WHY

Evolutionary factors matter?

Evolution

In the last class we have covered:

The Hardy-Weinberg principle

- Main concepts
- How to evaluate the HWE in a sample
- What are the main factors that affect HWE

Evolution

In this class we will cover:

The effect of the main evolutionary factors in a simple population model

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Alvo de estudo • Deriva • Selecção • Mutação • Migração • Outros

A composição genética das populações e os factores que determinam essa composição e as alterações ao longo do tempo

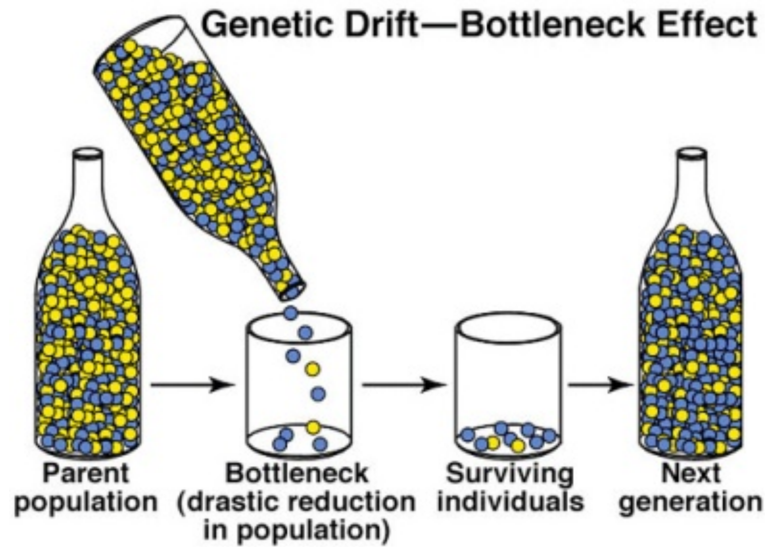
Modelos

Conjunto de hipóteses que especificam as relações matemáticas entre parâmetros num dado sistema:

- Expressão concisa entre parâmetros;
- Selecção dos parâmetros mais importantes;
- Orientação sobre colheita, organização e interpretação de dados;
- Previsão sobre o comportamento do sistema.

Deriva genética
Mutação
Migração (fluxo genético)
Selecção

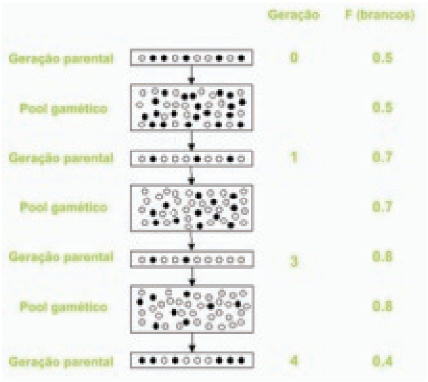
In genetic drift it is a case of being in the wrong place at the wrong time.

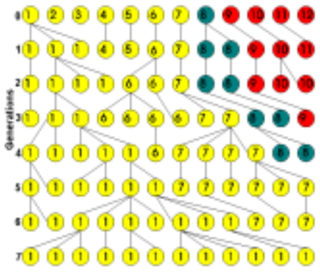


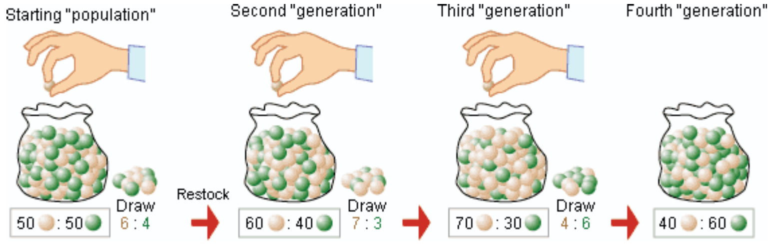
- **Na ausência de selecção, migração e mutação as frequências alélicas mantêm-se na próxima geração (HW).**
- **No entanto, sucessivas gerações são o resultado de uma amostragem ao acaso do pool genético parental.**

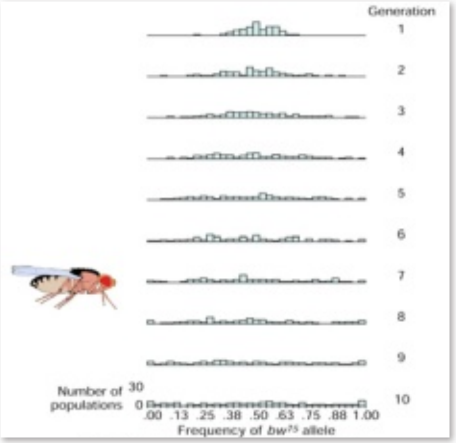
- **Assim, por erro de amostragem as frequências alélicas podem vir a ser diferentes das da geração anterior:**
  - **esperadas F_2: 9-3-3-1**
  - **observadas F_2:9-3-2.8-1.2**

- **Exemplo:**
  - **Cara ou coroa dez vezes em conjuntos de 20 moedas ou 4000 moedas.**
  - **A probabilidade de se obterem desvios à probabilidade esperada (50% cara, 50% coroa) *é maior* no conjunto de menos moedas.**

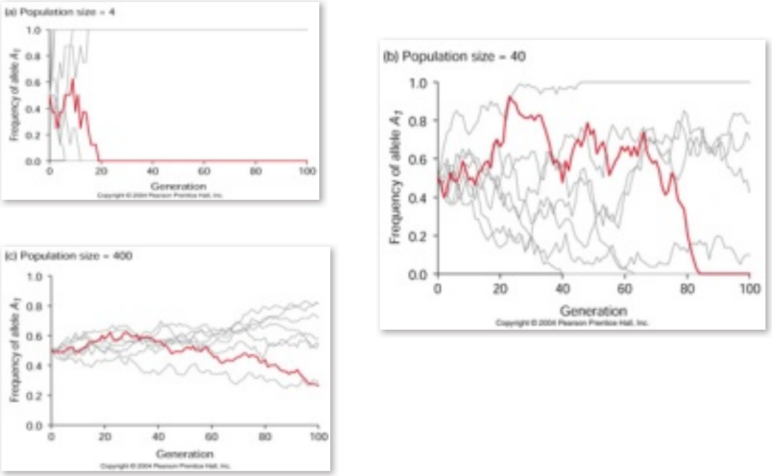


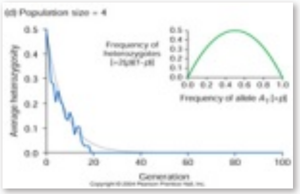


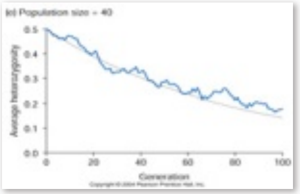


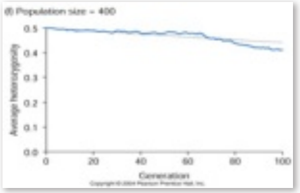


Initially identical subpopulations evolve by chance to become genetically different

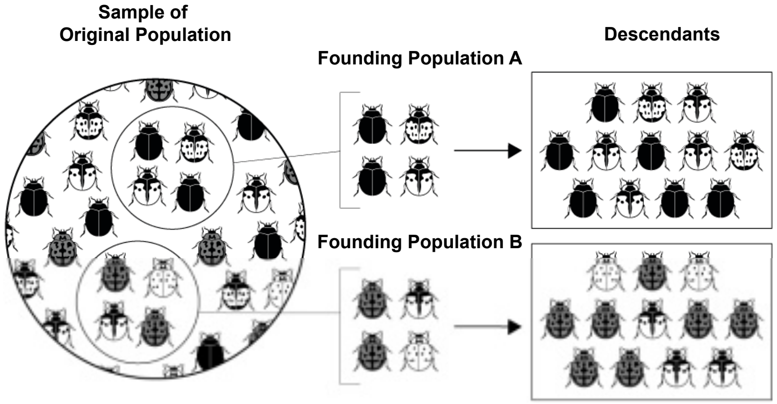






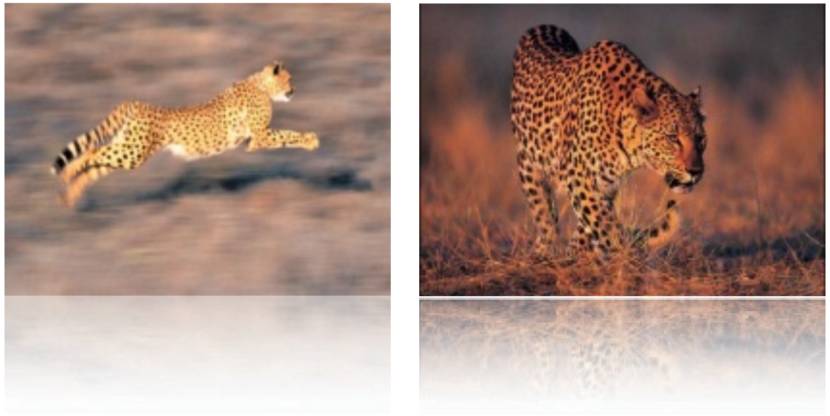


Drift leads to an inevitable decline in heterozygosity in finite populations over time. This happens faster in small populations.



- large US California and Baja California populations were hunted for oil and skins
- by end 1800's, ~ 20 individuals remained on Guadalupe Island off Baja California
- population rebounded to ~120,000 by 1980

- Bonnell and Selander (1974): ***no variation*** at 24 allozyme loci
- Hoelzel et al. (1993): extended survey to > 50 loci, still no variation
- severe genetic uniformity, despite "rapid" rebound to large population



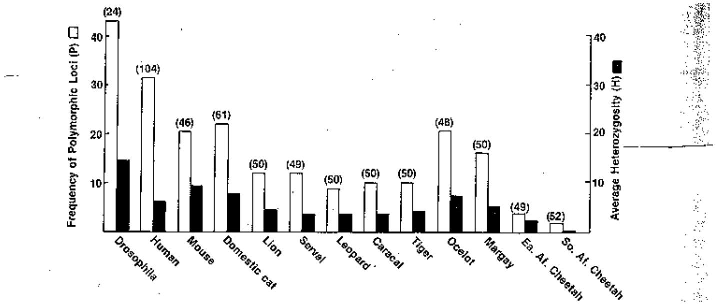


FIG. 1. Estimates of the extent of genetic variation based on allozyme electrophoretic surveys in the east African cheetah (*A. jubatus raineyi*), the south African cheetah (*A. jubatus jubatus*), eight additional feline species (8, 15), and three nonfelid species. The number in parentheses indicates the number of genetic loci that were considered in the estimate. For a review of over 250 such electrophoretic surveys see ref. 24. Enzymes typed and procedures used are as previously described (7, 8, 15).

in Fig. 1, the east African sample had a frequency of polymorphic loci (*P*) of 2/49 (4%) and an average heterozygosity

raineyi and *jubatus* subspecies, perhaps within the last 100 years, a second bottleneck or a chance-driven founder effect

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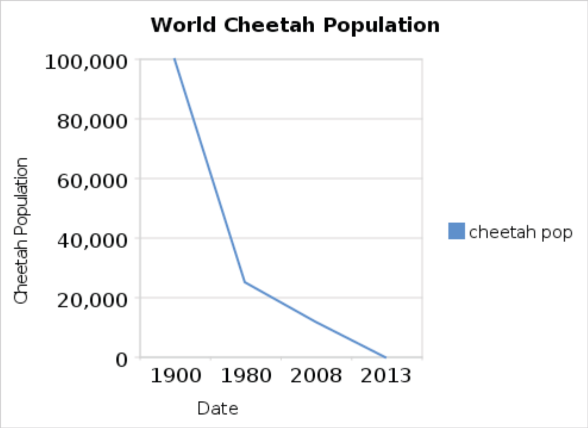
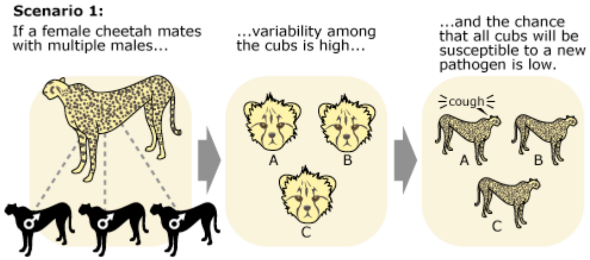
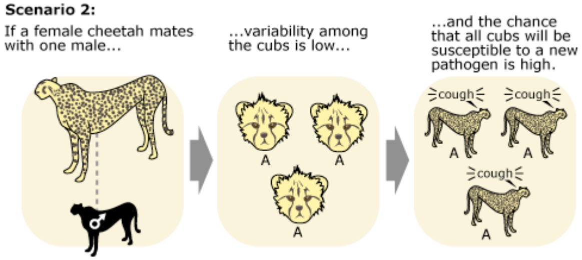




Scenario 1:
If a female cheetah mates with multiple males…

…variability among the cubs is high…

…and the chance that all cubs will be susceptible to a new pathogen is low.



Scenario 2:
If a female cheetah mates with one male…

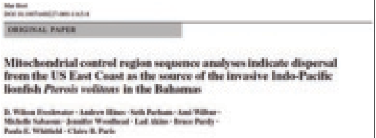
…variability among the cubs is low…

…and the chance that all cubs will be susceptible to a new pathogen is high.

Mirounga leonina

pinnepeds show low allozyme variation, but:

- southern elephant seals: not hunted, no bottleneck, much greater allozyme variation
- northern elephant seals: unusually low variation in highly variable DNA regions (microsatellite loci and mitochondrial DNA control region)
  - **2 different control region sequences in 40 northern elephant seals**
  - **southern species ~ 10X more variation**



ORIGINAL PAPER

Mitochondrial control region sequence analyses indicate dispersal from the US East Coast as the source of the invasive Indo-Pacific lionfish *Pterois volitans* in the Bahamas

D. Wilson Freshwater · Andrew Hines · Seth Parham · Ami Wilbur · Michelle Sabaoun · Jennifer Woodhead · Lad Akins · Bruce Purdy · Paula E. Whitfield · Claire B. Paris

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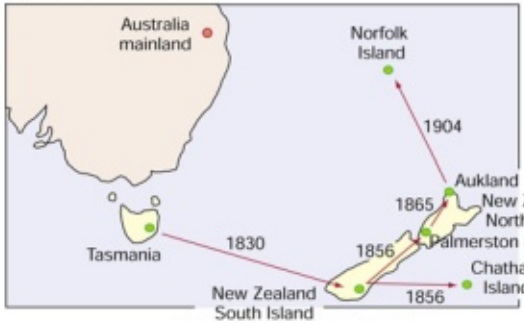
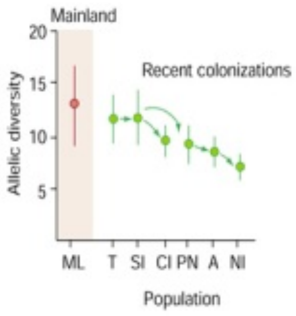
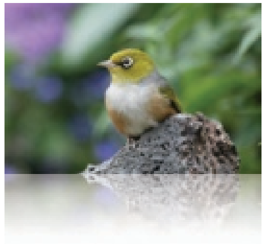
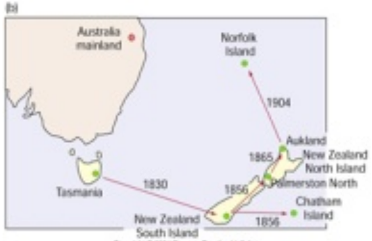


The history of colonization of Tasmania and New Zealand by the silvereye, *Zosterops lateralis*





ML = Australian mainland
T = Tasmania
SI = South Island New Zealand
CI = Chatham Island
PN = Palmerston North, south end of North Island New Zealand
A = Auckland, north end of North Island of New Zealand
NI = Norfolk Island



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- Tamanho da população
- Frequências alélicas iniciais
- Tempo disponível

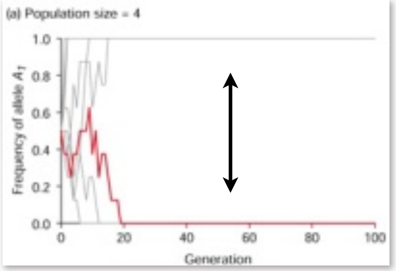
Simulações

A deriva genética:

- não tem direcção;
- provoca perda de variabilidade genética dentro de uma população;
- provoca aumento de variabilidade genética entre populações.

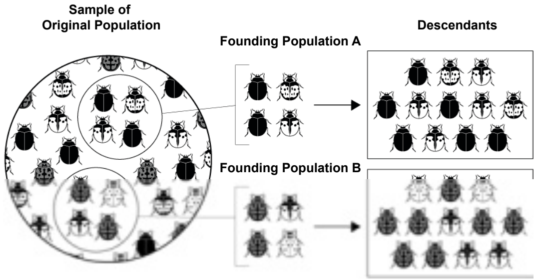
A deriva genética:

- não tem direcção



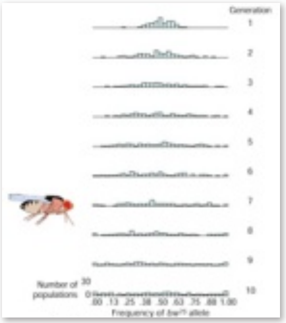
A deriva genética:

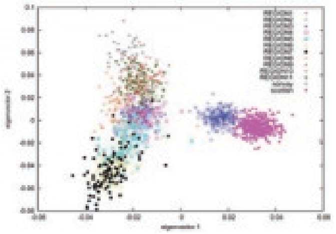
provoca perda de variabilidade genética dentro de uma população



A deriva genética:

provoca aumento de variabilidade genética entre populações

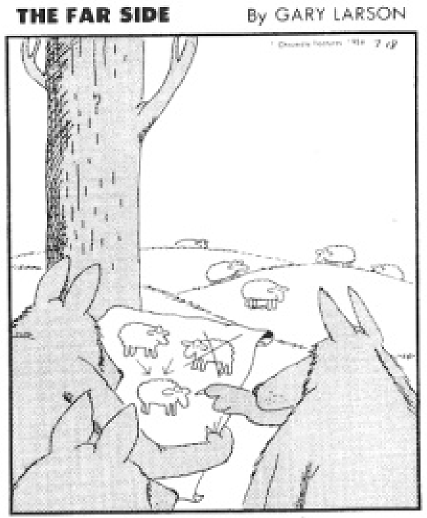




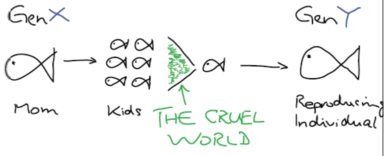
In genetic drift it is a case of being in the wrong place at the wrong time.

Natural Selection

Ahh, the first day teaching Natural Selection is always the best day.



Natural selection at work



A selecção natural baseia-se:

- Mais descendentes do que sobreviventes e reprodutores;
- Diferentes organismos ⇒ diferentes capacidades de sobrevivência e reprodução (reprodução diferencial);
- Parte da variação da capacidade de sobrevivência e reprodução é hereditária.

Certos genótipos estão *mais adaptados* a um dado ambiente que outros.

Dizem-se com maior ”fitness”.

= maior capacidade de sobrevivência e reprodução num dado ambiente.

O *número de descendentes* de cada genótipo é a expressão do seu valor adaptativo.

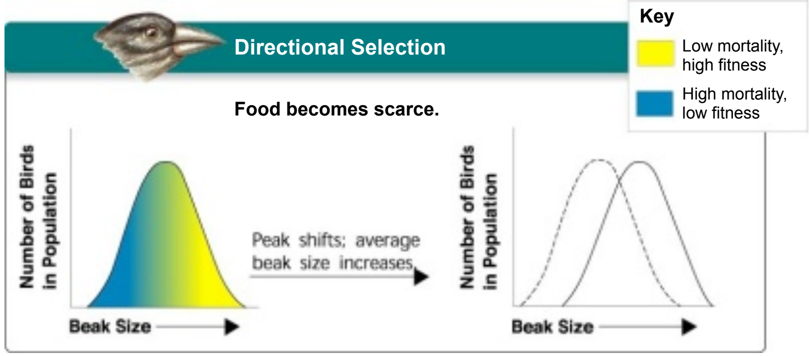
’Fitness' pode ser definido como:

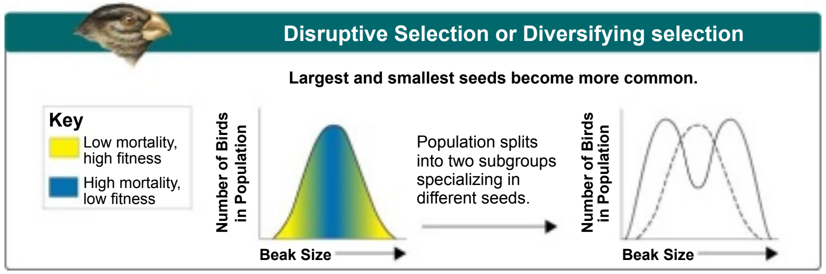
> **“a capacidade de os diferentes genótipos passarem alelos às gerações futuras”**

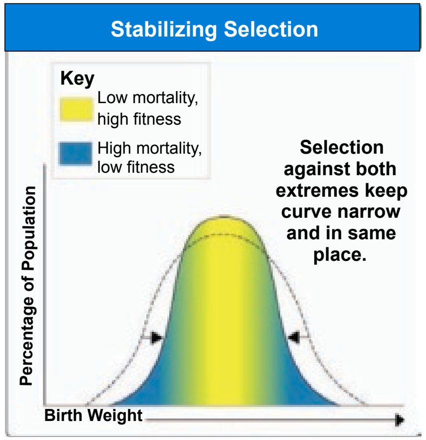
A selecção é a medida da diferença entre

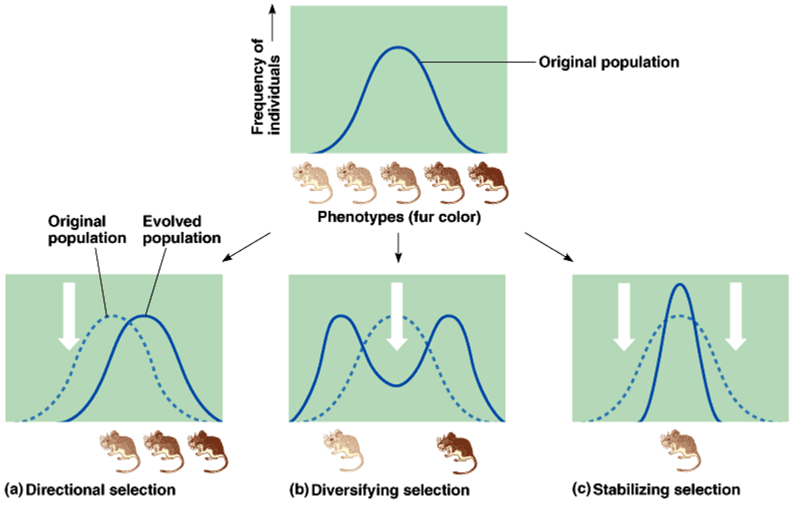
a frequência relativa dos descendentes prevista pela lei de Hardy-Weinberg e a frequência observada.

Tipos de selecção





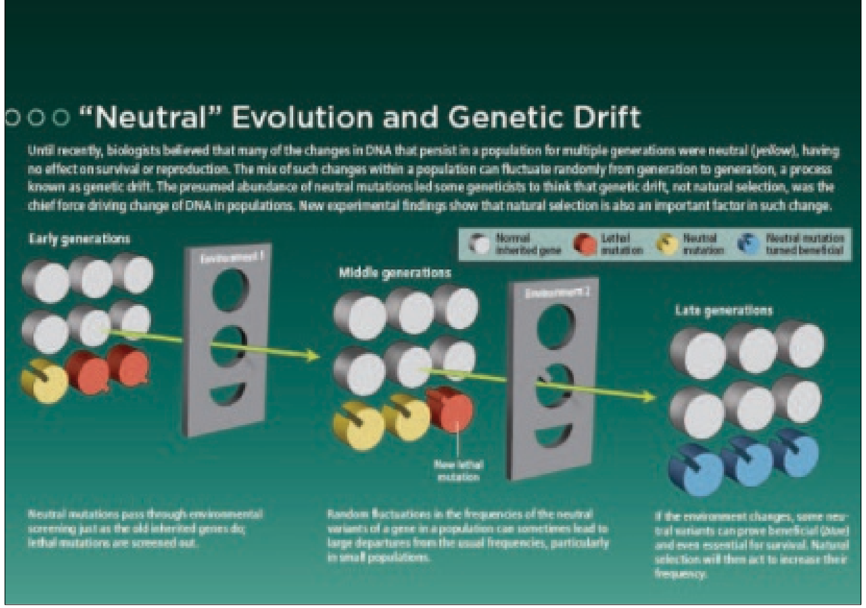




Efeitos básicos na variação genética:

quando favorece um alelo **em particular pode conduzir à redução da variação genética e à homozigotia do alelo favorecido;**

quando não favorece um alelo **em particular, pode manter a presença de 2 ou mais alelos na população.**



○○○ "Neutral" Evolution and Genetic Drift

Until recently, biologists believed that many of the changes in DNA that persist in a population for multiple generations were neutral (yellow), having no effect on survival or reproduction. The mix of such changes within a population can fluctuate randomly from generation to generation, a process known as genetic drift. The presumed abundance of neutral mutations led some geneticists to think that genetic drift, not natural selection, was the chief force driving change of DNA in populations. New experimental findings show that natural selection is also an important factor in such change.

Neutral mutations pass through environmental screening just as the old inherited genes do; lethal mutations are screened out.

Random fluctuations in the frequencies of the neutral variants of a gene in a population can sometimes lead to large departures from the usual frequencies, particularly in small populations.

If the environment changes, some neutral variants can prove beneficial (blue) and even essential for survival. Natural selection will then act to increase their frequency.

Individuals may differ in fitness because of their underlying genotype

Genotype	A_1A_1	A_1A_2	A_2A_2
Frequency	p^2	$2pq$	q^2
Fitness	w_{11}	w_{12}	w_{22}

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Genotype	A_1A_1	A_1A_2	A_2A_2
Frequency	p^2	$2pq$	q^2
Fitness	w_{11}	w_{12}	w_{22}

Next generation:

	Freq	$p^2 w_{11}$	$2pq\ w_{12}$	$q^2 w_{22}$
$w_{11} = 1.0$ $w_{12} = 1.0$ $w_{22} = 1.0$	**Freq**	p^2	$2pq$	q^2

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$w_{11} = 1.0$ $w_{12} = 1.0$ $w_{22} = 1.0$	**Freq**	p^2	$2pq$	q^2
$w_{11} = 1.0$ $w_{12} = 1.0$ $w_{22} = 0.8$	**Freq**	p^2	$2pq$	$q^2*0.8$

$A_1A_1 + A_1A_2 + A_2A_2 = 1$

$w_{11} = 1.0$ $w_{12} = 1.0$ $w_{22} = 1.0$	**Freq**	p^2 A_1A_1	$2pq$ A_1A_2	q^2 A_2A_2
		0.5 +	0.4 +	0.1 = 1

$w_{11} = 1.0$ $w_{12} = 1.0$ $w_{22} = 0.8$	**Freq**	p^2	$2pq$	$q^2*0.8$
		A_1A_1 +	A_1A_2 +	A_2A_2 < 1
		0.5 +	0.4 +	0.08 = 0.98!

Average fitness of the whole population:

$$\overline{W} = p^2w_{11} + 2pqw_{12} + q^2w_{22}$$

Average fitness of the whole population:

$$\overline{W} = p^2 w_{11} + 2pq w_{12} + q^2 w_{22}$$

Given variable fitness, frequencies after selection:

Genotype	A_1A_1	A_1A_2	A_2A_2
Freq	$\frac{p^2 w_{11}}{\overline{w}}$	$\frac{2pq\, w_{12}}{\overline{w}}$	$\frac{q^2 w_{22}}{\overline{w}}$

Given variable fitness, frequencies after selection:

Genotype	A_1A_1	A_1A_2	A_2A_2
Freq	$\frac{p^2 w_{11}}{\overline{w}}$	$\frac{2pq\, w_{12}}{\overline{w}}$	$\frac{q^2 w_{22}}{\overline{w}}$

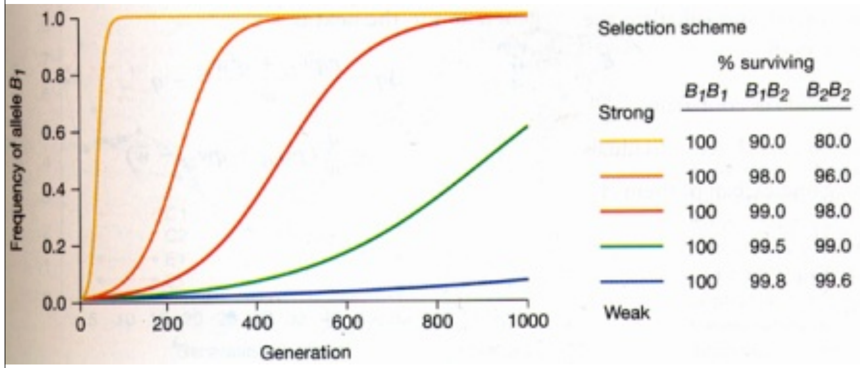
New allele frequencies after mating:

$$\frac{p^2 w_{11} + pq\, w_{12}}{\overline{W}} \qquad \frac{pq\, w_{12} + q^2 w_{22}}{\overline{W}}$$

New Frequency of A1

New Frequency of A2

Persistent Selection Changes Allele Frequencies



	% surviving		
	B_1B_1	B_1B_2	B_2B_2
🟨	100	90.0	80.0
🟧	100	98.0	96.0
🟥	100	99.0	98.0
🟩	100	99.5	99.0
🟦	100	99.8	99.6

Strength of selection is given by the magnitude of the selection differential

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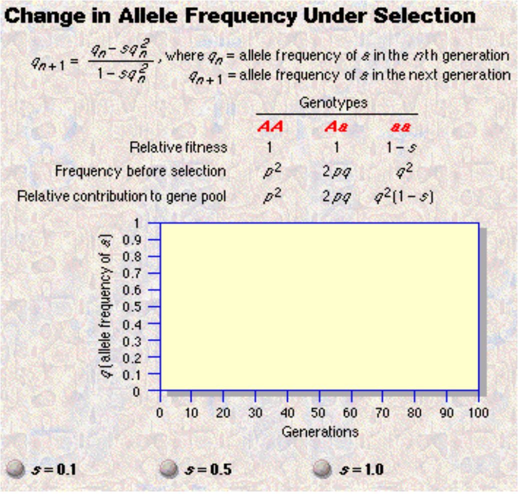
W=1-s

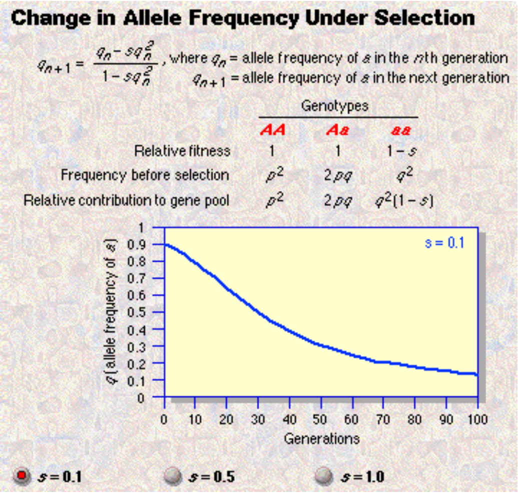


Change in Allele Frequency Under Selection

$q_{n+1} = \frac{q_n - s q_n^2}{1 - s q_n^2}$, where q_n = allele frequency of *a* in the *n*th generation
q_{n+1} = allele frequency of *a* in the next generation

	Genotypes		
	AA	*Aa*	*aa*
Relative fitness	1	1	$1 - s$
Frequency before selection	p^2	$2pq$	q^2
Relative contribution to gene pool	p^2	$2pq$	$q^2(1 - s)$

W=1-s



Change in Allele Frequency Under Selection

$q_{n+1} = \frac{q_n - sq_n^2}{1 - sq_n^2}$, where q_n = allele frequency of a in the nth generation

q_{n+1} = allele frequency of a in the next generation

	Genotypes		
	AA	Aa	aa
Relative fitness	1	1	$1 - s$
Frequency before selection	p^2	$2pq$	q^2
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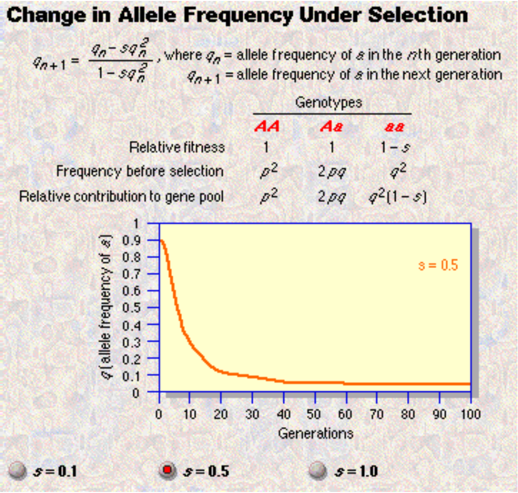
W=1-s



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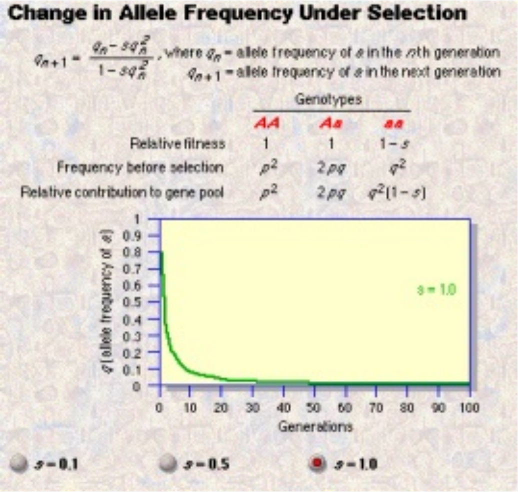

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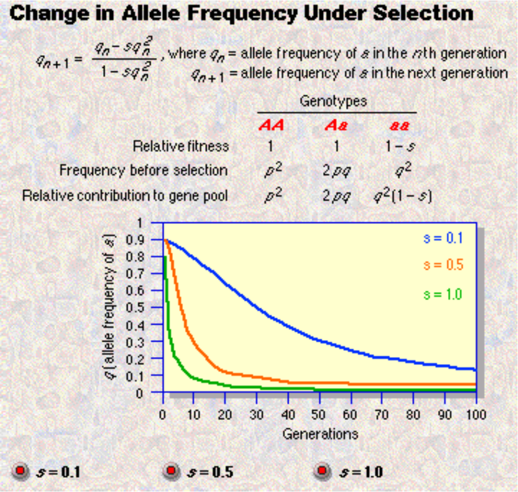

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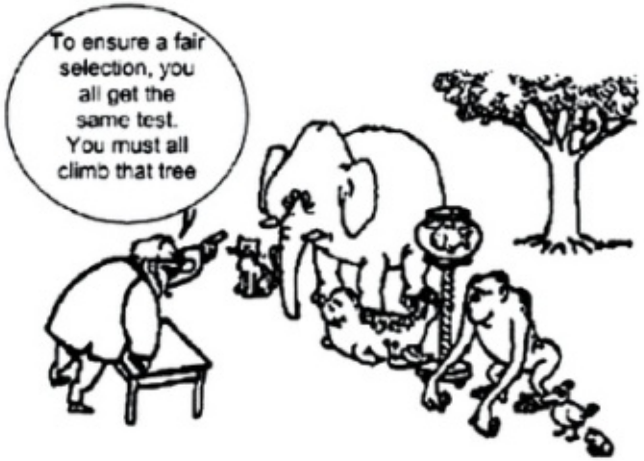
Relative contribution to gene pool	p^2	$2pq$	$q^2(1 - s)$


Mutation rate
Selection coefficient
Equilibrium allele frequency


To ensure a fair selection, you all get the same test. You must all climb that tree