Lecture 5: Population genetics I

The Hardy-Weinberg Principle & Selective forces

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What is Population Genetics?

Integrates:

- Darwin's Theory of Evolution by Natural Selection
- Mendelian laws of inheritance (ie, genetics)

Seeks to document and explain changes in:

- allele frequencies;
- genotype frequencies;
- phenotype frequencies.

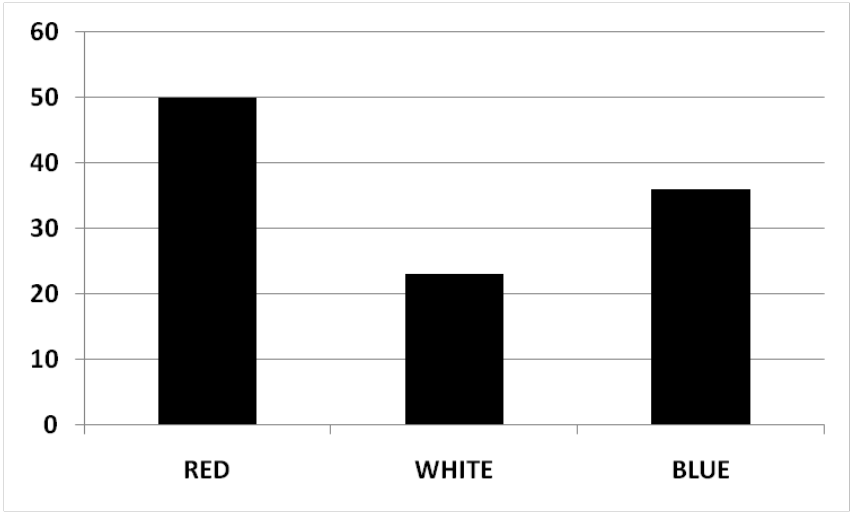
Offers quantifiable definition of evolution:

→ Evolution happens by allele frequency changes in the population

Types of characters

Discontinuous, discrete

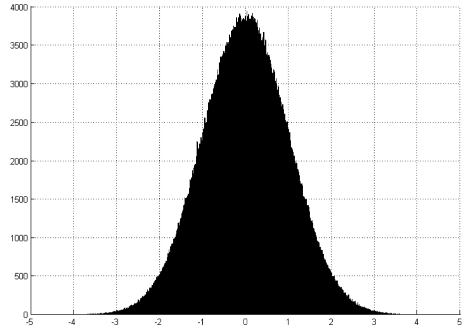
Count



Flower coloration

Types of characters

Continuous, quantitative

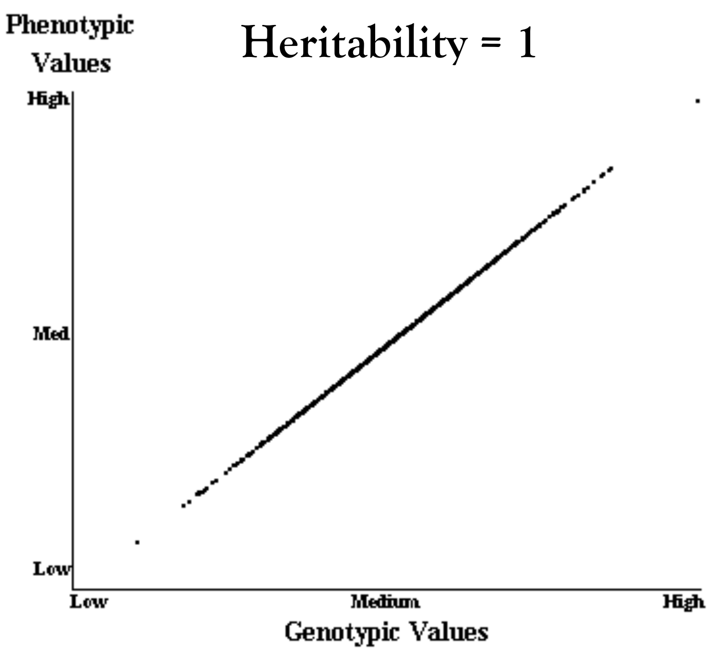


Yield

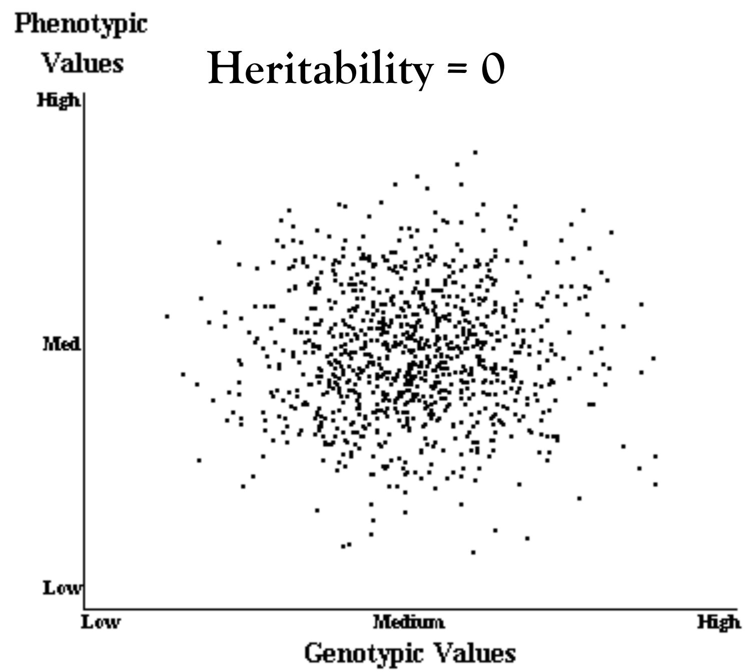
Heritability

Heritability = Variance (Genotype) / Variance (Phenotype) $h^2 = \frac{\sigma^2_G}{\sigma^2_P}$

Heritability = 1



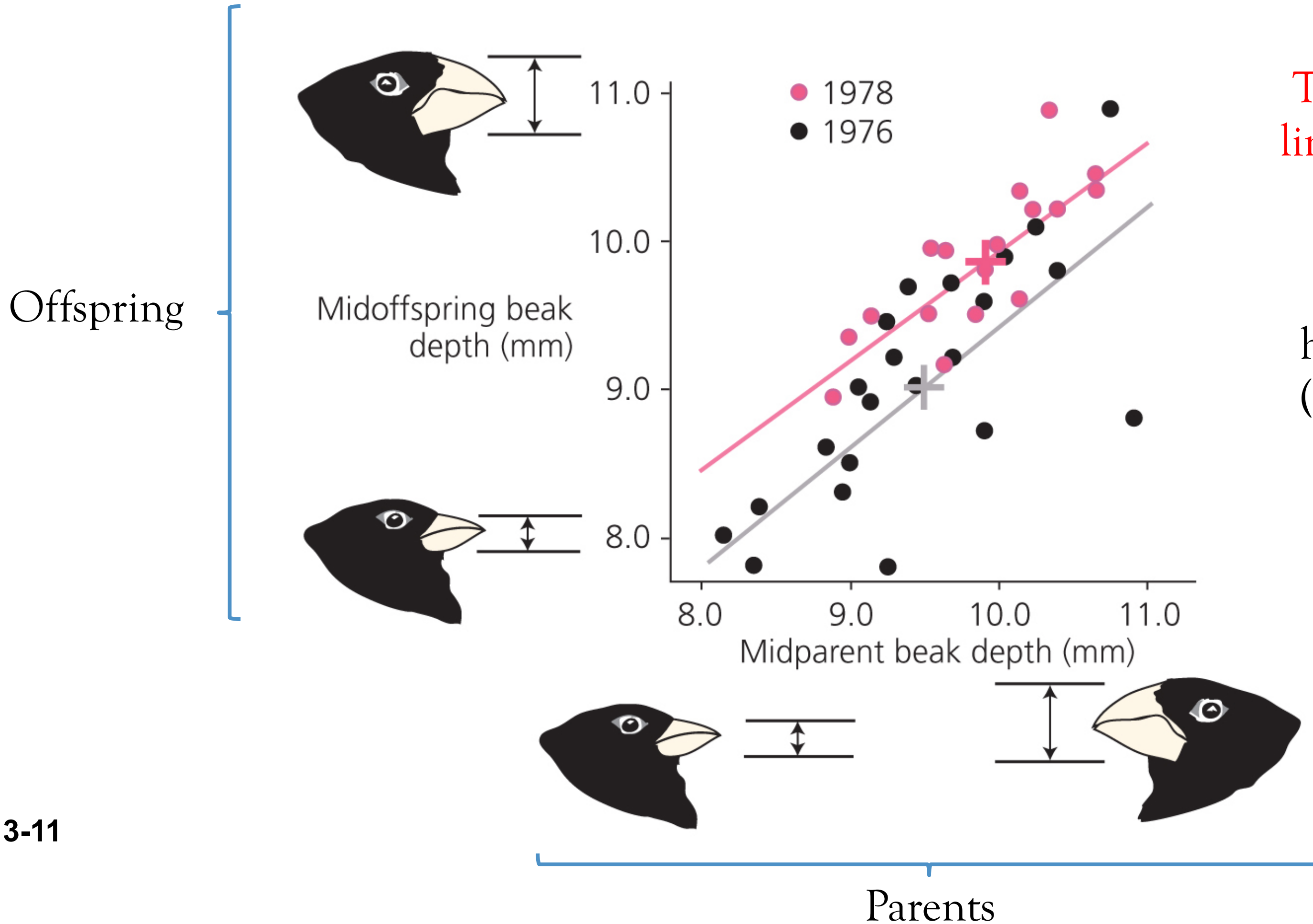
Heritability = 0



Influence of Environment → 0

Influence of Environment → 1

Remember from Class 2:
Parent-offspring regression

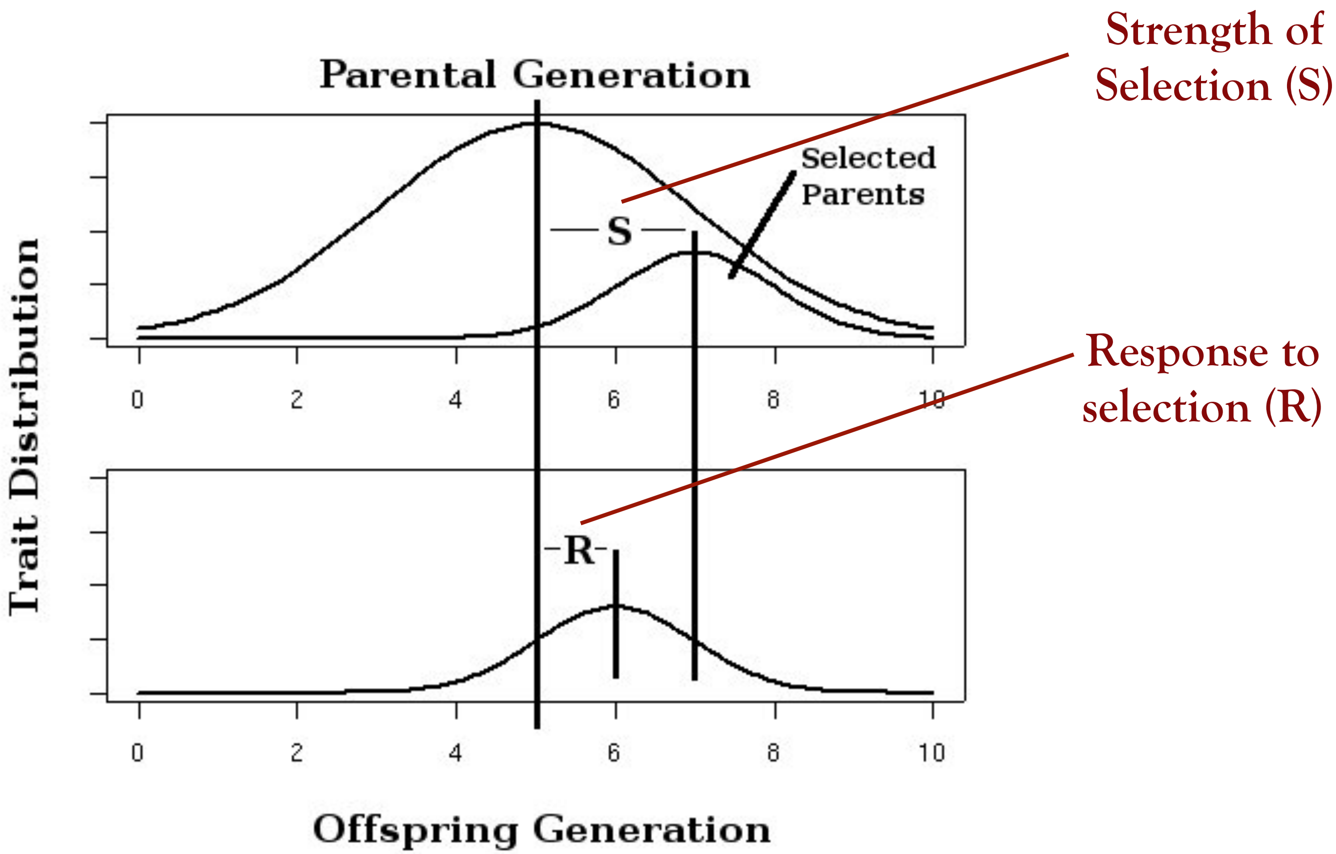


The slope of the line approximates the trait's heritability

h* = 65%
(Keller et al. 2001)

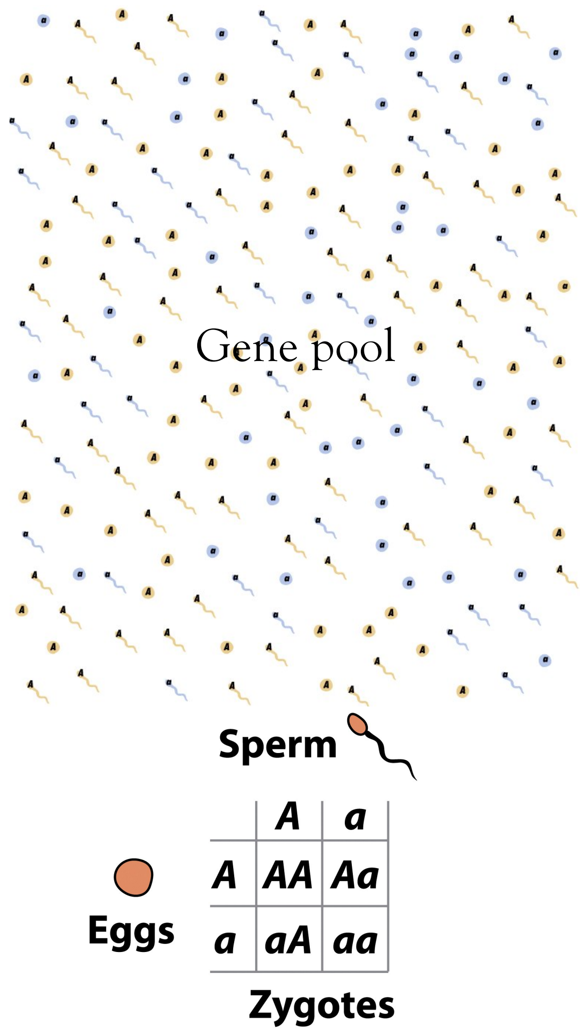
Figure 3-11

How else to determine Heritability (*H*)?
Selection experiments



H = R/S

The concept of the gene pool



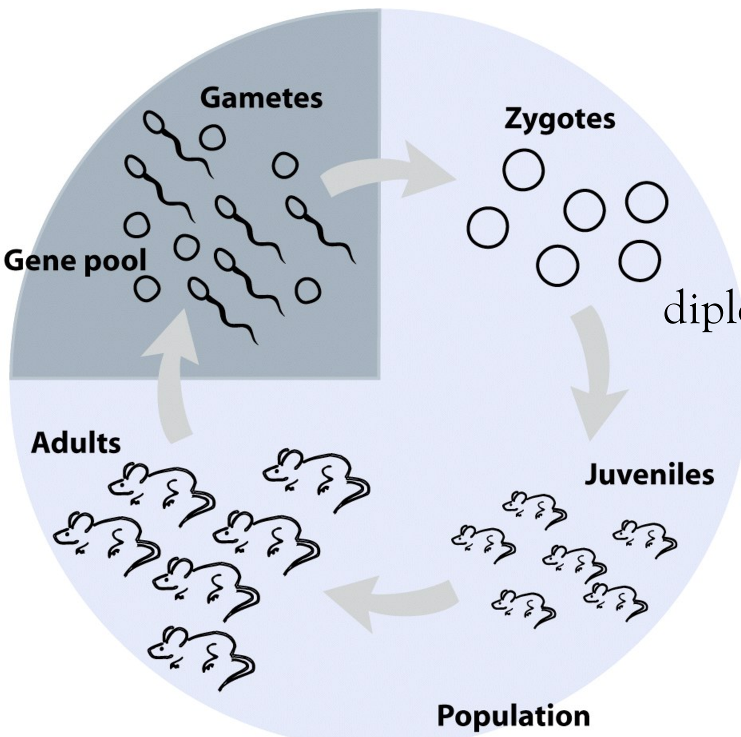
The metazoan sexual diploid life-cycle

haploid

diploid

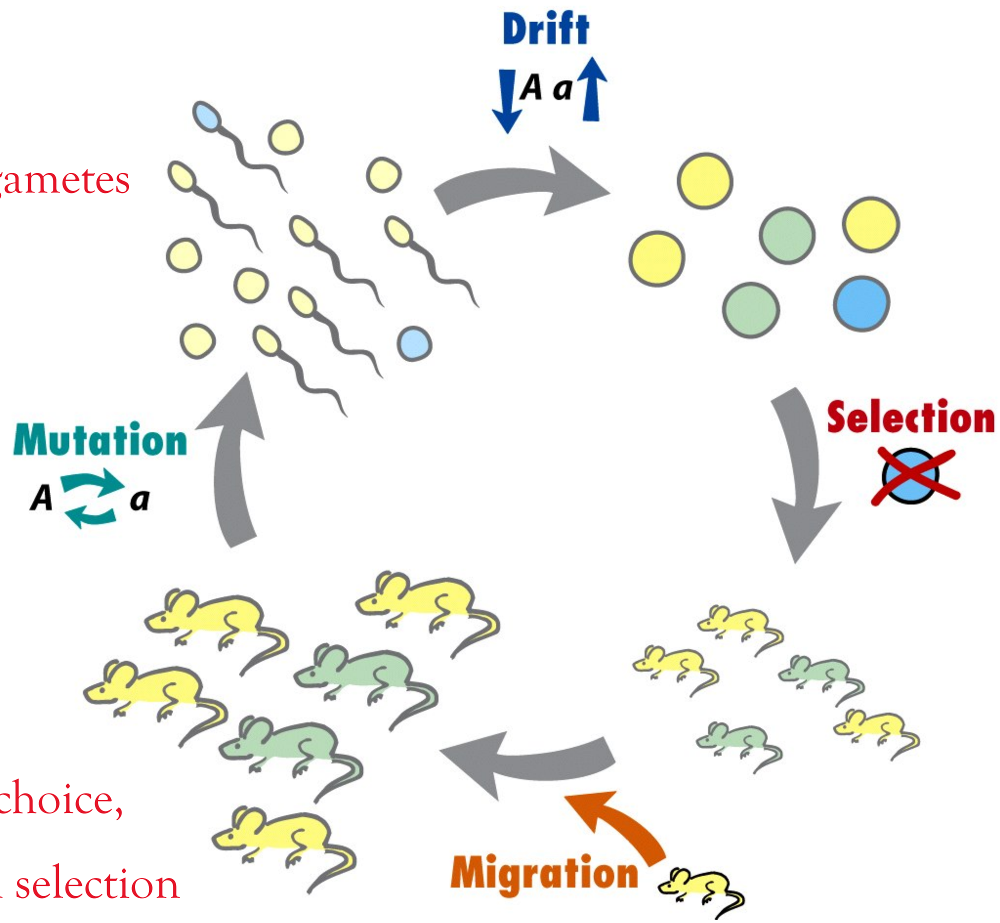
diploid

diploid



The forces causing evolution in real populations

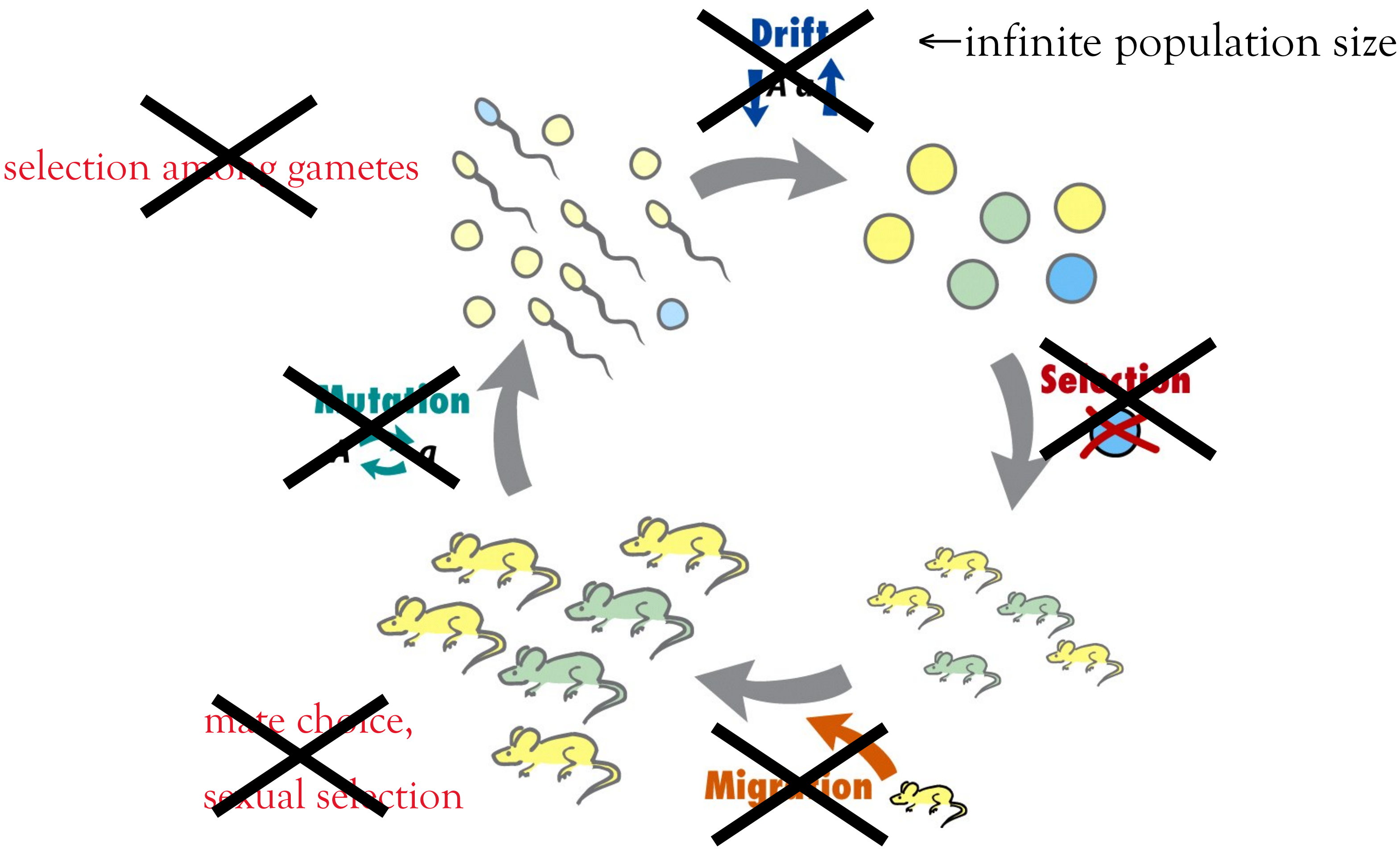
selection among gametes



mate choice,

sexual selection

The idealized population does not evolve



The concept of the ideal (Hardy-Weinberg) population

1. Infinite population size
2. Random mating among individuals in the population
3. No evolution:
    1. There is no selection
    2. There is no mutation
    3. There is no migration
    4. There is no chance evolution (genetic drift)

The Hardy-Weinberg Principle (HWP)

Does our population **evolve**?

If not: the population is in Hardy Weinberg Equilibrium

➔ The HWP is a null model against which we test if a population is evolving or not.

Calculating Genotype & Allele Frequencies

Genotypes:

Homzygous for A:	Heterozygous:	Homozygous for a:
AA	Aa	aa
36	48	16

N=100
N_allele=200

What are the **genotype frequencies** of AA, Aa and aa?

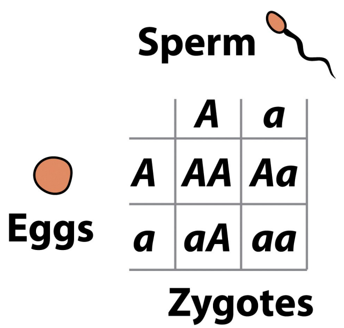


Figure 6-4 Evolutionary Analysis, 4/e © 2007 Pearson Prentice Hall, Inc.

What are the **allele frequencies** of A and a?

AA+AA+Aa/N_allele = (36+36+48)/200
Aa + aa + aa/N_allele= (48+16+16)/200

The Hardy Weinberg Equilibrium

$$p^2 + 2pq + q^2 = 1$$

Hardy (1908) and Weinberg (1908) demonstrated that in the absence of evolution any allele frequencies (summing up to 1.0) can be in equilibrium.

The Hardy-Weinberg Equlibrium (HWE)

- Simplest general case for illustration in a diploid organism:

  one gene (**A**) - two alleles (**A_1,A_2**) - three genotypes (**A_1A_1, A_1A_2, A_2A_2**)

Homozygote	**Heterozygote**	**Heterozygote**	**Homozygote**
$f(A_1A_1)$ +	$f(A_1A_2)$ +	$f(A_2A_1)$ +	$f(A_2A_2) = 1$
$p \bullet p$ +	$p \bullet q$ +	$q \bullet p$ +	$q \bullet q = 1$
p^2 +	$2pq$	+	$q^2 = 1$

Binomial equation: $(p+q)^2 = 1$

More general: $(SUM(p_n))^2 = 1$ **(with n = any number)**

The baseline hypothesis:

the ideal Hardy-Weinberg population

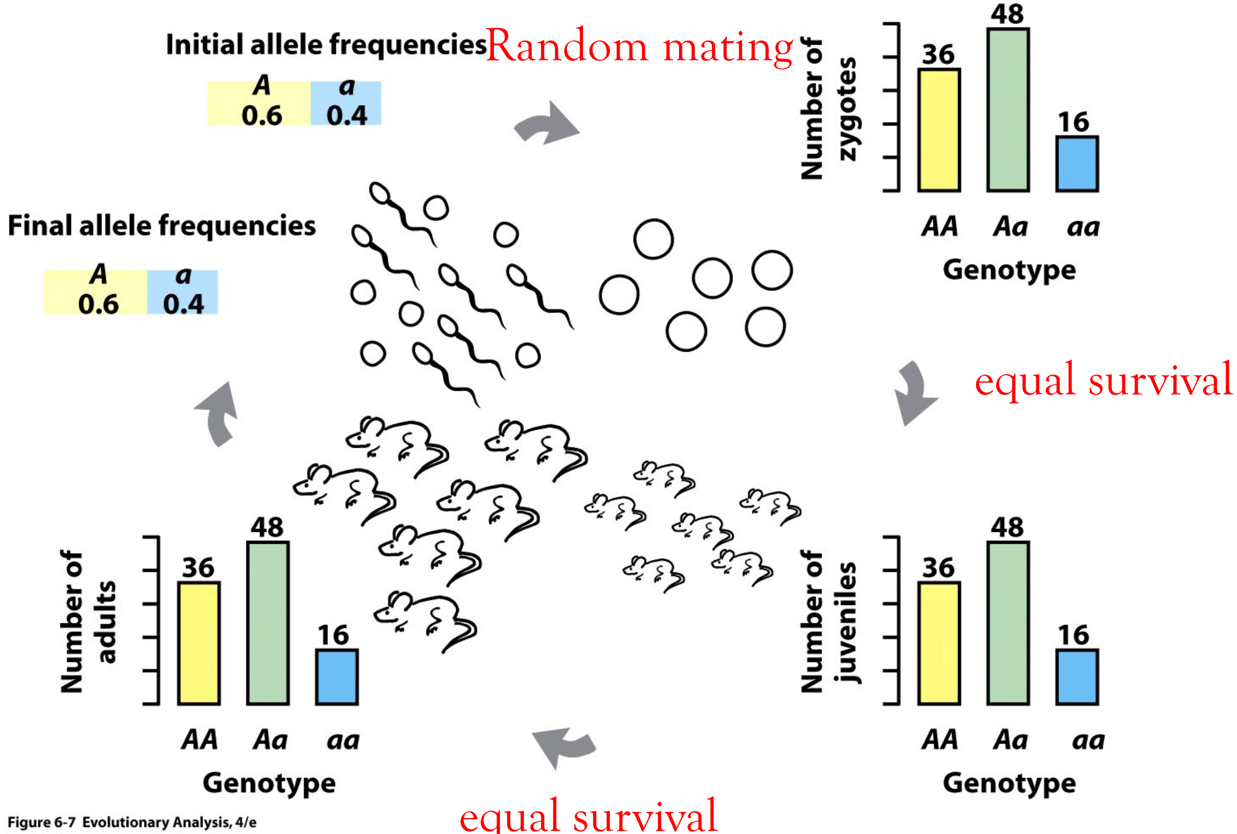


Figure 6-7 Evolutionary Analysis, 4/e

The Hardy-Weinberg Principle

- Does our population **evolve**?

  - **If not: the population is in Hardy Weinberg Equilibrium**
    - Populations follow the laws of Mendelian genetics
    - Allele frequencies do not change
    - Allele frequencies should be in an equilibrium after the first round of mating
    - Allele frequencies can be stable at any value between 0 and 1

  - **If yes:**
    - there is no Hardy-Weinberg Equilibrium (HWE)…

Take-home messages (Part 1)

1. **Population genetics** integrate **Darwin's theory of evolution** by natural selection with **Mendelian genetics** and seeks to **quantify evolutionary change over time.**

2. An **ideal (Hardy-Weinberg) population** is not limited in terms of population size and not impacted by evolution.

3. The **Hardy-Weinberg Equilibrium** postulates: $p^2 + 2pq + q^2 = 1$

Violations of the HWE

1) Allele frequencies change over time

$$f(A_1) = p \uparrow / \downarrow$$

$$f(A_2) = q \uparrow / \downarrow$$

2) Genotype frequencies do not meet expected values

$$f(A_1A_1) \neq p^2$$

$$f(A_1A_2) \neq 2pq$$

$$f(A_2A_2) \neq q^2$$

Testing if population is in Hardy-Weinberg-Equilibrium

Suppose we knew a sample of a population that is unaffected by a disease, while many others died, and that they have the following genotype frequencies:

AA	Aa	aa
4	23	3

allele frequencies:
f(A)= 31/60 =p
f(a)= 29/60 =q
use HWE equation to find 2pq
genotype frequencies:
g(AA)=4/34
g(Aa)=23/34
g(aa)=3/34
compare values from HWE with g(x)

1. Calculate the **observed allele** frequencies
2. Calculate the **expected number** of each genotype under HWE

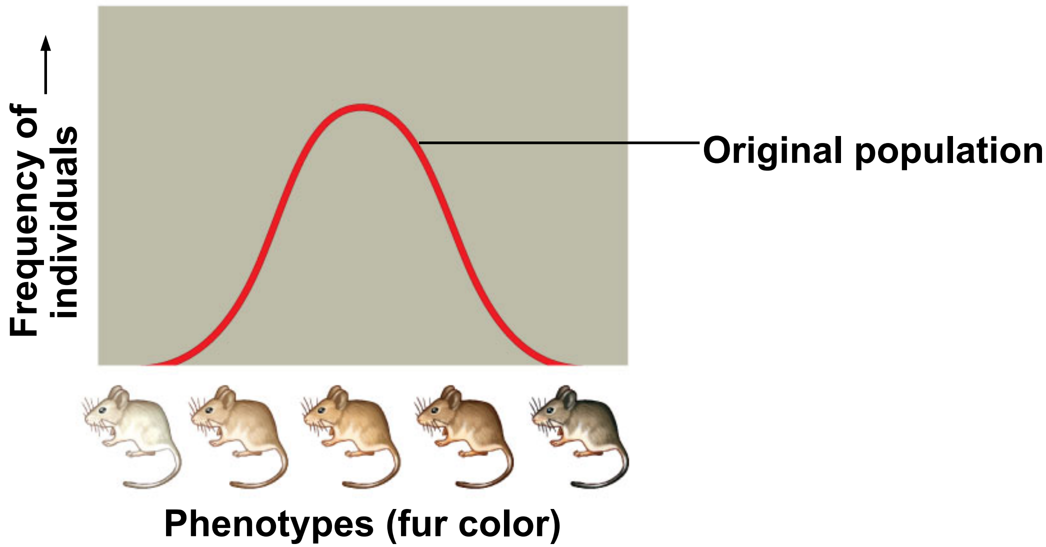
in this example: population not in HWE

3. Compare **expected and observed numbers, are they**
   - identical (HWE),
   - or not identical (no HWE)?
4. **Optional (more exact):** use a statistical chi-square test to test for significance

Lecture 5: Population genetics I

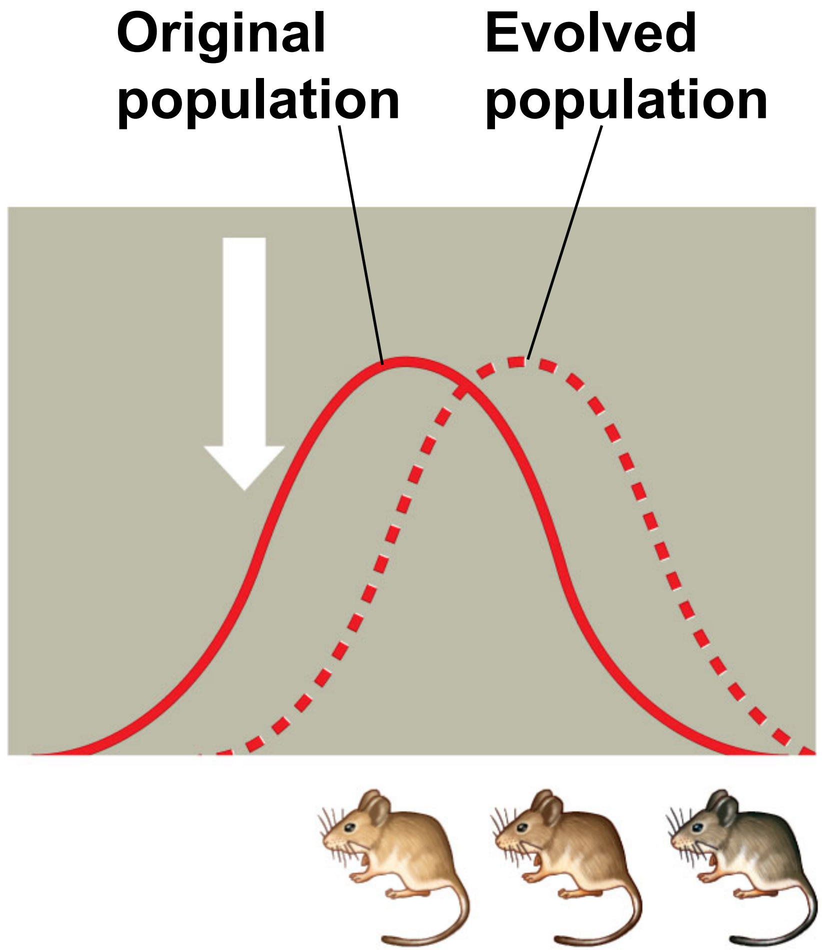
Selective forces

Figure 23.13



Frequency of individuals
Original population
Phenotypes (fur color)

Figure 23.13a

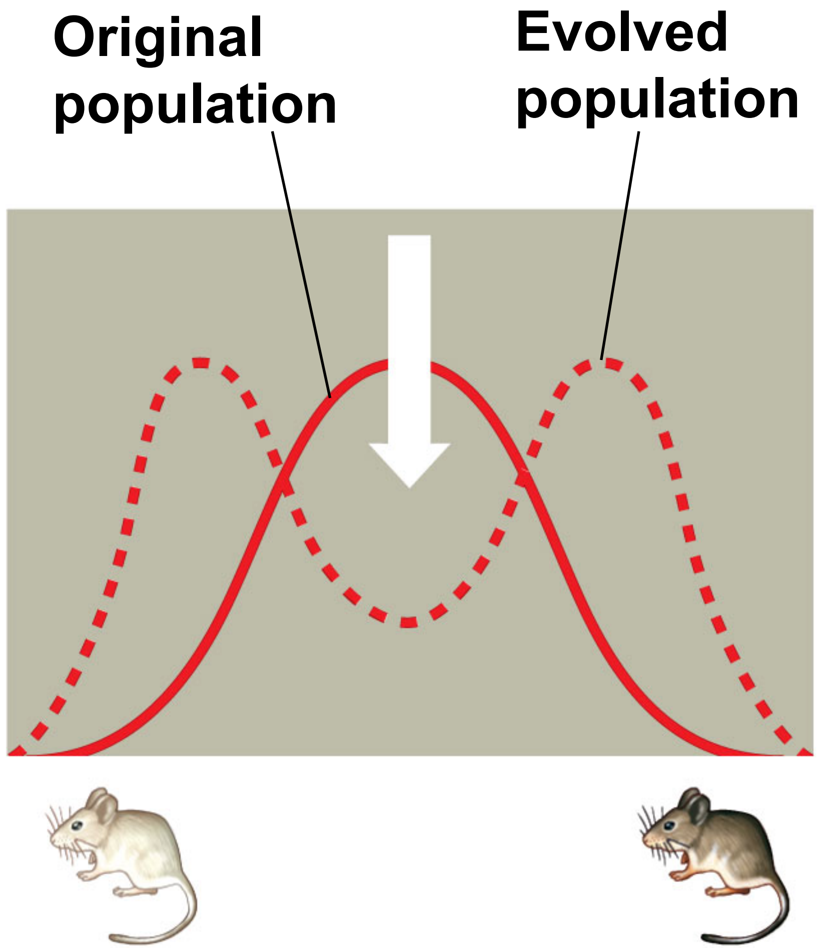


(a) Directional selection

Mean (trait) ↑

Variance (trait) ■

Figure 23.13b

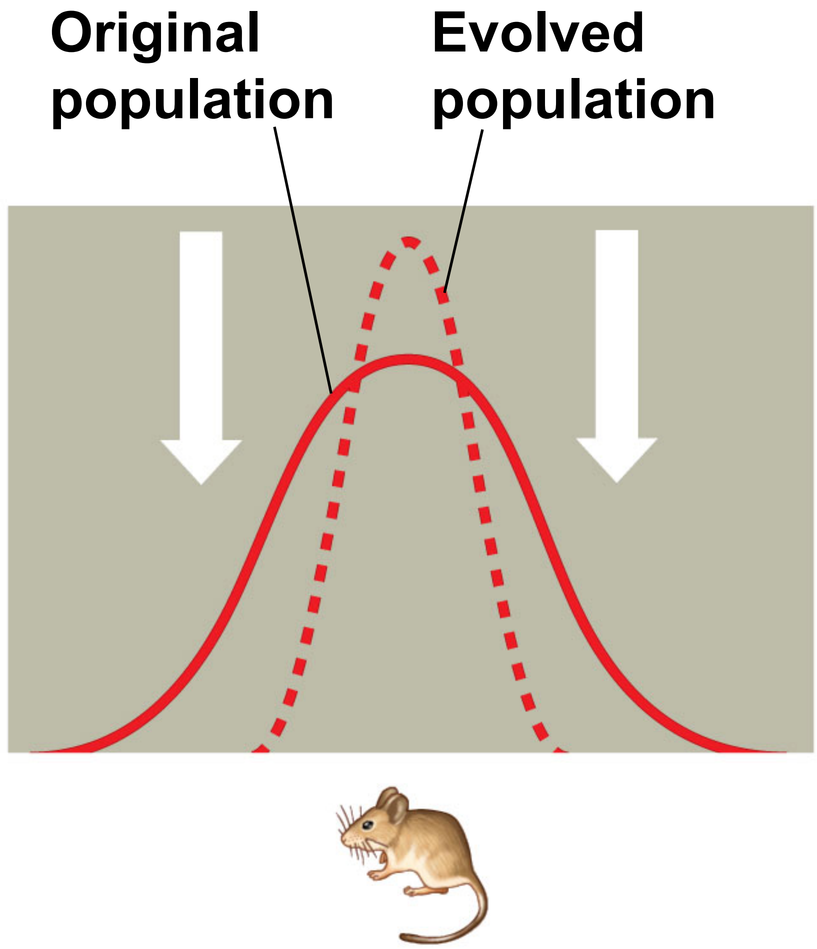


Mean (trait) ■

Variance (trait) ↑

(b) Disruptive selection

Figure 23.13c



Mean (trait) ■

Variance (trait) ↓

(c) Stabilizing selection

(d) Frequency-Dependent Selection

- In **frequency-dependent selection**, the fitness of a phenotype depends on its frequency in the population:
  - **positive FDS:** phenotype **increases** if it becomes most **common** in the population
  - **negative FDS:** phenotype **declines** if it becomes most **common** in the population

Negative Frequency-Dependent Selection

...when fitness depends on allele frequency.

Elderflower orchids cheat on bumblebees.

Color promises a nectar reward but none is given.

Why do both yellow and purple flowers exist?

Again, the question- **What maintains diversity?**

Negative Frequency-Dependent Selection

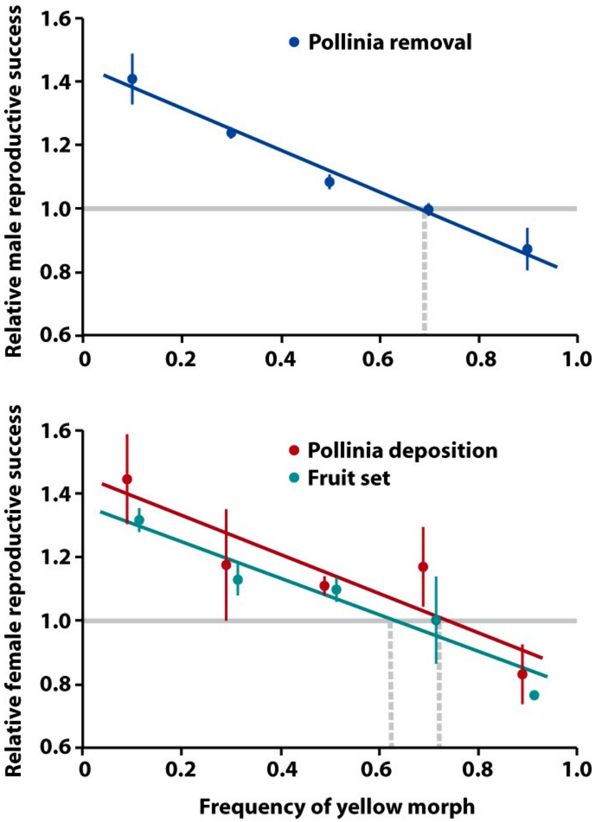


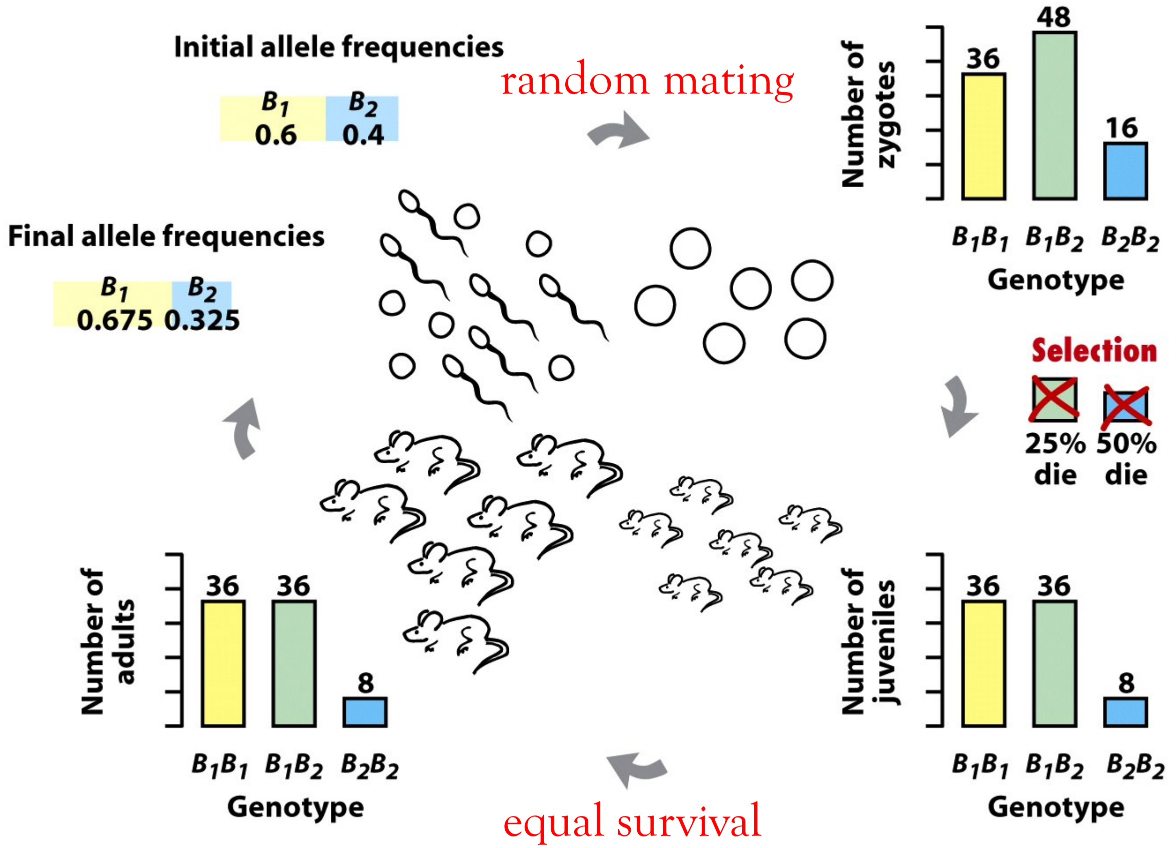
Figure 6-21bc Evolutionary Analysis, 4/e

Male function

Female function

Different modes of selection

- **Directional selection** favors individuals at one end of the phenotypic range
- **Disruptive selection** favors individuals at both extremes of the phenotypic range
- **Stabilizing selection** favors intermediate variants and acts against extreme phenotypes
- **Frequency-dependent selection: a) negative FDS,** favors the rarest variant and acts against the more common ones; **b) positive FDS,** the most common variant is favored over the rare ones


Initial allele frequencies
B1 0.6
B2 0.4
random mating
Number of zygotes
36
48
16
B1B1 B1B2 B2B2
Genotype
Selection
25% die
50% die
Number of juveniles
36
36
8
B1B1 B1B2 B2B2
Genotype
equal survival
Number of adults
36
36
8
B1B1 B1B2 B2B2
Genotype
Final allele frequencies
B1 0.675
B2 0.325

Selection coefficient (*s*)

Fitness (*w*) **[relative reproduction success]** is inversely correlated to the selection coefficient (*s*)

$$w = 1 - s$$

Fitness $(B_1B_1) = 100\% \Rightarrow s_{B1B1} = 0\%$

Fitness $(B_1B_2) = 75\% \Rightarrow s_{B1B2} = 25\%$

Fitness $(B_2B_2) = 50\% \Rightarrow s_{B2B2} = 50\%$

The effect of the selection coefficient

The larger the selection coefficient (s → **1):**

the faster allele frequencies change

Calculating changes in i) genotype and ii) allele frequencies due to selection

frequency of first allele: $p = f(A) = 0.5$
frequency of second allele: $q = f(a) = 0.5$
selection coefficient for a genotype: s

"Genotype fitness": $w = 1 - s$

$w(AA) = 1.0$
$w(Aa) = 0.7$
$w(aa) = 0.6$

1. Calculcate average fitness (w) for the whole population before selection:

$$w = p^2 \bullet w(AA) + 2pq \bullet w(Aa) + q^2 \bullet w(AA)$$

2. Calculate **genotype** frequencies **after** selection:

$f'(AA) = \boldsymbol{p^2} \bullet \boldsymbol{w(AA)}/\boldsymbol{w}$

$f'(Aa) = \boldsymbol{2pq} \bullet \boldsymbol{w(Aa)}/\boldsymbol{w}$

$f'(aa) = \boldsymbol{q^2} \bullet \boldsymbol{w(aa)}/\ \boldsymbol{w}$

2. Calculate **allele** frequencies among gametes after selection and random mating (p', q'):

$f'(A) = p' = f'(AA) + ½ \cdot f'(Aa)$

$f'(a) = q' = f'(aa) + ½ \cdot f'(Aa)$

$\Delta p = p' - p$

$\Delta q = q' - q$

Take-home messages (Part 2)

1. You can test if a population is in Hardy-Weinberg-Equilibrium

2. You can calculate the impact of selection on both genotype and allele frequencies

3. You understand the evolutionary trajectories that differential selection has on the various genotypes

Thanks for your attention!

… and don’t forget about next lecture:

Date: Wednesday, 25 Oct 2016

Location: HG F 3

Time: 8:00 - 10:00