

Bio 97

Population Genetics I

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Population/Gene Pool

- Population = group of organisms of the same species living in the same geographical area
- Gene pool = all alleles in population

Genotype & Allele Freqs

- Genotype frequency = proportion of individuals in a population with a specific genotype
- Allele frequency = proportion of alleles in a population

We'd like to know...

Relationship between allele
frequencies and genotype
frequencies

How to predict what AF's and
GF's will be in future generations

We need a theory

Hardy & Weinberg

Developed a simple
mathematical model of the
transmission of alleles from
generation to generation

To understand this model you need to know:

- $1 \times 1 = 1$
- $1 \times a = a$
- Simple high-school algebra

One gene (Arrogance), 2 alleles

A (dominant allele)

a (recessive allele)

Genotype

AA, Aa

aa

Phenotype

arrogant

humble

Hardy-Weinberg Equation

One gene, 2 alleles or wild-type

p = frequency of allele A (dominant allele)

q = " " " a (recessive allele)

Example - Gene pool with 700 A and 300 a alleles.

$$p = A/(A+a) = 700/(700+300) = 0.7$$

$$q = a/(A+a) = 300/(700+300) = 0.3$$

Allele
freqs

NOTE: $p + q = 1$

If we know the allele freqs in a
given generation

And we assume random mating

Then we can calculate the
genotype freqs in the next
generation

A population mates

Male gametes

p (A) ~~~~~ q (a)

Female gametes

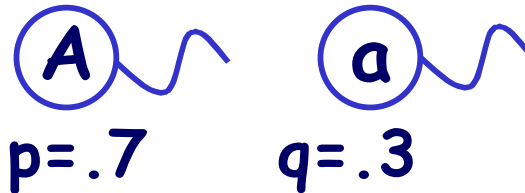
p (A)	p^2 AA	pq Aa
q (a)	pq Aa	q^2 aa

Genotype freq.
of aa genotype
is q^2

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

A population mates

Male gametes



Female gametes: A (frequency $p = .7$) and a (frequency $q = .3$)

p^2 AA .49	pq Aa .21
pq Aa .21	q^2 aa .09

Genotype freq. of aa genotype is q^2

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

AA Aa aa

That told us the genotype
freqs in the next generation

What are the allele freqs
in the next generation?

Allele counting

To determine the allele freq. in the next generation

Assume next generation has 100 people --> 200 alleles

p' , q' = allele freqs in next generation

AA 49	Aa 21
Aa 21	aa 9

$$p' = \frac{2 \times 49 + 21 + 21}{200} = .7$$

$$q' = \frac{2 \times 9 + 21 + 21}{200} = .3$$

Allele counting shortcut

$$\text{freq}(A) = \text{freq}(AA) + 1/2 \text{freq}(Aa)$$

$$\text{freq}(a) = \text{freq}(aa) + 1/2 \text{freq}(Aa)$$

AA .49	Aa .21
Aa .21	aa .09

$$p' = .49 + .21 = .7$$

$$q' = .09 + .21 = .3$$

Allele counting shortcut

$$\text{freq}(A) = \text{freq}(AA) + 1/2 \text{freq}(Aa)$$

p^2 AA .49	pq Aa .21
pq Aa .21	q^2 aa .09

$$p' = p^2 + pq = .49 + .21 = .7$$

$$q' = q^2 + pq = .09 + .21 = .3$$

$$p' = p^2 + pq = p(p+q) = p$$

$$q' = q^2 + pq = q(q+p) = q$$

Allele freqs don't change from one gen. to the next

HW says:

Allelic frequencies will remain constant over time if the following conditions are met:

- Mating is random
- Allelic frequencies are the same in males and females
- All genotypes have equivalent viability and fertility

HW Conditions II

- Mutation does not occur
- Migration into the population is absent
- Population is large so that allelic variations do not occur by chance

These idealized conditions are rarely met, but HW helps us learn why allele frequencies change.

Note...

If we know GF's in a generation of a pop, then we can always calculate AF's, whether or not HW equil.

If we know AF's, we can only calculate GF's (in this or the next generation) if the pop is in HW equil

At home exercise -

Try using both allele counting methods to determine the allele freqs in a pop of

10 people, consisting of

1 homozyg w.t,

6 hets and

3 homozyg recessive

Method 1: just count (there are 20 alleles total)

Method 2: $p = \text{freq}(AA) + 1/2 \text{freq}(Aa)$,

$q = \text{freq}(aa) + 1/2 \text{freq}(Aa)$,

HW

can be used to find the
freq of recessive
disease alleles in a
population and carrier
frequency

An autosomal recessive disease...

- Affects 1/100 people in a population
- (That is, it affects 1% of the population)
- (That is, the frequency of people with the aa genotype is 0.01)

What is the frequency of carriers (= heterozygotes), if the pop is in HW equilibrium ?

Answer...

- $q^2 = 0.01$ (given)
- $q = 0.1$ (take square root)
- $p = 1 - q = 0.9$ (since $p + q = 1$)
- $2pq = 0.18 =$ almost one in every five

X-linked HW

- X-linked genes are a special case (because males have only one X-chrm)
- Genotype frequencies among males are the same as allele frequencies:

Frequency of X^AY males = p

Frequency of X^aY males = q

Freq. of X^AX^A females = _____ Freq. of X^AX^a females = _____

Freq. of X^aX^a females = _____

Problem

One out of every 20 men has
red-green colorblindness

What is the incidence of **red-green colorblindness** among women?

Answer:

- $q = 1/20$ (given)
- $q^2 = 1/400 =$ incidence in females
- $2pq \sim 1/10 =$ female carrier freq.

Multiple allele HW

Hardy-Weinberg frequencies can be extended to multiple alleles:

Frequency of any homozygote
= square of allele frequency

Frequency of any heterozygote
= 2 X product of allele frequencies



**Ethical
Issue**

Genetic Testing and Genetic Discrimination

- **Do you have a right to know (to be tested)?**
- **Do you have a right NOT to know?**
- **Should your spouse/family know?**
- **Should your employer know?**
- **Should your insurance company know?**

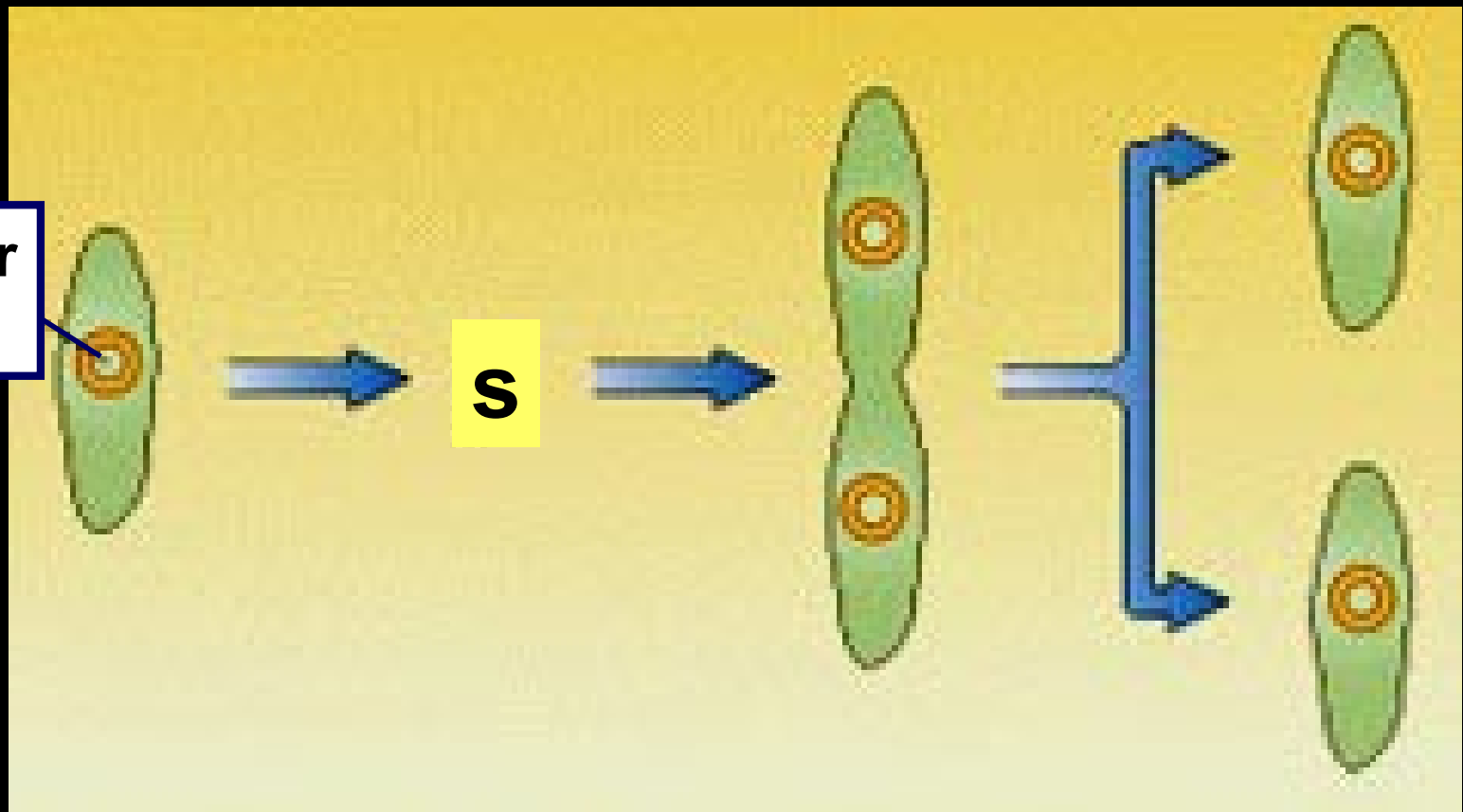
Forces that can cause deviation from HW ratios

1. Gene flow (e.g., migration)
2. Genetic drift
3. Mutation
4. Natural Selection
5. Non-random mating (e.g., inbreeding)

Let's Illustrate 1-3 with bacteria

Bacterial life cycle

Circular
chrom



Bacterial H-W Equilibrium

Generation

I	A	A	A	a	a	$p=0.6; q=0.4$
II	A	A	A	a	a	$p=0.6; q=0.4$
III	A	A	A	a	a	$p=0.6; q=0.4$
IV	A	A	A	a	a	$p=0.6; q=0.4$
⋮	⋮	⋮	⋮	⋮	⋮	
N	A	A	A	a	a	$p=0.6; q=0.4$

Migration in Bacteria

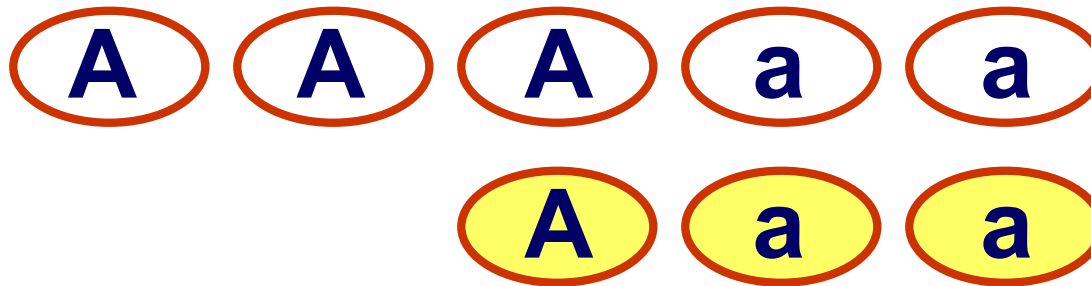
Generation

I



$p=0.6; q=0.4$

II



$p=0.5; q=0.5$

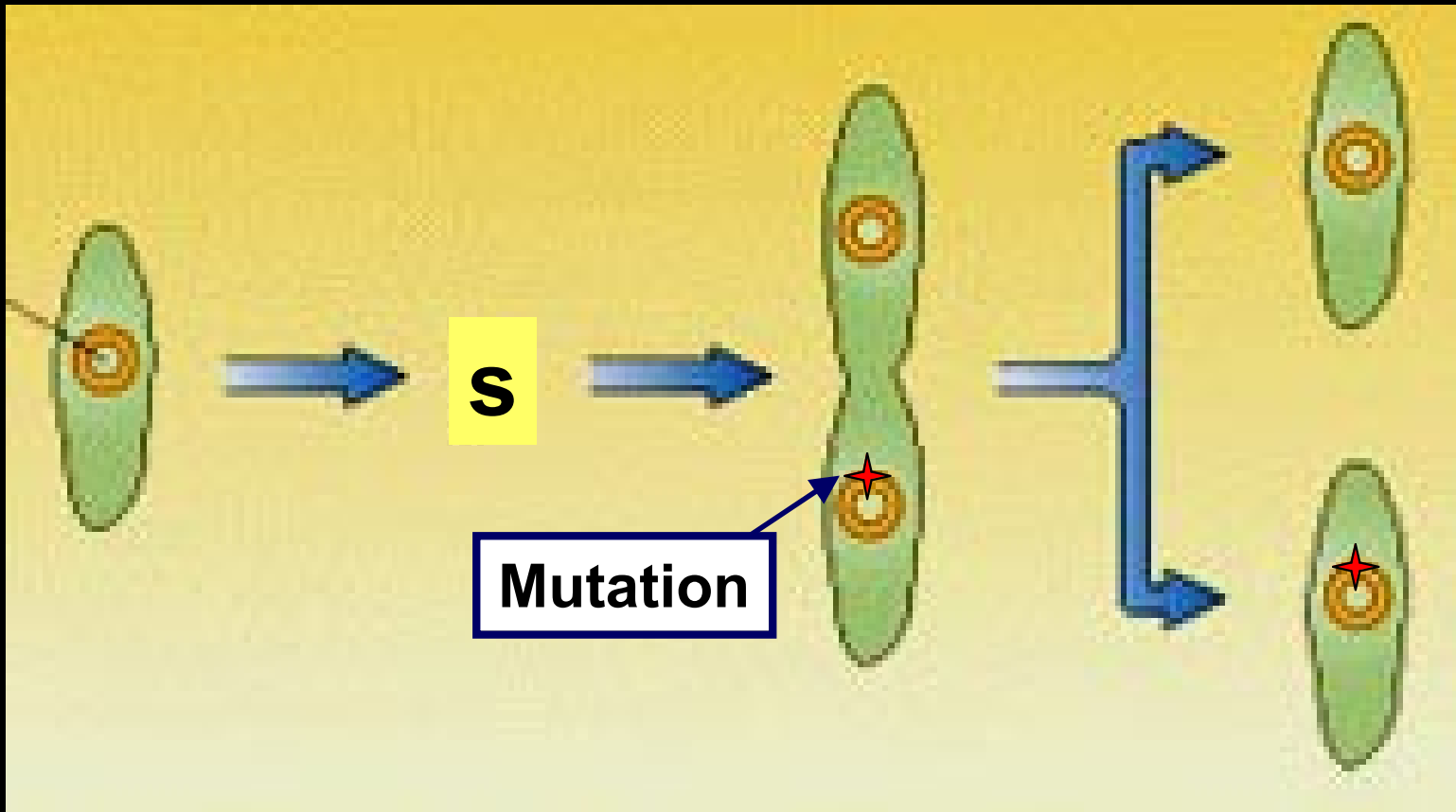
Genetic Drift

- Random changes in allele frequencies
- More important in small populations than in large ones
- In a small population, genetic drift, by itself, can cause an allele to become fixed (= the only allele)
- Even in large populations, new alleles are subject to drift

Drift in Bacteria

Generation

I	A	A	A	a	a	$p=0.6; q=0.4$
II		A	A	a	a	$p=0.5; q=0.5$
III	a	a	a	a	a	$p=0; q=1$
IV	a	a	a	a	a	$q=1$
⋮	⋮	⋮	⋮	⋮	⋮	
N	a	a	a	a	a	$q=1$



Mutation in Bacteria

Generation

I



II



Mutation

- A random mutation is far more likely to harm the function of a protein than to enhance it
- If mutation was the only force operating, then eventually most alleles would become nonfunctional

Natural Selection

- More organisms are produced each generation than can survive and reproduce
- Organisms differ in their ability to compete, based (in part) on their genotype
- Hence, genotypes that promote survival/ reproduction are favored, and are more likely to contribute alleles to the next generation's gene pool

Evolution

= long-lasting
changes in the gene
pool due to **natural
selection** and other
forces (mutation,
drift,...)