Gene Frequency and Evolution

POPULATION GENETICS:

The study of the rules governing the maintenance and transmission of genetic variation in natural populations

DARWINIAN EVOLUTION BY NATURAL SELECTION

- Many more individuals are born than survive (COMPETITION).
- Individuals within species are variable (VARIATION).
- Some of these variations are passed on to offspring (HERITABILITY).
- Survival and reproduction are not random. There must be a correlation between fitness and phenotype.

Mechanisms of Evolution: Mendelian Genetics in Populations

- Genetic variation is the raw material of evolutionary change: how do we measure it?
- What are the forces that cause genetic changes within populations? That is, what mechanisms cause evolutionary change?

Population Genetics

- Evolution can be defined as a change in gene frequencies through time.
- Population genetics tracks the fate, across generations, of Mendelian genes in populations.
- Population genetics is concerned with whether a particular allele or genotype will become more or less common over time.

Assumptions:

1) Diploid, autosomal locus with 2 alleles: **A** and **a**
2) Simple life cycle:

PARENTS	GAMETES	ZYGOTES
(DIPLIOD)	(HAPLOID)	(DIPLOID)

These parents produce a <u>large</u> gamete pool (Gene Pool) containing alleles **A** and **a**.

a A A a
a A A a A a a
a A A a a A a A A
a a A A a a a
a A a a A A
A a A

Gamete (allele) Frequencies:

Freq(A) = p

Freq(a) = q

$\Rightarrow$ $p + q = 1$

Genotype Frequencies of 3 Possible Zygotes:

AA Aa aa

$Freq\ (AA) = p_A \times p_A = p_A^2$

$Freq\ (Aa) = (p_A \times q_a) + (q_a \times p_A) = 2p_Aq_a$

$Freq\ (aa) = q_a \times q_a = q_a^2$

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

General Rule for Estimating Allele Frequencies from Genotype Frequencies:

Genotypes:	AA	Aa	aa
Frequency:	p^2	$2pq$	q^2

$\Rightarrow$ Frequency of the A allele:

$p = p^2 + ½ (2pq)$

$\Rightarrow$ Frequency of the a allele:

$q = q^2 + ½ (2pq)$

Sample Calculation: Allele Frequencies

Assume N = 200 indiv. in each of two populations 1 & 2

- Pop 1 : 90 AA   40 Aa   70 aa
- Pop 2 : 45 AA   130 Aa   25 aa

In Pop 1 :

- $p = p^2 + ½ (2pq) = 90/200 + ½ (40/200) = 0.45 + 0.10 = 0.55$
- $q = q^2 + ½ (2pq) = 70/200 + ½ (40/200) = 0.35 + 0.10 = 0.45$

In Pop 2 :

- $p = p^2 + ½ (2pq) = 45/200 + ½ (130/200) = 0.225 + 0.325 = 0.55$
- $q = q^2 + ½ (2pq) = 25/200 + ½ (130/200) = 0.125 + 0.325$ **= 0.45**

Main Points:

- $p + q = 1$ (more generally, the sum of the allele frequencies equals one)
- $p^2 + 2pq + q^2 = 1$ (more generally, the sum of the genotype frequencies equals one)
- Two populations with markedly different genotype frequencies can have the same allele frequencies

The Hardy-Castle-Weinberg Law

A single generation of random mating establishes H-W equilibrium genotype frequencies, and neither these frequencies nor the gene frequencies will change in subsequent generations.

$$\mathbf{p^2 + 2pq + q^2 = 1}$$

H-W ASSUMPTIONS:

1. Large population size
   - small populations have fluctuations in allele frequencies (e.g., fire, storm).
2. No migration
   - immigrants can change the frequency of an allele by bringing in new alleles to a population.
3. No net mutations
   - if alleles change from one to another, this will change the frequency of those alleles
4. Random mating
   - if certain traits are more desirable, then individuals with those traits will be selected and this will not allow for random mixing of alleles.
5. No natural selection
   - if some individuals survive and reproduce at a higher rate than others, then their offspring will carry those genes and the frequency will change for the next generation.

IMPLICATIONS OF THE H-W PRINCIPLE:

- A random mating population with no external forces acting on it will reach the equilibrium H-W frequencies in a single generation, and these frequencies remain constant there after.
- Any perturbation of the gene frequencies leads to a new equilibrium after random mating.
- The amount of heterozygosity is maximized when the gene frequencies are intermediate.

2pq has a maximum value of 0.5 when

p = q = 0.5

Modern Synthesis Theory

- Combines Darwinian selection and Mendelian inheritance
- Population genetics - study of genetic variation within a population
- Emphasis on quantitative characters (height, size …)
- 1940s – comprehensive theory of evolution (Modern Synthesis Theory)
- Introduced by Fisher & Wright
- Until then, many did not accept that Darwin's theory of natural selection could drive evolutionTODAY'S theory on evolution
- Recognizes that GENES are responsible for the inheritance of characteristics
- Recognizes that POPULATIONS, not individuals, evolve due to natural selection & genetic drift
- Recognizes that SPECIATION usually is due to the gradual accumulation of small genetic changes

Microevolution

- Changes occur in gene pools due to mutation, natural selection, genetic drift, etc.
- Gene pool changes cause more VARIATION in individuals in the population
- This process is called MICROEVOLUTION
- Example: Bacteria becoming unaffected by antibiotics (resistant)

Cause of microevolution

- **Genetic Drift**

- the change in the gene pool of a small population due to chance

- **Natural Selection**

- success in reproduction based on heritable traits results in selected alleles being passed to relatively more offspring (Darwinian inheritance)

- Cause ADAPTATION of Populations

- **Gene Flow**

-is genetic exchange due to the migration of fertile individuals or gametes between populations

• The Hardy-Weinberg theorem assumes that no individuals enter a population from the outside and that no individuals leave a population.

• Immigration or emigration upsets the Hardy-Weinberg equilibrium, resulting in changes in allelic frequency (evolution). Changes in relative allelic frequency from the migration of individuals are gene flow.

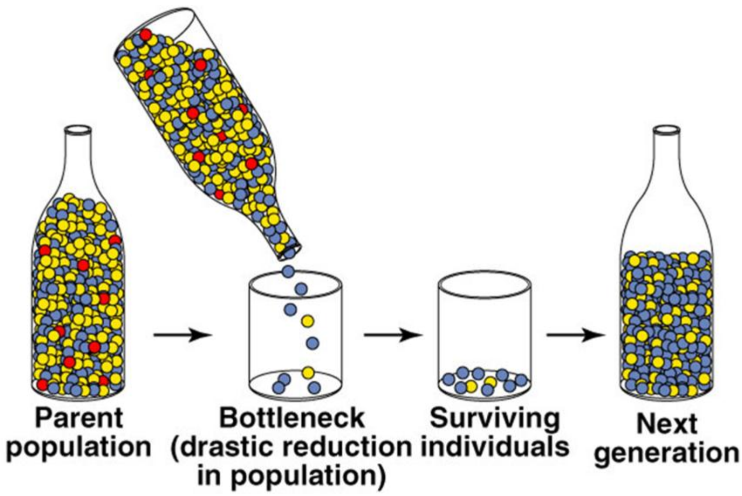
• The absence of gene flow can make change in the populations population less likely.

- **Mutation**
    - a change in an organism's DNA
    - Mutations can be transmitted in gametes to offspring
- **Non-random mating**

- Mates are chosen on the basis of the best traits

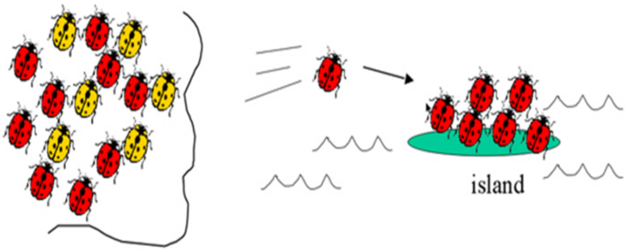
Genetic drift

- **Bottleneck Effect**
    - a drastic reduction in population (volcanoes, earthquakes, landslides …)
    - Reduced genetic variation
    - Smaller population may not be able to adapt to new selection pressures



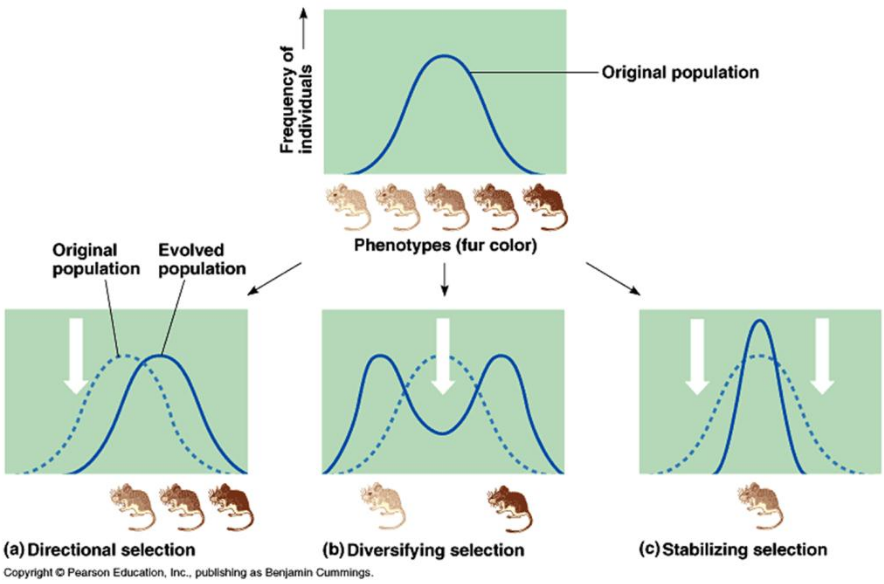
Founder Effect

- occurs when a new colony is started by a few members of the original population
- Reduced genetic variation
- May lead to speciation



Modes of Natural Selection

- **Directional Selection**
    - Favors individuals at one end of the phenotypic range
    - Most common during times of environmental change or when moving to new habitats
- **Disruptive selection**
    - Favors extreme over intermediate phenotypes
    - Occurs when environmental change favors an extreme phenotype
- **Stabilizing Selection**
    - Favors intermediate over extreme phenotypes
    - Reduces variation and maintains the current average
    - Example: Human birth weight



Geographic Variations

- Variation in a species due to climate or another geographical condition
- Populations live in different locations
- Example: Finches of Galapagos Islands & South America

Other Sources of Variation

- **Mutations**
  - In stable environments, mutations often result in little or no benefit to an organism, or are often harmful
  - Mutations are more beneficial (rare) in changing environments (Example: HIV resistance to antiviral drugs)
- **Genetic Recombination**
  - source of most genetic differences between individuals in a population
- **Co-evolution**
  - Often occurs between parasite & host and flowers & their pollinators

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