odstranění škodlivých mutací
- → zdánlivé rozdíly fitness genotypů neutrálního lokusu

# Background selection

- Odstraňování škodlivých mutací
- Zánik linií (i těch v blízkosti, co nesou neutrální nebo slabě výhodné mutace)
- Snížení $N_E \rightarrow$ rychlejší fixace



# Pozor existuje i

- **Sexual selection**
  - Assortative mating
  - Male competition
  - Female choice
  - Sexually antagonistic genes
  - …
- **Gametic selection**
  - Drive
  - Self-incompatibility alleles

# Závěr

- Evoluce = (někdy) změna frekvence alel (a genotypů)

- Ideální populace
  - HW (jedna generace)
  - Linkage equilibrium (delší doba)

- Reálná populace
  - Hierarchická struktura
  - Drift
  - Mutace
  - Migrace
  - Odchylky od „random mating“
    - inbreeding, assortative mating
  - Selekce
    - ovlivní i další lokusy